

D2.3: Survey of REC approaches and codes for genomics

[WP2 – Genomics: ethical, legal and social analysis]

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Abstract

This report describes the outcome of task 2.3, Current coverage of ethical guidelines by professional organisations, ethics advisory groups, and research ethics committees for human genetics and genomics. For this task the SIENNA partners searched for documents which could give normative guidance (excluding legislation) for stakeholders in human genetic and genomics. Three kinds of documents were searched in different EU countries and internationally:

1. professional ethics codes
2. documents from professional groups and ethics advisory groups, and
3. guidance documents on how to write research ethics protocols in different EU countries and internationally.

Furthermore, representatives of research ethics committees have been asked for the following information in an online survey:

- to what extent are they aware of Human Genomics technologies and ethical issues associated with them,
- how do they currently approach these issues and do they have plans to more explicitly address them.

This report is a descriptive document, which includes summaries and detailed lists of normative document searches in human genetics and genomics per country and internationally, as well as preliminary insights into REC members' awareness and views of normative documents. This informative tool includes a list of over 150 documents addressing genetics and/or genomics in some normative capacity. These will be used to help inform work for task 2.7 (ethical frameworks) and work for WP5 (normative proposals by the consortium).

Document history

Version	Date	Description	Reason for change	Distribution
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Information in this report that may influence other SIENNA tasks

Linked task	Points of relevance
Task 3.3, Task 4.3 – Current coverage by research ethics committees and in ethical codes	Tasks 3.3 and 4.3 are strongly connected to task 2.3. The same methodology for the national and international searches was used and the opinions and knowledge of REC members was examined via one online survey for all three SIENNA areas.
Task 2.2, 3.2, 4.2 – Analysis of legal and human rights requirements in and outside the EU	Ethical frameworks or normative rules are sometimes regulated as soft law. Therefore, there might be overlapping's between the 2.3 and the 2.2 tasks. Although the 2.2 tasks focused only on normative frameworks (not on legal binding documents).
Task 2.4, 3.4, 4.4 – Analysis of current and future ethical issues	The results of the 2.3 tasks will also be useful for the 2.4 tasks in which partners will conduct a review existing ethical theories and approaches regarding the three fields.
WP2.7	The outcome of the 2.3 task will help us develop the ethical frameworks.
WP5	The outcome of the 2.3 tasks will be the basis for the work in WP5, in which operational guidelines, ethics codes and proposals for improved ethical and legal frameworks will be developed for genetic and genomics.



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Executive summary

What the reader should know about the general context behind the development of this report

This report was developed in the context of a European Commission (EC) funded SWAFS¹ project called [SIENNA](#), which began in October 2017. The SWAFS-18-2016² call developed by the EC and categorised as research and innovation action outlined the areas of technologies to be studied and the specific challenges it had identified as important for study. In response to this call, the SIENNA project was developed and ultimately chosen for funding. The partners of the SIENNA project are studying the ethical, legal and social issues (ELSI) of three areas of technologies: Human Genomics, AI/Robotics, and Human Enhancement (as dictated by the EC SWAFs call). The report you are currently reading is the second deliverable completed for Work Package (WP) 2 which addresses the ELSI of Human Genomics (HG). We report herein how and to what extent different ethical normative documents explicitly or implicitly address genomic technologies. In particular we studied normative documents issued by three different types of groups: i) professional organisations, ii) (ethical) advisory groups, and iii) research ethics committees. The same research has been conducted for Human Enhancement (WP3, D3.3) and AI & Robotics (WP4, D4.3).

Furthermore, this report also informs about the results of an online survey completed by representatives of research ethics committees (RECs) in Europe. The survey was conducted to determine to what extent the representatives of RECs are aware of the three SIENNA areas of technologies and ethical issues associated with them, how they currently approach them, and if there are plans to more explicitly feature them.

What are the aims and use of this report?

This report gives an overview of the search for documents which could give normative guidance for stakeholders in HG (mostly clinicians and researchers but also anyone involved in the offer of genetic testing). Three kinds of documents were searched for across different countries, both in the EU and internationally. Based on the findings, the partners will decide which elements should be part of ethical frameworks and which are missing. That means that this report will be used as a source of information which will help conduct the work for task 2.7 (ethical framework) and WP5 (e.g. the development of elements that complement operational guidelines for research ethics committees, the development of central elements of a code of responsible conduct for researchers in genomics).

Preliminary analysis of international documents reveals different approaches in providing guidance, for example, specific vs. general guidance, scope of applications to which it is relevant, attributed role. Furthermore, preliminary analysis of national documents suggests that some issues may have been receiving more attention in guidelines than others.

¹ SWAFS = Science with and for Society,

<https://ec.europa.eu/research/participants/portal/desktop/en/opportunities/h2020/calls/h2020-swafs-2018-2020.html#%c,topics=callIdentifier/t/H2020-SwafS-2018-2020/1/1/1/default-group&callStatus/t/Forthcoming/1/1/0/default-group&callStatus/t/Open/1/1/0/default-group&callStatus/t/Closed/1/1/0/default-group&+identifier/desc>

² <https://ec.europa.eu/research/participants/portal/desktop/en/opportunities/h2020/topics/swafs-18-2016.html>



It is important to understand that given the large number of documents retrieved (over 160)³ and all the different languages involved, it is impossible in the time provided for this task (4 person-months total, including for all the country-specific work) to conduct a thorough content analysis of all documents beyond the questions answered in the summary tables (Annex 1) and arrive at any specific conclusion. Indeed, this deliverable is meant as resource in and of itself, somewhat similarly to how we would use a dictionary or an encyclopaedia, as a resource to help for larger tasks.

Since task 2.3 is very strongly connected with task 3.3 and 4.3 this report contains text modules which are also used in D3.3 and D4.3.

What are the conclusions and “take-home-messages” of this report?

While our analysis still remains somewhat preliminary (due to the time restraints and sheer volume of data, see above and footnote 3 below), we can report that there is a large variation in the provision of ethical codes specific to genetics or genomics in the partner countries. From the UK, France and Greece which have upward of two dozen documents reported, to South Africa which identified only two (with the rest of the partners reporting somewhere around a dozen documents specific to genetics or genomics).

Now the questions remain:

- 1- Are any of these documents particularly helpful in identifying gaps and/or important guideline areas in the current analysis of ELSI for Human Genetics and Genomics?
- 2- Which of these documents address the most important current and future ELSI questions and dilemmas in Human Genetics and Human Genomics?
- 3- How can these documents be used to further the work in SIENNA regarding the proposal of an ethical framework for HG (Task 2.7) and the development of (enhanced) codes (Tasks 5.2, 5.5) and operational guidelines for RECs (5.1)?

While the (preliminary) analysis of all documents herein give us some ideas on the answers to these questions, the answers will become more concrete, valid and definitive only as we go forward with the ethical analysis in Task 2.4 (deliverable month 21/ 2019) and Task 2.7 (deliverable month 30/2020) as well as the work in WP5, which will begin shortly and be completed at month 40 (2021). Indeed, one of the greatest challenges with HG will be to identify which particular areas we want to address in the frameworks/guidelines? How general do we want or can we make these? Herein lies the beauty and complexity of the SIENNA project.

As for the self-reported awareness and views of Research Ethics Committee members regarding guidance in human genomics, there seems to be moderate to low knowledge and guidance specific to ELSI of HG. Fittingly, an overwhelming majority of respondents feel that REC members would benefit from additional education around the ELSI of HG. This subject will be further addressed in WP5.

³ After all the preparations for the tasks, searches, and synthesis of materials already done for this task, and for which we have already gone beyond the paid time, we would still need many more weeks to conduct any type of rigorous content analysis to answer very specific questions.



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List of acronyms/abbreviations

Abbreviation	Explanation
AI & R	Artificial Intelligence & Robotics
D	Deliverable
DoA	Description of Action
ELSI	Ethical, Legal and Social Implications
EU	European Union
HE	Human Enhancement
HG	Human Genomics
NAEG	National Advisory/Ethics Groups
PEC	Professional Ethics Codes
REC	Research Ethics Committee
REP	Research Ethics Protocol
SOP	Standard Operating Procedure
WP	Work package

Table 1: List of acronyms/abbreviations

Glossary of terms

Term	Explanation
Ethics advisory bodies	Independent groups of ethics experts giving advice to a researcher, or research group on specific ethical, regulatory, social or philosophical issues raised by science.



Professional ethics codes	Guidelines to help members, workers, management or researchers conduct themselves in accordance with common values and/or ethical standards.
Professional organizations/groups	Organizations or groups usually bring together people working for a special profession to represent their interests.
Research ethics committees	Committees that review research applications and give opinions about whether research is ethical.
Research ethics protocols	Sets out how a study or project will deal with issues that are challenging from an ethical perspective.

Table 2: Glossary of terms



1. Introduction

1.1 Background and objectives

One of the main goals of SIENNA is the development of ethical frameworks (WPX.7) and codes (WP5.2-4) for the three technology areas: human genomics (HG), human enhancement (HE) and artificial intelligence and robotics (AI&R). As a basis for this work, we need to have a good overview of relevant existing documents. We need to understand to what extent HG, HE and AI&R are already addressed by guidance documents for research ethics protocols and by professional organisations and by national ethics' groups, e.g., the (ethics) codes they have developed, in different EU countries and internationally.

To get an overview of already existing relevant documents, we took the following steps:

1. **National search:** With the help of our partners we conducted a national search in 13 countries. Every partner searched in his/her own country in his/her own language for three types of documents:
 - professional ethics codes
 - guidance documents or recommendations from professional groups and ethics advisory groups
 - guidance documents on how to write research ethics protocols.
2. **International search:** We also searched for the same three types of documents internationally for all three SIENNA fields. This work was conducted by UU for Human Genomics (task 2.3), by EUREC for Human Enhancement (task 3.3) and by TRI for AI & Robotics (task 4.3).
3. **Online survey with REC representatives:** Via a survey UU and EUREC addressed representatives of research ethics committees (RECs) to determine to what extent they are aware of these technologies and ethical issues associated with them, how they currently approach them, and if there are plans to more explicitly feature them. The EUREC network served as basis for this. REC members from 11 different countries responded.

1.2 Structure of the report

The main part of this report informs about the research questions, which guided our work and about the methodology and key results of our analyses and surveys (national, international analyses and online surveys). The key results are described in section 3. In the three annexes the reader will find detailed information concerning our methodology and results. In Annex 1 the reader can find the guidance provided to our partners to assist them in undertaking the survey and detailed results from the national searches. In Annex 2 we have listed all relevant international documents for genetics and genomics and gave further information on these documents. In Annex 3 we provide a detailed overview of the online survey (questions and detailed responses).

1.3 Scope and limitations

We aimed to obtain a wide range of normative documents to capture as many of the different types of salient normative statements as possible. This goal, along with the time allotted to this task, means that we cannot claim to have conducted a strictly systematic and search nor that we retrieved all existing normative documents. However, our search approach aimed at revealing the most important/influential documents to guide us further in tasks WPX.7 and WP5.



2. Research questions

The work for the X.3 tasks was guided by the following types of questions (their development, innovation and use, depends on the area of technology). Please note that the goal was not to answer each question in detail; rather the idea was that these questions would lead us to identify documents that could give answers either obviously/explicitly and/or via a more extensive analysis (to be done outside the scope of this task).

- A. Are there *professional organisations that have developed professional ethics codes (PECs)* that specifically address the three areas of technology/research?
- B. Are there documents from *national advisory/ethics groups (NAEGs)* that address specifically the three areas?
- C. Are there *guidance documents on research ethics protocols (GDREP)* that specifically address research in the three areas?
 - a. Human Genomics:
 - i. research using HG?
 - ii. research developing technologies in the three areas?
 - b. Human Enhancement:
 - i. research that explicitly focuses on HE?
 - ii. research that can have HE as side effect (as many clinical studies do) – how is that mirrored in guidance documents, if at all?
 - c. Artificial Intelligence & Robotics:
 - i. research that focuses on development and/or use of AI?
 - ii. research that focuses on development and/or use of Robotics?
- D. How are HG/HE/AI&R addressed in these documents?
 - a. in professional ethics codes (PECs)?
 - b. in documents from national advisory/ethics groups (NAEGs)?
 - c. in guidance documents on how to write research ethics protocols (*GDREPs*)?
- E. If the specific area (HG, HE, AI&R) is not addressed specifically, how could existing guidance documents also apply to these areas of technologies?
- F. To what extent are REC representatives aware of these technologies?
- G. To what extent are REC representatives aware of ethical issues associated with technologies developed in the three areas?
- H. How do REC representatives think/know/are aware that the ELSI of tech X is currently approached?
- I. Do REC representatives think/know/are aware of plans to more explicitly feature ethical issues in the three areas?

Based on these questions we developed the methodology for the national search, the international search and the online survey with REC members.



3. Methods

3.1 Normative document search

3.1.1 Methodology used for the national search in 13 countries

The national search was conducted by the partners. The task leaders from UU and EUREC developed a guidance document on how to conduct the search in collaboration with TRI and UT (the guidance instructions for the national search can be found in Annex 1). This document was developed for the search across all three SIENNA areas (HG, HE and AI&R). We provided our partners a plan on how to proceed with the search and reporting, although we informed them, that this methodology can be changed (provided that all changes, e.g. change of keywords for the search etc., must be documented). We prepared a presentation which we showed in a Skype call with the partners, to give an overview what we expect from them for the X.3 tasks. During the whole working time on the X.3 tasks, UU and EUREC as task leaders answered questions via email and phone. In a second Skype call partners exchanged their experiences with the tasks and had the possibility to clarify their questions. The detailed guidance on how to proceed for the national search can be found in Annex 1 of this document

The national search was conducted in 13 countries by 13 different SIENNA partners (in alphabetical order):

- Brazil
- China
- France
- Germany
- Greece
- Japan
- Netherlands
- Poland
- South Africa
- Spain
- Sweden
- UK
- USA

Japan and the USA partners, however, did not have budget allocated to this task. Therefore, Japan partner performed only basic search. The summary of the relevant documents in the USA was prepared by the Uppsala University team based on their expertise.

Every partner wrote a summary of their findings for HG/HE/AI&R. The summaries were guided by the following two questions: What are the most important “take-away” lessons from your search? What are the gaps? These summaries are listed below. In the annex the reader will find tables of all relevant documents the partners identified in their searches and further information and tables in which the most relevant documents are described in more detail.

3.1.1.1 Challenges during the national searches

In our methodology we defined that we exclude documents written by single authors or author groups. Included were only documents from professional organisations or groups and/or national



(ethics) advisory groups. One of the main challenges in the national searches was that it was not always clear what constitutes a professional organisation. This was difficult for HG, but particularly so for HE and AI&R, since in these fields it is not really clear what a professional group might be. We recommended the partners to make use of the column “comments” in these cases and write down their uncertainties regarding these issues there.

Another challenge occurred during the search for guidance documents from research ethics committees (GDREPs), since it turned out that in all three SIENNA areas there are very few of these documents available. In some countries the search showed no results. Indeed, the analysis and conclusions regarding documents from RECs/GDREPs is very thin for both the national and international analyses (for example, we did not find any international RECs that we could study).

3.1.2 International Document Search

We followed the following strategies to retrieve relevant documents:

- 1) We searched for the documents in a database HumGen - international database of laws and policies in human genetics (<http://www.humgen.org>), which is run by researchers at McGill University. We set the criteria to international, and we did not restrict the search by any search terms – all documents relevant to genetics were retrieved. The database contains documents issued between 1990 and 2017.

The following documents irrelevant to our analysis were excluded:

- reports which did not provide guidance or recommendations, e.g. “OECD Science, Technology and Industry Scoreboard 2015: Innovation for Growth and Society”
 - documents not concerning humans directly (for example about gene editing in other organisms)
 - documents which were not obviously relevant to genetics/genomics, for example the search retrieved “Digital Security Risk Management for Economic and Social Prosperity: OECD Recommendation and Companion Document”.
- 2) We searched the same database to get insight into how many documents in various categories are available. We searched documents with assigned key terms such as biobanks, prenatal, carrier status etc. Please see results section for the full list.
 - 3) We grouped the documents into those referring to activities of exploring the genome, modifying the genome, or both. These groups correspond to three tables in the results section (Annex 2). We further analysed the document with regard to the following features:
 - whether it was issued by an international or regional organization
 - whether it is relevant to research or clinical contexts
 - whether it is specific to genetics or genomics.

Additional characteristics, especially when deemed relevant to SIENNA objectives, such as format, audience, scope of applications have also been briefly discussed.



3.2 Survey of Research Ethic Committees

To determine to what extent representatives of RECs are aware of the three SIENNA areas of technologies and ethical issues associated with them, how they currently approach them, and if there are plans to more explicitly feature them (in guidelines for researchers), we developed and sent out an online survey. We decided to develop one survey for all three SIENNA areas together and worked out 18 questions. Most of them could be answered by multiple choice. The full list of questions and answers can be found in this document in annex 3.

Based on the experience that there is a much higher response rate by online surveys, compared to questions sent via email or in a document, we created an online survey via Google forms. With the time allotted to this task in view, we decided to send the online survey first only to the members of the European network of research ethics committees (EUREC). EUREC members have a broad expertise in research ethics and come from different European countries, which guarantees a good geographic distribution. Although the RECs represented in EUREC are all focused on medical and bioethics. Sending the survey to the EUREC members can be seen as a first round. At a later point we may send this survey, or a revised version, to members of other research ethics committees or experts, which will focus also on other research fields.

The online survey was developed with Google forms in June/July 2018 and distributed by EUREC to its members in August 2018 by sending a link via email. Unfortunately, August is for many people summer holiday time, which is likely to have impacted on the response rate. However, due to the deliverable deadline of 1st October, we were not able to send out the survey earlier or later. The mail was initially sent to 30 EUREC members and reminder email was also sent.



4. Key results and related discussion

In this section the reader can find information about the key results of the national search, the international search and the online REC members' survey. Further detailed results and information on these can be found in annex 1, 2 and 3.

4.1 National search

We received the information from 13 countries, 11 of which followed the methodology outlined above. The search and analysis of two remaining countries – Japan and USA is limited in its scope due to limited resources that could be allocated to this task by partners from that countries. These two countries will not be discussed herein further.

4.1.1 Summaries of the national searches

The following summaries were prepared by:

Country	Institution	Contributor(s)
Brazil	Universidade Federal do Rio de Janeiro (UFRJ)	Marcelo de Araujo, Clara Dias
China	Dalian University of Technology (DUT)	LIU Hongzuo
France	Sciences Po Paris	Anaïs Rességuier
Germany	European Network of Research Ethics Committees (EUREC)	Lisa Tambornino, Dirk Lanzerath
Greece	Ionian University	Maria Bottis
Japan	Chuo	Hiroshi Miyashia
the Netherlands	University of Twente (UT)	Tanne Ditzel, Philip Jansen
Poland	Helsinki Foundation for Human Rights (HFHR)	Zuzanna Warso
South Africa	University of Cape Town (UCT)	Jantina de Vries
Spain	University of Granada (UGR)	Javier Valls
Sweden and USA	University of Uppsala	Heidi Howard, Emilia Niemiec
UK	Trilateral Research Ltd (TRI)	Rowena Rodrigues, David Wright

Table 3: List of contributors for the national searches



4.1.1.1 Brazil

Some of the documents related to genomics listed in this report refer to the Brazilian Federal Law nº 11.105, from 24 March 2005.⁴ This piece of legislation, also known as the “Biosecurity Law”, governs a variety of issues related to the generation, use, and commerce of genetically modified organisms in Brazil. It also rules the work of government bodies that supervise compliance with this law and enforce sanctions, as the case may be. Research involving the use of biobanks, gene-editing technologies, IVF and related techniques ultimately must comply with this piece of legislation. After Law nº 11.105 was enacted, some resolutions had to be issued on a number of occasions so as to provide researchers and entrepreneurs with more specific guidelines as to how to comply with the law. New resolutions are often necessary as a result of scientific advancements in biotechnology. And new advancements may, in turn, give rise to new social practices such as, for instance, “shared pregnancy” and “social egg freezing”, which are not directly addressed by Law nº 11.105. Some of these resolutions include, for instance, *Resolution CFM nº2.168/2017*, which establishes rules for the practice of assisted human reproduction and addresses issues related to both social and oncological cryopreservation of gametes and embryos. *Resolution CFM nº 2168/2017* allows the selection of biological traits only as far as the avoidance of genetic diseases is concerned; it does not allow sex selection, eye colour selection or further non-therapeutic biotechnological interventions in human cells. The report issued by ANVISA in 2017 (*1st report on seminal samples for assisted human reproduction*) provides an in-depth insight into the current practice of importing human semen from foreign sperm banks for the purpose of assisted human reproduction. Interestingly, although Law nº 11.105 forbids the commerce of human reproductive cells within Brazilian territory, it does not prohibit their importation from sperm banks abroad. According to the ANVISA report, between 2011 and 2016 the importation of seminal samples for assisted reproduction increased in 2.625% in Brazil.

4.1.1.2 China

In general, China's genetic technology has developed rapidly, and gene technology has been applied to many fields of research. It has brought great changes to human health, agriculture and environmental protection. In the field of clinical medicine, gene technology has been used in non-invasive prenatal gene detection, neonatal screening, tumor examination, individualized cancer treatment, cardiovascular physical examination and other fields. In the future, genome technology will be combined with the latest research results and traditional clinical medicine to form a precise medical treatment, which will bring revolutionary changes to the diagnosis, treatment and clinical decision of the disease. Within the gene industry, many gene technology companies have been launched, such as BGI, Berry Genomics and ANOROAD and so forth. In the field of scientific research, in addition to the aforementioned gene technology companies, scientific research institutions related to genetics include central and various local medical institutions (mainly the Three-level A-rate hospitals) and scientific research institutions (including institutions of higher education and research institutes). Typical organizations such as Beijing Institute of Genome Research Affiliated Chinese Academy of Sciences, National Institute of Genetic Research of the Chinese Academy of Sciences, Shenzhen National Gene Bank, China National Human Genome Center (Shanghai & Beijing), etc.

In contrast, in terms of gene technology, the legislative supervision and ethical review at the government level appear to be lagging behind. In the available documents on gene technology, several are professional technical guidelines for genetic technology and its applications. In other

⁴ BRAZIL. “Lei nº 11.105, de 24 de março de 2005”, available at: http://www.planalto.gov.br/ccivil_03/ato2004-2006/2005/lei/l11105.htm.



words, there are few documents on the ELSI of genetic technology. In particular, some PEC/NAEG/GDREC files, which are directly related to genetic technology, are still numbered. At the same time, the areas directly involving in gene technology are limited, such as gene sequencing, gene patents and pharmacogenomics, etc.

However, this does not mean that all sectors of the Chinese government do not pay attention to ELSI caused by genetic technology. In fact, the state has set up a number of regulatory agencies, introduced a series of relevant policies and some industry and technology associations, put forward many technical norms and ethical principles, and formulated a great deal of detailed ethical codes to regulate genetic technology in many ways. It should be emphasized that although many documents do not clearly point out that the document is directly referred to gene technology, it is equally binding on genetic technology. For example, it is clearly pointed out that "*the clinical application of medical technology should be guided by scientific, safe, standardized, effective, economic and ethical principles*". In addition, it is clearly defined that gene therapy technology should be classified into the third categories of medical technologies as a major ethical problem, and the safety and effectiveness of medical technology need to be further verified by standardized clinical trials. Finally, it is clearly pointed out that the technology of gene chip diagnosis and treatment must also be specially administrated and supervised. In the field of legal supervision, Ministry of Science and Technology of the People's Republic of China, National Health Commission of the People's Republic of China, State Administration of Market Supervision, and its subordinate agencies, have formulated ethical guidelines and ethical review rules related to the research and application of gene technology (local governments at different levels have also formulated some local laws and regulations and ethical review rules in line with local realities).

For institutions of professional ethics and ethical review, they mainly include the Medical Ethics Committee Affiliated the National Health and Family Planning Commission, the local expert committee on bioethics, ethical research organizations within universities and other research institutions, the non-profit social organizations in accordance with the law (for example, Chinese Medical Biotechnology Association, Chinese Medical Association), and the ethical committee set up within the gene related companies, etc.

We predict that all of these documents provide a good reference value for the SIENNA project, as well as reflecting Chinese characteristics to a certain extent.

4.1.1.3 France

Research in human genomics is generating much interest in France, as exemplified by the two major reports commissioned by the government and prepared by the Parliamentary Office for the Evaluation of Scientific and Technological Choices (in 2010 and 2014). Ethical considerations occupy a central position in these reports.

As reflected by the series of documents that I have collected for this task, the main groups addressing ethical issues regarding this technology in France are the National Ethics Consultative Committee for Life Sciences and Health (CCNE) and the Ethics committee of the National Institute of Health and Medical Research (INSERM).

Furthermore, genomics is regulated by the Bioethics law that is updated every seven years (a new consultation took place this year and results will be reported in 2019). As part of this consultation, an important national debate took place in France in 2018. It brought together experts (scientists, jurists, ethicists, etc.), policy makers and the general public around ethical questions posed by new technological developments in relation to health and life sciences. Three overarching themes that



structured the debate were directly relevant to genomics: genetic tests and genomic medicine; health data; stem cells and research on embryos.

4.1.1.4 Germany

Genetic testing and genetic diagnosis in Germany are regulated mainly by the Genetic Diagnosis Act from 2010. There are different German national advisory groups, ethics groups and professional groups, which were important for the development of this act and furthermore are still publishing recommendations and guidelines on human genomics which are likely to have an influence on the public and political debate and on legal developments. The German Medical Association (Bundesärztekammer) and its central ethics committee (ZEKO) and the Office of Technology Assessment at the German Bundestag (TAB) published relevant recommendations and position papers or guidelines on research in human genomics, which are partially implemented as soft law. The German Ethics Council (Deutscher Ethikrat), the working Group of Medical Ethics Committees in the Federal Republic of Germany (Arbeitskreis medizinischer Ethik-Kommissionen in der Bundesrepublik Deutschland), the German research foundation (DFG), and the national academy of sciences Leopoldina published recommendations which are important for the German debate and developments, although they have no legal basis. The same applies for documents published by human genetic specific professional organisations, e.g. the German Society of Human Genetics, the Robert-Koch-Institute and the Paul-Ehrlich-Institute.

The search for professional ethics codes dealing with research on human genomics showed no direct results. The most relevant professional group in Germany is the German Society of Human Genetics, which developed and published relevant guidelines and recommendations, but no professional ethics code (see in the table in the annex). The same applies for the German Ethics Council, which is one of the most important German ethics advisory groups. There are professional ethics codes which may be relevant for research in human genomics, although these are not specifically developed for this technology area, e.g. the professional code (Berufsordnung)⁵ for physicians developed by the German Medical Association or the “Research Code of Conduct”⁶ developed by the University of Cologne.

The search for relevant documents from professional groups, national advisory and ethics groups showed many documents, the most relevant are listed in the tables under 5.4 (annex 1). The German Ethics Council published a document about “the future of genetic diagnosis – from research to clinical practice”⁷ in 2013 and recommendations on “germline intervention in the human embryo” in 2017. Both documents are important for the German debate, although they include no legal binding advises. The German Medical Association published recommendations on biobanks⁸ and a guideline

⁵ German Medical Association: professional code for physicians.

https://www.bundesaerztekammer.de/fileadmin/user_upload/downloads/pdf-Ordner/MBO/MBO-AE.pdf
(only German)

⁶ University of Cologne: research Code of Conduct. https://verwaltung.uni-koeln.de/forschungsmanagement/content/e12474/e160886/018_Research_Code_of_Conduct_neu_D_ger.pdf
(only German)

⁷ German Ethics Council: The future of genetic diagnosis – from research to clinical practice. 2013.
<https://www.ethikrat.org/fileadmin/Publikationen/Stellungnahmen/englisch/opinion-the-future-of-genetic-diagnosis.pdf>

⁸ German Medical Association: Medical, ethical and legal aspects of biobanks. 2017.
https://www.bundesaerztekammer.de/fileadmin/user_upload/downloads/pdf-Ordner/WB/Biobanken.pdf



about the extraction and transmission of human cells within assisted reproduction in 2017. From relevance is furthermore the statement on genome sequencing, written by the German Research Foundation (the professional organisation of German scientists) in 2016⁹ and a document published by EURAT on genome sequencing¹⁰.

The search for guidance documents on how to write research ethics protocols for research in human genomics showed that there are guidance documents from universities on how to write a research proposal or a research protocol in general. However, these documents are very broad and did not focus on the ethical issues or components. The documents developed by the working Group of Medical Ethics Committees in the Federal Republic of Germany (Arbeitskreis medizinischer Ethik-Kommissionen in der Bundesrepublik Deutschland - AMEK) are very useful for our purposes. The AMEK developed templates for the informed consent process for different study types (e.g. biobanks, clinical studies, paediatric research) and also recommendations for members of ethics committees on how to assess research projects. Unfortunately most of the AMEK documents are only available in German (see here: [AMEK Website](#)).

4.1.1.5 Greece

At the centre of the authoritative texts on bioethics, genetics and genomics is the important work of the National Bioethics Commission. Since 2001, the Commission has given us a series of opinions, reports, recommendations and informative texts, all available on the commission's website, which cover all the main issues in bioethics today, including work relating to the SIENNA themes of genomics/genetics and human enhancement. The reports are texts with scientific data upon which the opinion pieces are based. Informative texts basically solely contain information. Two Codes of Ethics are also relevant here: the Code of Medical Ethics and the Code of Ethics for Medically-Assisted Reproduction also cover genetics/genomics ethical issues relevant to SIENNA. Quite importantly, these Codes do not come strictly from a professional organization, but have been introduced, discussed and voted as with any other statute, this one containing ethical rules binding as law. This is not new, as in 1955 the first Code of Medical Ethics was implemented as a presidential decree (hence, having the force of a statute). In Greece, the National Association of Medical Geneticists has not discussed or produced a code of ethics (<http://www.sige.gr/index.php/gr/>). The National Authority for Medically Assisted Reproduction recently ceased its work due to immense bureaucratic problems, so its important work had to start anew, under a new Chair and members. As such, they produced the Code of Medical Ethics for MAR which also makes rulings on issues relating to SIENNA genomics/genetics. Greece also has Codes of Ethics in research in universities, such as the National Kapodestrian University of Athens. Although these are not detailed, they refer to the main laws (internal and international, such as Conventions etc) on bioethics. Many Universities have formed Research Ethics Committees, to approved research protocols also on bioethics, and therefore on any research on genetics/genomics. In 2018, a new law (4521/2018) made it obligatory for the universities to form Ethics Committees to control research protocols etc. Since then, universities have initiated calls for the expression of interest from faculty members to become members of these Committees. It is too early to determine how these Committees will deal with research on SIENNA themes such as genomics/genetics, but we can safely declare that they will follow international and

⁹ German Research Foundation: Statement on genome sequencing. 2016.

http://www.dfg.de/download/pdf/dfg_im_profil/reden_stellungnahmen/2016/160801_stellungnahme_humane_genomsequenzierung.pdf

¹⁰ https://www.uni-heidelberg.de/md/totalsequenzierung/informationen/mk_eurat_stellungnahme_2013.pdf



internal legal and ethical standards. Some universities had already formed an Ethics Committee prior to 2018, as have many private medical centres and hospitals.

4.1.1.6 Japan

Due to the fact that Japanese partner did not have budget assigned to this task, only basic search and no further analysis was performed for the relevant documents.

The following documents were found:

<http://www.sci.go.jp/ja/info/kohyo/pdf/kohyo-23-t251-1-en.pdf>

http://www.lifescience.mext.go.jp/files/pdf/n796_00.pdf

http://jams.med.or.jp/guideline/genetics-diagnosis_e.pdf

http://jshg.jp/wp-content/uploads/2017/08/10academies_e.pdf

4.1.1.7 The Netherlands

There are two professional organisations in the Netherlands for genetics and genomics professionals: the Dutch Association of Clinical Geneticists (Vereniging Klinische Genetica Nederland, VKGN), which unites medical specialists in clinical genetics; and the Association of Clinical Genetic Diagnostic Laboratories (Vereniging Klinisch Genetische Laboratoriumdiagnostiek, VKGL), which includes recognised clinical genetics laboratory specialists. Neither of these associations has developed an official ethics code. The former, however, offers sets of technical guidelines, some of which are of an ethical nature. It has also recently supported the opening of an online service desk for ELSI issues in Personalized medicine and next generation sequencing (Servicedesk voor Ethische, Juridische en Maatschappelijke Vraagstukken Over Personalized Medicine & Next Generation Sequencing). More general codes for healthcare professionals for the most part do not focus on issues in genetics/genomics. One exception is the code of conduct for responsible use of human tissue by the Federation of Dutch Medical Scientific Societies, which focuses on the responsible use of human tissue in scientific research, including the genetic information derived from it.

There are a number of organisations that provide policy-makers and the general public with information, ethical guidelines and other (policy) recommendations regarding the development and application of a variety of genomics technologies and procedures. These include the Rathenau Institute, the Netherlands Centre for Ethics and Health (CEG), the Netherlands Commission on Genetic Modification (COGEM), the Health Council of the Netherlands (Gezondheidsraad) and the Royal Dutch Academy of Sciences (KNAW). They have published reports containing ethical analyses and guidelines and recommendations for genetic and genomic testing and screening and genome editing. The older reports (pre-2010) focus mostly genetic testing and screening, whereas the more recent reports also discuss the implications of developments in whole genome sequencing and somatic and germline gene-editing.

This search has not turned up any guidance documents on research ethics protocols that address issues in genomics.

4.1.1.8 Poland

In Poland there is no professional ethics code that specifically address HG. Such a document has not been established by the Polish Society of Human Genetics, nor any other institution. The Society has



however issued statements that address ethical issues related to HG. In addition some provisions that explicitly address HG can be found in the general Code of Medical Ethics.

As far as national advisory groups are concerned, two institutions addressed relevant topics: the Bioethics Committee by the Polish Academy of Sciences (PAS) and the Office of the Commissioner for Human Rights (it is important to note that the Bioethics Committee by PAS is not formally a National Ethics Committee). These two bodies have issued statements and repeatedly draw attention to the need to establish legal standards that would address ethical challenges related to HG. Despite their recommendations, however, no legal action has been taken, which suggests that there is very little political interest nor willingness to address these concerns.

A draft code of conduct on processing personal data for research purposes has been recently circulated for public consultations.

It seems pertinent to point out that the word “genomics” (*pol. genomika*) or phrases “genomic testing” or “genomic screening” are not used in the documents analysed. We have made the assumption that whenever “genetic” (e.g. genetic testing in the context of the need to introduce laws or adhere to ethical standards) is mentioned, both genetics and genomics are being referred to. Therefore, it was at times challenging to precisely categorize the documents in table 5. That being said, none of the documents identified refers specifically and solely to high throughput genomics (*pol. wysokowydajne sekwencjonowanie genomu, wysokowydajna genomika*).

In addition, in Poland there is no National Ethics Committee. In a way, the Bioethics Committee by the Polish Academy of Science tries to fill that gap, however it has a limited capacity and authority.

4.1.1.9 South Africa

As is obvious from this analysis, there is a scarcity of ethics guidelines, policies and other regulatory documents pertaining to human genetics or genomics research, with a real regulatory gap in these fields. There certainly is considerable scope for improvement. That said, however, it seems clear to me that there are a number of ethical values that stand out in the consideration of research ethics, and that should inform the development of the Sienna frameworks in all three fields. The most important of these is a consideration of how new technologies can be used to reduce inequality across domains, ranging from health inequality to socio-economic inequality. The focus of technology development and research should be on reducing these, both at the level of South African society but also internationally. The threat – articulated by South African stakeholders in ethics and policy documents but also in more informal deliberations I’ve had with people in the context of the SIENNA project and more broadly – is that new technologies will be appropriated by the rich and used to promote their interests, with the result that technology will increase and not reduce inequality. There are several ways in which this has been approached in ethics guidelines broadly, and arguably for biobanking more specifically (in the Biobanking Policy of Wits University), and that is to place important focus on questions of social value and benefit of research, with researchers being pressed to specify how their research is going to impact on the lives of South Africa’s poorest people.

Another value that seems to me to be of increasing importance is ‘reciprocity’. You may know that many societies and communities in Africa are organised on principles of solidarity (in South Africa called ‘ubuntu’). On the one hand, such a worldview seems to be supportive of policies of sharing – because people are quite open to contributing their bit to the benefit of others – but on the other hand, there are also strong reciprocity obligations. I often come across this in terms of ‘I will help you fulfil your need but I equally expect you to recognise my need and help where you can’. So participants seem to be motivated by altruism to donate samples but do so in the expectations that



they, and their dignity, would be considered in return. This is not an absolute requirement – i.e. a researcher wouldn't absolutely be expected to 'pay' for samples – but rather an expectation that if you can, you will help. This is manifested quite clearly in relation to feedback of results – participants expect that the minimum a researcher could do is share with them any results pertinent to their health – but sometimes also w.r.t. expecting researchers to be able to help them with basic drugs for instance.

W.r.t. gene editing: South Africa does have a GMO Act, and Executive and Advisory Councils that give effect to the act. The GMO Act gives effect to the Cartagena Protocol, which South Africa ratified in 2003. But the GMO Act and the advisory councils are administered by the Department of Agriculture. Whilst including 'gene therapy' in its mandate, the Act specifically excludes 'human gene therapy', leaving a regulatory gap in terms of human gene therapy.

There has been some discussion about whether gene editing techniques used in crops should be regulated under the GMO Act, with the Academy of Sciences of South Africa advising that it should (in a report entitled "The regulatory implications of the new breeding techniques", see <http://research.assaf.org.za/handle/20.500.11911/29>), advise which the African Centre for Biodiversity objects to (in a report entitled "Deception or Dishonesty", see https://acbio.org.za/wp-content/uploads/2017/06/ASSAF_Paper_Web.pdf). But neither of these two reports concern gene editing in humans and so I have not included them here.

Please note that I am a member of a Consensus Panel by the African Academy of Sciences which was tasked with developing a report on 'Ethical issues in Genomics', with the stated objective that the report would be used to outline gaps in regulation w.r.t. human genomics to guide the development of legislation and guidelines nationally. The report is currently with reviewers but not publicly available and so I cannot include it here. The panel is quite strict about due process. The report outlines many of the regulatory gaps in the country.

4.1.1.10 Spain

The professional organization that have developed professional ethics codes (PECs) in Spain have not specifically addressed the area of human genomics.

However, there are documents from national advisory / ethics groups (NAEGs) that specifically address the area of human genomics. Nevertheless, there are many more groups and research projects on this subject than ethical protocols.

The search on guidance documents on how to write research ethics protocols (GDREP), identified a document "Recommendations of the bioethics committee of Spain in relation to the impulse and implantation of good scientific practices in Spain", although it contains no specific content on how to make codes in genomics but rather it is about a very general guide that covers scientific practice in general.

From a global point of view it can be affirmed that the specific protocols on the ethical aspects of human genomics are scarce. The publication of research articles on this subject is very broad, and the presence of groups and research projects whose areas of research are focused on different aspects of human genomics is also especially representative. The data to highlight is that this broad professional dedication in research has little representation in documents / protocols that deal with the ethical aspects.



4.1.1.11 Sweden

We found two main associations/bodies who issue recommendations related to genetics and genomics: Swedish Association of Medical Genetics and Genomics and SMER: the Swedish National Council on Medical Ethics.

Swedish Association of Medical Genetics and Genomics provides a number of guidelines on various issues in clinical genetics and genomics. These are addressed to clinicians and contain very concrete indications on how to proceed in specific situations, e.g. screening for specific cancers etc. We found sections with guidelines on cancer genetics, quality assurance in NGS, and genetics in cardiology. They do not seem to discuss ethical issues related to described situations. These are addressed rather implicitly in the recommendations, e.g. indicated responsibilities of physicians. Interestingly one of the documents (analysed in this report) did not mention the issue of informed consent, i.e. the document on testing of deceased in sudden death and screening his facility. This is potentially a gap which should be addressed.

Meanwhile SMER i.e. the Swedish National Council on Medical Ethics addressed emerging ethical issues in short reports. One of these reports focused on CRISPR-Cas9 reporting on recent discussions among academics. SMER reported on, among others, the article by Baltimore (2015), which includes a call for a moratorium. SMER agreed with the suggestions included in this publication. The other document of SMER analysed herein concerns personalized medicine. SMER's document (Table 5B 5, Annex 1) comments on the document: "Personalized Medicine for the European Citizen - Towards more precise medicine for the diagnosis, treatment and prevention of disease (iPM). November 2012. Report from European Science Foundation (ESF). <http://www.esf.org/index.php?id=7988>". SMER points out that the claims expressed in this document seem to be exaggerated. Recently, in the beginning of 2018, SMER published a letter in which the authors recommend the development of a framework and legislation for the application of genetic technologies in research and in the clinic. For some issues, SMER gives quite specific suggestions as to how the legislation should be changed, e.g. it should be considered whether mitochondrial replacement procedure should be allowed (for the details and other suggestions please see Table 5B 4 for Sweden, Annex 1). The attachment supporting their statement discusses each genomic technology and some related ethical issues. Unlike the former two documents, which to some extent are commentaries on foreign reports containing some recommendations, the latter document recommends more explicitly and concretely changes in the Swedish legislation.

4.1.1.12 UK

Using the SIENNA prescribed steps, we researched professional codes and documents from national advisory and ethics bodies in the UK to find relevant documents in human genomics. We first searched for professional organisations, national advisory bodies and ethics bodies using specified search terms¹¹ using search engines such as DuckDuckGo, Google and the European Society of Human Genetics website. Next, we searched¹² the websites of the identified organisations for ethics codes, guidance documents. Inclusion criteria included ethics, ethical aspects (such as consent, confidentiality) or ELSI being considered in the documents. Articles that were written by individual authors not part of an official or recognised group, professional organisation or advisory body were

¹¹ Example search terms used: UK+professional organisation/national advisory body/ethics body/ethics council+human enhancement/ neuro-enhancement/smart drugs

¹² Example search terms used: enhancement/human enhancement/augmentation/ Code/Guidance/Guidelines/Recommendations/Policy



excluded. One of the key challenges was that there a wide variety of documents specially by organisations such as think tanks that are influential actors but did not strictly fit into the set categories i.e., professional organisation or national ethics group – some of these we included due to their impact and wide acceptability. We found 10 PECs, 12 NAEG documents and 2 GDRECs.

As seen from the research, there are a number of professional and other organisations active in genetics/genomics in the UK (focussed on genetics, cytogenetics) developing Codes of ethics, Codes of Practice, policies, principles, frameworks and guidelines. The nature of these vary from being very broad (ethical guidance for doctors) to covering very specific aspects of genetics (e.g., postnatal best practice, direct-to-consumer genetic testing services, biobank ethics). There are also other relevant organisations than those covered here, e.g., [British Society for Genetic Medicine \(BSGM\)](#) and the Cancer Genetics Group.

Our research for NAEG documents revealed a large number of documents addressing a variety of topics and audiences. The range and nature of identified NAEGs varies. Some of these are not ‘official’ advisory bodies but are highly influential, e.g., Nuffield Council on Bioethics has wide acceptability in relation to identifying and defining ethical issues in biological and medical research (often seen as ‘the UK ethics body’); the PHG Foundation is a non-profit think tank with a special focus on genomics and other emerging health technologies. Genomics England is wholly owned by the Department of Health & Social Care; the Royal Colleges¹³ are professional membership organisations; POST is Parliament's in-house source of independent, balanced and accessible analysis of public policy issues related to science and technology.”

We found only two relevant *guidance documents on research ethics protocols* (GDREC) that might apply but do not specifically mention genomics or genetics.

Of late, there has been a lot of media attention¹⁴ and negative criticism of a new report by the Nuffield Council on Bioethics on “Genome editing and human reproduction: social and ethical issues”,¹⁵ published in July 2018. The report concluded “that the use of heritable genome editing interventions to influence the characteristics of future generations could be ethically acceptable in some circumstances, provided: it is intended to secure, and is consistent with, the welfare of a person who may be born as a consequence of interventions using genome edited cells; and it upholds principles of social justice and solidarity, i.e. it should not be expected to increase disadvantage, discrimination, or division in society.”¹⁶ Even though the conclusions are conditional, this has been seen by some as an approval of sorts for “designer babies”¹⁷ (and generated discussion on this¹⁸) and even “a red carpet for unrestricted use of inheritable genetic engineering, and a gilded

¹³ Note, we did not look for general Codes of ethics of these organisations.

¹⁴ See <http://nuffieldbioethics.org/project/genome-editing-human-reproduction/media-coverage>

¹⁵ <http://nuffieldbioethics.org/project/genome-editing-human-reproduction>

¹⁶ <http://nuffieldbioethics.org/project/genome-editing-human-reproduction>

¹⁷ Knapton, Sarah, “Designer babies on horizon as ethics council gives green light to genetically edited embryos”, The Telegraph, 17 July 2018. <https://www.telegraph.co.uk/science/2018/07/16/designer-babies-horizon-ethics-council-gives-green-light-genetically/>

¹⁸ See <https://www.express.co.uk/life-style/life/992794/designer-babies-gene-editing-eradicate-diseases-eugenics> ; <https://www.independent.co.uk/voices/genome-editing-human-genes-designer-babies-illness-traits-diseases-disabilities-nuffield-council-a8451491.html>



age in which some are treated as genetic ‘haves’ and the rest of us as ‘have-nots’”.¹⁹ This discussion is relevant to consider in the development of Codes and guidelines in SIENNA.

4.1.1.13 USA

The USA SIENNA partner did not have resources allocated for the task 2.3. General overview of the guidance available for human genomics in the USA below was prepared by Uppsala University team.

There are two main professional organizations providing guidelines and recommendations specifically for human genetics and genomics in the USA: American College of Medical Genetics and Genomics (ACMG) and American Society of Human Genetics (ASHG). ASHG gathers professionals and has its own Codes of Ethics (<http://www.ashg.org/about/ethics.shtml>); it issues commentaries and policy statements on relevant issues (<http://www.ashg.org/policy/research.shtml>). Similarly ACMG provides recommendations related to the practice of clinical genetics (<https://www.acmg.net/ACMG/Advocacy/Policy-Statements/ACMG/Advocacy/Policy-Statements.aspx?key=31d4ab23-4888-412f-953e-b5a2be3af63d>). Both organizations are rather active and address in their work variety of problems encountered in human genomics.

There are also professional organizations, which are not focused specifically on genetics or genomics, but which issue guidelines which are sometimes relevant to human genetics/genomics, for example American College of Obstetricians and Gynaecologists.

Regarding the national advisory ethics committees, the Presidential Commission for the Study of Bioethical Issues (PCSB) is likely the most prominent in the USA. The PCBI, as explained on its website, is:

“an advisory panel of the nation’s leaders in medicine, science, ethics, religion, law, and engineering. The Bioethics Commission advises the President on bioethical issues arising from advances in biomedicine and related areas of science and technology. The Bioethics Commission seeks to identify and promote policies and practices that ensure scientific research, health care delivery, and technological innovation are conducted in a socially and ethically responsible manner.”
(<https://bioethicsarchive.georgetown.edu/pcsbi/about.html>)

Among their guidelines, there are some, which address issues pertaining to genomics specifically, for example, “Privacy and Progress in Whole Genome Sequencing” (<https://bioethicsarchive.georgetown.edu/pcsbi/node/764.html>).

4.1.2 Preliminary analysis and discussion

We received the information from 13 countries, 11 of which followed the methodology outlined above. The search and analysis of two remaining countries – Japan and USA is limited in its scope due to limited resources that could be allocated to this task by partners from that countries. These two countries will not be discussed herein further.

Most of the partners found documents in all three categories : professional ethics codes, national advisory/ethics groups, guidance documents on how to write research ethics protocols. Greek and

¹⁹ <https://www.theguardian.com/science/2018/jul/17/genetically-modified-babies-given-go-ahead-by-uk-ethics-body>



Dutch partners did not report any guidance documents on how to write research ethics protocols. The South African partner found documents only in the “professional ethics codes” category.

Documents which are specific to human genomics or genetics were found in all 11 countries (Table 4, based on Tables 5A in Annex 1). Table 4 shows the number of documents found in a few categories. In some countries there were no documents addressing only high throughput genomics. However, all countries have some documents related to genetics (and genomics). This may indicate that the ethical issues related to high-throughput sequencing have not been yet been addressed adequately. These may be considered as requiring specific further attention in SIENNA.

Herein we do not provide thorough analysis of all results obtained in the study due to time constrains. Yet, the findings presented will be further analysed and will be considered in doing further work in SIENNA, in particular for Task 2.7 (proposal for an ethical framework) and for WP5 (Task 5.1: Develop elements to help RECS ; Task 5.2 elaboration of code of responsible conduct ; and Task 5.5: enhancement of ethical frameworks). The specific issues that we will decide to focus on for these tasks (e.g. gene editing or high-throughput sequencing) may be explored further in the project as needed.

Country name	How many documents did you find from each of these sources that specifically address human genetics or genomics (question 2a, Table 5A)	How many documents specifically ONLY address high throughput genomics? By high throughput genomics, we mean using sequencing of larger parts of the genome, or exome (including large gene panels)? (question 3a, Table 5A)	How many documents only address genetics (so testing or screening but without a lot of sequencing)? (question 4a, Table 5A)	How many documents specifically address Genome Modification, gene editing, crispr, or gene therapy (question 6a, Table 5A)
Brazil	17	1	1	2
China	16	4	3	3
France	24	3	3	5
Germany	12	0	9	3
Greece	28	0	7	2
The Netherlands	13	1	3	4
Poland	10	0	9	1
South Africa	2	0	2	0



Spain	9	Not specified	5	3
Sweden	9	1	1	3
UK	24	1	9	6

Table 4 Summary of the national documents

4.2 International search

4.2.1 Results and preliminary discussion

4.2.1.1 Number of documents retrieved

- 1) In the initial search we found 316 documents issued between 1990-2017. Not all of them were relevant to our analysis (see exclusion criteria above). The documents were reviewed, starting from the most recent, and the relevant documents have included in the tables below (Tables 4.1-4.3). Due to time constraints we limited our analysis to years 2014-2017; we found 22 relevant documents issued in this period (8 in the category “exploring genome” – Table 3.2.1, 8 in “modifying genome” – Table 3.2.2, 6 relevant to both categories – Table 3.2.3; the tables can be found in the Annex 2)
- 2) In the search for documents with specific key terms we obtained the following number of international documents (please note that not all of these may be relevant to our analysis, exclusion criteria were not applied to these documents, therefore the numbers below are only indicative):
 - biobank: 71
 - carrier status: 4
 - commercialization: 52
 - communication of results: 31
 - conflict of interest: 9
 - consumer: 1
 - counselling: 42
 - databases: 28
 - deceased: 9
 - discrimination: 58
 - duty to recontact: 9
 - embryos (created for research): 10
 - ethical review: 31
 - eugenics: 17
 - genetic engineering: 17



- germline therapy: 20
- incompetent adult: 20
- integrity: 16
- intellectual property: 47
- minor/child: 42
- newborn: 21
- open source: 4
- patents: 48
- pharmacogenomics: 12
- pre-implantation: 8
- preconception: 17
- prenatal: 22
- respect for human life: 44
- surplus embryos: 11
- umbilical cord blood: 2

These numbers suggest that ethical and legal issues pertaining to human genetics and genomics have been the focus of many documents and have been on the agenda of many international organizations. The list above clearly shows that many various aspects of human genetics and genomics were addressed by guidelines, as well as various principles and values are brought in the documents. Clearly some issues seem to be more popular, such as patents, biobanks; other, e.g. carrier status, much less. This may be related to many factors such as international, economical relevance.

4.2.1.2 Geographical origin: international and regional organizations

The documents were issued either by international organizations gathering stakeholders from all over the world (Organisation for Economic Co-operation and Development, OECD) or regional, gathering members of a particular geographical region (e.g. European Society of Human Genetics). It is important to keep in mind that in both types of organizations not all countries are represented and among the member countries there may be an imbalance with respect to the power they hold in a given organization.

4.2.1.3 Authors and audience

The characteristics of the organizations issuing guidelines relevant to human genetic and genomics is diverse. We found documents issued by organizations focused on economics, such as OECD, through health organizations such as World Health Organization, World Medical Association, to professional organizations strictly focused on genetics and/or genomics (e.g. European Society of Human Genetics). We also found guidelines issued by professional associations, with a focus other than genetics, for example, International Conference of Data Protection and Privacy Commissioners. Interestingly, there are also guidelines coming directly from industry stakeholders, for example the Industry Pharmacogenomics Working Group. Importantly, when analysing such guidelines, the



interests of the stakeholders and potential conflict of interest should be kept in mind. Some guidelines are endorsed by a few organizations; see for example, “Whole-genome sequencing in newborn screening? A Statement on the continued importance of targeted approaches in newborn screening programmes”, which was issued by the European Society of Human Genetics and endorsed by a few other genetics associations. Such endorsement may potentially increase impact of such recommendations.

Importantly, there are a number of organizations which, despite being issued at the national level, such as the UK Nuffield Council or the American College of Medical Genetics, may have worldwide recognition and/or impact. This is related to the language in which they are issued as well as to the fact that these countries are leaders in implementation of new technologies. Their guidelines if are first addressing a given issue, are often cited or discussed by professionals in the field.

The documents were addressed to wide range of stakeholders. For example, the OECD document “Recommendation on Health Data Governance 2017” explicitly addresses some recommendations to governments. Meanwhile, the “Report of the International Bioethics Committee on Updating Its Reflection on the Human Genome and Human Rights” by UNESCO gives specific recommendations to each groups of stakeholders, such as states and governments, the community of scientists, media and educators, and for-profit companies. Guidelines issued by professional associations are usually addressed to a specific group of professionals, for example geneticists.

4.2.1.4 General principles vs. specific dos and don'ts

Notably, the documents are also diverse with regard to how specific the guidance they provide. Some of them discuss in general ethical issues and provide very general guidance on how to approach them (e.g. Global Health Ethics: Key Issues by the World Health Organization). Meanwhile, others offer rather concrete suggestions of ‘dos and don'ts’ (e.g. “Prenatal screening and diagnosis of chromosomal and genetic abnormalities in the foetus in pregnancy” by the Royal Australian and New Zealand College of Obstetricians and Gynaecologists). These approaches to ethical guidance often depend on the audience to which it is addressed and goals of the authors. Some documents provide both, general principles and specific recommendations, for example, the “Report of the International Bioethics Committee on Updating Its Reflection on the Human Genome and Human Rights (UNESCO)”, explains the guiding principles, after which it provides specific recommendations in various applications of genetic and genomics, addressing specifically various stakeholders.

The function or goal of a document is sometimes supported by its format, for example. of a “manual”. “Medical Ethics Manual by World Medical Association, 2015” gives explanation what medical ethics is, its principles, and explains ethical aspects in various contexts of practice as well as discusses case studies. Another document, with similarly explanatory format was “Global Health Ethics: Key Issues” by World Health Organization, whose authors explained:

“This document aims to assist policy-makers, health care providers and researchers to understand key concepts in health ethics and to identify basic ethical questions surrounding health and health care.”

Indeed, the format of the document, being organized around questions which are then explained in rather short chapters and paragraphs, seems to facilitate its understanding and may increase its impact.



4.2.1.5 Scope of application

Clearly, the scope of the documents also varies. Some are very general outlining principles which should guide medical practice in general, such as “WMA Declaration of Lisbon on the Rights of the Patient”. Meanwhile other focus on very specific application of genomic technologies (see European Society of Human Genetic recommendations on newborn screening). Another group of documents addresses a range of ethical issues in genomics in a specific context e.g. of human reproduction (e.g. “Ethical Issues in Obstetrics and Gynecology” of International Federation of Gynecology and Obstetrics).

4.2.1.6 Attributed role/importance

Some documents specify the role and importance which is or should be attributed to them. For example, the OECD Council in the “Recommendation on Health Data Governance 2017” explains that

“OECD Recommendations are not legally binding, but practice accords them great moral force as representing the political will of Member countries and there is an expectation that Member countries will do their utmost to fully implement a Recommendation. The OECD will monitor progress in the implementation of this Recommendation.”

Meanwhile, the Council for International Organization of Medical Sciences (CIOMS) in the document “International Ethical Guidelines for Health-related Research Involving Humans” claims that:

“The ethical principles set forth in these Guidelines should be upheld in the ethical review of research protocols. The ethical principles are regarded as universal. Moreover, the Guidelines should be read and interpreted as a whole.”

The believe that the ethical principles outlined in this document are universal seems to be confirmed by strong language used in these recommendations, for example:

“Therefore, researchers, sponsors, research ethics committees, and health authorities, must ensure that proposed studies are scientifically sound, build on an adequate prior knowledge base, and are likely to generate valuable information”

Indeed, some of these guidelines may be followed relatively widely, based on the importance of the body that issued them. Yet it is not always straightforward to assess the acceptability and impact of the recommendations; the importance of the authoring organization not always translates into recommendations’ recognition and/or impact of the guidance provided. Furthermore, the impact of the guidelines may not be easy to measure. It is also important to keep in mind that some recommendations may spark discussion, rather than change the practice, due to their controversial nature. For example, American College of Medical Genetics issued recommendations on secondary findings in genomic sequencing, which were vividly discussed and later on were updated²⁰.

²⁰ Green et al., “ACMG Recommendations for Reporting of Incidental Findings in Clinical Exome and Genome Sequencing.”; ACMG Board of Directors, “ACMG Policy Statement: Updated Recommendations Regarding Analysis and Reporting of Secondary Findings in Clinical Genome-Scale Sequencing.”



4.2.2 Relevance to SIENNA

Our search and preliminary analysis of the retrieved international documents in human genetics and genomics show that there is variety of ethical guidance provided with respect to the character of the issuing organization, geographical origin, the audience, scope of application, specificity of guidance and attributed role and importance.

This preliminary analysis may serve as a material for reflection and help with regard to type of guidance and ethical framework, which are going to be designed in SIENNA. Based on the various features we found in the document studies, the following questions may be posed:

- To which, and how broad an audience is the SIENNA ethical framework going to be posed?
- Are we going to provide general principles or more specific recommendations to be followed?
- Which applications of human genetics and genomics will be covered? Will they concern research, clinic or both?
- How strictly should these recommendations be followed?
- What format will our guidelines have? e.g. an explanatory “manual” aimed at professionals or more of an “essay” elaborating more on the values and principles suggested?

Among the documents we found, there are good examples for guidelines, which fall in various categories, which may serve as help and inspiration for completion of task 2.7.

Due to time constrains we were not able to provide more detailed analysis of guidelines, yet some studies reviewing and summarizing international (and national) guidelines concerning specific issue are available in the literature. For example:

- Table 1: Summary of Recommendations in Major Group, Organizational, and Government Statements Related to Human Germline Gene Editing in “Human Germline Genome Editing” by K. Ormond et al. (2017)²¹
- Communicating genetic information in families – a review of guidelines and position papers” Forrest et al. (2007)²²
- “Carrier testing in minors: a systematic review of guidelines and position papers” Borry et al. (2006)²³
- “Presymptomatic and predictive genetic testing in minors: a systematic review of guidelines and position papers” Borry et al. (2006)²⁴
- “What is ideal genetic counselling? A survey of current international guidelines” Rantanen et al. (2008)²⁵
- “Direct to consumer genetic testing: a systematic review of position statements, policies ad recommendations” Skirton et al. (2012)²⁶

²¹ Ormond et al., “Human Germline Genome Editing”.

²² Forrest et al., “Communicating Genetic Information in Families - A Review of Guidelines and Position Papers”.

²³ Borry et al., “Carrier Testing in Minors: A Systematic Review of Guidelines and Position Papers”.

²⁴ Borry et al., “Presymptomatic and Predictive Genetic Testing in Minors: A Systematic Review of Guidelines and Position Papers”.

²⁵ Rantanen et al., “What Is Ideal Genetic Counselling? A Survey of Current International Guidelines”.



If we will decide to address particular area of applications in genetics in genomics, such reviews may be helpful in identifying gaps and issues to address.

²⁶ Skirton et al., “Direct to Consumer Genetic Testing: A Systematic Review of Position Statements, Policies and Recommendations”.



4.3 Online survey with REC members

We received 13 completed online surveys out of the 30 RECs to which invitations were sent (Box 1 has the specific results for Human Genomics). The majority of the respondents were slightly aware of technologies in Human Genomics, Human Enhancement and AI & Robotics. A few REC members indicated they were fully aware of technologies in HG. No one was more than slightly aware of technologies in HE and AI&R. Furthermore, the majority of respondents were slightly aware of the ELSI relating to HG, HE and AI. Only a few of the REC members who participated described themselves as experts in all three SIENNA areas. The answers to the question “Does your REC address or offer any special guidance for researchers working in HG/HE/AI&R” showed that most RECs offer no special guidance (HG: 75% no, HE: 93% no, AI&R: 86% no).

One REC member stressed that guidance documents on how to assess research ethics protocols are not needed, since general principles of research ethics would apply in any case. Other members pointed to specific documents that have been developed by (national) committees on genomics.

Box 1 Specific Summary of Results from the survey for questions on human genomics

What did respondents from 13 RECs have to say about about human genomics and ELSI:

- their awareness regarding existing and potentially needed normative guidance for HG: roughly one third reported that they were fully informed about genetic and genomic technologies, with the majority (56%) saying they were slightly aware.
- their awareness of ethical, legal and social issues of HG: 12% stated that they were experts in the field, one third stated they were fully aware and 56% stated they were slightly aware.
- whether they currently offer specific guidance on the ELSI HG to researchers: there was a clear majority of 75% of RECs reporting that they do not offer specific guidance on human genomic research.
- More specific guidance from the REC: 18% had plans to offer more specific guidance, 18% had no plans and 62% reported not being sure about future plans for guidance on HG.
- The need to offer more training on the ELSI of HG to REC members: 88% said yes, and the remainder were not sure

To the question if there are any future plans to deal with the ELSI of HG, HE and AI&R we received the following answers. Please note that identifying details such as locations have been coded:

Human Genomics:

- “RECs normally have limited control on the researches submitted to them. The region around the [X] presents itself as the [important health area]. This is therefore likely that there will be increased research activities in this field. The REC will then adapt itself to this evolution (see remarks above).”
- “we have published guidelines”
- “We plan to help researchers to balance health needs and risks of high expectations, exploitation”

Human Enhancement:

- “It all depends on what is meant by human enhancement. Advance research is done on exoskeleton and repairing brain damages. There is also a lot of activities around doping. This is therefore likely that there will more activities in this field in the future (see remark on human genomics).”



AI & Robotics:

- “RECs normally have limited control on the researches submitted to them. The region around the [X] presents itself as the “important health area”. This is therefore likely that there will be increased research activities in this field. The REC will then adapt itself to this evolution (see remarks above).”
- “[Y] is organising a symposium, specifically designed for members of the [country] ethics committees, on ethical, legal and social issues of artificial intelligence, in [Z] in 2018”
- “Topics like data protection and validation of research are more important in big data”

To the question if the REC members think there is a need to offer additional guidance for people doing research in HG/HE/AI&R we received the following feedback:

Human Genomics:

- *“As recent (and past) history, most abuses do not happen due to a lack of norms but rather a lack of consideration for them and their underlying principles. Producing more norms has been a trend in research ethics and regulation since WWII. As Jay Katz said in 1969: “The proliferation of such codes testifies to the difficulty of promulgating a set of rules that does not immediately raise more questions than it answers. At this stage of our confusion, it is unlikely that codes will resolve many of the problems, though they may serve a useful function later. Even the much endorsed Declaration of Helsinki – praised, perhaps, because it is the newest and therefore the least examined – will create problems for those who wish to implement it”.* There has been limited progress in raising the ethical mentality within research institutions. Of course, this would be less lucrative for ethics centers as the industry and others are less likely to finance virtues behaviour rather than workshops and other publications.”
- “There is a need for the informed consent in this field”
- “risk management, realistic expectations”
- “Ethically difficult issue with rapid development”

Human Enhancement:

- “Not only the same remarks apply than for human genomics, but the very concept of “human enhancement” is at best confusing, at worst the entry door to totalitarianism. The very idea that humans need to be enhanced is worrying, especially if you refer to the previous time in history when similar proposals were formulated and, even worst, tested. As Hans Jonas said in 1969 (again), ““Let us not forget that progress is an optional goal, not an unconditional commitment, and that its tempo in particular, compulsive as it may become, has nothing sacred about it”. The best guidance in fact would be to explain to researchers why “human enhancement” should be banned as a concept.”
- “a guide for REC members regarding the ethical concerns such research projects may raise and possible approaches to deal with them could be useful”
- “The knowledge about these issues and their development is scarce. Identifying the ethical problems they pose is the first step”
- “risk Management, use and abuse,”
- “It is necessary to draw a line between enhancement and mere addiction to anything new”

AI & Robotics:

- “a guide for REC members regarding the ethical concerns such research projects may raise and possible approaches to deal with them could be useful”
- “The knowledge about these issues and their development is scarce. Identifying the ethical problems they pose is the first step”



- “consequences of automated decision support”
- “Quite dangerous research with unpredictable progress”

Furthermore, our last set of questions showed us that almost all respondents think that there is a need to offer additional education and training for REC members to learn more about the ELSI in HG, HE and AI&R.

Conclusion and steps forward

Indeed, from our limited survey it appears that we can suggest that there may be a trend where there is a lack of specific documents specific to RECs (for researchers) on the three technologies addressed in SIENNA. There are indications however, that RECs are aware of specific documents for genomics. The fact that we have aimed the survey at RECs who are mostly for biomedical research may explain why there are no specific documents for AI/R and HE. However, given the overlap of all three areas of technologies, we could argue that even such biomedical RECs should have specific documents for the latter two technology areas.

As with the (further) analysis of the documents mentioned above, the results of this survey will also be deepened where possible (and where needed) in order to contribute further to the task 2.7 and the tasks in WP5.

Sending the survey to EUREC members can be seen as a first round. For task 5.1 (the development of operational guidelines for RECs beyond biomedical research) EUREC will conduct semi-structured interviews with REC members, which will focus also on other research fields than biomedical. EUREC will include the most important questions from the online survey in these semi-structured interviews. This shall lead to further insights

5. Conclusion

In total, one hundred and sixty-four documents which specifically address human genetics and/or genomics were retrieved from 11 countries where country studies were specifically performed: Brazil, China, France, Germany, Greece, The Netherlands, Poland, South Africa, Spain, Sweden, and the United Kingdom. In addition, 22 documents from “international groups” like the OECD, or HUGO were also identified (between 2014-2017) (Annex 2); this includes documents from the USA since despite being issued at the national level, they often have worldwide recognition and/or impact. There was a large range of number of documents with the UK, France and Greece reporting mid to high twenties, and South Africa only reporting two documents addressing human genetics or genomics. Of the documents reported, most addressed genetic (testing or screening, excluding sequencing/genomics; range 1-9), then a smaller portion addressed gene therapy or gene editing (range 0-6) and even less addressed high throughput genomics/sequencing (range 0-4) (Table 4). This is not completely surprising since the latter two areas (re)emerged only recently in comparison to more traditional genetic testing and screening. That being said, having the concrete data to support our focus (in all of WP2 to date) on genomic sequencing and gene editing is valuable; it gives us evidence that we are working in areas where additional guidance is necessary. Indeed, this turns out to be somewhat different than what was initially described in the grant call (i.e. to focus on genetic testing and screening among other things). However, the way we have organized the tasks, we nevertheless still include these subjects in our work, yet give more focus on ELSI of genomic sequencing and gene editing.



Furthermore, the responses from the survey with REC members' also indicate that addressing the ELSI of genomics is not only needed, but would be supported by the large majority of respondents in the context of research ethics. Respondents were in agreement that additional education on the ELSI of HG would be helpful.

In conclusion, this report provides information supporting our course of action in WP2 on ELSI of Human Genetics and Genomics, and it also offers a resource or tool to better inform the work required in task 2.7 (ethical framework in HG; e.g. what are the specific gaps? Are there documents we can use as a starting point?) and WP5 (consortium proposal for RECs and stakeholders).



Annex 1: National search

6. Annex 1: Guidance for the national search

The national search was guided by the following phases and steps, which were shared with the partners.

Persons conducting the search who are not familiar with each area of technology were asked to do some preparatory work before starting with the search: they may want to re-read part of the Deliverables 2.1, 3.1, 4.1 (State-of-the-art review of the three SIENNA areas), as needed and/or use Wikipedia in the country’s language(s) and in English to help with the translation of some of the key words mentioned below.

The following methodology was suggested to all partners:

PHASE 1: Conduct the search for **professional ethics codes** and documents from **national advisory/ethics groups** and identify the most relevant documents. From these documents you will fill in the tables at the end (in this report we will present the results documented in the tables).

#	Step name	Details
Step 1	Search via national associations/societies Note to all: This approach may be best suited to Genomics. So if you find nothing for HE or AI& R, please move on to Step 2 (search via Google/Database)	A. Find national professional associations for the area of technology <ul style="list-style-type: none"> ▪ for Human Genomics see national associations here: https://www.eshg.org/76.0.html (choose 1-3 groups depending on how many exist) ▪ for HE: none per se, but if you like, you can look at National Neuroscience groups etc. ▪ for AI&R: see SATORI reports on how ethics assessment is carried out in different countries: http://satoriproject.eu/work_packages/comparative-analysis-of-ethics-assessment-practices/ (Annex 4) or see The European Association of AI: https://eurai.org/organisation/member-societies or see the International Federation for Robotics: https://ifr.org/members-list B. Find national advisory groups, or national ethics groups that offer ethical guidance for these areas of technology <ol style="list-style-type: none"> i. For example, in Sweden, there is “SMER” The Swedish National Council on Medical Ethics which puts out guidance documents. http://www.smer.se/ ii. You can find these in different ways: <ol style="list-style-type: none"> 1. Search for “name of country + national + group or council or synonym + ethics or advisory or synonym”



		<p>C. Once at the website look for documents for each field of technology</p>
		<p>D. If you don't find anything through A or B, go directly to Database search below in Step 2</p>
		<p>E. For all documents, save all relevant documents (up to 10 per tech) as PDF or word file in a folder (which we will ask you to place on SharePoint once you are done), save as Name of doc + organisation, topic, year. List all Professional Ethics Codes (PECs) documents you find in TABLE 2</p>



<p>Step 2</p>	<p>Search via Google/Database search</p>	<p>A. Databases to search:</p> <ul style="list-style-type: none"> ▪ Google as main database (use google.com or your national google page) ▪ Log out of your personal google account ▪ Use specific databases if available <ul style="list-style-type: none"> ○ e.g. specific database for genetics is POPGEN http://www.popgen.info/ ○ e.g. in Sweden we also have CODEX, which is a compilation of important ELSI guidelines and laws. You may have something similar in your country? http://www.codex.vr.se/en/index.shtml ▪ pages/search results to be read: Look at first 50 results ▪ exclude individual authors, the documents must be from a formal/recognized group or organization. ▪ Time period: No time period restrictions but for Genomics and HE focus on 1998-2018, for AI/R focus on most recent ▪ Keywords: make sure to note these in your report in your language <ul style="list-style-type: none"> • HG: <ul style="list-style-type: none"> → genomics or Genetics or biobanks or registries or pharmacogenomics or pharmacogenetics, or genetic patents or gene editing AND “recommendations” or “points to consider” or “guidelines” or “guidance” or “code” or “policy” AND your country → only if you find no documents with the above searches, then use “biomedicine” or “biomedical research” • HE: <ul style="list-style-type: none"> → “enhancement” or “human enhancement” or “neuro-enhancement” or “human augmentation” AND “recommendations” or “points to consider” or “guidelines” or “guidance” or “code” or “policy” AND your country → “lifestyle drugs” or “biohacking” or “non-therapeutic” or “beyond therapy” or “physical performance enhancement” or “image and performance enhancing drugs” or “nootropics” or “smart-drugs” or “designer drugs” → if none, then use “biomedical research” and check whether there is implicit mentioning • AI&R: <ul style="list-style-type: none"> → for Robotics: “robotics” or “robot” or “robots” or “automation” or “machine” or “machines” or “unmanned” or “driverless” or “pilotless” or “drones” AND “recommendations” or “points to
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		<p>consider” or “guidelines” or “guidance” or “code” or “policy” AND your country</p> <p>→ for Artificial intelligence: “AI” or “artificial intelligence” or “intelligent agents” or “automation” or “smart systems” or smart information systems” or “big data” AND “recommendations” or “points to consider” or “guidelines” or “guidance” or “code” or “policy” AND your country</p> <ul style="list-style-type: none">▪ Languages: local (only English if needed)▪ inclusion/exclusion criteria:<ul style="list-style-type: none">○ inclusion: any ELSI (ethics must be in it, but it can be more applied)○ exclude anything that does not address ELSI, e.g. just practical SOPs○ exclude articles that are written by individual authors who are not part of an official /recognised group/professional organisation/advisory body. If, in your country, individual author recommendations are important, you can include these but flag them and explain why they are important.
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		<p>B. Save all relevant documents in SharePoint (max 15 per tech) as PDF or word, save as Document name + organisation, topic, year.</p> <p>C. List all PECs in TABLE 2 and all NAEGs in TABLE 3, and if, per chance, you found any guidance documents on how to complete an ethics application for a research ethics committee you would put these in TABLE 4 (GDREC). Estimated time allotted per tech: roughly half a day</p>
<p>Step 3 OPTIONAL</p>	<p>Contact experts in your country who work in each field (PLEASE NOTE THAT THIS STEP IS OPTIONAL)</p>	<p>A. If you decide to do this step, you can also do this step in parallel with steps 1 and 2 above.</p> <p>B. Via a search of the literature, or Google, or your professional contacts, contact via email or phone, researchers in your country who work on the ELSI of the different areas of technologies</p> <p>C. Ask them if they are aware of any ELSI guidance documents from: national professional organisations and/or from national ethics groups, and/or if they know of guidance docs for research ethics protocols in your country.</p> <p>D. Indeed, we should not get too bogged down with categories of documents, if the source is too complicated to explain/understand, you may want to simply ask for any important ELSI guidance in your country on each area of tech.</p> <p>E. See example email on the last page of this document.</p> <p>F. Again, save all documents, see how above.</p> <p>G. Fill in the appropriate table based on document type.</p>



<p>Step 4</p>	<p>Document Analysis</p> <p>Note to all: we allot on average 1.5 - 2 hours per document to analyse. If you deem more time needed to analyse more general documents this is fine to have less documents analysed, just explain it. For example, we expect more specific docs for human genomics where it may be easier to grasp the scope faster than more general documents addressing larger areas.</p>	<p>A. Choose for each area 5-15 document that is specific for the area (PEC or NAEG)</p> <ol style="list-style-type: none"> a. If you need to select from a large number of documents, we would like you to select based on the importance of the document in addressing issues that are particularly relevant in your country for that area of technology. b. For Genomics, we ask you to choose one document each for: genetic testing, genetic screening, pharmacogenetics/pharmacogenomics, databases/bioabanks, patents, and gene editing etc.... See instructions for tables 5 <p>B. Answer for these documents the questions asked in the TABLES 5-7.</p>
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Table 5 Guidance for the national search for professional ethics codes and documents from national advisory/ethics groups

Phase 2: Conduct the search for documents for **guidance on writing research ethics protocols (REPs)** and identify the most relevant documents

#	Name	Description
<p>Step 1</p>	<p>Find national REC in your country</p>	<ul style="list-style-type: none"> – if it is a European country, you can use: http://www.eurecnet.org/information/index.html – read on the REC system(s) in your country. – Based on that, choose a REC that is national or most over-reaching/inlfuential. – If your country does not have national RECs, then you can choose a regional REC. Please note that. – for non EU countries (or in case you found nothing on the EUREC page, you will have to search via Google and/or ask someone at your university to explain a bit more about the RECs in your country.



Step 2	Search for guidance on national REC page	Look for documents that offer guidance/information on how to fill out an ethics application.
Step 3	Document Analysis	Choose for each area one document that is specific for the tech area + ELSI. For each relevant REC doc, keep doc as word or PDF, REC name+ year + subject and list it in TABLE 4 Answer for these three documents the questions asked in the TABLE 5-7

Table 6 Guidance on search for documents with guidance on writing research ethics protocols (REPs)

7. Annex 1: Detailed results of the national search in the different countries

In the following you find the detailed results of the national search as they were prepared by the SIENNA partners.

7.1 Brazil

TABLE 1: INDIVIDUAL AND COUNTRY INFORMATION

Names and emails of persons who did the work (if different from above)	Dr. Marcelo de Araujo / Dr. Maria Clara Dias
Your organisation	UFRJ – Universidade Federal do Rio de Janeiro
Your country (again)	Brazil
Search conducted in which language	Portuguese
Acknowledgements (any researcher who helped you to complete this task)	Fabiana Pompermayer (UFRJ, Doctoral Candidate in Bioethics & Health Policy)

Brazil

TABLE 2: LIST OF ALL RELEVANT PROFESSIONAL ETHICS CODES

SIENNA area	Title of document (original + English translation)	URL	Year	Author/organisation	Stated audience	comments
HG	Estatuto <i>(By-Law)</i>	http://www.abbi.org.br/pt/estatuto/ [web page]	2014	ABBI – Associação Brasileira de Biotecnologia Industrial http://www.abbi.org.br/pt/home_pt_br/ <i>(Brazilian Association of Industrial Biotechnology)</i>	Associated members	Very general.

Brazil

<p>HG / HE / AI&R</p>	<p>Rigor e Integridade na Condução da Pesquisa Científica - Guia de Recomendações de Práticas Responsáveis</p> <p><i>(Rigour and Integrity in the Pursuit of Scientific Research. A Guide with Recommendations for Responsible Practices)</i></p>	<p>http://www.abc.org.br/IMG/pdf/doc-4559.pdf [13p.]</p>	<p>2013</p>	<p>ABC – Academia Brasileira de Ciências http://www.abc.org.br <i>(Brazilian Academy of Sciences)</i></p>	<p>Researchers; doctoral and postdoctoral students</p>	<p>Very general. The document does not address specific question relative to the responsible use of HG, HE, AI&R.</p>
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Brazil

<p>HG / AI&R</p>	<p>Código de Conduta <i>(Code of Conduct)</i></p>	<p>https://www.abimed.org.br/files/etica/abimed_codigo_conduta_2018_pt.pdf [21p.] <i>[Text in Portuguese]</i></p> <p>https://www.abimed.org.br/files/etica/abimed_codigo_conduta_2018_en.pdf [21p.] <i>[Text in English]</i></p>	<p>2018</p>	<p>ABIMED – Associação Brasileira da Indústria de Alta Tecnologia de Produtos para Saúde https://www.abimed.org.br</p> <p><i>(Brazilian Association of of Industries of High Technology Health Products)</i></p>	<p>Computer scientists; telemedicine professionals; researchers; entrepreneurs</p>	<p>The code is quite comprehensive and shows some concern over the storage of personal data, but does not specifically address AI&R.</p>
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Brazil

HG / HE	<p>Resolução nº 2, de 5 de março de 2002 - “Aprova o Código de Ética do Profissional Biólogo”</p> <p><i>(Resolution nº 2, 5 March 2002 “Approves the Ethical Code of the Professional Biologist)</i></p>	<p>http://www.cfbio.gov.br/artigos/RESOLUCAO-N%C2%BA-2-DE-5-DE-MARCO-DE-2002 [web page]</p>	2002	<p>Conselho Federal de Biologia – CFBio</p> <p><i>(Brazilian Council of Biology)</i></p>	Biologists; biology researchers	<p>The document is very general, but explicitly forbids eugenic experiments (Chapter 5, art. 22). The document also recommends the use of the “precautionary principle” with genetically modified organisms (Chapter 5, art. 21).</p>
HG / HE	<p>Código de Ética da Profissão de Biomédico</p> <p><i>(Ethics Code of the Biomedicine Professionals)</i></p>	<p>http://cfbm.gov.br/legisacao/codigo-de-etica-da-profissao-de-biomedico/ [web page]</p>	2011	<p>CFBM – Conselho Federal de Biomedicina</p> <p><i>(Brazilian Federal Council of Biomedicine)</i></p>	Biomedicine professionals	<p>The document is very general, but explicitly forbids eugenic experiments (Chapter 3, xi)</p>

Brazil

<p>HG / HE</p>	<p>Código de Ética Médica. (Code of Medical Ethics)</p>	<p>https://portal.cfm.org.br/images/stories/biblioteca/codigo%20de%20etica%20medica.pdf [74p.]</p> <p>Also available at: http://www.portalmedico.org.br/novocodigo/integra_1.asp [web page]</p>	<p>2009</p>	<p>CFM – Conselho Federal de Medicina http://portal.cfm.org.br (Brazilian Federal Council of Medicine)</p>	<p>Physicians, including clinicians; surgeons; psychiatrists</p>	<p>This is an important document. It specifically addresses issues relative to informed consent, eugenics, and genetic modification of human embryos (Chapter 3, art. 15). It forbids non-therapeutic gene editing. It also forbids germline modification for whatever purposes (Chapter 3, art. 16).</p>
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Brazil

<p>HG / AI&R</p>	<p>O Código de Ética da IMIA para Profissionais de Informática em Saúde</p> <p><i>(The Code of Ethics of the IMIA for Health Information Professionals)</i></p>	<p>http://sbis.org.br/images/ProTics/Codigo_Etica_IMIA_Brasil.pdf [11p.]</p> <p>NB: This document is the Brazilian version of this international ethics code:</p> <p><i>The IMIA Code of Ethics for Health Information Professionals</i></p> <p>http://imia-medinfo.org/wp/wp-content/uploads/2015/07/IMIA-Code-of-Ethics-2016.pdf</p>	<p><i>not infor med</i></p>	<p>SBIS – Sociedade Brasileira de Informática em Saúde</p> <p>http://www.sbis.org.br/index.php</p> <p><i>(Brazilian Society of Informatics and Health)</i></p>	<p>Health professionals; computer scientists; physicians; telemedicine professionals</p>	<p>The document is very general.</p>
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TABLE 3: LIST OF ALL RELEVANT DOCUMENTS FROM NATIONAL ADVISORY/ETHICS GROUPS

SIENNA area	Title of document (original + English translation)	URL	Year	Author/organization	Stated audience	comments
HG / HE	1° Relatório de Amostras Seminais para uso em Reprodução Humana Assistida (1 st report on seminal samples for assisted human reproduction)	http://portal.anvisa.gov.br/documents/33840/3484451/1%C2%B0+Relat%C3%B3rio+de+Importa%C3%A7%C3%A3o+de+Amostras+Seminais+para+uso+em+Reprodu%C3%A7%C3%A3o+Humana+Assistida/33c91fcf-18bb-4825-b659-a8a45053113f [19p.]	2017	ANVISA – Agência Nacional de Vigilância Sanitária http://portal.anvisa.gov.br (National Sanitary Surveillance Agency)	Policy makers; clinicians; fertility clinic; press; public at large	The document shows that between 2011 and 2016 the importation of seminal samples for assisted reproduction increased in 2.625% in Brazil.

Brazil

HG / HE	<p>Resolução CFM nº2.168/2017 <i>(Resolution CFM nº2.168/2017)</i></p>	<p>https://sistemas.cfm.org.br/normas/visualizar/resolucoes/BR/2017/2168 [10p.]</p>	2017	<p>CFM – Conselho Federal de Medicina http://portal.cfm.org.br <i>(Brazilian Federal Council of Medicine)</i></p>	Physicians; professionals working in fertility clinics	<p>This is an important document. It establishes ethical guidelines for assisted human reproduction. It forbids non-therapeutic use of IVF and sex selection (see Part 1, §2, art. 5).</p>
HG / HE	<p>Resolução nº 466, de 12 de dezembro de 2012 <i>(Resolution nº 466, 12 December 2012)</i></p>	<p>http://conselho.saude.gov.br/resolucoes/2012/Reso466.pdf [12p.]</p>	2012	<p>CNS – Conselho Nacional de Saúde http://conselho.saude.gov.br <i>(Brazilian National Council of Health)</i></p>	Physicians; clinicians; surgeons; researchers	<p>The document establishes ethical guidelines for research with human beings, including research on assisted human reproduction, IVF, biobanks, iPSCs. It does not directly address HE.</p>

Brazil

<p>HG / HE</p>	<p>Resolução Normativa nº 16, de 15 de janeiro de 2018</p> <p><i>(Normative Resolution nº 16, January, 2018)</i></p>	<p>http://www.mctic.gov.br/mctic/opencvms/legislacao/outros atos/resolucoes/Resolucao Normativa CTNBio n 16 de 1501 2018.html [web page]</p> <p>also available at: https://goo.gl/KtveFN [web page]</p>	<p>2018</p>	<p>CTNBio – Comissão Técnica Nacional de Biossegurança</p> <p>http://ctnbio.mcti.gov.br</p> <p><i>(National Technical Board on Biosecurity)</i></p>	<p>Researchers and entrepreneurs in the domain of biotechnology</p>	<p>This document establishes rules for the use genome editing tools such as CRISPR.</p>
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Brazil

<p>HG / AI&R</p>	<p>Amicus curiae: Coleta de Material Genético no STF</p> <p>(<i>Amicus Curiae at STF [Brazilian Supreme Federal Court]: Collection of DNA sample</i>)</p>	<p>https://itsrio.org/wp-content/uploads/2018/05/Petição-Memorial-Amicus-Perfis-Geneticos250917-final.pdf [27p.]</p> <p>Source page: https://itsrio.org/pt/publicacoes/amicus-curiae-coleta-de-material-genetico-no-stf/</p>	<p>2017</p>	<p>ITS Rio – Institute for Technology and Society of Rio de Janeiro https://itsrio.org/pt/home/</p>	<p>Brazilian Supreme Federal Court (also known as STF)</p>	<p>Amicus Curiae on Federal Law nº 12.654 (http://www.planalto.gov.br/ccivil_03/ Ato2011-2014/2012/Lei/L12654.htm#art3) on forced collection and electronic storage of DNA samples of convicted criminals. The document does not directly address AI.</p>
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TABLE 4: LIST OF ALL RELEVANT GUIDANCE DOCUMENTS ON HOW TO WRITE RESEARCH ETHICS PROTOCOLS

Name of national REC	Title of document (original + English translation)	URL	Year	Stated audience	comments
<p>Sistema CEP-CONEP http://plataformabrasil.saude.gov.br/login.jsf (CEP-CONEP-System)</p> <p>CEP = Comitê de Ética na Pesquisa (Research Ethics Committee)</p> <p>CONEP = Comissão Nacional de Ética em Pesquisa (National Board for Research Ethics)</p>	<p>Norma Operacional nº 001/2013</p> <p>(Operational Norm nº 001/2013)</p>	<p>http://conselho.saude.gov.br/Web_comissoes/conep/aquivos/CNS%20%20Norma%20Operacional%2001%20-%20conep%20finalizada%2030-09.pdf [17p.]</p>	2013	<p>CNS – Conselho Nacional de Saúde http://conselho.saude.gov.br</p> <p>(Brazilian National Council of Health)</p>	<p>This document describes procedures that govern work performed within the CEP-CONEP-System as a whole. This document builds on Resolution nº 446, August, 2011, which is listed below.</p>

Brazil

<p>The local CEPs are ruled by CONEP, which is a section of the CNS – Conselho Nacional de Saúde http://conselho.saude.gov.br <i>(Brazilian National Council of Health)</i></p>	<p>Manual Operacional para Comitês de Ética em Pesquisa <i>(Operational Handbook for Research Ethics Committees)</i></p>	<p>http://conselho.saude.gov.br/biblioteca/livros/manual_operacional_miolo.pdf [138p.]</p>	<p>2007</p>	<p>CNS – Conselho Nacional de Saúde http://conselho.saude.gov.br <i>(Brazilian National Council of Health)</i></p>	<p>This document is a handbook of principles and procedures for writing ethics protocols.</p>
<p>CNS is a section of the “Ministério da Saúde” <i>(Ministry of Health)</i> http://portalms.saude.gov.br</p> <p>An updated directory of current RECs in Brazil can be accessed by following the link “Consultar Comitê de Ética em Pesquisa” at: http://plataformabrasil.sa</p>	<p>Resolução CNS nº 441, de 12 de maio de 2011. <i>(Resolution CNS nº 441, May 2011)</i></p>	<p>http://conselho.saude.gov.br/resolucoes/2011/Reso441.pdf [4p.]</p>	<p>2011</p>	<p>CNS – Conselho Nacional de Saúde http://conselho.saude.gov.br <i>(Brazilian National Council of Health)</i></p>	<p>This document establishes ethical guidelines for the ethical assessment of research projects that involve the use of biobanks.</p>

Brazil

<p>ude.gov.br/login.jsf</p>	<p>Resolução nº 446, de 11 de agosto de 2011</p> <p>(Resolution nº 446, August, 2011)</p>	<p>http://plataformabrasil.saude.gov.br/login.jsf [6p.]</p> <p><i>[There is no direct link to the PDF file. Please follow link for "Resolução e Normativas]</i></p>	<p>2011</p>	<p>CNS – Conselho Nacional de Saúde</p> <p>http://conselho.saude.gov.br</p> <p>(Brazilian National Council of Health)</p>	<p>This document describes procedures that govern work performed within the CEP-CONEP-System as a whole.</p>
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Brazil

	<p>Resolução nº 340, de 8 de julho de 2004</p> <p>(Resolution nº 340, de 8 de julho de 2004)</p>	<p>http://plataformabrasil.saude.gov.br/login.jsf</p> <p>[5p.]</p> <p><i>[There is no direct link to the PDF file. Please follow link for "Resolução e Normativas]</i></p>	<p>2004</p>	<p>CNS – Conselho Nacional de Saúde</p> <p>http://conselho.saude.gov.br</p> <p><i>(Brazilian National Council of Health)</i></p>	<p>This document establishes ethical guidelines for the ethical assessment of research projects in genomics.</p>
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TABLE 5A: General questions on your document search in Human Genomics

<p>1- Did you find guidance documents specific to human genomics or genetics?</p>	<p><i>Yes</i></p>
<p>2a How many documents did you find from each of these sources that specifically address human genetics or genomics</p> <p>2b Did you find more documents than you had the time to analyse in this task?</p>	<p>PECs / 7 NAEG / 5 GDREC / 5 Total: 17</p> <p><i>No</i></p>

Brazil

<p>3- a) How many documents specifically ONLY address high throughput genomics? By high throughput genomics, we mean using sequencing of larger parts of the genome, or exome (including large gene panels).</p>	<p>PECs / None</p> <p>NAEG / 1: (“Normative Resolution nº 16, January, 2018” at: https://goo.gl/KtveFN)</p> <p>GDREC / None</p> <p>Genomics used for diagnostics in the clinic (genomic testing) / 1: (Resolution CFM nº2.168/2017)</p> <p>Genomics used for research / 7: (Resolution CFM nº2.168/2017); all documents in Table 4; (Resolution nº 2, 5 March 2002 “Approves the Ethical Code of the Professional Biologist)</p>
<p>3b Please list the year of publication of each documents here: 2018</p>	<p>Consent in genomics in the clinic / 1: (Resolution CFM nº2.168/2017)</p> <p>Consent in genomics in research / 9: (Code of Medical Ethics); (Resolution CFM nº2.168/2017); (Resolution nº 466, 12 December 2012); All documents in Table 4; (Resolution CNS nº 441, May 2011)</p>
<p>3c please specify the topics addressed explicitly, ideally indicate the number of documents that address each, place an X beside the topic and add the number of docs</p>	<p>Return of results in genomics in the clinic /</p> <p>Return of results in genomics in research /</p> <p>Reinterpretation and recontact in genomics in the clinic /</p> <p>Reinterpretation and recontact in genomics in the clinic /</p> <p>Genomics for newborn screening /</p> <p>Genomics for prenatal screening or testing / 1: 1: (Resolution CFM nº2.168/2017)</p> <p>Non-invasive prenatal testing/screening /</p> <p>Discrimination / 3: (Ethics Code of the Biomedicine Professionals); (Resolution nº 2, 5 March 2002 “Approves the Ethical Code of the Professional Biologist); (Code of Medical Ethics)</p> <p>Eugenics / (Ethics Code of the Biomedicine Professionals); (Resolution nº 2, 5 March 2002 “Approves the Ethical Code of the Professional Biologist); (Code of Medical Ethics)</p> <p>Justice / (Ethics Code of the Biomedicine Professionals); (Resolution nº 2, 5 March 2002 “Approves the Ethical Code of the Professional Biologist); (Code of Medical Ethics)</p> <p>Genetic exceptionalism /</p>

Brazil

4 a How many documents only address genetics (so testing or screening but without a lot of sequencing)?

PECs / *
NAEG / 1
GDREC / *

** Basically all documents only deal with genetics, except for one document viz: The Normative Resolution nº 16, January, 2018 issued by CTNBio – the National Technical Board on Biosecurity, available: <https://goo.gl/KtveFN>.*

4b List the years of publications of each document here: 2018

Genetics used for diagnostics in the clinic (genetic testing) ____

Genetics used for research ____

Consent in genetics in the clinic ____

3c please specify the topics addressed explicitly, ideally indicate the number of documents that address each, place an X beside the topic and add the number of docs. [See please 3c above]

Consent in genetics in research ____

Return of results in genetics in the clinic ____

Return of results in genetics in research ____

Reinterpretation and recontact in genetics in the clinic ____

Reinterpretation and recontact in genetics in the clinic ____

newborn screening ____

prenatal screening or testing ____

Non-invasive prenatal testing/screening ____

Discrimination ____

Eugenics ____

Justice ____

Genetic exceptionalism ____

Brazil

<p>5. How many documents address both genetics and genomics?</p>	<p>Specify docs or not? <i>Basically all documents only deal with genetics, except for one document viz: The Normative Resolution nº 16, January, 2018 issued by CTNBio – the National Technical Board on Biosecurity, available at: https://goo.gl/KtveFN.</i></p>
<p>6 a) How many documents specifically address Genome Modification, gene editing, crispr, or gene therapy 6b) do they address somatic or germline editing or both?</p>	<p>6a) <i>The Normative Resolution nº 16, January, 2018 issued by CTNBio – the National Technical Board on Biosecurity. This documents deals with gene editing, though it aims chiefly at the use of CRISPR in the agriculture sector.</i> 6b) <i>The Code of Medical Ethics issued by CFM – Brazilian Federal Council of Medicine explicitly forbids non-therapeutic gene editing. It also forbids germline modification for whatever purposes (Chapter 3, art. 16).</i></p>
<p>How many documents specifically address (so in the title of the document)</p>	<p>Genetic or genomic testing / 1: <i>(Resolution CFM nº2.168/2017)</i> Genetic or genomic screening Biobanks or databases / 1: <i>(Resolution CNS nº 441, May 2011)</i> Pharmacogenomics or pharmacogenetics Patents / <i>None</i></p>

Table 5B RELEVANT PEC or NAEG DOCUMENTS IN HUMAN GENOMICS

Document 1

Document found via (national associations or google or another database)	<i>Google</i>
Title of document	<i>Resolução CNS nº 441, de 12 de maio de 2011 (Resolution CNS nº 441, May 2011)</i>
Scope/ main topic	<i>Biobanks</i>
Kind of document (PEC or NAEG or other)	<i>GDREC</i>
Document developed by whom (organisation, profession)?	<i>CNS – Conselho Nacional de Saúde Brazilian National Council of Health</i>
Year the document was published	<i>2011</i>
Document saved in folder as	<i>SIENNA X.3 BRAZIL - Table 4 - CNS 2011 - Resolution 441.pdf</i>
Who is the stated audience, if none specified, write not stated (NS)	<i>NS</i>
For Clinical or research or both or not specified	<i>Not specified</i>

Brazil

<p>What level of guideline is provided?</p>	<p>Very broad: i.e.: on many aspects of genomics or genetics, including many issues all together e.g. Very specific: only focuses on 1- 2 issues More Theoretical (values, principles) More practical (a practitioner could apply them)</p>
<p>What is the scope of the document, and what are the main recommendations?</p>	<p><i>Address issues related to informed consent. Every 5 years the institution that is responsible for the biobanks has to submit a report to Brazilian National Council of Health. Human cells may be stored for up to 12 years. The report must address issues related, but not limited to, discarding of human cells.</i></p>
<p>Which life stage is addressed in the document?</p>	<p>Adults Minors (excluding newborns) Newborns Prenatal</p> <p><i>Not specified</i></p>
<p>Which ethical challenges are addressed in the document? (you can see lists above for ideas of types)</p>	<p><i>Not specified</i></p>

Brazil

<p>How are the ethical issues addressed? (practically, theoretically etc? Any comment on this is fine</p> <p>Are solutions offered? If so, which ones? (depending on the document, this may be redundant with above)</p>	<p><i>Not specified</i></p>
<p>How is the document structured? e.g. Which format is used in the document (checklist, continuous text, other)?</p>	<p><i>Sequence of numbered paragraphs and articles.</i></p>
<p>Is the document clearly understandable?</p>	<p><i>Yes</i></p>
<p>Why is the document important/useful for your country? why did you choose this doc? TELL US WHY IMPORTANT for country and why for SIENNA</p>	<p><i>There is a growing demand for biobanks in Brazil. This document rules research and services that involve the use of biobanks.</i></p>
<p>Is the document useful for the development of the SIENNA codes and other ethical frameworks? If yes, please explain.</p>	<p><i>Yes. The document may be compared to other documents, which rule the work of biobanks in other countries. This will allow SIENNA partners to have a broader view on how this topic has been addressed in different legal cultures.</i></p>

Document 2

Document found via (national associations or google or another database)	<i>Google</i>
Title of document	<i>Resolução CFM nº2.168/2017 (Resolution CFM nº2.168/2017)</i>
Scope/ main topic	<i>Assisted human reproduction</i>
Kind of document (PEC or NAEG or other)	<i>NAEG</i>
Document developed by whom (organisation, profession)?	<i>CFM – Conselho Federal de Medicina (Brazilian Federal Council of Medicine)</i>
Year the document was published	<i>2017</i>
Document saved in folder as	<i>SIENNA X.3 BRAZIL - Table 3 - CFM 2017 - Technical Standards Assisted Human Reproduction.pdf</i>
Who is the stated audience, if none specified, write not stated (NS)	<i>NS</i>
For Clinical or research or both or not specified	<i>Both</i>

Brazil

<p>What level of guideline is provided?</p>	<p>Very broad: i.e.: on many aspects of genomics or genetics, including many issues all together e.g. Very specific: only focuses on 1- 2 issues More Theoretical (values, principles) More practical (a practitioner could apply them)</p>
<p>What is the scope of the document, and what are the main recommendations?</p>	<p><i>The document establishes ethical guidelines for assisted human reproduction. It forbids non-therapeutic use of IVF and sex selection (see Part 1, §2, art. 5). It allows practices that were not yet clearly regulated such as, for instance, shared pregnancy.</i></p>
<p>Which life stage is addressed in the document?</p>	<p>Adults Minors (excluding newborns) Newborns Prenatal</p>
<p>Which ethical challenges are addressed in the document? (you can see lists above for ideas of types)</p>	<p><i>The commercialization of human semen is not allowed in Brazil, but its importation from foreign sperm banks is allowed. The documents established rules for the importation of human semen.</i></p>

Brazil

<p>How are the ethical issues addressed? (practically, theoretically etc? Any comment on this is fine</p> <p>Are solutions offered? If so, which ones? (depending on the document, this may be redundant with above)</p>	<p><i>The document adopts a more inclusive conception of “family” by allowing lesbian couples to share a pregnancy, i.e. the egg of one woman is extracted and fertilized in vitro with the sperm of an anonymous donor, then the fertilized egg is transferred to the womb of the other woman.</i></p>
<p>How is the document structured? e.g. Which format is used in the document (checklist, continuous text, other)?</p>	<p><i>Sequence of numbered paragraphs and articles.</i></p>
<p>Is the document clearly understandable?</p>	<p><i>Yes.</i></p>
<p>Why is the document important/useful for your country? why did you choose this doc? TELL US WHY IMPORTANT for country and why for SIENNA</p>	<p><i>This topic was not clearly regulated in Brazil before this document was issued.</i></p>
<p>Is the document useful for the development of the SIENNA codes and other ethical frameworks? If yes, please explain.</p>	<p><i>Yes. The document may be compared to documents that rule the work of fertility clinics in other countries. This will allow SIENNA partners to have a broader view on how this topic has been addressed in different legal cultures.</i></p>

7.2 China

TABLE 1: INDIVIDUAL AND COUNTRY INFORMATION

Names and emails of persons who did the work (if different from above)	LIU Hongzuo
Your organisation	Dalian University of Technology
Your country (again)	China
Search conducted in which language	Chinese and English
Acknowledgements (any researcher who helped you to complete this task)	WANG Qian

TABLE 2: LIST OF ALL RELEVANT PROFESSIONAL ETHICS CODES

SIENNA area	Title of document	URL	Year	Author/organisation	Stated audience	comments
HG	Construction and Management Method of Biological Gene Information Database (SZDB /Z 92—2014)	http://www.szmqz.gov.cn/xxgk/qt/ztlm/szb/szsdfbz_szb/201412/W020141202347715842445.pdf	2014	Shenzhen Municipal Administration of Market Supervision	Public and Information professionals in Shenzhen	<p>This regulation is formulated against the background that there are no any national standards, industry standards, and local standards for the construction of biological gene information databases. Therefore, although it is not a document at the national level, it is also necessary to list it.</p> <p>It specifies the basic requirements for the equipment and environment related to the construction of biological genetic information database, as well as the processing methods and principles of biological information data. The document clearly points out that "in the process of building biological gene information databases, an ethical review committee should be set up to establish the ethical criteria of biological gene information databases, and to conduct ethical review on the sources, usage and sharing of biological gene information data and other ethical issues. (6.1.1) ."</p>

HG	Management Method of Human Assisted Reproductive Technology	http://www.nhfpc.gov.cn/mohzcfgs/s6729/200804/29342.shtml	2001	National Health and Family Planning Commission of People's Republic of China	Public information professionals and	Although the document does not explicitly refer to HG, due to the close relation with human reproductive technology, the HG is also applied with this regulation. The fourteenth article states that: "the implementation of human assisted reproductive technology should follow the principle of informed consent and sign informed consent. Those involving ethical issues should be referred to the medical ethics committee for discussion.
HG	Ethical Principles of Human Assisted Reproductive Technology and Human Sperm Bank	http://www.nhfpc.gov.cn/qjjys/s3581/200805/f69a925d55b44be2a9b4ada7fcdec835.shtml	2003	Ministry of Health, PRC	Public, medical institutions, policymakers	This document makes no specific reference to HG, but the document clearly identifies a number of ethical codes that are very relevant to genetic technology. Furthermore, these ethical norms are crucial to the SIENNA project, so we consider it makes sense to list it. There is no doubt that these ethical norms play an important role in the safe, effective and rational application of human assisted reproductive technology, the protection of the health and interests of individuals, families, and future generations, and the maintenance of social public welfare.

HG	Management Method of Clinical Application of Medical Technology	http://www.nhfpc.gov.cn/mohbgt/s9507/200903/39511.shtml	2009	Ministry of Health, PRC	Public, medical institutions, policymakers	This document stipulates that the clinical application of medical technology should follow the principles of science, safety, standardization, effectiveness, economy and ethics. And If there is an absence of ethical justification for one medical technology, it should immediately stop the clinical application of the medical technology (article 41). And in the annex, it is clearly pointed out that gene therapy is related to ethical issues. Its safety and effectiveness are still one of the medical technologies that need to be further verified by standardized clinical trials.
HG	Human Gene Therapy Research and Guiding Principles for Quality Control of Preparations	http://www.cde.org.cn/zdyz.do?method=largePage&id=37	2003	China Food and Drug Administration	Biomedical researchers and related research institutions	This document points out the basic principles of the production of gene therapy preparations. Moreover, it is emphasized that researchers should strengthen consultation and demonstration, and propose a scientific and feasible research protocol to ultimately ensure safe and effective gene therapy products. In addition, full attention should be paid to the codes of ethics: before the implementation of this programme, it is necessary to explain to the patient that the treatment is in the experimental stage, and its potential risks and benefits. At the same time, it is ensured that the patient has the right to either participate or withdraw the trials. And it is necessary to protect the right of patients to have other treatments once the trial is terminated ; To protect the patient's privacy strictly; only after the patient and his family fully understand and give the consent, the treatment can be started.

HG	Management Method of Stem Cell Clinical Research (Trial implementation)	http://www.nhfpc.gov.cn/qjjys/s3581/201508/28635ef99c5743e294f45e8b29c72309.shtml	2015	National health Commission of the People's Republic of China 、 China Food and Drug Administration	Public information and professionals	<p>This document makes no specific reference to HG, but in fact, in the field of biomedical research and clinical treatment, gene technology is closely related to stem cells. So we consider that some of the ethical norms in this document are also of reference significance.</p> <p>The document lists the principles that stem cell research needs to follow, and emphasizes the need to establish a committee of experts on stem cell clinical research and a committee of ethical experts to provide technical support and ethical guidance for the standardized management of stem cell clinical research.</p>
HG	Management Specification for Gene Chip Diagnostic Technology (Trial Implementation)	http://www.nhfpc.gov.cn/yzygj/s3585u/200911/2de81ba07d914819a4fe5e2253254e0e.shtml	2009	Ministry of Health, PRC	Public information and professionals, especially gene chip technology related health technicians and medical institutions	<p>This document is the basis for technical audit institutions to apply for clinical application of gene chip diagnostic technology in medical institutions. It is the minimum requirement for medical institutions and medical technicians to carry out gene chip diagnosis technology. It clearly points out that "all gene chip diagnostic technologies should be approved and oversight by the ethicscommittee for clinical application. "</p>

TABLE 3: LIST OF ALL RELEVANT DOCUMENTS FROM NATIONAL ADVISORY/ETHICS GROUPS

SIENNA area	Title of document	URL	Year	Author/organization	Stated audience	comments
HG	Interim Management Method of Human Genetic Resources	http://www.most.gov.cn/bszn/new/rlyc/wjxz/200512/t20051226_55327.htm	1998	General Office of the State Council of the People's Republic of China	Public and Information professionals	This document is the first normative document of China's comprehensive administration of human genetic resources. Regulations on the administration system of human genetic resources and the procedures for examination and approval of international cooperation and outbound activities in China's human genetic resources have been stipulated. It has become an important basis for the administration of human genetic resources in China.

HG	Regulations on the Management of Human Genetic Resources(Sent for Review by the Ministry of Science and Technology)	http://www.gov.cn/gzdt/2012-10/31/content_2254379.htm	2016	State Council of the People's Republic of China	Public and Information professionals	This document provides an administrative licensing system for collection activities of human genetic resources, international cooperation and outbound activities. Specific procedures and requirements are specified for the exit of human genetic resources. It also strengthened supervision and inspection of human genetic resources administration activities.
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HG	Ethical Guiding Principles for Human Embryonic Stem Cell Research	http://www.most.gov.cn/fggw/zfwj/zfwj2003/200512/t20051214_54948.htm	2003	Ministry of Science and Technology of People's Republic of China	Public and Information professionals	This document makes no specific reference to HG, but in fact, in the field of biomedical research and clinical treatment, gene technology is closely related to stem cells. So we consider that some of the ethical norms in this document are also of reference significance (As mentioned above) . The promulgation of this document enables the research of human embryonic stem cells in the field of biomedicine to conform to the bioethics norms. It has ensured the internationally recognized bioethical principles and the relevant regulations in China have been respected and observed, and promoted the healthy development of human embryonic stem cell research.
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HG	Management Method of Human Sperm Bank	http://www.nhfpc.gov.cn/mohzcfgs/pgz/200804/29615.shtml	2001	Ministry of Health, PRC	of Public and Information professionals	This document makes no specific reference to HG, but in fact, with the development of technology, reproductive technology based on gene technology has been able to intervene in the whole process of human reproduction. So we consider that some of the ethical norms in this document are also of reference significance. The document sets up a management standard for human sperm bank, which ensures the safe, effective application and healthy development of human assisted reproductive technology. The document clearly states that the collection and provision of sperm should comply with the voluntary and ethical principles of the parties. Moreover, medical institutions applying for the establishment of human sperm banks should establish medical ethics committees.
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HG	Ethical Review of Biomedical Research Involving Human Beings	http://www.nhfpc.gov.cn/fzs/s3576/201610/84b33b81d8e747eaaf048f68b174f829.shtml	2016	National health Commission of the People's Republic of China	Public, Researchers and Information professionals	This approach standardizes the ethical review of human biomedical research. It is of great significance for respecting and protecting the legitimate rights and interests of the subjects, protecting human life and health and safeguarding human dignity. It clearly stipulates that ethical review should abide by the provisions of national laws and regulations, respect the voluntary willingness of the subjects in the study, and abide by the principles of being beneficial, doing no harm and justice.
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HG	Proposal for the Implementation of Ethical Standards for Chinese Pharmaceutical Enterprises	http://www.cmba.org.cn/common/index.aspx?nodeid=275	2015	Chinese Medical Biotechnology Association	Various member units and pharmaceutical and commercial enterprises	The proposal does not specify the gene technology, but it puts forward common moral standards for genetic technology practitioners and genetic technology companies, in a broader sense, all the stakeholders in the bio pharmaceutical industry (including companies, industry associations, professional organizations, management units and anti-corruption units) which are also of reference to the SIENNA project. It ensures the maximization of the interests of the patients in the medical activities, and is of great significance to the strengthening of drug safety supervision, the fight against commercial bribery and the improvement of the commercial moral behavior among the stakeholders.
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HG	Guiding Principles for Ethical Review of Drug Clinical Trials	http://www. zs-hospital. sh. cn/lcsy/101119. htm	2010	China Food and Drug Administration	Public, Researchers and information professionals	<p>This document does not specifically target gene technology, but it also applies to genetic technology. This guideline plays an important role in strengthening the guidance and supervision of ethical review of drug clinical trials, standardizing ethical review procedures of drug clinical trials, and safeguarding the clinical trials meet the requirements of scientific and ethical requirements. This document clearly points out that the ethics committee should review the scientificity and ethical rationality of the drug clinical trial projects to ensure the dignity, safety and rights of the subjects, to promote the scientific and healthy development of drug clinical trials and to enhance public trust and support for drug clinical trials. Moreover, the ethical review conference should pay special attention to the science, safety, fairness, the protection of the subjects, the documents of informed consent and the process of informed consent, and the conflict of interests (Article 13). The above contents also have reference significance for SIENNA projects.</p>
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HG	Management Method of Prenatal Diagnosis Technology	http://www.moh.gov.cn/mohzcfgs/s3577/200804/17612.shtml	2003	Ministry of Health, PRC	Public, medical workers and information professionals	This document does not specifically target gene technology, but it also applies to gene technology. It is of great significance to protect the health of mother and child, improve the quality of the birth population, safeguard the safety and effectiveness of prenatal diagnosis technology, and standardize the supervision and management of prenatal diagnosis technology.
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TABLE 4: LIST OF ALL RELEVANT GUIDANCE DOCUMENTS ON HOW TO WRITE RESEARCH ETHICS PROTOCOLS

Name of national REC	Title of document (original + English translation)	Ethical issues addressed in which SIENNA area?	URL	Stated audience	comments
National Health Commission of the People's Republic of China	Ethical Review of Biomedical Research Involving Human Beings	HG	http://www.nhfpc.gov.cn/fzs/s3576/201610/84b33b81d8e747eaaf048f68b174f829.shtml	Public, Researchers and Information professionals	<p>This document makes no specific reference to how to write research ethics protocols on HG. But it lists general principles of professional conduct of biomedical research.</p> <p>For example, "medical and health institutions having no set up ethical committees should not carry out biomedical research involving human subjects. "</p> <p>(Article 7): "the responsibility of the ethics committee is to protect the legitimate rights and interests of the subjects, to maintain the dignity of the subjects and to promote the standard of biomedical research; (Article 8); the basic criteria for the approval of the research project by the ethics committee are((Article 22):</p> <ul style="list-style-type: none"> - adherence to the social value of bioethics;

China

					<ul style="list-style-type: none"> - scientific research protocol ; - fair enrollment of the subjects; - favorable risk-benefit ratio; - valid consent; - respect for the subjects; - compliance with the integrity of scientific research.
Ministry of Science and Technology of People's Republic of China	Ethical Guiding Principles for Human Embryonic Stem Cell Research	HG	http://www.most.gov.cn/fggw/zfwj/zfwj2003/200512/t20051214_54948.htm	Ministry of Science and Technology , PRC ; Ministry of Health, PRC	This code makes no specific reference to HG, but in fact, in the field of biomedical research and clinical treatment, gene technology is closely related to stem cells (As mentioned above) . The document clearly points out a number of codes of conduct that must be observed in human embryonic stem cell research.

3a) How many documents specifically ONLY address high throughput genomics? By high throughput genomics, we mean using sequencing of larger parts of the genome, or exome (including large gene panels).

3b Please list the year of publication of each documents here:

-Management Method of Human Sperm Bank (2001)

-Human Gene Therapy Research and Guiding Principles for Quality Control of Preparations (2003)

-Management Method of Clinical Application of Medical Technology (2009)

-Construction and Management Method of Biological Gene Information Database (2014)

3c please specify the topics addressed explicitly, ideally indicate the number of documents that address each, place an X beside the topic and add the number of docs

PECS-2
NAEG-2
GDREC-0

- Genomics used for diagnostics in the clinic (genomic testing) 4
- Genomics used for research 1
- Consent in genomics in the clinic 1
- Consent in genomics in research 1
- Return of results in genomics in the clinic 1
- Return of results in genomics in research 1
- Reinterpretation and recontact in genomics in the clinic
- Reinterpretation and recontact in genomics in the research
- Genomics for newborn screening 0
- Genomics for prenatal screening or testing 1
- Non-invasive prenatal testing/screening 1
- Discrimination 1
- Eugenics 0
- Justice 0
- Genetic exceptionalism 0

4 a How many documents only address genetics (so testing or screening but without a lot of sequencing)?

PECS-1
NAEG-2
GDREC-0

4b List the years of publications of each document here

-Interim Management Method of Human Genetic resources

(1998)

-Management Method of Human Assisted Reproductive Technology

(2001)

-Regulations on the Management of Human Genetic Resources(Sent for Review by the Ministry of Science and Technology) (2016)

3c please specify the topics addressed explicitly, ideally indicate the number of documents that address each, place an X beside the topic and add the number of docs

- Genetics used for diagnostics in the clinic (genetic testing) 1
- Genetics used for research 0
- Consent in genetics in the clinic 0
- Consent in genetics in research 0
- Return of results in genetics in the clinic 0
- Return of results in genetics in research 0
- Reinterpretation and recontact in genetics in the clini 0
- Reinterpretation and recontact in genetics in the clinic 0
- newborn screening 0
- prenatal screening or testing 0
- Non-invasive prenatal testing/screening 0
- Discrimination 1
- Eugenics 1
- Justice 1
- Genetic exceptionalism 1

<p>5.How many documents address both genetics and genomics?</p>	<p>Two documents but not specification. <i>-Management Method of Prenatal Diagnosis Technology (2003)</i> <i>-Ethical Review of Biomedical Research Involving Human Beings (2016)</i></p>
<p>6 a) How many documents specifically address Genome Modification, gene editing, crispr, or gene therapy 6b) do they address somatic or germline editing or both?</p>	<p>Three documents but only specifically address gene therapy <i>-Human Gene Therapy Research and Guiding Principles for Quality Control of Preparations (2003)</i> <i>-Management Method of Clinical Application of Medical Technology (2009)</i> <i>-Management Specification for Gene Chip Diagnostic Technology (Trial Implementation) (2009)</i></p> <p>The first documents only address somatic editing. The others are not mentioned.</p>

How many documents specifically address (so in the title of the document)

Genetic or genomic testing-3 docs:

-Construction and Management Method of Biological Gene Information Database (2014)

-Management Specification for Gene Chip Diagnostic Technology (Trial Implementation) (2009)

-Human Gene Therapy Research and Guiding Principles for Quality Control of Preparations (2003)

Genetic or genomic screening-3 docs:

-Management Method of Prenatal Diagnosis Technology (2003)

-Human Gene Therapy Research and Guiding Principles for Quality Control of Preparations (2003)

-Management Specification for Gene Chip Diagnostic Technology (Trial Implementation) (2014)

Biobanks or databases-3 docs:

-Management Method of Human Assisted Reproductive Technology (2003)

-Management Method of Human Sperm Bank (2001)

-Construction and Management Method of Biological Gene Information Database (2014)

Pharmacogenomics or pharmacogenetics-0 doc.

Patents-1 doc:

-Human Gene Therapy Research and Guiding Principles for Quality Control of Preparations (2003)

Table 5B RELEVANT PEC or NAEG DOCUMENTS IN HUMAN GENOMICS, copy the table for as many documents as you have to describe. Up to 5-10 Please fill one table out for each document addressing

5Bi: genomic testing (if none on genomics, use genetic testing)

5Bii genomic screening (if none on genomics, use genetic screening)

5Biii biobanks, databases or registries

5Biv pharmacogenomics or pharmacogenetics

5Bv patents

5Bvi gene editing, crispr, or gene therapy

Document 1 – Human Gene Therapy Research and Guiding Principles for Quality Control of Preparations

Document found via (national associations or google or another database)	National association (CFDA)
Title of document	Human Gene Therapy Research and Guiding Principles for Quality Control of Preparations
Scope/ main topic	(choose from list above, 5Bi-5Bvi, write out the topic) Gene therapy, Patents
Kind of document (PEC or NAEG or other)	PEC
Document developed by whom (organisation, profession)?	CFDA

Year the document was published	2003
Document saved in folder as	Human Gene Therapy Research and Guiding Principles for Quality Control of Preparations .doc
Who is the stated audience, if none specified, write not stated (NS)	Biomedical researchers and related research institutions
For Clinical or research or both or not specified	Both for clinical practice and research
What level of guideline is provided?	<p>Very broad: i.e.: on many aspects of genomics or genetics, including many issues all together e.g.</p> <p><input checked="" type="checkbox"/> Very specific: only focuses on 1- 2 issues</p> <p>More Theoretical (values, principles)</p> <p><input checked="" type="checkbox"/> More practical (a practitioner could apply them)</p>

<p>What is the scope of the document, and what are the main recommendations?</p>	<p>Scope:</p> <ul style="list-style-type: none"> -Research on human gene therapy -Quality control technology of preparation -Technical specifications -Ethical principles 	<p>Main recommendations:</p> <p>Basic principles of research and production of gene therapy preparations:</p> <p>Firstly, we must ensure safety and effectiveness. We should fully estimate the risk that may be encountered and propose corresponding quality control requirements.</p> <p>Secondly, we should promote the research of gene therapy and strengthen innovation. The characteristics of gene therapy should be noted. When applying for clinical trials to the State Administration of Drug Administration, the following materials should be prepared in addition to "research content and product quality control" in accordance with the guiding principles, as well as the following materials:</p> <ol style="list-style-type: none"> (1) review of the status and progress of research at home and abroad; (2) the intellectual property rights of this research or product; (3) to closely monitor the whole process of clinical research based on the complexity and risk of genetic technology. (4) to attach full importance to the principles of ethics, such as autonomy, informed consent and privacy protection.
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Which life stage is addressed in the document?	<input checked="" type="checkbox"/> Adults Minors (excluding newborns) Newborns Prenatal
Which ethical challenges are addressed in the document? (you can see lists above for ideas of types)	This document focuses on ethical challenges brought about by gene therapy and gene sequencing. This document focuses on ethical challenges brought about by gene therapy and gene patents. Based on the complexity and risks of gene technology, there are many unknown situations in gene therapy, so it is necessary to ensure the operation of technical specification and the rights and interests of patients. In addition, patent disputes are also an important aspect.

<p>How are the ethical issues addressed? (practically, theoretically etc? Any comment on this is fine</p> <p>Are solutions offered? If so, which ones? (depending on the document, this may be redundant with above)</p>	<p>Both theoretical and practical norms are included: it is also necessary to formulate detailed technical specifications and elaborate ethical considerations, and to provide relevant materials in the application of clinical trials to the State Drug Administration. For intellectual property rights, the document clearly stipulates that research institutions need to provide intellectual property retrieval reports and search results for this research or product. Obviously, these measures are of positive significance for the eventual assurance of safe and effective gene therapy products.</p> <p>Solutions: Requirements held in this documents should also pay full attention to the principles of ethics, and specifically in accordance with the relevant provisions of the State Drug Administration strict implementation. This includes explaining to the patient that the treatment is in experimental phase, its potential benefits and risks, ensuring that the patient has the right to participate and withdraw the treatment, and to secure the right to receive other treatments once the trial is withdrawn. To protect patients' privacy strictly. After the patient and his family fully understand and give consent, the treatment can begin.</p>
<p>How is the document structured?</p> <p>a. g. Which format is used in the document (checklist, continuous text, other)?</p>	<p>Chapters & sections</p> <p>continuous text</p>

<p>Is the document clearly understandable?</p>	<p>Clearly but not easy to understand, because this document involves a lot of technical terms related to genetic technology, we need a profound knowledge reserve. However, the ethical principles involved are very clear.</p>
<p>Why is the document important/useful for your country? why did you choose this doc? TELL US WHY IMPORTANT for country and why for SIENNA</p>	<p>For China: The document has made clear norms for the research of human gene therapy and the guidelines for the quality control of preparations, and the relevant intellectual property rights and the ethical issues needing attention are made clear, which is of great significance to the development of Chinese gene therapy research and preparation quality control technology.</p> <p>For SIENNA: The technical specifications and the ethical proposal put forward in the document have a good reference value for SIENNA.</p>
<p>Is the document useful for the development of the SIENNA codes and other ethical frameworks? If yes, please explain.</p>	<p>Yes.The technical rules and regulations in the document, the countermeasures for moral regulation of patent disputes and the proposed ethical schemes have a certain universal adaptability, so it has good reference value for SIENNA.</p>

Document 2-Management Specification for Gene Chip Diagnostic Technology (Trial Implementation)

Document found via (national associations or google or another database)	National association (Ministry of Health, PRC)
Title of document	Management Specification for Gene Chip Diagnostic Technology (Trial Implementation)
Scope/ main topic	(choose from list above, 5Bi-5Bvi, write out the topic) Genomic testing, Gene therapy
Kind of document (PEC or NAEG or other)	PEC
Document developed by whom (organisation, profession)?	Ministry of Health, PRC
Year the document was published	2009
Document saved in folder as	Management Specification for Gene Chip Diagnostic Technology (Trial Implementation).doc
Who is the stated audience, if none specified, write not stated (NS)	Gene chip technology related health technicians and medical institutions
For Clinical or research or both or not specified	For clinical

<p>What level of guideline is provided?</p>	<p>Very broad: i.e.: on many aspects of genomics or genetics, including many issues all together e. g. <input checked="" type="checkbox"/>Very specific: only focuses on 1- 2 issues <input checked="" type="checkbox"/>More Theoretical (values, principles) More practical (a practitioner could apply them)</p>	
<p>What is the scope of the document, and what are the main recommendations?</p>	<p>The scope of the document: -Genomic testing -Gene therapy -Management criterion and ethical issues related to gene chip technology</p>	<p>The main recommendations: This specification is the minimum requirement for medical institutions and their doctors and medical technicians to carry out gene chip diagnostic technology. It emphasizes that all gene chip diagnostic technologies need to be approved and supervised by the clinical application ethics committee. In addition, before using gene chip diagnosis, the patient should be informed of the purpose, technical reliability, reference value, objective evaluation and attention of the gene chip diagnosis, as well as the potential economic and psychological burden.</p>
<p>Which life stage is addressed in the document?</p>	<p><input checked="" type="checkbox"/> Adults Minors (excluding newborns) Newborns Prenatal</p>	

<p>Which ethical challenges are addressed in the document? (you can see lists above for ideas of types)</p>	<p>This document focuses on ethical challenges brought about by gene therapy and gene sequencing. The quality control system of gene chip diagnostic technology, the corresponding standard operating procedures and related ethical issues.</p>
<p>How are the ethical issues addressed? (practically, theoretically etc? Any comment on this is fine)</p> <p>Are solutions offered? If so, which ones? (depending on the document, this may be redundant with above)</p>	<p>The ethical issues are addressed in both practically and theoretically: The document clearly stipulates the three aspects of the diagnosis of gene chip involving medical institutions, practitioners and technical regulations. It also gives a high degree of concern and response to the ethical problems that may arise. Obviously, this is effective and therefore worthy of reference.</p> <p>Solutions involving ethical issues: First, all the gene chip diagnostic technologies need to be approved and supervised by the clinical application ethics committee of the hospital gene chip diagnosis technology. In addition, before using gene chip diagnosis, the patient should be informed of the purpose, technical reliability, reference value, objective evaluation and attention of the gene chip diagnosis, as well as the potential economic and psychological burden.</p>
<p>How is the document structured? b. g. Which format is used in the document (checklist, continuous text, other)?</p>	<p>Chapters & sections continuous text</p>
<p>Is the document clearly understandable?</p>	<p>Yes, it is clearly understandable.</p>

<p>Why is the document important/useful for your country? why did you choose this doc? TELL US WHY IMPORTANT for country and why for SIENNA</p>	<p>For China: The document standardizes the clinical application of gene chip diagnosis technology, which provides the legal basis for ensuring medical quality and medical safety, and pays attention to the potential ethical problems.</p> <p>For SIENNA: The technical management specifications and the ethical proposal put forward in the document have a good reference value for SIENNA.</p>
<p>Is the document useful for the development of the SIENNA codes and other ethical frameworks? If yes, please explain.</p>	<p>Yes. Some of the technical management guidelines and ethical proposals put forward in the document are of considerable reference value to other medical technology fields.</p>

Document 3-Construction and Management Method of Biological Gene Information Database

<p>Document found via (national associations or google or another database)</p>	<p>National association (Shenzhen Municipal Administration of Market Supervision)</p>
<p>Title of document</p>	<p>Construction and Management Method of Biological Gene Information Database</p>
<p>Scope/ main topic</p>	<p>(choose from list above, 5Bi-5Bvi, write out the topic) Biobanks, Databases, Genomic screening (Only a bit of gene sequencing is involved)</p>
<p>Kind of document (PEC or NAEG or other)</p>	<p>PEC</p>

Document developed by whom (organisation, profession)?	Shenzhen Municipal Administration of Market Supervision	
Year the document was published	2014	
Document saved in folder as	Construction and Management Method of Biological Gene Information Database. PDF	
Who is the stated audience, if none specified, write not stated (NS)	People and organizations related to biologic information database in Shenzhen	
For Clinical or research or both or not specified	Not specified	
What level of guideline is provided?	<p>Very broad: i.e.: on many aspects of genomics or genetics, including many issues all together e. g.</p> <p><input checked="" type="checkbox"/>Very specific: only focuses on 1- 2 issues</p> <p>More Theoretical (values, principles)</p> <p><input checked="" type="checkbox"/>More practical (a practitioner could apply them)</p>	
What is the scope of the document, and what are the main recommendations?	<p>The scope of the document:</p> <ul style="list-style-type: none"> -Biological gene information database -Genome sequencing 	<p>The main recommendations:</p> <p>In the construction of biological gene information database, ethical review committee and scientific review committee should be set up to better guide the standardization of genetic information database.</p>

Which life stage is addressed in the document?	<input checked="" type="checkbox"/> Adults Minors (excluding newborns) Newborns Prenatal
Which ethical challenges are addressed in the document? (you can see lists above for ideas of types)	This document focuses on ethical challenges brought about by biological databases and gene sequencing. With the completion of the sequencing of the human genome, the gene sequencing technology has developed rapidly, especially the emergence of the second generation high-throughput sequencing technology, producing a large number of biological genetic information data. However, at present, there is no national standard, industry standard and local standard in China, which leads to lack of basis for collecting and sorting genetic data resources. It is difficult to ensure the accuracy, integrity and safety of the data in the construction of biological gene information database (the privacy rights involved in data).

<p>How are the ethical issues addressed? (practically, theoretically etc? Any comment on this is fine</p> <p>Are solutions offered? If so, which ones? (depending on the document, this may be redundant with above)</p>	<p>The ethical issues are addressed both practically and theoretically. This regulation has better solved the technical standard and ethical problems encountered in the construction of biological genetic information database. In view of the ethical issues designed for the construction of genetic information database, the standard clearly points out that "the ethical review committee should be set up in the construction of biological genetic information database, the ethical standards for establishing the biological genetic information database, the use and sharing of biological gene information data, and the use and sharing of biological gene information data." Ethical censorship of ethical issues (Clause 6.1.1) . "</p>
<p>How is the document structured?</p> <p>c. g. Which format is used in the document (checklist, continuous text, other)?</p>	<p>Chapters, sections, glossary, and normative documents.</p> <p>continuous text</p>
<p>Is the document clearly understandable?</p>	<p>Yes. In addition to the difficult part of professional terms, the interpretation of ethical principles is very clear to understand.</p>

Why is the document important/useful for your country? why did you choose this doc?
TELL US WHY IMPORTANT for country and why for SIENNA

For China:

There is no doubt that genetic information database is very important. At present, there are no national standards, industry standards and local standards in China. This document ensures that the collection and sorting of genetic data resources can be evidence-based (mainly aimed in Shenzhen), and the accuracy, integrity and safety of the data are guaranteed very well in the construction of the biological gene information database.

Therefore, standardizing the construction of biological gene information database by standardized means (including ethical standards) can better guide the standardization of genetic information database, promote the sharing and utilization of genetic data resources, and promote the rapid and healthy development of the biological industry in China.

For SIENNA:

This standard provides an ethical review of the ethical issues involving the sources of biological genetic information data, the use and sharing of biological genetic information data, and the membership of the ethical review committee should involve multidisciplinary members, who have the ability to review and evaluate all the ethical questions. This is of good reference for SIENNA. At the same time, it also embodies the Chinese characteristics.

<p>Is the document useful for the development of the SIENNA codes and other ethical frameworks? If yes, please explain.</p>	<p>This standard stipulates the basic requirements of the equipment and environment related to the construction of biological genetic information database, the processing methods and ethical principles of biological information data. These specifications have a certain prospective value, and the ethical principles are of considerable universal value. It has a good reference value for the SIENNA project.</p>
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Document 4-Management Method of Clinical Application of Medical Technology

<p>Document found via (national associations or google or another database)</p>	<p>National association (Ministry of Health, PRC)</p>
<p>Title of document</p>	<p>Management Method of Clinical Application of Medical Technology</p>
<p>Scope/ main topic</p>	<p>(choose from list above, 5Bi-5Bvi, write out the topic) Gene therapy</p>
<p>Kind of document (PEC or NAEG or other)</p>	<p>PEC</p>
<p>Document developed by whom (organisation, profession)?</p>	<p>Ministry of Health, PRC</p>
<p>Year the document was published</p>	<p>2009</p>
<p>Document saved in folder as</p>	<p>Webpage-http://www.nhfpc.gov.cn/mohbgt/s9507/200903/39511.shtml</p>

Who is the stated audience, if none specified, write not stated (NS)	Public, medical institutions, policymakers	
For Clinical or research or both or not specified	For Clinical	
What level of guideline is provided?	<p>Very broad: i.e.: on many aspects of genomics or genetics, including many issues all together e. g.</p> <p><input checked="" type="checkbox"/> Very specific: only focuses on 1- 2 issues</p> <p><input checked="" type="checkbox"/> More Theoretical (values, principles)</p> <p>More practical (a practitioner could apply them)</p>	
What is the scope of the document, and what are the main recommendations?	<p>The scope of the document:</p> <p>Gene therapy</p>	<p>The main recommendations:</p> <p>The document clearly points out that medical technology refers to one of the following situations: medical technology that needs strict control and management by the health administration department:</p> <p>(1) Involving major ethical issues;</p> <p>(2) High risk;</p> <p>(3) Further validation for the safety and effectiveness by standardized clinical trials is still needed;</p> <p>(4) The need to use scarce resources;</p> <p>(5) Other medical technologies that require special management under the Ministry of Health.</p>

<p>Which life stage is addressed in the document?</p>	<p><input checked="" type="checkbox"/> Adults Minors (excluding newborns) Newborns Prenatal</p>
<p>Which ethical challenges are addressed in the document? (you can see lists above for ideas of types)</p>	<p>This document focuses on the ethical challenges of medical technology, including gene therapy technology. It mainly includes the second and third kinds of medical technology, that is, the ethical risks involved in a certain ethical problem or a higher risk of medical technology.</p>
<p>How are the ethical issues addressed? (practically, theoretically etc? Any comment on this is fine)</p> <p>Are solutions offered? If so, which ones? (depending on the document, this may be redundant with above)</p>	<p>The ethical issues are addressed both practically and theoretically, which has responded well to the ethical problems brought about by medical technology (including genetic technology).</p> <p>Related solutions are offered as follows:</p> <ul style="list-style-type: none"> - the clinical application of medical technology should follow the principles of science, safety, standardization, efficiency, economy and ethics. - for the second and third types of medical technology stipulated in the document, the health administrative departments must be strictly controlled and managed. - The second type of medical technology and the third type of medical technology must be implemented before the application of the third party technology audit system.
<p>How is the document structured?</p> <p>d. g. Which format is used in the document (checklist, continuous text, other)?</p>	<p>Chapters, sections, glossary, appendix and annex</p> <p>continuous text</p>

Is the document clearly understandable?	Yes.The language and the structure of the document are clear and logical.
Why is the document important/useful for your country? why did you choose this doc? TELL US WHY IMPORTANT for country and why for SIENNA	<p>For China: This document has played an important role in strengthening the management of clinical application of medical technology in China, establishing medical technology access and management system, promoting the development of medical science and medical technology, improving medical quality and ensuring medical safety.</p> <p>For SIENNA: The document stipulates that the clinical application of medical technology (which is equally effective for gene therapy) should follow scientific, safe, normative, effective, economic, and ethical principles. In addition, according to the different safety and ethical risks of different medical technologies, this regulation establishes the admittance and management system of clinical application of medical technology, and classification management of medical technology. So these practices have a good reference value for SIENNA projects. At the same time, it also embodies the Chinese characteristics.</p>

<p>Is the document useful for the development of the SIENNA codes and other ethical frameworks? If yes, please explain.</p>	<p>Yes. A number of technical classification management principles and the clinical application of medical technology established by the document should follow scientific, safe, normative, effective, economical and ethical principles. It also has considerable reference value for the development and formulation of other ethical codes in the field of medical technology.</p>
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Document 5-Ethical Principles of Human Assisted Reproductive Technology and Human Sperm Bank

<p>Document found via (national associations or google or another database)</p>	<p>National association (Ministry of Health, PRC)</p>
<p>Title of document</p>	<p>Ethical Principles of Human Assisted Reproductive Technology and Human Sperm Bank</p>
<p>Scope/ main topic</p>	<p>(choose from list above, 5Bi-5Bvi, write out the topic) Gene therapy(The document does not directly refer to gene technology, but it also has a restriction on genetic technology.)</p>
<p>Kind of document (PEC or NAEG or other)</p>	<p>PEC</p>
<p>Document developed by whom (organisation, profession)?</p>	<p>Ministry of Health, PRC</p>
<p>Year the document was published</p>	<p>2003</p>

Document saved in folder as	Ethical Principles of Human Assisted Reproductive Technology and Human Sperm Bank.doc	
Who is the stated audience, if none specified, write not stated (NS)	Public, medical institutions, policymakers	
For Clinical or research or both or not specified	For clinical	
What level of guideline is provided?	<p>Very broad: i. e. : on many aspects of genomics or genetics, including many issues all together e. g.</p> <input checked="" type="checkbox"/> Very specific: only focuses on 1- 2 issues <input checked="" type="checkbox"/> More Theoretical (values, principles) More practical (a practitioner could apply them)	
What is the scope of the document, and what are the main recommendations?	<p>The scope of the document:</p> <ul style="list-style-type: none"> -Gene therapy -Genomic testing&Genetic testing 	<p>The main recommendations:</p> <p>This document clearly defines the ethical principles that human assisted reproductive technology and human sperm banks must follow.</p>
Which life stage is addressed in the document?	<input checked="" type="checkbox"/> Adults Minors (excluding newborns) Newborns Prenatal	

Which ethical challenges are addressed in the document? (you can see lists above for ideas of types)

The document focuses on the ethical challenges of human assisted reproductive technology and the ethical principles of human sperm banks (including the application of gene therapy), including human assisted reproductive technology and the establishment of human sperm banks involved in the health and benefits of individuals, families, and later generations, as well as many of social impacts and ethical risks.

<p>How are the ethical issues addressed? (practically, theoretically etc? Any comment on this is fine</p> <p>Are solutions offered? If so, which ones? (depending on the document, this may be redundant with above)</p>	<p>The ethical issues are addressed both practically and theoretically. A number of ethical principles have been established in the document, which respond well to the ethical issues brought about by human assisted reproductive technology and human sperm bank (including gene technology).</p> <p>Related solutions are offered in the document as follows:</p> <p>The ethical principles of human assisted reproductive technology include:</p> <ul style="list-style-type: none"> - the principle of being beneficial to the patient; - the principle of informed consent; - the principle of protecting descendants; - the principle of social public welfare; - the principle of confidentiality; - the principle of anti-commercialization; -the principle of ethical supervision. <p>The ethical principles of human sperm bank include:</p> <ul style="list-style-type: none"> - the principle of being beneficial to the donor; - the principle of informed consent; - the principle of protecting descendants; - the principle of confidentiality; - the principle of anti-commercialization; - the principle of ethical supervision.
<p>How is the document structured? e. g. Which format is used in the document (checklist, continuous text, other)?</p>	<p>Chapters& sections</p> <p>continuous text</p>
<p>Is the document clearly understandable?</p>	<p>Yes.The language and the structure of the document are clear and logical.</p>

Why is the document important/useful for your country? why did you choose this doc?
TELL US WHY IMPORTANT for country and why for SIENNA

For China:

This regulation plays an important role in promoting the human sperm bank safely, effectively and rationally collecting, preserving and providing sperm, protecting the health and rights of the individuals, families and descendants of the sperm donors and recipients, and carrying out the human assisted reproductive technology in a safe, effective and reasonable way and maintaining social public welfare.

For SIENNA:

This document makes no specific reference to HG, but the document clearly identifies a number of ethical codes in two aspects that are closely relevant to genetic technology. Furthermore, these ethical norms are crucial to SIENNA project ,so it is necessary to list it.

Is the document useful for the development of the SIENNA codes and other ethical frameworks? If yes, please explain.

Yes.The document presents the ethical principles of human assisted reproductive technology and human sperm bank.For example, the principles of informed consent, protecting the offsprings, social public welfare, confidentiality, anti-commercialization, and ethical supervision, etc.These are also of great reference value for the development and formulation of ethical rules in other fields.

Table 5C GDREC DOCUMENTS for HUMAN GENOMICS

Document 1-Ethical Review of Biomedical Research Involving Human Beings

Document found via (national associations or google or another database)	National association (National Health and Family Planning Commission)
Title of document	Ethical Review of Biomedical Research Involving Human Beings
Scope/ main topic	(choose from list above, 5Bi-5Bvi, write out the topic Gene therapy (Thorough, but no direct guidelines for HG)
Kind of document (PEC or NAEG or other)	NAEG
Document developed by whom (organisation, profession)?	National Health and Family Planning Commission
Year the document was published	2016
Document saved in folder as	Webpage- http://www.nhfpc.gov.cn/fzs/s3576/201610/84b33b81d8e747eaaaf048f68b174f829.shtml
Who is the stated audience, if none specified, write not stated (NS)	Public, Researchers and Information professionals
For Clinical or research or both or not specified	For both clinical and research

<p>What level of guideline is provided?</p>	<p>Very broad: i.e.: on many aspects of genomics or genetics, including many issues all together e. g.</p> <p><input checked="" type="checkbox"/>Very specific: only focuses on 1- 2 issues</p> <p><input checked="" type="checkbox"/>More Theoretical (values, principles)</p> <p>More practical (a practitioner could apply them)</p>	
<p>What is the scope of the document, and what are the main recommendations?</p>	<p>The scope of the document:</p> <p>-Gene therapy</p> <p>-Genomic testing&Genetic testing</p> <p>(The two documents are indirectly involved in HG)</p>	<p>The main recommendations:</p> <p>The document specifies that biomedical research involving human beings should conform to the following ethical principles:</p> <ul style="list-style-type: none"> - the principle of informed consent; - the principle of controlling risk; - the principle of being free of charge and compensation; - the principle of protecting privacy; - the principle of compensation in accordance with the law; - the principle of special protection.
<p>Which life stage is addressed in the document?</p>	<p><input checked="" type="checkbox"/> Adults</p> <p>Minors (excluding newborns)</p> <p>Newborns</p> <p>Prenatal</p>	

<p>Which ethical challenges are addressed in the document? (you can see lists above for ideas of types)</p>	<p>The document focuses on the ethical challenges of biomedical research involving human beings (including the application of gene therapy indirectly). The biomedical research involving in human beings has brought many negative effects and ethical risks for protecting the human life and health, maintaining human dignity, respecting and protecting the legitimate rights and interests of the subjects.</p>
<p>How are the ethical issues addressed? (practically, theoretically etc? Any comment on this is fine)</p> <p>Are solutions offered? If so, which ones? (depending on the document, this may be redundant with above)</p>	<p>These ethical problems are more theoretically solved, and the ethical principles put forward are enlightening and insightful.</p> <p>Related solutions offered in the document as follows: This document not only puts forward ethical principles that must be followed in human biomedical research activities. (as mentioned above) Moreover, the basic criteria for the approval of the ethics committee are:</p> <ul style="list-style-type: none"> - adherence to the social value of bioethics; - scientific research protocol; - fair enrollment of the subjects; - favorable risk-benefit ratio; - valid consent; - respect for the subjects; - compliance with the integrity of scientific research.
<p>How is the document structured? e. g. Which format is used in the document (checklist, continuous text, other)?</p>	<p>Chapters& sections</p> <p>continuous text</p>

Is the document clearly understandable?	Yes. The language and the structure of the document are clear and logical.
Why is the document important/useful for your country? why did you choose this doc? TELL US WHY IMPORTANT for country and why for SIENNA	<p>For China: This document provides a basis for protecting human life and health, safeguarding human dignity, respecting and protecting the legitimate rights and interests of the subjects, and regulating the ethical review of biomedical research involving human subjects.</p> <p>For SIENNA: This approach puts forward the ethical principles that must be followed by the biomedical research institutes and the basic standards approved by the ethics committee for the research project. These rules have good reference value for SIENNA project and also reflect Chinese characteristics.</p>
Is the document useful for the development of the SIENNA codes and other ethical frameworks? If yes, please explain.	The document proposes ethical principles that must be followed by human biomedical research institute and the basic standards approved by the ethics committee for research projects, such as the principles of informed consent, risk control, free charging and compensation, privacy protection, special protection, and so on. It has good universal implication . The ethical rules in other fields also have considerable reference value.

Document 2-Ethical Guiding Principles for Human Embryonic Stem Cell Research

Document found via (national associations or google or another database)	National association(Ministry of Science and Technology, PRC; Ministry of Health, PRC)
Title of document	Ethical Guiding Principles for Human Embryonic Stem Cell Research
Scope/ main topic	(choose from list above, 5Bi-5Bvi, write out the topic)
Kind of document (PEC or NAEG or other)	GDREC
Document developed by whom (organisation, profession)?	Ministry of Science and Technology of the People's Republic of China Ministry of Health, PRC
Year the document was published	2003
Document saved in folder as	Webpage- http://www. most. gov. cn/fggw/zfwj/zfwj2003/200512/t20051214_54948. htm
Who is the stated audience, if none specified, write not stated (NS)	All provinces, autonomous regions, municipalities directly under the central government and all relevant departments of the State Council, etc. , all relevant institutions
For Clinical or research or both or not specified	For research

<p>What level of guideline is provided?</p>	<p>Very broad: i. e. : on many aspects of genomics or genetics, including many issues all together e. g.</p> <p><input checked="" type="checkbox"/>Very specific: only focuses on 1- 2 issues</p> <p><input checked="" type="checkbox"/>More Theoretical (values, principles)</p> <p>More practical (a practitioner could apply them)</p>	
<p>What is the scope of the document, and what are the main recommendations?</p>	<p>The scope of the document:</p> <p>-Gene therapy -Genomic testing&Genetic testing</p>	<p>The main recommendations:</p> <p>The document clearly sets out the codes of conduct for human embryonic stem cell research, includingbut not limited to, any research on the prohibition of reproductive cloning of human beings.</p>
<p>Which life stage is addressed in the document?</p>	<p>Adults Minors (excluding newborns) Newborns <input checked="" type="checkbox"/>Prenatal</p>	

Which ethical challenges are addressed in the document? (you can see lists above for ideas of types)

This document is mainly aimed at ethical challenges brought about by human embryonic stem cell research (indirectly including gene therapy technology). It involves human biomedical research in protecting human life and health, maintaining human dignity, respecting and protecting the legitimate rights and interests of human beings, ensuring that the internationally recognized ethical standards for life and the relevant regulations in China are respected and observed.

<p>How are the ethical issues addressed? (practically, theoretically etc? Any comment on this is fine</p> <p>Are solutions offered? If so, which ones? (depending on the document, this may be redundant with above)</p>	<p>These ethical problems are more theoretically solved, and the ethical principles put forward are enlightening and insightful.</p> <p>Related solutions offered in the document are as follows:</p> <p>This document clearly stipulates that human embryonic stem cell research must comply with the following norms:</p> <ul style="list-style-type: none"> -When a blastula is obtained by in vitro fertilization, somatic cell nuclear transfer technique, monosexual reproduction technique or genetic modification, the culture period in vitro cannot be more than 14 days since fertilization or nucleus transfer. -The implantation of the human blastula which has been used for research into human or other animal's reproductive system is prohibited. - The hybrid between human germ cells and germ cells of other species is prohibited. <p>Buying and selling human gamete, fertilized egg, embryo and fetal tissue are prohibited. The principle of informed consent and informed choice, the signing of informed consent form and protection of subject's privacy must be adhered in the conduct of human embryonic stem cell research.</p>
<p>How is the document structured?</p> <p>e. g. Which format is used in the document (checklist, continuous text, other)?</p>	<p>Chapters& sections</p> <p>continuous text</p>
<p>Is the document clearly understandable?</p>	<p>Yes, the document is clearly understandable, owing to the language of the document is succinct, and its structure is clear and logical.</p>

<p>Why is the document important/useful for your country? why did you choose this doc? TELL US WHY IMPORTANT for country and why for SIENNA</p>	<p>For China: This document provides an important basis for ensuring the research activities of human embryonic stem cells in the field of biomedicine, respecting the relevant regulations of our country, respecting the internationally recognized standards of bioethics, and promoting the healthy development of human embryonic stem cell research.</p> <p>For SIENNA: The document puts forward the ethical principles that must be followed in the research activities of human embryonic stem cells in biomedical field, including behavior norms that must be observed in human embryonic stem cell research, which have good reference value for SIENNA project, and also reflect Chinese characteristics.</p>
<p>Is the document useful for the development of the SIENNA codes and other ethical frameworks? If yes, please explain.</p>	<p>The document provides ethical guidelines for the research activities of human embryonic stem cells in biomedical field. For example, "the research on human embryonic stem cells must conscientiously implement the principles of informed consent and informed choice, sign informed consent form and protect the privacy of the subjects (Article 8)", "Research on the reproductive cloning of human beings (fourth articles)" is prohibited. They not only have good universal implications, but also reflect the ethical concerns with Chinese characteristics. They also have considerable reference value for developing and formulating ethical rules in other fields.</p>

7.3 France

TABLE 1: INDIVIDUAL AND COUNTRY INFORMATION

Names and emails of persons who did the work	Anaïs Rességuier anais.resseguier@sciencespo.fr
Your organisation	Sciences Po Paris
Your country (again)	France
Search conducted in which language	French
Acknowledgements (any researcher who helped you to complete this task)	Robert Gianni and Bernard Reber

TABLE 2: LIST OF ALL RELEVANT PROFESSIONAL ETHICS CODES

Ethical issues addressed in which SIENNA area (HG, HE, AI&R)?	Title of document (original + English translation)	URL	Year	Author/organisation	Stated audience	comments
HG	<p>“Charte Ethique BIOBANQUES”</p> <p><i>“Ethics Charter of BIOBANQUES Infrastructures”</i></p>	<p>http://www.biobanques.eu/fr/offre-de-services/outils-et-documentation</p>	2014	Biobanques	Biobanques staff	
Medical research in general	<p>“Charte d’éthique de l’Institut Pasteur”</p> <p><i>“Pasteur Institute Ethics Charter”</i></p>	<p>https://www.pasteur.fr/fr/institut-pasteur/engagements</p>	2012	Pasteur Institute	Researchers at Pasteur Institute	

Research general	in	<p>“Charte Nationale de Déontologie”</p> <p><i>“French National Charter for Research Integrity”</i></p>	<p>http://www.cnrs.fr/comets/spip.php?article183</p>	2015	A consortium of research institutes	Researchers	
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TABLE 3: LIST OF ALL RELEVANT DOCUMENTS FROM NATIONAL ADVISORY/ETHICS GROUPS

SIENNA area	Title of document (original + English translation)	URL	Year	Author/organization	Stated audience	comments
Research in general	<p>“Pratiquer une recherche intègre et responsable. Un guide”</p> <p><i>“Conducting honest and responsible research. A guide”</i></p>	<p>http://www.cnrs.fr/comets/IMG/pdf/pratiquer_une_recherche_integre_et_responsable_un_guide_05.12.2016.pdf</p>	2016	National Centre for Scientific Research (CNRS) and the Conference of University Presidents (CPU)	Researchers in general	

Research in general	<p>“Promouvoir une recherche intègre et responsable. Un guide”</p> <p><i>“Promoting honest and responsible research. A guide”</i></p>	<p>http://www.cnrs.fr/comets/IMG/pdf/guide_promouvoir_une_recherche_integre_et_responsable_8septembre2014.pdf</p>	2014	COMETS: Ethics Committee of the National Centre for Scientific Research (CNRS)	Researchers in general	
HG	<p>“Migration, filiation et identification par empreintes génétiques”</p> <p><i>“Migration, filiation, and identification through genetic fingerprint”</i></p>	<p>http://www.ccne-ethique.fr/fr/publications/migration-filiation-et-identification-par-empreintes-genetiques</p>	2007	National Ethics Consultative Committee for Life Sciences and Health (CCNE)	Researchers in life sciences and health, policy makers, and the general public.	

HG	<p>“Questions éthiques posées par la délivrance de l’information génétique néonatale à l’occasion du dépistage de maladies génétiques (exemples de la mucoviscidose et de la drépanocytose)”</p> <p><i>“Ethical issues raised through access to neonatal genetic information at genetic illness screening (examples of the cystic fibrosis and the sickle cell anemia)”</i></p>	<p>http://www.ccne-ethique.fr/fr/publications/questions-ethiques-posees-par-la-delivrance-de-linformation-genetique-neonatale</p>	2007	National Ethics Consultative Committee for Life Sciences and Health (CCNE)	Researchers in life sciences and health, policy makers, and the general public.	
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HG	<p>“Avis sur les problèmes éthiques liés aux diagnostics anténatals : le diagnostic prénatal (DPN) et le diagnostic préimplantatoire (DPI)”</p> <p><i>“Perspective on ethical issues related to antenatal diagnostic: the prenatal diagnostic and the preimplantation diagnostic”</i></p>	<p>http://www.ccne-ethique.fr/fr/publications/avis-sur-les-problemes-ethiques-lies-aux-diagnostics-antenatals-le-diagnostic-prenatal</p>	2009	National Ethics Consultative Committee for Life Sciences and Health (CCNE)	Researchers in life sciences and health, policy makers, and the general public.	
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HG	<p>“Une réflexion éthique sur la recherche sur les cellules d’origine embryonnaire humaine, et la recherche sur l’embryon humain in vitro”</p> <p><i>“Ethical reflection on research on cells taken from human embryo and research on human embryo in vitro”</i></p>	<p>http://www.ccne-ethique.fr/fr/publications/une-reflexion-ethique-sur-la-recherche-sur-les-cellules-dorigine-embryonnaire-humaine</p>	2010	National Ethics Consultative Committee for Life Sciences and Health (CCNE)	Researchers in life sciences and health, policy makers, and the general public.	
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HG	<p>“Utilisation des cellules souches issues du sang de cordon ombilical, du cordon lui-même et du placenta et leur conservation en biobanques. Questionnement éthique.”</p> <p><i>“Use of stem cells drawn from the blood of the umbilical cord, from the umbilical cord itself and from the placenta and their conservation in biobanks. Ethical reflection.”</i></p>	<p>http://www.ccne-ethique.fr/fr/publications/utilisation-des-cellules-souches-issues-du-sang-de-cordon-ombilical</p>	2012	National Ethics Consultative Committee for Life Sciences and Health (CCNE)	Researchers in life sciences and health, policy makers, and the general public.	
HG	<p>“Questions éthiques associées au développement des tests génétiques fœtaux sur sang maternel”</p> <p><i>“Ethical issues related to the development of foetal genetic testing on maternal blood”</i></p>	<p>http://www.ccne-ethique.fr/sites/default/files/publications/avis-120.pdf</p>	2013	National Ethics Consultative Committee for Life Sciences and Health (CCNE)	Researchers in life sciences and health, policy makers, and the general public.	

HG	<p>“Réflexion éthique sur l'évolution des tests génétiques liée au séquençage de l'ADN humain à très haut débit”</p> <p><i>“Ethical reflection on the evolution of genetic tests in relation to very high throughput sequencing of human DNA”</i></p>	<p>http://www.ccne-ethique.fr/sites/default/files/publications/ccne_avis_124.pdf</p>	2016	National Ethics Consultative Committee for Life Sciences and Health (CCNE)	Researchers in life sciences and health, policy makers, and the general public.	
HG	<p>“Saisine concernant les questions liées au développement de la technologie CRISPR (clustered regularly interspaced short palindromic repeat)-Cas9. Note du Comité d'éthique”</p> <p><i>“Seisin concerning questions related to the development of the CRISPR (clustered regularly interspaced short palindromic repeat)-Cas9 technology. Note from the ethics committee.”</i></p>	<p>https://www.inserm.fr/sites/default/files/2017-10/Inserm_Saisine_ComiteEthique_Crispr-Cas9_Fevrier2016.pdf</p>	2016	Ethics committee of the National institute of health and medical research (INSERM)	The director of the National institute of health and researchers in life sciences and health, policy makers, and the general public.	

HG	<p>“Rapport sur la recherche sur les cellules souches”</p> <p><i>“Report on stem cell research”</i></p>	<p>http://www.senat.fr/rap/r09-652/r09-6521.pdf</p>	2010	OPECST: Parliamentary office for the evaluation of scientific and technological choices	The Parliament and researchers in life sciences and health, policy makers, and the general public.	
HG	<p>“Les progrès de la génétique : vers une médecine de précision ? Les enjeux scientifiques, technologiques, sociaux et éthiques de la médecine personnalisée”</p> <p><i>“Progresses in genetics: toward a precision medicine? Scientific, technological, social and ethical stakes of personalised medicine”</i></p>	<p>https://www.senat.fr/notice-rapport/2013/r13-306-notice.html</p>	2014	OPECST: Parliamentary office for the evaluation of scientific and technological choices	The Parliament and researchers in life sciences and health, policy makers, and the general public.	

HG	<p>“Ethique et réglementation des biobanques de recherche”</p> <p><i>“Ethics and regulation of research biobanks”</i></p>	<p>http://www.biobanques.ues.eu/fr/publications/publications-de-biobanques</p>	2015	Biobanques and the National institute of health and medical research	For professionals working in or with biobanks	
HG	<p>“Etat de la recherche sur l’embryon humain et propositions (2ème partie)”</p> <p><i>“State of research on human embryo and propositions” (2nd part)”</i></p>	<p>https://www.inserm.fr/sites/default/files/media/entity_documents/Inserm_Note_ComiteEthique_GroupEmbryon_juin2015.pdf</p>	2015	Ethics committee of the National institute of health and medical research (INSERM)	Researchers in life sciences and health, policy makers, and the general public.	

HG	<p>“France médecine génomique 2015”</p> <p>“France genomic medicine”</p>	<p>https://www.gouvernement.fr/sites/default/files/document/document/2016/06/22.06.2016_remise_du_rapport_dyves_levy_-_france_medecine_génomique_2025.pdf</p>	2016	AVIESAN: National Alliance for Life and Health Sciences	<p>Researchers in life sciences and health, policy makers, and the general public.</p>	
HG	<p>“De la recherche à la thérapie embryonnaire”</p> <p><i>“From research to embryonal therapy”</i></p>	<p>https://www.inserm.fr/recherche-inserm/ethique/comite-ethique-inserm-cei/groupes-reflexion-thematique-comite-ethique</p>	2017	Ethics committee of the National institute of health and medical research (INSERM)	<p>Researchers in life sciences and health, policy makers, and the general public.</p>	

HG	<i>Les données génétiques</i> <i>Genetic data</i>	http://www.ladocumentationfrancaise.fr/ouvrages/9782111451872-les-donnees-genetiques?xtor=AL-2858	2017	CNIL: National Commission Information Technologies and Liberties	Researchers in life sciences and health, policy makers, and the general public.	This has been published as a book. It is not freely available online.
AI&R and HG	"Biométrie, données identifiantes et droits de l'homme" <i>"Biometry, identifying data and human rights"</i>	http://www.ccne-ethique.fr/fr/publications/biometrie-donnees-identifiantes-et-droits-de-lhomme	2007	National Ethics Consultative Committee for Life Sciences and Health (CCNE)	General public	

AIR and HE	<p>“Le corps, nouvel objet connecté. Du quantified self à la M-santé: les nouveaux territoires de la mise en données du monde”</p> <p><i>“The body, new connected object. From the quantified self to mobile health: the new territories of the datafication of the world”</i></p>	<p>https://www.cnil.fr/sites/default/files/typo/document/CNIL_CA_HIERS_IP2_WEB.pdf</p>	2014	CNIL: National Commission Information Technologies and Liberties	Researchers, funding bodies, policy makers and the general public.	
<p><i>Note from Science Po: The five documents below have been produced as part of the national consultation in relation to the revision of the Bioethics Law in 2018. I have included them in this table because, though these documents do not represent the view of a particular NAEG, they have been produced by one, the CCNE, and deal with ethical questions in relation to new technologies that SIENNA examines.</i></p>						
HG, HE and AI&R	<p>“Rapport de synthèse du comité consultatif national d’éthique”</p> <p><i>“Synthesis report from the national consultative committee for life science and health”</i></p>	<p>https://etatsgenerauxdelabioethique.fr/blog/le-rapport-des-etats-generaux-de-la-bioethique-2018-est-en-ligne</p>	2018	CCNE: National Ethics Consultative Committee for Life Sciences and Health	General public, policy makers, health professionals, researchers and lawyers	

HG	<p>“Examens génétiques et médecine génomique”</p> <p><i>“Genetic texts and genomic medicine”</i></p>	https://etatsgenerauxdelabioethique.fr/pages/examens-genetiques-et-medecine-genomique	2018	National Ethics Consultative Committee for Life Sciences and Health (CCNE)	<p>Researchers in life sciences and health, policy makers, and the general public.</p>	
HG	<p>“Cellules souches et recherche sur l’embryon”</p> <p><i>“Stem cells and research on the embryo”</i></p>	https://etatsgenerauxdelabioethique.fr/pages/cellules-souches-et-recherche-sur-l-embryon	2018	National Ethics Consultative Committee for Life Sciences and Health (CCNE)	<p>Researchers in life sciences and health, policy makers, and the general public.</p>	

HG and AI&R	“Données de santé” <i>“Health data”</i>	https://etatsgenerauxdelabioethique.fr/pages/donnees-de-sante	2018	National Ethics Consultative Committee for Life Sciences and Health (CCNE)	Researchers in life sciences and health, policy makers, and the general public.	
HG, HE and A&IR	“Intelligence artificielle et robotique” <i>“Artificial intelligence and robotique”</i>	https://etatsgenerauxdelabioethique.fr/pages/intelligence-artificielle-et-robotisation	2018	National Ethics Consultative Committee for Life Sciences and Health (CCNE)	General public	

TABLE 4: LIST OF ALL RELEVANT GUIDANCE DOCUMENTS ON HOW TO WRITE RESEARCH ETHICS PROTOCOLS

Name of national REC	Title of document (original + English translation)	Ethical issues addressed in which SIENNA area (HG, HE, AI&R)?	URL	Stated audience	comments
ANSM	<p>“Courrier de demande d’autorisation d’une recherche impliquant la personne humaine mentionnée au 1° de l’article L. 1121-1 du code de la santé publique portant sur le médicament”</p> <p><i>“Request for authorising research involving the human person as mentioned in 1° of article L. 1121-1 of the public health code relating to drugs”</i></p>	Concerns medical research in general	https://ansm.sante.fr/Mediatheque/Publications/Formulaires-et-demarches-Essais-cliniques	Researchers	
N/A	<p>Rédaction d’un protocole médicamenteux</p> <p><i>Template for writing a medicinal research protocol</i></p>	Concerns research on drugs	http://urcest.com/essais-cliniques/documents-essaiscliniques	Researchers in health research	

N/A	<p>Rédaction d'un protocole pour un dispositif médicamenteux</p> <p><i>Template for writing a research protocol for a medicinal dispositive</i></p>	<p>Concerns research on drugs</p>	<p>http://urcest.com/essais-cliniques/documents-essaiscliniques</p>	<p>Researchers in health research</p>	
INSERM	<p>Instruction de remplissage du format du protocole</p> <p><i>Guide to complete the protocol template</i></p>	<p>Concerns medical research involving the human person</p>	<p>https://www.inserm.fr/professionnels-recherche/recherche-sur-personnes/soumission-projets-impliquant-personne-humaine</p>	<p>Researchers in health research</p>	

<p>CNIL: National Commission Information Technologies and Liberties</p>	<p>MR-001 "Recherches dans le domaine de la santé avec recueil du consentement" "<i>Research in the health area with consent</i>"</p>	<p>Concerns research</p> <p>medical</p>	<p>https://www.cnil.fr/sites/default/files/atoms/files/mr-001.pdf</p>	<p>Researchers research</p> <p>in health</p>	<p>Concerns research that involve collection and management of private data as part of the research.</p>
<p>CNIL: National Commission Information Technologies and Liberties</p>	<p>"Demande d'autorisation d'un traitement de recherche dans le monde de la santé" "<i>Request for authorising a research treatment in the health sector</i>"</p>	<p>Concerns research</p> <p>medical</p>	<p>https://www.formulaires.modernisation.gouv.fr/gf/cerfa_10769.d</p>	<p>Researchers research</p> <p>in health</p>	<p>Concerns research that involve collection and management of private data as part of the research.</p>

<p>CERNI (Ethics Committees for noninvasive research) at the University of Paris-Saclay</p>	<p>“Formulaire de soumission au CERNI”</p>	<p>Concerns medical research</p>		<p>Researchers in non invasive research at the University of Paris-Saclay</p>	
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CERNI (Ethics Committees for noninvasive research) Grenoble Alpes	"Guide de soumission" "Submission guide"	Concerns research	medical http://www.grenoblecognition.fr/index.php/ethique/ethique-soumettre-un-dossier	Researchers in Grenoble area	Guide to proceed to the evaluation of research protocols in particular in relation to ethical aspects.
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CERNI (Ethics Committees for noninvasive research) of the Federal University of Toulouse	"Formulaire de soumission au CERNI" "CERNI application form"	Concerns medical research	http://www.univ-toulouse.fr/actualites/comite-d-ethique-de-recherche-cer EN version also available.	Researchers in non invasive research at the Federal University of Toulouse.	Guide to proceed to the evaluation of research protocols in particular in relation to ethical aspects.
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Note from Science Po:

My understanding is that the system concerning research ethics committees and the authorisation of research with regards to their ethical character is quite different from what happens in the Anglo-Saxon world. For this reason, I thought that a few words about the French system could be useful here.

To begin with, it appears that there are no research ethics protocols specifically designed to address particular technologies, such as those SIENNA studies. Furthermore, the role of ethics committees in France, such as the CCNE (Consultative committee for health and life sciences) or the ethics committee of the French national center for research (Comets) is primarily to nourish the public debate on ethics and new technologies. They do research, publish and organize events; however, they do not evaluate the ethical conformity of a research protocol.

In France research protocols involving the human person are assessed by the **CPP (Comité de Protection des Personnes: Committees for the protection of persons)**. This Committee is recognised in French law but does not correspond to what is recognised as a Research Ethics Committee internationally. This is

problematic for researchers in France when they want their research to reach international audience. For instance, international journals might require the studies that they publish to have gone through the evaluation by a research ethics committee. This is the reason why Research Ethics Committee as defined by the Council of Europe and registered in the Office for Human Research Protection in the US have been created recently in France. They are called **Institutional Review Board (IRB)** (in English). These committees do not have a juridical status in France but are useful for French research to reach international audiences as they provide proof of ethical validity.

In addition, research protocols might require to go through:

- the **Autorité Nationale de Sécurité du Médicament (ANSM)** (National Authority for Drug Safety) in case the research involves the human person and is invasive, or
- the **Comité National de l'Informatique et des Libertés (CNIL)** - National IT and Liberties Committee: in case the research involves personal data.

In addition, a number of French universities have recently put in place **CERNI ("Comités d'éthique pour les recherches non-interventionnelles": Ethics Committees for non-invasive research)** that advise on research ethics. I have included in the table above a couple of submission guides prepared by these committees.

Finally, authorisation to conduct research on human stem cells or on the human embryos is given by the **Agency of Biomedicine** (<https://www.agence-biomedecine.fr>). As the relevant webpage on the "Etats généraux" notes, as of 31 december 2017, 90 protocols of research had been authorised including 19 on the embryo and 8 had been refused (<https://etatsgenerauxdelabioethique.fr/pages/cellules-souches-et-recherche-sur-l-embryon>).

TABLE 5A: General questions on your document search in Human Genomics

1- Did you find guidance documents specific to human genomics or genetics?	Yes
2a How many documents did you find from each of these sources that specifically address human genetics or genomics 2b Did you find more documents than you had the time to analyse in this task?	PECs: 1 NAEG: 23 GDREC: 0 Total: 24 I have skimmed through all these documents and looked in more depth in the documents analysed in the tables below (5B).
3- a) How many documents specifically ONLY address high throughput genomics? By high throughput genomics, we mean using sequencing of larger parts of the genome, or exome (including large gene panels). 3b Please list the year of publication of each documents here: 3c please specify the topics addressed explicitly, ideally indicate the number of documents that address each, place an X beside the topic and add the number of docs	PECs: 0 NAEG: 3 GDREC: 0 2013, 2016, 2016 Genomics used for diagnostics in the clinic (genomic testing): X3 Genomics used for research: X3 Consent in genomics in the clinic: X2

	<p>Consent in genomics in research: X2</p> <p>Return of results in genomics in the clinic: X2</p> <p>Return of results in genomics in research: X2</p> <p>Reinterpretation and recontact in genomics in the clinic: X1</p> <p>Reinterpretation and recontact in genomics in research: X1</p> <p>Genomics for newborn screening: X2</p> <p>Genomics for prenatal screening or testing: X2</p> <p>Non-invasive prenatal testing/screening: X2</p> <p>Discrimination: X2</p> <p>Eugenics: X2</p> <p>Justice: X2</p> <p>Genetic exceptionalism: X1</p>
<p>4 a How many documents only address genetics (so testing or screening but without a lot of sequencing)?</p> <p>4b List the years of publications of each document here</p> <p>3c please specify the topics addressed explicitly, ideally indicate the number of documents that address each, place an X beside the topic and</p>	<p>PECs</p> <p>NAEG: 3</p> <p>GDREC</p> <p>2007, 2007, 2014</p> <p>Genetics used for diagnostics in the clinic (genetic testing): 2</p> <p>Genetics used for research: 1</p>

<p>add the number of docs</p>	<p>Consent in genetics in the clinic; 2</p> <p>Consent in genetics in research: 1</p> <p>Return of results in genetics in the clinic; 1</p> <p>Return of results in genetics in research: 1</p> <p>Reinterpretation and recontact in genetics in the clinic: 1</p> <p>Reinterpretation and recontact in genetics in research: 1</p> <p>newborn screening; 2</p> <p>prenatal screening or testing; 2</p> <p>Non-invasive prenatal testing/screening: 1</p> <p>Discrimination: 2</p> <p>Eugenics: 1</p> <p>Justice; 1</p> <p>Genetic exceptionalism_____</p>
<p>5. How many documents address both genetics and genomics?</p>	<p>Specify docs or not?</p> <p>5 documents:</p> <ul style="list-style-type: none"> - AVIESAN, 2016, "France Génomique 2015" - CCNE, 2013, "Questions éthiques associées au développement des tests génétiques foetaux sur sang maternel" (<i>"Ethical issues related to the development of foetal genetic testing on maternal blood"</i>) - CCNE, 2016, "Réflexion éthique sur l'évolution des tests génétiques liée au séquençage de l'ADN humain à très haut débit" (<i>"Ethical reflection on the evolution of genetic tests in relation to very high throughput sequencing of human DNA"</i>) - INSERM, 2016, "Saisine concernant les questions liées au développement de la technologie CRISPR (clustered regularly interspaced short palindromic repeat)-Cas9. Note

	<p>du Comité d'éthique" (<i>"Seisin concerning questions related to the development of the CRISPR (clustered regularly interspaced short palindromic repeat)-Cas9 technology. Note from the ethics committee."</i>)</p> <ul style="list-style-type: none"> - OPECST, 2014, "Les progrès de la génétique : vers une médecine de précision ? Les enjeux scientifiques, technologiques, sociaux et éthiques de la médecine personnalisée" (<i>"Progresses in genetics: toward a precision medicine? Scientific, technological, social and ethical stakes of personalised medicine"</i>)
<p>6 a) How many documents specifically address Genome Modification, gene editing, crispr, or gene therapy</p> <p>6b) do they address somatic or germline editing or both?</p>	<p>4</p> <p><i>I'm sorry but I'm not really sure as I struggle with these terms.</i></p>
<p>How many documents specifically address (so in the title of the document)</p>	<p>Genetic or genomic testing: 5</p> <p>Genetic or genomic screening: 2</p> <p>Biobanks or databases: 3</p> <p>Pharmacogenomics or pharmacogenetics patents</p>

Table 5B RELEVANT PEC or NAEG DOCUMENTS IN HUMAN GENOMICS**5Bi: genomic testing (if none on genomics, use genetic testing)****5Bii genomic screening (if none on genomics, use genetic screening)****5Biii biobanks, databases or registries****5Biv pharmacogenomics or pharmacogenetics****5Bv patents****5Bvi gene editing, crispr, or gene therapy**

Document found via (national associations or google or another database)	National associations
Title of document	“Questions éthiques associées au développement des tests génétiques fœtaux sur sang maternel” <i>“Ethical issues related to the development of foetal genetic testing on maternal blood”</i>
Scope/ main topic	5Bi: genomic testing and screening
Kind of document (PEC or NAEG or other)	NAEG
Document developed by whom (organisation, profession)?	National Ethics Consultative Committee for Life Sciences and Health (CCNE)
Year the document was published	2013

Document saved in folder as	HG_CCNE_2013_Questions éthiques associées au développement des tests génétiques foetaux sur sang maternel
Who is the stated audience, if none specified, write not stated (NS)	NS but most likely for researchers in life sciences and health, policy makers, and the general public.
For Clinical or research or both or not specified	Clinical
What level of guideline is provided?	Very broad: i.e.: on many aspects of genomics or genetics, including many issues all together e.g. Very specific: only focuses on 1- 2 issues More Theoretical (values, principles) More practical (a practitioner could apply them)
What is the scope of the document, and what are the main recommendations?	It proposes an ethical reflection on the ethical issues related to foetal genetic testing on maternal blood following recent technological developments in the area. It particularly focuses on trisomy illness and genetical handicaps and illnesses. It starts by presenting the scientific and technological context, then highlight the particular ethical issues and presents a series of propositions/recommendations (for these, see pp. 37-40). a

Which life stage is addressed in the document?	Adults Minors (excluding newborns) Newborns Prenatal
Which ethical challenges are addressed in the document? (you can see lists above for ideas of types)	It addresses ethical challenges in relation to foetal genetic testing on maternal blood.
How are the ethical issues addressed? (practically, theoretically etc? Any comment on this is fine) Are solutions offered? If so, which ones? (depending on the document, this may be redundant with above)	It addresses them both practically and theoretically. As mentioned above, the recommendations made this document can be found pp. 37-40.
How is the document structured? e.g. Which format is used in the document (checklist, continuous text, other)?	The document is 50 pages long of continuous text divided in 4 sections.
Is the document clearly understandable?	Yes it is, though quite technical.

<p>Why is the document important/useful for your country? why did you choose this doc? TELL US WHY IMPORTANT for country and why for SIENNA</p>	<p>It has been written by a highly recognised institution that provides opinion on ethical issues regarding health and life sciences, the CCNE. I chose this document because, as I understand it, it is the first one published by the CCNE that engaged with genomics.</p>
<p>Is the document useful for the development of the SIENNA codes and other ethical frameworks? If yes, please explain.</p>	<p>Yes it is as it provides the view on genomics of the CCNE which is a very important institution in France on ethics related to health issues.</p>

<p>Document found via (national associations or google or another database)</p>	<p>National associations</p>
<p>Title of document</p>	<p>“Réflexion éthique sur l'évolution des tests génétiques liée au séquençage de l'ADN humain à très haut débit” <i>“Ethical reflection on the evolution of genetic tests in relation to very high throughput sequencing of human DNA”</i></p>
<p>Scope/ main topic</p>	<p>5Bi: genomic testing and screening</p>
<p>Kind of document (PEC or NAEG or other)</p>	<p>NAEG</p>

Document developed by whom (organisation, profession)?	National Ethics Consultative Committee for Life Sciences and Health (CCNE) With an interdisciplinary team of researchers
Year the document was published	2016
Document saved in folder as	HG_CCNE_2016_Réflexion éthique sur l'évolution des tests génétiques liée au séquençage de l'ADN humain à très haut débit
Who is the stated audience, if none specified, write not stated (NS)	NS
For Clinical or research or both or not specified	Both
What level of guideline is provided?	Very broad: i.e.: on many aspects of genomics or genetics, including many issues all together e.g. Very specific: only focuses on 1- 2 issues More Theoretical (values, principles) More practical (a practitioner could apply them)

<p>What is the scope of the document, and what are the main recommendations?</p>	<p>It proposes an ethical reflection on the evolution of genetic tests following technological developments on high throughput sequencing of human DNA. The 4th section (out of 5) of the document presents the “main questions” drawn from the description of the state of research presented in the previous sections. These concern:</p> <ul style="list-style-type: none"> - the place of genetics in medical practice - the need for a reflection on the respect and protection of private life - the risk for genetics to overtake prevention in public health.
<p>Which life stage is addressed in the document?</p>	<p>Adults Minors (excluding newborns) Newborns Prenatal</p>
<p>Which ethical challenges are addressed in the document? (you can see lists above for ideas of types)</p>	<p>It addresses ethical challenges due to genomic testing and screening quite broadly.</p>

<p>How are the ethical issues addressed? (practically, theoretically etc? Any comment on this is fine</p> <p>Are solutions offered? If so, which ones? (depending on the document, this may be redundant with above)</p>	<p>It addresses them both practically and theoretically.</p> <p>Primarily the need to engage in a reflection on these issues.</p>
<p>How is the document structured? e.g. Which format is used in the document (checklist, continuous text, other)?</p>	<p>The document is 84 pages divided in 5 sections of continuous text.</p>
<p>Is the document clearly understandable?</p>	<p>Yes it is.</p>
<p>Why is the document important/useful for your country? why did you choose this doc? TELL US WHY IMPORTANT for country and why for SIENNA</p>	<p>It has been written by a highly recognised institution that provides opinion on ethical issues regarding health and life sciences, the CCNE.</p> <p>I chose this document as it provides an up-to-date (as of 2016) overview by this institution on the new developments regarding genomics.</p>
<p>Is the document useful for the development of the SIENNA codes and other ethical frameworks? If yes, please explain.</p>	<p>Yes it is as it provides the view on genomics of the CCNE which is a very important institution in France on ethics related to health issues.</p>

Document found via (national associations or google or another database)	National associations
Title of document	“Ethics Charter of Biobanques Infrastructure”
Scope/ main topic	5Biii biobanks, databases or registries
Kind of document (PEC or NAEG or other)	PEC
Document developed by whom (organisation, profession)?	Biobanques Infrastructure: a “distributed Infrastructure dedicated to research in biology and health areas using biological resources (samples and associated data).” http://www.biobanques.eu/fr/
Year the document was published	2014
Document saved in folder as	HG_Biobanques_2014_Charter Ethics Biobanques
Who is the stated audience, if none specified, write not stated (NS)	NS explicitly but most likely for employees from Biobanques Infrastructure and researchers who use their samples and expertise.
For Clinical or research or both or not specified	Research

What level of guideline is provided?	<p>Very broad: i.e.: on many aspects of genomics or genetics, including many issues all together e.g.</p> <p>Very specific: only focuses on 1- 2 issues</p> <p>More Theoretical (values, principles)</p> <p>More practical (a practitioner could apply them)</p>
What is the scope of the document, and what are the main recommendations?	<p>The document consists in a series (10) of principles that Biobanques Infrastructure respects. These principles primarily concern the use of biological resources made available by the Infrastructure.</p>
Which life stage is addressed in the document?	<p>Adults</p> <p>Minors (excluding newborns)</p> <p>Newborns</p> <p>Prenatal</p> <p><i>Not specified</i></p>
Which ethical challenges are addressed in the document? (you can see lists above for ideas of types)	<p>N/A</p>

<p>How are the ethical issues addressed? (practically, theoretically etc? Any comment on this is fine</p> <p>Are solutions offered? If so, which ones? (depending on the document, this may be redundant with above)</p>	<p>General principles to address potential issues are highlighted.</p>
<p>How is the document structured? e.g. Which format is used in the document (checklist, continuous text, other)?</p>	<p>The document introduces the organisation, the aim of the document and then present the 10 principles with a paragraph for each principle in order to define them.</p>
<p>Is the document clearly understandable?</p>	<p>Yes it is.</p>
<p>Why is the document important/useful for your country? why did you choose this doc? TELL US WHY IMPORTANT for country and why for SIENNA</p>	<p>It is important as Biobanques Infrastructure is an important organisation on the question of use for research of biological samples.</p>
<p>Is the document useful for the development of the SIENNA codes and other ethical frameworks? If yes, please explain.</p>	<p>It might be useful for SIENNA in order to help the project identify key principles in relation to the use of biological samples for research.</p>

Document found via (national associations or google or another database)	National associations
Title of document	“De la recherche à la thérapie embryonnaire” “From research to embryonic therapy”
Scope/ main topic	5Bvi gene editing, crispr, or gene therapy
Kind of document (PEC or NAEG or other)	NAEG
Document developed by whom (organisation, profession)?	By the ethics committee of the National institute of health and medical research (INSERM)
Year the document was published	2017
Document saved in folder as	HG_Inserm_2017_Note_ComiteEthique_GroupeEmbryon
Who is the stated audience, if none specified, write not stated (NS)	NS but most likely for researchers and health professionals.
For Clinical or research or both or not specified	Both

What level of guideline is provided?	<p>Very broad: i.e.: on many aspects of genomics or genetics, including many issues all together e.g.</p> <p>Very specific: only focuses on 1- 2 issues</p> <p>More Theoretical (values, principles)</p> <p>More practical (a practitioner could apply them)</p>
What is the scope of the document, and what are the main recommendations?	<p>It covers embryo research and therapy and particularly focuses on two techniques: mitochondrial transfer and the treatment of triploidy.</p> <p>In terms of ethical issues, it focuses on questions related to safety and to principles.</p> <p>The document concludes by recommending that more research be conducted in France on these techniques in order to ensure that they are conducted with high ethical rigour.</p>
Which life stage is addressed in the document?	<p>Adults</p> <p>Minors (excluding newborns)</p> <p>Newborns</p> <p>Prenatal</p>
Which ethical challenges are addressed in the document? (you can see lists above for ideas of types)	<p>Those related to mitochondrial transfer and the treatment of triploidy.</p>

<p>How are the ethical issues addressed? (practically, theoretically etc? Any comment on this is fine</p> <p>Are solutions offered? If so, which ones? (depending on the document, this may be redundant with above)</p>	Both practically and theoretically.
<p>How is the document structured? e.g. Which format is used in the document (checklist, continuous text, other)?</p>	It is a continuous text 13 pages long.
<p>Is the document clearly understandable?</p>	Yes, though it is quite technical.
<p>Why is the document important/useful for your country? why did you choose this doc? TELL US WHY IMPORTANT for country and why for SIENNA</p>	It is important as it is written by the ethics committee of the main medical research institution in France, the INSERM.
<p>Is the document useful for the development of the SIENNA codes and other ethical frameworks? If yes, please explain.</p>	It might be useful to SIENNA as it focuses specifically on two techniques and proposes the view of the ethics committee of a major French institution on these.

Document found via (national associations or google or another database)	Google
Title of document	France Médecine Génomique 2025
Scope/ main topic	Genomic testing and screening and pharmacogenetics
Kind of document (PEC or NAEG or other)	Document commissioned by the Prime Minister in view of launching a national strategy for the following 10 years in relational to genomic medicine.
Document developed by whom (organisation, profession)?	Document developed by AVIESAN: National Alliance for Life and Health Sciences.
Year the document was published	2016
Document saved in folder as	HG_Aviesan_2016_France_medecine_genomique_2025
Who is the stated audience, if none specified, write not stated (NS)	NS but commissioned by the Prime Minister at the time, Manuel Valls, and can be of interest to policy makers, health professionals and the general public.
For Clinical or research or both or not specified	Both

What level of guideline is provided?	<p>Very broad: i.e.: on many aspects of genomics or genetics, including many issues all together e.g.</p> <p>Very specific: only focuses on 1- 2 issues</p> <p>More Theoretical (values, principles)</p> <p>More practical (a practitioner could apply them)</p>
What is the scope of the document, and what are the main recommendations?	<p>It is a document proposing a 10-year national plan regarding genomic medicine in France. It proposes 14 measures for France to develop this technology.</p>
Which life stage is addressed in the document?	<p>Adults</p> <p>Minors (excluding newborns)</p> <p>Newborns</p> <p>Prenatal</p> <p><i>Not specified.</i></p>
Which ethical challenges are addressed in the document? (you can see lists above for ideas of types)	<p>Measure 8 relates to ethical aspects pertaining to the gathering, the conservation, and the use of clinical and genomic data and to guarantee a good quality and safe care.</p> <p>Annexe 12 is dedicated to questions of ethics and regulations.</p>

<p>How are the ethical issues addressed? (practically, theoretically etc? Any comment on this is fine</p> <p>Are solutions offered? If so, which ones? (depending on the document, this may be redundant with above)</p>	<p>It is a practical and programmatic document.</p>
<p>How is the document structured? e.g. Which format is used in the document (checklist, continuous text, other)?</p>	<p>It is a 170-pages long document of continuous text broken down into 14 measures, to this is added a glossary and a series of annexes.</p>
<p>Is the document clearly understandable?</p>	<p>Yes it is.</p>
<p>Why is the document important/useful for your country? why did you choose this doc? TELL US WHY IMPORTANT for country and why for SIENNA</p>	<p>It is important for Sienna as it presents what is recommended to French policy makers to develop the country's strategy for genomic medicine.</p>
<p>Is the document useful for the development of the SIENNA codes and other ethical frameworks? If yes, please explain.</p>	<p>It is useful more in terms of the general background it gives on the French context.</p>

Document found via (national associations or google or another database)	Google
Title of document	<p>“Les progrès de la génétique : vers une médecine de précision ? Les enjeux scientifiques, technologiques, sociaux et éthiques de la médecine personnalisée”</p> <p><i>“Progresses in genetics: toward a precision medicine? Scientific, technological, social and ethical stakes of personalised medicine”</i></p>
Scope/ main topic	Genomic testing, genetic screening, pharmacogenetics
Kind of document (PEC or NAEG or other)	Report produced by the Parliamentary office for the evaluation of scientific and technological choices and more precisely by two deputies: M. Alain CLAEYS et Jean-Sébastien VIALATTE.
Document developed by whom (organisation, profession)?	This parliamentary office is composed of deputies and senators.
Year the document was published	2014
Document saved in folder as	HG_OPECST_2014_Les progrès de la génétique_Vers une médecine de précision?

Who is the stated audience, if none specified, write not stated (NS)	The two French assemblies: the National Assembly and the Senate, but it might also be of interest to policy makers, researchers, and the general public.
For Clinical or research or both or not specified	Both
What level of guideline is provided?	Very broad: i.e.: on many aspects of genomics or genetics, including many issues all together e.g. Very specific: only focuses on 1- 2 issues More Theoretical (values, principles) More practical (a practitioner could apply them)
What is the scope of the document, and what are the main recommendations?	The document is very broad in its scope and seeks to identify the scientific, technological, social, and ethical stakes of personalised medicine in relation to advances in genetics.
Which life stage is addressed in the document?	Adults Minors (excluding newborns) Newborns Prenatal
Which ethical challenges are addressed in the document? (you can see lists above for ideas of types)	In terms of ethical challenges, it particularly addresses: <ul style="list-style-type: none"> - the impact of genetic testing - protection and management of personal data - fair access to care for all and restricting medico-surveillance

<p>How are the ethical issues addressed? (practically, theoretically etc? Any comment on this is fine</p> <p>Are solutions offered? If so, which ones? (depending on the document, this may be redundant with above)</p>	<p>Ethical issues are addressed quite closely and covered over about 40 pages of the report.</p>
<p>How is the document structured? e.g. Which format is used in the document (checklist, continuous text, other)?</p>	<p>Document structured in 3 main parts: the two identifies the scientific context, the second socio-economical aspects, and the third the ethical issues.</p> <p>It is a continuous text.</p>
<p>Is the document clearly understandable?</p>	<p>Yes it is.</p>
<p>Why is the document important/useful for your country? why did you choose this doc? TELL US WHY IMPORTANT for country and why for SIENNA</p>	<p>It is an important official document and constitutes a reference on these issues. It might be useful to Sienna for this reason.</p>
<p>Is the document useful for the development of the SIENNA codes and other ethical frameworks? If yes, please explain.</p>	<p>It might be helpful to Sienna rather to understand France's context and the current (as of 2014) state of the debate.</p>

7.4 Germany

TABLE 1: INDIVIDUAL AND COUNTRY INFORMATION

Names and emails of persons who did the work (if different from above)	Lisa Tambornino, tambornino@eurecnet.eu Dirk Lanzerath, mailto:lanzerath@eurecnet.org
Your organisation	EUREC
Your country (again)	Germany
Search conducted in which language	German
Acknowledgements (any researcher who helped you to complete this task)	

TABLE 2: LIST OF ALL RELEVANT PROFESSIONAL ETHICS CODES

SIENNA area	Title of document (original + English translation)	URL	Year	organisation	stated audience	comments
HG, HE, AI&R	Research Code of Conduct	https://verwaltung.uni-koeln.de/forschungsmanagement/content/e12474/e160886/018_Research_Code_of_Conduct_neu_Deutschland.pdf		University of Cologne	researchers	Very broad. value based approach

Germany

HG, HE	Berufsordnung Ärztinnen und Ärzte	für	https://www.bundesärztekammer.de/fileadmin/user_upload/downloads/pdf-Ordner/MBO/MBO-AE.pdf (only German)	Version from 2018	Bundesärztekammer German Medical Association	physicians	Very broad
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TABLE 3: LIST OF ALL RELEVANT DOCUMENTS FROM NATIONAL ADVISORY/ETHICS GROUPS AND PROFESSIONAL GROUPS

SIENNA area	Title of document (original + English translation)	URL	Year	organization	Stated audience	comments
HG, HE, AI&R	<p>Wissenschaftsfreiheit und Wissenschaftsverantwortung Empfehlungen zum Umgang mit sicherheitsrelevanter Forschung</p> <p>Scientific Freedom and Scientific Responsibility Recommendations for Handling Security-Relevant Research</p>	http://www.dfg.de/download/pdf/dfg_im_profil/reden_stellungnahmen/2014/dfg-leopoldina_forschungsrisiken_de_en.pdf	2014	<p>DFG</p> <p>German Research Foundation + Leopoldina</p>	Professional organizations and universities, policy makers, researchers, public	Very broad “The recommendations offer assistance in answering ethical questions, thus contributing to defining standards and codes of conduct beyond statutory norms for scientists dealing with security-relevant research.”

HG	<p>Die Zukunft der genetischen Diagnostik – von der Forschung in die klinische Anwendung</p> <p>The future of genetic diagnosis – from research to clinical practice</p>	<p>German version: https://www.ethikrat.org/fileadmin/Publikationen/Stellungnahmen/deutsch/stellungnahme-zukunft-der-genetischen-diagnostik.pdf</p> <p>English version: https://www.ethikrat.org/fileadmin/Publikationen/Stellungnahmen/englisch/opinion-the-future-of-genetic-diagnosis.pdf</p>	2013	<p>Deutscher Ethikrat</p> <p>German Ethics Council</p>	Policy makers, professionals	
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HG	<p>Keimbahneingriffe am menschlichen Embryo: Deutscher Ethikrat fordert globalen politischen Diskurs und internationale Regulierung</p> <p>Germline intervention in the human embryo: German Ethics Council calls for global political debate and international regulation</p>	<p>German version: https://www.ethikrat.org/fileadmin/Publikationen/Ad-hoc-Empfehlungen/deutsch/empfehlung-keimbahneingriff-am-menschlichen-embryo.pdf</p> <p>English version: https://www.ethikrat.org/fileadmin/Publikationen/Ad-hoc-Empfehlungen/englisch/recommendation-germline-intervention-in-the-human-embryo.pdf</p>	2017	Deutscher Ethikrat German Ethics Council	Policy makers, professionals	
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HG	<p>Medizinische, ethische und rechtliche Aspekte von Biobanken</p> <p>Medical, ethical and legal aspects of biobanks</p>	<p>https://www.bundesärztekammer.de/fileadmin/user_upload/downloads/pdf-Ordner/WB/Biobanken.pdf</p>	2017	<p>Bundesärztekammer</p> <p>German Medical Association</p>	<p>Society, researchers, donors</p> <p>physicians, potential</p>	
HG	<p>Richtlinie zur Entnahme und Übertragung von menschlichen Keimzellen im Rahmen der assistierten Reproduktion</p> <p>Guideline about the extraction and transmission of human cells within assisted reproduction</p>	<p>https://www.aerzteblatt.de/down.asp?id=21066</p>	2017	<p>Bundesärztekammer</p> <p>German Medical Association</p>	<p>Society, patients, policy makers</p> <p>physicians,</p>	

HG	<p>Stellungnahme. Eckpunkte für eine Heidelberger Praxis der Ganzgenomsequenzierung</p> <p>Statement. Key points for sequencing the whole genome.</p>	<p>https://www.uni-heidelberg.de/m/d/totalsequenzierung/information/en/mk_eurat_stellungnahme_2013.pdf</p>	2013	<p>Projektgruppe EURAT „Ethische und Rechtliche Aspekte der Totalsequenzierung des menschlichen Genoms“</p>	Physicians, researchers	
HG	<p>Humane Genomsequenzierung – Herausforderungen für eine verantwortungsvolle Anwendung in der Wissenschaft</p> <p>Statement on genome sequencing</p>	<p>http://www.dfg.de/download/pdf/dfg_im_profil/reden_stellungnahmen/2016/160801_stellungnahme_humane_genomsequenzierung.pdf</p>	2016	<p>Deutsche Forschungsgemeinschaft (DFG)</p> <p>German Research Foundation</p>	Policy makers, researchers, physicians	

HG	<p>S2-Leitlinie Humangenetische Diagnostik und genetische Beratung</p> <p>Guidance on genetic diagnosis and genetic counselling</p>	<p>https://www.bvdh.de/download/LL_ST/2011_06_24_S2_LL_Human_genetik.pdf</p>	2011	<p>Deutsche Gesellschaft für Humangenetik e.V. (GfH) German society on human genetics</p>	<p>Researchers, scientists, physicians,</p>	<p>Different modules</p>
HG	<p>Leitlinien zur genetischen Beratung</p> <p>Guidance on genetic counselling</p>	<p>https://www.medgenetik.de/sonderdruck/1996-3-1.PDF</p>	2001	<p>Berufsverband Medizinische Genetik e.V.</p>	<p>professionals</p>	

HG	<p>Chancen und Grenzen des genome editing</p> <p>The opportunities and limits of genome editing</p>	<p>https://www.leopoldina.org/uploads/tx_leopublication/2015_3Akad_Stellungnahme_Genome_Editing.pdf</p>	2015	<p>Leopoldina</p> <p>German National Academy of Sciences</p> <p>DFG German Research Foundation</p> <p>Achatec German national academy of science and engineering</p> <p>UNION German umbrella organization of german academies</p>	<p>Professionals, policy makers</p> <p>society,</p>	
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HG	Präzise Technik? Kritik an Genome Editing Statement on genome editing	http://www.gen-ethisches-netzwerk.de/files/Genome_Editing_Stellungnahme-GeN.pdf	2017	Gen-ethisches Netzwerk	Professionals, policy makers	society,	
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HG	<p>Stellungnahme. Forschungsklonen mit dem Ziel therapeutischer Anwendungen</p> <p>Position paper Therapeutic research cloning</p>	<p>https://www.zentrale-ethikkommission.de/fileadmin/user_upload/downloads/pdf-Ordner/Zeko/Klonen.pdf</p>	2006	<p>Zentrale Ethikkommission bei der Bundesärztekammer (ZEKO)</p> <p>Central Ethics Committee for Observance of Ethical Principles in Medicine and its Adjacent Fields of the German Medical Association (Bundesärztekammer)</p>	Professionals, policy makers, society,	
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HG	<p>Stellungnahme der Zentralen Ethikkommission zur Stammzellforschung</p> <p>Statement on stem cell research</p>	<p>https://www.zentrale-ethikkommission.de/fileadmin/user_upload/downloads/pdf-Ordner/Zeko/Stammzell.pdf</p>	2002	<p>Zentrale Ethikkommission bei der Bundesärztekammer (ZEKO)</p> <p>Central Ethics Committee for Observance of Ethical Principles in Medicine and its Adjacent Fields of the German Medical Association (Bundesärztekammer)</p>	<p>Professionals, policy makers</p> <p>society,</p>	
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HE, HG	<p>Gendoping Wissenschaftliche Grundlagen – Einfallstore – Kontrolle</p> <p>Gene Doping Scientific Basis – Gateways – Monitoring</p>	<p>https://www.tab-beim-bundestag.de/de/pdf/publikationen/buecher/gerli-nger-et-al-2008-124.pdf</p> <p>English version: https://www.bundestag.de/blob/190958/8638bc0aa98521f057e22e542259fe85/gene_doping_data.pdf</p>	2008	<p>Büro für Technikfolgen- abschätzung beim Deutschen Bundestag (TAB)</p> <p>Office of Technology Assessment at the German Bundestag</p>	professionals	
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TABLE 4: LIST OF ALL RELEVANT GUIDANCE DOCUMENTS ON HOW TO WRITE RESEARCH ETHICS PROTOCOLS

Name of national REC	Title of document (original + English translation)	SIENNA area	URL	stated audience	comments
Ethics committee of the German Society for Nursing Science	Fragen zur ethischen Reflexion Guidance for ethical reflexion	all	https://dg-pflegewissenschaft.de/wp-content/uploads/2017/05/FragenEthReflexion.pdf	researchers	Very broad
Arbeitskreis Medizinischer Ethikkommissionen in der Bundesrepublik Deutschland e.V. (AMEK) Working group of medical ethics committees in Germany	Checkliste für die Probandeninformation zur Erlangung der Einwilligung in die wissenschaftliche Verwendung von Blut- bzw. Gewebeprobe Checklist for getting informed consent for studys with blood or tissue samples	HG	https://www.uni-due.de/imperia/md/content/ethikkommission/berufsrecht_checkliste_probandeninformation.pdf	researchers	

	<p>Checkliste: Erforderliche Antragsunterlagen für Studien nach AMG</p> <p>Checklist: Required application documents for studies according to AMG</p>	HE, HG	<p>https://www.uniklinik-freiburg.de/fileadmin/mediapool/10_andere/ethikkommission/sonstiges/checklisteamg.doc</p>	researchers	
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	<p>Mustertext zur Information und Einwilligung in die Verwendung von Biomaterialien und zugehöriger Daten in Biobanken</p> <p>Template For informed consent concerning the donation, storage, and utilization of biological materials as well as collecting, processing, and usage of (related) data in biobanks</p>	HG	<p>https://www.ak-med-ethik-komm.de/docs/MustertextBiobanken.docx</p> <p>https://www.ak-med-ethik-komm.de/docs/Template-for-informed-consent.docx</p>	researchers	
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	<p>Mustertext zur Information und Einwilligung bei einer optionalen zusätzlichen Sammlung von Biomaterialien anlässlich einer klinischen Arzneimittelprüfung zur Nutzung außerhalb des Prüfplans</p> <p>Template for information and consent to an optional additional collection of biomaterials for a clinical proving out of schedule</p>	HG	https://www.ak-med-ethik-komm.de/docs/PharmakogenetikalsZusatzzuAMG.docx		
	<p>Recommendation For the Assessment of Research-related Human Biobanks by Ethics Committees</p>	HG	https://www.ak-med-ethik-komm.de/docs/Recommendations2016_draft2016_09_07.pdf	<p>For Ethics Committee members !!!!</p>	

TABLE 5A: General questions on your document search in Human Genomics

1- Did you find guidance documents specific to human genomics or genetics?	Yes
2a How many documents did you find from each of these sources that specifically address human genetics or genomics 2b Did you find more documents than you had the time to analyse in this task?	PECs: 1 NAEG: 11 GDREC: 0 Total: 12 No
3- a) How many documents specifically ONLY address high throughput genomics? By high throughput genomics, we mean using sequencing of larger parts of the genome, or exome (including large gene panels). 3b Please list the year of publication of each documents here: 3c please specify the topics addressed explicitly, ideally indicate the number of documents that address each, place an X beside the topic and add the number of docs	PECs: 0 NAEG: 0 GDREC: 0 Genomics used for diagnostics in the clinic (genomic testing) Genomics used for research Consent in genomics in the clinic Consent in genomics in research Return of results in genomics in the clinic

	<p>Return of results in genomics in research</p> <p>Reinterpretation and recontact in genomics in the clinic</p> <p>Reinterpretation and recontact in genomics in the clinic</p> <p>Genomics for newborn screening</p> <p>Genomics for prenatal screening or testing</p> <p>Non-invasive prenatal testing/screening</p> <p>Discrimination</p> <p>Eugenics</p> <p>Justice</p> <p>Genetic exceptionalism</p>
<p>4 a How many documents only address genetics (so testing or screening but without a lot of sequencing)?</p> <p>4b List the years of publications of each document here</p> <p>4c please specify the topics addressed explicitly, ideally indicate the number of documents that address each, place an X beside the topic and add the number of docs</p>	<p>PECs: 0</p> <p>NAEG: 9</p> <p>GDREC: 0</p> <p>2013, 2017,2017, 2017, 2011, 2001, 2015, 2008, 2017</p> <p>Genetics used for diagnostics in the clinic (genetic testing) _1__</p> <p>Genetics used for research____</p> <p>Consent in genetics in the clinic____</p> <p>Consent in genetics in research____</p>

	<p>Return of results in genetics in the clinic __3_</p> <p>Return of results in genetics in research __1__</p> <p>Reinterpretation and recontact in genetics in the clinic ____</p> <p>Reinterpretation and recontact in genetics in the clinic ____</p> <p>newborn screening ____</p> <p>prenatal screening or testing __1__</p> <p>Non-invasive prenatal testing/screening ____</p> <p>Discrimination ____</p> <p>Eugenics ____</p> <p>Justice ____</p> <p>Genetic exceptionalism ____</p>
5. How many documents address both genetics and genomics?	3
6 a) How many documents specifically address Genome Modification, gene editing, crispr, or gene therapy	3
6b) do they address somatic or germline editing or both?	both
How many documents specifically address (so in the title of the document)	<p>Genetic or genomic testing: 2</p> <p>Genetic or genomic screening</p> <p>Biobanks or databases: 1</p> <p>Pharmacogenomics or pharmacogenetics</p>

	patents
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Table 5B RELEVANT PEC or NAEG DOCUMENTS IN HUMAN GENOMICS, copy the table for as many documents as you have to describe.

5Bi: genomic testing (if none on genomics, use genetic testing)

5Bii genomic screening (if none on genomics, use genetic screening)

5Biii biobanks, databases or registries

5Biv pharmacogenomics or pharmacogenetics

5Bv patents

5Bvi gene editing, crispr, or gene therapy

Document 1:

Document found via (national associations or google or another database)	Google
Title of document	The future of genetic diagnosis – from research to clinical practice
Scope/ main topic	Genomic testing
Kind of document (PEC or NAEG or other)	NAEG
Document developed by whom (organisation, profession)?	German Ethics Council

Year the document was published	2013
Document saved in folder as	German Ethics Council_The future of genetic diagnosis_NAEG_2013
Who is the stated audience, if none specified, write not stated (NS)	NS
For Clinical or research or both or not specified	Both
What level of guideline is provided?	Very broad: i.e.: on many aspects of genomics or genetics, including many issues all together e.g. Very specific: only focuses on 1- 2 issues More Theoretical (values, principles) More practical (a practitioner could apply them)

What is the scope of the document, and what are the main recommendations?	<p>“The German Ethics Council calls for improvements in the provision of information to the public, in the training of healthcare professionals and in the quality of genetic tests and their funding within the healthcare system. Furthermore, it recommends a number of amendments to the Genetic Diagnosis Act concerning the screening of newborns and the conduct of genetic tests for non-medical purposes. It also calls for improved, EU-wide measures to provide for independent consumer information and for patient and consumer protection.</p> <p>Considering prenatal diagnosis in cases of increased risk of a disorder of genetic origin, the German Ethics Council could not make a unanimous decision. Nonetheless, the council calls for taking account of the particular emotional situation of the pregnant woman and making it easier for parents of children with disabilities to access possibilities of support.”</p>
Which life stage is addressed in the document?	Adults Minors (excluding newborns) Newborns Prenatal

Which ethical challenges are addressed in the document? (you can see lists above for ideas of types)

- possible effects of predictive genetic diagnosis on the understanding of disease and health (Genes as determinants of illness, Interim status between health and disease)
- effects on the autonomy, self-determination and responsibility of users of genetic tests
- the difficulty of appropriately communicating complex information for varying target groups
- cultural differences as challenge in Germany
- discrimination: will the new developments of genetic diagnosis result in the discrimination and stigmatization of people with particular genetic characteristics?
- justice: question of access to genetic tests

How are the ethical issues addressed? (practically, theoretically etc? Any comment on this is fine

Are solutions offered? If so, which ones? (depending on the document, this may be redundant with above)

The German Ethics Council recommends 23 points on genetic diagnosis in general (A1-A23) and 9 points on prenatal diagnosis (B1-B9). For both see page 159-166 in the document:

- improvements in the provision of information to the public
- training of healthcare professionals in relation to the available genetic tests
- in the future medical explanation and counselling should be mandatory even in the case of genetic tests conducted for non-medical purposes, as such tests too may yield medically relevant results.
- mitigate the risks of direct-to-consumer genetic tests and the possible psychological burden of their results
- recommendations on whether and to what extent the healthcare system should cover the cost of diagnostic procedures used in the context of therapy, on technical quality assurance, on the funding of research and on research and health policy

Prenatal diagnosis

- parents who decide to go ahead with the birth of a disabled child should be highly valued in society
- “The majority of the members of the Ethics Council in addition consider that prenatal genetic diagnosis should be permitted only where an increased risk of a disorder of genetic origin exists. In their view it is essential to ensure that no genetic information concerning the unborn child that is unconnected with pathology and no details of possible carrier status that are irrelevant to the health of the child itself are disclosed.”

How is the document structured? e.g. Which format is used in the document (checklist, continuous text, other)?	191 pages, including glossary. around 20 pages recommendations at the end
Is the document clearly understandable?	Yes
Why is the document important/useful for your country? why did you choose this doc?	The German ethics council is a very important institution for the ethical debate in Germany. The Council offers recommendations for law makers and politicians, and furthermore stimulates the public debate on ethical challenges of genetic testing and diagnosis.
Is the document useful for the development of the SIENNA codes and other ethical frameworks? If yes, please explain.	Yes, the document gives a good overview of ethical challenges and possible solutions.

Document 2:

Document found via (national associations or google or another database)	Google
Title of document	The opportunities and limits of genome editing
Scope/ main topic	5Bvi gene editing, crispr
Kind of document (PEC or NAEG or other)	NAEG

Document developed by whom (organisation, profession)?	<ul style="list-style-type: none"> – Leopoldina. German National Academy of Sciences – DFG German Research Foundation – Achatec German national academy of science and engineering – UNION German umbrella organization of german academies
Year the document was published	2015
Document saved in folder as	Leopoldina_DFG_genome editing_HG-NAEG.pdf
Who is the stated audience, if none specified, write not stated (NS)	Society, researchers, policy makers, industry
For Clinical or research or both or not specified	Both
What level of guideline is provided?	<p>Very broad: i.e.: on many aspects of genomics or genetics, including many issues all together e.g.</p> <p>Very specific: only focuses on 1- 2 issues</p> <p>More Theoretical (values, principles)</p> <p>More practical (a practitioner could apply them)</p>
What is the scope of the document, and what are the main recommendations?	scientific potential, advantages and ethical issues of genome editing

Which life stage is addressed in the document?	Adults Minors (excluding newborns) Newborns Prenatal
Which ethical challenges are addressed in the document? (you can see lists above for ideas of types)	Challenges and opprotunities of genome editing Off-target mutations low efficiency and numerous off-target mutations of the CRISPR-Cas9 technique
How are the ethical issues addressed? (practically, theoretically etc? Any comment on this is fine) Are solutions offered? If so, which ones? (depending on the document, this may be redundant with above)	“scientists should remain realistic about the potential applications and not raise unfounded fears or exaggerated hopes”
How is the document structured? e.g. Which format is used in the document (checklist, continuous text, other)?	1. Background to the Statement 2. Principles of genome editing 3. Application of genome editing 4. Consequences and recommendations
Is the document clearly understandable?	Yes

<p>Why is the document important/useful for your country? why did you choose this doc? TELL US WHY IMPORTANT for country and why for SIENNA</p>	
<p>Is the document useful for the development of the SIENNA codes and other ethical frameworks? If yes, please explain.</p>	<p>No, too broad</p>

7.5 Greece

TABLE 1: INDIVIDUAL AND COUNTRY INFORMATION

Names and emails of persons who did the work	Maria Bottis
Your organisation	Ionian University
Your country	Greece
Search conducted in which language	Greek and English
Acknowledgements	Fereniki Panagopoulou

TABLE 2: LIST OF ALL RELEVANT PROFESSIONAL ETHICS CODES and OF ALL RELEVANT DOCUMENTS FROM NATIONAL ADVISORY/ETHICS GROUPS**Note: Only the first three documents are PECs.**

SIENNA area	Title of document	URL	Year	Author/ Organisation	Stated audience	Comments
HG, HE	Κώδικας Ιατρικής Δεοντολογίας (Code of Medical Ethics)	https://www.lawspot.gr/nomikes-plirofories/nomothesia/nomos-3418-2005	2015 (latest version)	Greek Parliament (The Parliament codified medical ethics rules also as a statute)	Physician, Nurses, general public, policy makers	This is a code on medical ethics, which has been ratified also by law. The code makes reference to the possible amendment to the human genome, which is expressly forbidden, except for particular purposes, under art. 34 of the Code, par. 1 and 2. Also, the Code deals with consent to plastic surgery.

HG, HE	Code of Ethics for the Medically Assisted Reproduction Κώδικας Δεοντολογίας Ιατρικώς Υποβοηθούμενης Αναπαραγωγής	http://eaiya.gov.gr/wp-content/uploads/2016/12/KODIKASSITE.pdf	2017	National Committee for the Medically Assisted Reproduction Εθνική Επιτροπή για την Ιατρικά Υποβοηθούμενη Αναπαραγωγή/ The Greek Parliament /Βουλή των Ελλήνων	Physician, Geneticists, researchers, policy makers, general public	This Code covers the ethical rules on medically assisted reproduction. It is also a statute voted by the Greek Parliament.
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HG	Opinion-Latest developments on the amendment of human genome (Γνώμη - τελευταίες εξελίξεις για την τροποποίηση του ανθρώπινου γονιδιώματος)	file:///C:/Users/user/Desktop/OPINION_gene%20editing_Final_GR.pdf at www.bioethics.gr	2016	National Bioethics Commission (Εθνική Επιτροπή Βιοηθικής)	Physicians, Geneticists, General Public, Policy makers, Geneticists	The Opinion contains recommendations on the amendment of the human genome, tied to the latest scientific developments on the subject
HG	Report, Genetic Processing of the Genome (Γενετική επεξεργασία του γονιδιώματος)	file:///C:/Users/user/Desktop/REPORT_Gene_editing_FINAL_GR.pdf www.bioethics.gr	2016	National Bioethics Commission Ελληνική Επιτροπή Βιοηθικής	Physicians, Geneticists, General public and policy-makers	-

HG	Replacement of Mitochondria to avoid mitochondria related diseases Αντικατάσταση μιτοχονδρίων για την αποφυγή μιτοχονδριακών νοσημάτων	www.bioethics.gr	2017	National Bioethics Committee Εθνική Επιτροπή Βιοηθικής	Physicians, Geneticists, General public and policy-makers	An Opinion on the replacement of mitochondria to avoid relevant diseases
HG	Creation of Chimeras and Hybrids Δημιουργία Χιμαιρών και Υβριδικών Οργανισμών	http://www.bioethics.gr/images/pdf/GNOMES/ENHMEROTIKO_KEI_MENO_hybrids_chimeras_FINAL_GR.pdf www.bioethics.gr	2017	National Bioethics Committee Ελληνική Επιτροπή Βιοηθικής	Physicists, Geneticists, General public and policy-makers	An informative text on the possibility of the creation of Chimeras and hybrid organisms, related to the developments in the European Commission

HG	Opinion,Recent development in genome editing Πρόσφατες εξελίξεις στην τροποποίηση του ανθρώπινου γονιδιώματος, εκδοτική επεξεργασία	http://www.bioethics.gr/index.php/el/gnomes/1271-genome-editing	2017	National Bioethics Committee Ελληνική Επιτροπή Βιοηθικής	Physicians, Geneticists, General public and policy-makers	Opinion on genome editing
HG	Report, Recent development in genome editing Πρόσφατες εξελίξεις στην τροποποίηση του ανθρώπινου γονιδιώματος, εκδοτική επεξεργασία	http://www.bioethics.gr/images/pdf/GNOMES/REPORT_Gene_editing_FINAL_GR.pdf	2017	National Bioethics Committee Εθνική Επιτροπή Βιοηθικής	Physicians,Geneticists, General public and policy-makers	Report upon which the relevant Opinion on genome editing was based

HG	Current matters on human reproduction Σύγχρονα Ζητήματα Αναπαραγωγής	http://www.bioethics.gr/index.php/el/gnomes/717-2014-09-01-08-33-10	2017	National Bioethics Commission Εθνική Επιτροπή Βιοηθικής	Physicians, Geneticists, General public and policy-makers	Opinion on current matters of human reproduction: preimplantation, mitochondria replacement and selection of gametes before fertilization
HG	Opinion, Current Matters on 'Choice' in Human Reproduction, Σύγχρονα Ζητήματα 'Επιλογής' στην Αναπαραγωγή	http://www.bioethics.gr/images/pdf/GNOMES/OPINION_ISSUES_I N_FERTILISATION_FIN AL_GR.pdf	2014	National Bioethics Committee/ Εθνική Επιτροπή Βιοηθικής Physicians, Geneticists, General public, policy makers		The Opinion deals with matters of choice in eugenics, on the standards used to select embryos for reproduction, mitochondrial diseases etc. It clarifies the 2007 Opinion on the same matters.

HG	Opinion, Prenatal and preimplantation diagnosis and treatment of the fetus, Γνώμη για την προγεννητική και προεμφυτευτική διάγνωση και τη μεταχείριση του εμβρύου	http://www.bioethics.gr/images/pdf/GNOMES/rec_pd_gr.pdf	2007	National Bioethics Committee/ Εθνική Επιτροπή Βιοηθικής Physicians, Geneticists, General Public, Policy makers		The Opinion discusses the standards for preimplantation and prenatal diagnosis and treatment of the fetus (prenatal diagnosis of genetic diseases and legal termination of pregnancy, choice of sex etc)
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HG	Opinion, The use of genetic data in private insurance, Γνώμη, Η χρήση των γενετικών δεδομένων στην ιδιωτική ασφάλιση	http://www.bioethics.gr/index.php/el/gnomes/85-genetika-dedomena-sthn-idiotikh-asfalish http://www.bioethics.gr/images/pdf/GNOMES/ins_opinion_gr.pdf	2008	National Bioethics Committee/ Εθνική Επιτροπή Βιοηθικής Physicians, Geneticists, Insurance companies, general public, workers organizations		Opinion on the use of genetic data in the private insurance sector
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HG	Report, The use of genetic data in private insurance, Έκθεση, Η χρήση γενετικών δεδομένων στην ιδιωτική ασφάλιση	http://www.bioethics.gr/images/pdf/GNOMES/insur_report_gr.pdf	2008	National Bioethics Committee/ Εθνική Επιτροπή Βιοηθικής Physicians, Geneticists, insurance companies, general public, workers organizations		Report with data on the use of genetic data in the private insurance sector
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HG	Opinion, Patents in biotechnology, Γνώμη, Πατέντες στην βιοτεχνολογία	http://www.bioethics.gr/index.php/el/gnomes/115-patentes-sth-biotexnologia	2003	National Bioethics Committee/ Εθνική Επιτροπή Βιοηθικής Physicians, Geneticists, insurance companies, general public, workers organizations		Opinion on patents and biotechnology
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HG	Report, Patents in biotechnology Έκθεση, Πατέντες στην βιοτεχνολογία	http://www.bioethics.gr/images/pdf/GNOMES/patents_report_gr.pdf	2003	National Bioethics Committee/ Εθνική Επιτροπή Βιοηθικής Physicians, Geneticists, insurance companies, general public, workers organizations	Report on patents and biotechnology
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HG	Fundamental Principles of Ethics and Bioethics, Βασικές Αρχές Ηθικής και Βιοηθικής	file:///C:/Users/user/Desktop/VasikesArxesDeontologiasKaiVioithikis15062016.pdf	2003-The Senate of the National and Kapodestrian University in Athens Σύγκλητος ΕΚΠΑ	Senate of the National and Kapodestrian University of Athens Physicians, Faculty Members, researchers, Geneticists, general public		A text with the main principles on ethics and bioethics to serve as a guide in the University of Athens, voted by the Senate of the University
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TABLE 5A: General questions on your document search in Human Genomics

1- Did you find guidance documents specific to human genomics or genetics?	Yes
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2a How many documents did you find from each of these sources that specifically address human genetics or genomics	PECs: 2 NAEG: 12 Total: 14
2b Did you find more documents than you had the time to analyse in this task?	No
3- a) How many documents specifically ONLY address high throughput genomics? By high throughput genomics, we mean using sequencing of larger parts of the genome, or exome (including large gene panels).	None

<p>4 a How many documents only address genetics (so testing or screening but without a lot of sequencing)?</p> <p>4b List the years of publications of each document here</p> <p>3c please specify the topics addressed explicitly</p>	<p>PECs two NAEG 5</p> <p>2005, 2007, 2008, 2014, 2015, 2016, 2017</p> <p>Genetics used for diagnostics in the clinic (genetic testing) Genetics used for research Consent in genetics in the clinic Consent in genetics in research newborn screening prenatal screening or testing Non-invasive prenatal testing/screening</p>
<p>5. How many documents address both genetics and genomics?</p>	<p>4</p>

<p>6 a) How many documents specifically address Genome Modification, gene editing, crispr, or gene therapy</p> <p>6b) do they address somatic or germline editing or both?</p>	<p>2</p> <p>Both</p>
<p>How many documents specifically address (so in the title of the document)</p>	<p>Genetic or genomic testing 2</p> <p>Genetic or genomic screening 2</p> <p>Biobanks or databases 0</p> <p>Pharmacogenomics or pharmacogenetics 0</p> <p>Patents 2</p>

Table 5B RELEVANT PEC or NAEG DOCUMENTS IN HUMAN GENOMICS

<p>Document found via (national associations or google or another database)</p>	<p>National Bioethics Committee, website</p>
<p>Title of document</p>	<p>Current matters of 'choice' in reproduction</p>
<p>Scope/ main topic</p>	<p>Genetic testing</p>
<p>Kind of document (PEC or NAEG or other)</p>	<p>NAEG</p>

Document developed by whom (organisation, profession)?	National Bioethics Committee
Year the document was published	2014
Document saved in folder as	
Who is the stated audience, if none specified, write not stated (NS)	Not stated NS
For Clinical or research or both or not specified	Not specified
What level of guideline is provided?	Specific More Theoretical (values, principles) More practical (a practitioner could apply them)

<p>What is the scope of the document, and what are the main recommendations?</p>	<p>The document is an Opinion to guide current matters of 'choice 'in reproduction. It follows a previous relevant Opinion on preimplantation and prenatal diagnosis of the fetus in 2006. The Committee rejects positive eugenics with the choice of gametes with desired characteristics. The Committee recommends that a choice of embryos for health reasons should be accepted in the case of grave monogenetic diseases and also, to detect a high chance of cancer of the breast/ovaries by checking for BRCA1 and BRCA2, NHPCC or Lynch syndrome (genes MLH1 and MSH2) and syndrome Li-Fraumeni (gene 53), A list of severe genetic diseases should be compiled in accordance to the developments in genetics. The Clinics should adhere to the list of the Human Fertilization and Embryology Authority (HFEA), since no Greek accepted list exists in the case of prenatal diagnosis/preimplantation. The Committee accepts savior sibling choice in genetics. The Committee, finally, rejects the replacement of mitochondria until further medical evidence.</p>	
<p>Which life stage is addressed in the document?</p>	<p>Prenatal</p>	
<p>Which ethical challenges are addressed in the document? (you can see lists above for ideas of types)</p>	<p>The challenges are the freedom of people to reproduce at will, and to accept the 'creation' of an suitable healthy embryo and the right to life of embryos possibly carrying a severe genetic disease. Informed consent issues are also dealt with, protecting autonomy in recreation.</p>	
<p>How are the ethical issues addressed?</p>	<p>The issues are addressed practically and theoretically (not in depth in this case)</p>	

How is the document structured? e.g. Which format is used in the document (checklist, continuous text, other)?	Continuous text.
Is the document clearly understandable?	Yes
Why is the document important/useful for your country?	The document is the formal guidance document on the issues it deals with in Greece, by the National Bioethics Committee.
Is the document useful for the development of the SIENNA codes and other ethical frameworks? If yes, please explain.	The document could be useful for the development of the SIENNA codes etc as it deals with these matters.
Document found via (national associations or google or another database)	National Bioethics Committee website
Title of document	Opinion, Recent developments on the amendment of human genome: Genome editing
Scope/ main topic	Genome editing
Kind of document (PEC or NAEG or other)	NAEG
Document developed by whom (organisation, profession)?	National Bioethics Committee

Year the document was published	2016
Document saved in folder as	
Who is the stated audience, if none specified, write not stated (NS)	NS
For Clinical or research or both or not specified	Not specified
What level of guideline is provided?	Specific More Theoretical (values, principles) More practical (a practitioner could apply them)

<p>What is the scope of the document, and what are the main recommendations?</p>	<p>The National Bioethics Committee dealt with the ethical challenges arising from the new methods of CRISPR/Cas9. The methods are explained in the Opinion. The Opinion analyses the questions of safety, use of these methods, and recommends that the medical community must design safe clinical trials which will give us safe results. The scientific community has to prove the costs and benefits of these new technologies of genome editing, at first on body cells, as parts of a genome therapy. These clinical trials are encouraged and the Ministry of Health is also urged to design in its research designs these research protocols. Under the Committee, the use of human gametes and of embryo in vitro is legitimate only for basic research purposes, which means that these embryos and these gametes must not be used in implantation to be born. The Committee proposes that the Oviedo Convention needs a clarification in this case, a positive prohibition of the application of genome amendments for reasons of positive eugenics.</p>
<p>Which life stage is addressed in the document?</p>	<p>Prenatal</p>
<p>Which ethical challenges are addressed in the document?</p>	<p>Safety of people, right to health, autonomy</p>

How is the document structured? e.g. Which format is used in the document (checklist, continuous text, other)?	Continuous text
Is the document clearly understandable?	Yes
Why is the document important/useful for your country?	Yes
Is the document useful for the development of the SIENNA codes and other ethical frameworks? If yes, please explain.	Yes (shows the Greek position on genome editing by the most authoritative source).

Document found via	National Bioethics Committee website
Title of document	Recommendation, Replacement of Mitochondria to avoid mitochondria diseases
Scope/ main topic	Mitochondria replacement
Kind of document (PEC or NAEG or other)	NAEG

Document developed by whom (organisation, profession)?	National Bioethics Committee
Year the document was published	2017
Document saved in folder as	
Who is the stated audience, if none specified, write not stated (NS)	NS
For Clinical or research or both or not specified	Not specified
What level of guideline is provided?	Specific More Theoretical (values, principles) More practical (a practitioner could apply them)
What is the scope of the document, and what are the main recommendations?	The National Bioethics Committee dealt with the ethical challenges arising from the new methods of replacing mitochondria (MRT) to avoid mitochondria diseases. It recommends that the new method not be applied in Greece clinically because of the lack of appropriate and full evidence of their safe use. Clinical trials in the future should be reserved for particular cases in special clinical centres. In the case we have positive results from thee trials, an amendment of the statute on medically assisted reproduction will be necessary to include the authority of the National Authority for Medically assisted Reproduction to license particular clinics to apply the methods.

Which life stage is addressed in the document?	Prenatal
Which ethical challenges are addressed in the document?	Safety of people, right to health, autonomy
How is the document structured? e.g. Which format is used in the document (checklist, continuous text, other)?	Continuous text
Is the document clearly understandable?	Yes
Why is the document important/useful for your country?	Yes
Is the document useful for the development of the SIENNA codes and other ethical frameworks? If yes, please explain.	Yes (shows the Greek position on MRT by the most authoritative source).

Document found via (national associations or google or another database)	National Bioethics Committee website
Title of document	Opinion on the Use of Genetic Data in Private Insurance
Scope/ main topic	Genetic data
Kind of document (PEC or NAEG or other)	NAEG
Document developed by whom (organisation, profession)?	National Bioethics Committee
Year the document was published	2008
Document saved in folder as	
Who is the stated audience, if none specified, write not stated (NS)	NS
For Clinical or research or both or not specified	Not specified
What level of guideline is provided?	Specific More Theoretical (values, principles) More practical (a practitioner could apply them)

<p>What is the scope of the document, and what are the main recommendations?</p>	<p>The National Bioethics Committee dealt with the ethical challenges arising from the use of genetic data in private insurance. The Opinion describes the ethical challenges from the need to protect the workers right to personality and also, their economic freedom, and the danger of 'genetic determinism', in connection with the value of genetic data. It also notes that insurance is a basic value for the people and cannot become an object of commercialization, and that genetic data reflect a statistical possibility to fall ill, and not a certainty. Research on human genome has to serve health as a human right and it should be encouraged. In view of all the above, there is a need to regulate the use of genetic data in insurance. Genetic data are not seen as very important in the estimate of the insurance risk. To protect the interests of the insurers not to fall victims to scam, a period of moratorium is necessary, in which time the insurers will promise not to make use of any genetic data, but also that while this moratorium stands, the Greek State will engage in a specific legislative regulation on this matter. During the same time, all genetics laboratories need to be licensed so that all genetic counselling and other genetic services be ascertained as to their quality and safety towards their customers.</p>
<p>Which life stage is addressed in the document?</p>	<p>Adults</p>
<p>Which ethical challenges are addressed in the document?</p>	<p>Safety of people, right to health, autonomy, right to insurance, economic rights</p>

How is the document structured? e.g. Which format is used in the document (checklist, continuous text, other)?	Continuous text
Is the document clearly understandable?	Yes
Why is the document important/useful for your country?	Yes
Is the document useful for the development of the SIENNA codes and other ethical frameworks? If yes, please explain.	Yes (shows the Greek position on genetic data and insurance by the most authoritative source).

Document found via (national associations or Google or another database)	National Bioethics Committee website
Title of document	Opinion, Consent in the doctor/patient relationship
Scope/ main topic	Consent (covers also consent of genetic/genome treatment issues)

Kind of document (PEC or NAEG or other)	NAEG
Document developed by whom (organisation, profession)?	National Bioethics Committee
Year the document was published	2010
Document saved in folder as	
Who is the stated audience, if none specified, write not stated (NS)	NS
For Clinical or research or both or not specified	Not specified
What level of guideline is provided?	Specific More Theoretical (values, principles) More practical (a practitioner could apply them)
What is the scope of the document, and what are the main recommendations?	The National Bioethics Committee dealt with the ethical challenges arising from consent in the doctor/patient relationship. The Committee notes that the old paternalistic standard of the doctor/patient relationship has changed and it has been replaced by the need to respect the patient's autonomy. Consent in treatment expresses this autonomy. Consent cannot be a merely bureaucratic procedure. The Committee aims at contributing to the best application of the principle of consent with specific recommendations.

Which life stage is addressed in the document?	Adult Minors
Which ethical challenges are addressed in the document?	right to health, autonomy, self determination, right to information
How is the document structured? e.g. Which format is used in the document (checklist, continuous text, other)?	Continuous text
Is the document clearly understandable?	Yes
Why is the document important/useful for your country?	Yes
Is the document useful for the development of the SIENNA codes and other ethical frameworks? If yes, please explain.	Yes (shows the Greek position on informed consent to treatment by the most authoritative source).

Document found via (national associations or google or another database)	The Greek Parliament/The National Authority for Medically Assisted Reproduction
Title of document	Code of Ethics for Medically Assisted Reproduction
Scope/ main topic	Genetics, genomics
Kind of document (PEC or NAEG or other)	PEC/The Greek Parliament
	National Authority for the Medically Assisted Reproduction/The Greek Parliament
Year the document was published	2017
Document saved in folder as	
Who is the stated audience, if none specified, write not stated (NS)	NS
For Clinical or research or both or not specified	Not specified
What level of guideline is provided?	Specific More Theoretical (values, principles) More practical (a practitioner could apply them)

<p>What is the scope of the document, and what are the main recommendations?</p>	<p>The Code of Ethics for the Medically Assisted Reproduction (MAR) is a comprehensive document covering the widest possible area of medically assisted reproduction issues in Greece. It was drafted by the National Authority for Medically Assisted Reproduction and then, it was voted as a Code by the Greek Parliament and has the value of a statute. The Code provides for the conditions of medically assisted reproduction, such as the accepted methods of MAR, the respect of human rights in MAR, the information necessary to be given to the persons who want to participate in MAR, consent in MAR, special obligations arising from preimplantation genetic diagnosis, genetic research and MAR when pregnancy is not sought necessarily, genetic research in the case of MAR leading to a pregnancy, and other matters. Cloning is forbidden. Choice of sex is also forbidden as a rule, unless sex is related to a severe genetic inheritable disease which can be avoided. PID is allowed with the consent of the persons involved and under the license of the Authority.</p>
<p>Which life stage is addressed in the document?</p>	<p>Prenatal Adult (consent to MAR, PID etc)</p>
<p>Which ethical challenges are addressed in the document?</p>	<p>right to health, right to procreation, autonomy, right to form a family</p>
<p>How is the document structured? e.g. Which format is used in the document (checklist, continuous text, other)?</p>	<p>Continuous text</p>

Is the document clearly understandable?	Yes
Why is the document important/useful for your country?	Yes
Is the document useful for the development of the SIENNA codes and other ethical frameworks? If yes, please explain.	Yes (shows the Greek position on MAR and genetic diagnosis PID etc by the most authoritative source, having the validity of a statute).

Document found via (national associations or google or another database)	Google search
Title of document	Code of Medical Ethics
Scope/ main topic	Ethics in medicine
Kind of document (PEC or NAEG or other)	PEC
Document developed by whom (organisation, profession)?	The Greek Parliament (code voted as a statute)
Year the document was published	2005

Document saved in folder as	
Who is the stated audience, if none specified, write not stated (NS)	NS
For Clinical or research or both or not specified	Not specified
What level of guideline is provided?	Specific More Theoretical (values, principles) More practical (a practitioner could apply them)
What is the scope of the document, and what are the main recommendations?	The Code of Medical Ethics covers all major issues in medical ethics, such as informed consent, consent to research, non therapeutic biomedical research, medically assisted reproduction etc. The Code prohibits the amendment of the human genome and safeguards genetic identity of the people.
Which life stage is addressed in the document?	Prenatal Adults
Which ethical challenges are addressed in the document?	Safety of people, right to health, autonomy, right to procreation

How is the document structured? e.g. Which format is used in the document (checklist, continuous text, other)?	Continuous text
Is the document clearly understandable?	Yes
Why is the document important/useful for your country?	Yes
Is the document useful for the development of the SIENNA codes and other ethical frameworks? If yes, please explain.	Yes (shows the Greek position on the amendment of the human genome and the protection of genetic identity by the most authoritative source, a statute also).

Document found via	National Bioethics Committee website
Title of document	Opinion on patent rights in biotechnology inventions
Scope/ main topic	Biotechnology patents
Kind of document (PEC or NAEG or other)	NAEG
Document developed by whom (organisation, profession)?	National Bioethics Committee

Year the document was published	2003
Document saved in folder as	
Who is the stated audience, if none specified, write not stated (NS)	NS
For Clinical or research or both or not specified	Not specified
What level of guideline is provided?	Specific More Theoretical (values, principles) More practical (a practitioner could apply them)

<p>What is the scope of the document, and what are the main recommendations?</p>	<p>The National Bioethics Committee dealt with the ethical challenges arising from patenting in biotechnology. The Committee accepts that patents reward inventions and not discoveries. This main principle is also applicable in the case of biotechnology patents. The enhancement of human life via biotechnology patents is a valid purpose but only in the frame of the protection of human rights. The Committee states that the human genome or selected sequences of human genome, tissues, cells, organisms and their functions cannot form a valid basis for a patent, to the degree that they are discoveries and not inventions. This principle stems also from the Directive 98/44/EC, which has been incorporated into the Greek legal order. But patents over living matter, which is not human, can stand, as there is no 'person' to protect (only animals and plants), having in mind though that animals have legally protected moral status. Any matter not separately existing in nature under the above conditions can be a subject of a patent right. Genetically amended (edited) cell sequences can form the subject of a patent right. The amended biological system, though, should be deposited into a properly authorized biological matter database, according to the spirit of the Directive 98/44/EC (art 11). Biotechnological methods can be patented. Their products are not patentable. Methods to describe or separate biological systems or their parts can be patented, irrespective of whether they come from humans, animals or plants.</p>
<p>Which life stage is addressed in the document?</p>	<p>Prenatal Adult</p>
<p>Which ethical challenges are addressed in the document?</p>	<p>Safety of people, right to health, autonomy, interests of animals, human dignity</p>

How is the document structured? e.g. Which format is used in the document (checklist, continuous text, other)?	Continuous text
Is the document clearly understandable?	Yes
Why is the document important/useful for your country?	Yes
Is the document useful for the development of the SIENNA codes and other ethical frameworks? If yes, please explain.	Yes (shows the Greek position on biotechnology patents by the most authoritative source).

7.6 Japan

Due to the fact that Japanese partner did not have budget assigned to this task, only basic search and no further analysis was performed for the relevant documents.

The following documents were found:

<http://www.sci.go.jp/ja/info/kohyo/pdf/kohyo-23-t251-1-en.pdf>

http://www.lifescience.mext.go.jp/files/pdf/n796_00.pdf

http://jams.med.or.jp/guideline/genetics-diagnosis_e.pdf

http://ishg.jp/wp-content/uploads/2017/08/10academies_e.pdf

7.7 the Netherlands

TABLE 1: INDIVIDUAL AND COUNTRY INFORMATION

Names and emails of persons who did the work (if different from above)	Tanne Ditzel (t.f.ditzel@student.utwente.nl), Philip Jansen
Your organisation	University of Twente (NL)
Your country (again)	The Netherlands
Search conducted in which language	Dutch / English
Acknowledgements (any researcher who helped you to complete this task)	

TABLE 2: LIST OF ALL RELEVANT PROFESSIONAL ETHICS CODES

SIENNA area	Title document (original English translation)	of +	URL	Year	Author/organisation	Stated audience	comments
HG	Richtlijnen en protocollen (Guidelines and protocols for clinical genetics)	en +	http://www.vkgn.org/vakinformatie/richtlijnen-en-protocollen/	2018	Vereniging klinische genetica nederlands VKGN (Dutch Association for Clinical Genetics)	Practitioners in the field of genetics/genomics	No ethics code. These guidelines for the most part focus on controlling the technical quality of the work performed by practitioners. In a few instances, it can be said they are of a more ethical nature.

Netherlands

HG	Ethisch manifest (ethical manifesto of the Association for Collaborating Parent and Patient Organisations)	https://vsop.nl/media/uploads/file/position%20papers/Ethisch%20Manifest%20VVSOP.pdf	1997	Vereniging Samenwerkende Ouderen Patiëntenorganisaties (VSOP) (Association for Collaborating Parent and Patient Organisations)	Physicians, professionals, patients and relatives of patients	This ethical manifesto focuses on particular issues in genetics/genomics
HG	Gedragsregels voor artsen / ethical code of conduct for physicians	https://www.knmg.nl/web/file?uuid=7d93d97b-5c50-4368-99f6-c250f4459c53&owner=5c945405-d6ca-4deb-aa16-7af2088aa173&contentid=412&elementid=1889271	2013	Koninklijke Nederlandsche Maatschappij tot bevordering der Geneeskunst (KNMG) (Royal Dutch Society for the Advancement of Medical Science)	Practitioners/members	This code of conduct for physicians makes no specific mention of genetics or genomics research

Netherlands

HG	Gedragcode Medische hulpmiddelen 2018 / Code of Conduct Medical Devices 2018	http://www.gmh.nu/images/Gedragcode_GMH_-_english_January_2018.pdf	2018	Gedragcode medische hulpmiddelen (GMH)	Developers medical devices and healthcare professionals	This code of conduct for the use of medical devices makes no specific mention of genetics or genomics research
HG	Human Tissue and Medical Research: Code of Conduct for responsible use (2011)	https://www.elsi.health-ri.nl/sites/elsi/files/ELSI/Code%20Goed%20Gebruik%20Engelstalig.pdf	2011	Federation of Dutch Medical Scientific Societies	Healthcare professionals	This code focuses on particular issues in genetics/genomics
HG	Dutch code of conduct for medical research	https://www.federa.org/sites/default/files/bijlagen/coreon/code_of_conduct_for_medical_research_1.pdf	2004	Federation of Dutch Medical Scientific Societies	Healthcare professionals	Makes no specific mention of genetics or genomics research

Netherlands

HG	<p>Handreiking voor het constateren van, omgaan met en informeren over nevenbevindingen voor biobanken in BBMRI-NL (Guidelines and best practices for biobanks for identifying, dealing with, and informing patients/clients about incidental findings)</p>	<p>https://www.elsi.health-ri.nl/sites/elsi/files/ELSI/Handreiking_nevenbevindingen_Erasmus_MC.pdf</p>	2017	Erasmus Medical Centre (& Biobanking and BioMolecular resources Research Infrastructure The Netherlands)	Practitioners in biobanking	Contains guidelines relevant for geneticists in biobanking
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TABLE 3: LIST OF ALL RELEVANT DOCUMENTS FROM NATIONAL ADVISORY/ETHICS GROUPS

SIENNA area	Title of document (original + English translation)	URL	Year	Author/organization	Stated audience	comments
HG	Ingrijpen in het DNA van de mens (intervening in human DNA)	https://www.gezondheidsraad.nl/sites/default/files/grpublication/cogem-gezondheidsraad_kiembaanmodificatie.pdf	2017	Gezondheidsraad (Health Council) & COGEM (The Netherlands Commission on Genetic Modification)	Government, public	
HG	Trendanalyse biotechnologie (Biotechnology Analysis) Trend	https://www.cogem.net/index.cfm/nl/publicaties/publicatie/trendanalyse-biotechnologie-2016-regelgeving-ontregeld	2016	Gezondheidsraad (Health Council) & COGEM (The Netherlands Commission on Genetic Modification)	Government, public	

Netherlands

HG	Genome Editing Visiedocument (genome editing vision document)	https://www.knaw.nl/shared/resources/actueel/publicaties/pdf/genome-editing-visiedocument-knaw	2016	KNAW (Royal Dutch Academy of Sciences)	Government, policy-makers, scientists, professionals, physicians	
HG	IVF: afrondende advisering (IVF complete advice policy)	https://www.gezondheidsraad.nl/sites/default/files/9808n.pdf	1998	Gezondheidsraad (Health Council)	Government, public, physicians	
HG	Handelingen met geslachtscellen en embryo's (proceedings with reproductive cells and embryos)	https://ceg.nl/uploads/publicaties/Hoofdstuk1handelingen.pdf	2003	Gezondheidsraad (Health Council)	Government, public, physicians	
HG	Screening en de rol van de overheid (Screening and the Role of the Government)	https://www.raadrvs.nl/publicaties/item/screening-en-de-rol-van-de-overheid	2018	Raad voor Volksgezondheid en Samenleving (The Council for Health and Society)	Government, general public	

Netherlands

HG	Het 'duizend dollar genoom': een ethische verkenning	https://ceg.nl/uploads/publicaties/signalement-duizenddollargenoom.pdf	2010	Gezondheidsraad (Health Council)	Government, public, physicians	
HG	Zorg voor het ongeboren kind (care for the unborn child)	https://ceg.nl/uploads/publicaties/signalement-zorgongeborenkind.pdf	2009	Gezondheidsraad (Health Council)	Government, general public, physicians	
HG	Regels voor het digitale mensenpark (rules and restrictions for the digital "human park")	https://www.rathenau.nl/sites/default/files/2018-02/Regels%20voor%20het%20digitale%20mensenpark%20-%20Rathenau%20Instituut%20-.pdf	2017	Rathenau Institute	Policy-makers	
HG	Screening: tussen hoop en hype (Screening: Between Hope and Hype)	https://www.gezondheidsraad.nl/sites/default/files/200805.pdf	2008	Gezondheidsraad (Health Council of the Netherlands)	Government	

Netherlands

HG	Embryonale stamcellen zonder morele pijn? (embryonic stem cells without moral "hurting"?)	https://ceg.nl/uploads/publicaties/si_gn_2005_h1.pdf	2005	CEG & gezondheidsraad	Government, physicians	Somewhat less relevant (not included in analysis)
HG	Hematopoietische stamcellen (hematopoietic stem cells)	https://www.gezondheidsraad.nl/sites/default/files/0317n.pdf	2003	Gezondheidsraad	Government, physicians	Somewhat less relevant (not included in analysis)
HG	Stamcellen voor weefselherstel: Onderzoek naar therapie met somatische en embryonale stamcellen (stem cells for tissue recovery)	https://www.gezondheidsraad.nl/sites/default/files/0209n.pdf	2002	Gezondheidsraad	Government, public, physicians	Somewhat less relevant (not included in analysis)

Netherlands

HG	Geslachtscellen uit het lab (reproductive cells from the lab)	https://www.ceg.nl/uploads/publicaties/WEB_103726_Signalement_CEG_Gametogenese.pdf	2017	Centrum voor Ethiek en Gezondheid (CEG)	Policy-makers	Somewhat less relevant (not included in analysis)
HG	Embryoselectie (embryoselection)	https://vsop.nl/media/uploads/file/Position_paper_Embryoselectie.pdf	2012	Vereniging Samenwerkende Ouder- en Patiëntenorganisaties (VSOP) (Association for Collaborating Parent and Patient Organisations)	Physicians, professionals, patients and relatives of patients	Strictly speaking not a national advisory body (not included in analysis)
HG	Genetische screening bij volwassenen (genetic screening for adults)	https://vsop.nl/media/uploads/file/Position_paper_Genetische_screening_bij_volwassenen_het_psychischaspect.pdf	2012	Vereniging Samenwerkende Ouder- en Patiëntenorganisaties (VSOP) (Association for Collaborating Parent and Patient Organisations)	Physicians, professionals, patients and relatives of patients	Strictly speaking not a national advisory body (not included in analysis)

TABLE 5A: General questions on your document search in Human Genomics

1- Did you find guidance documents specific to human genomics or genetics?	Yes
2a How many documents did you find from each of these sources that specifically address human genetics or genomics 2b Did you find more documents than you had the time to analyse in this task?	PECs = 3 NAEG = 10 GDREC = 0 Total: 13 Yes, roughly how many more documents than those listed here did you find? _____ No

3- a) How many documents specifically ONLY address high throughput genomics? By high throughput genomics, we mean using sequencing of larger parts of the genome, or exome (including large gene panels).

PECs = 0
 NAEG = 1 (only focusing on genome sequencing, not also editing the genome)
 GDREC = 0

3b Please list the year of publication of each documents here:

2010

3c please specify the topics addressed explicitly, ideally indicate the number of documents that address each, place an X beside the topic and add the number of docs (This included docs that not only focus on genome sequencing)

X Genomics used for diagnostics in the clinic (genomic testing) 3
 X Genomics used for research 3
 X Consent in genomics in the clinic 3
 X Consent in genomics in research 3
 Return of results in genomics in the clinic
 X Return of results in genomics in research 3
 X Reinterpretation and recontact in genomics in the clinic 2
 Reinterpretation and recontact in genomics in the clinic
 X Genomics for newborn screening 2
 X Genomics for prenatal screening or testing 2
 Non-invasive prenatal testing/screening
 X Discrimination 2
 X Eugenics 1
 X Justice 2
 X Genetic exceptionalism 1

<p>4 a How many documents only address genetics (so testing or screening but without a lot of sequencing)?</p> <p>4b List the years of publications of each document here</p> <p>3c please specify the topics addressed explicitly, ideally indicate the number of documents that address each, place an X beside the topic and add the number of docs</p>	<p>PECs 0 NAEG 3 GDREC 0</p> <p>1998, 2003, 2009</p> <p>Genetics used for diagnostics in the clinic (genetic testing) ____3 Genetics used for research____ Consent in genetics in the clinic ____3 Consent in genetics in research____ Return of results in genetics in the clinic ____2 Return of results in genetics in research____ Reinterpretation and recontact in genetics in the clinic ____2 Reinterpretation and recontact in genetics in the clinic____ newborn screening____2 prenatal screening or testing____2 Non-invasive prenatal testing/screening____ Discrimination____1 Eugenics____1 Justice____1 Genetic exceptionalism____1</p>
<p>5. How many documents address both genetics and genomics?</p>	<p>5</p>

<p>6 a) How many documents specifically address Genome Modification, gene editing, crispr, or gene therapy</p> <p>6b) do they address somatic or germline editing or both?</p>	<p>4</p> <p>2 address only germline editing, 2 address both</p>
<p>How many documents specifically address (so in the title of the document)</p>	<p>Genetic or genomic testing 8</p> <p>Genetic or genomic screening 5</p> <p>Biobanks or databases 3</p> <p>Pharmacogenomics or pharmacogenetics 3</p> <p>Patents 0</p>

Table 5B RELEVANT PEC or NAEG DOCUMENTS IN HUMAN GENOMICS

5Bi: genomic testing (if none on genomics, use genetic testing)

5Bii genomic screening (if none on genomics, use genetic screening)

5Biii biobanks, databases or registries

5Biv pharmacogenomics or pharmacogenetics

5Bv patents

5Bvi gene editing, crispr, or gene therapy

Netherlands

Document found via (national associations or google or another database)	Google
Title of document	Human Tissue and Medical Research: Code of Conduct for responsible use (2011)
Scope/ main topic	5Biii biobanks, databases or registries
Kind of document (PEC or NAEG or other)	NAEG
Document developed by whom (organisation, profession)?	Dutch Federation of Medical Scientific Societies
Year the document was published	2017
Document saved in folder as	Code of conduct for responsible use of human tissue 2011.pdf
Who is the stated audience, if none specified, write not stated (NS)	Medical professionals/researchers
For Clinical or research or both or not specified	Not specified
What level of guideline is provided?	<p>Very broad: i.e.: on many aspects of genomics or genetics, including many issues all together e.g.</p> <p>X Very specific: only focuses on 1- 2 issues</p> <p>More Theoretical (values, principles)</p> <p>More practical (a practitioner could apply them)</p>

<p>What is the scope of the document, and what are the main recommendations?</p>	<p>This code of conduct focuses on the responsible use of human tissue in scientific research, including the genetic information derived from such material. It does not apply to foetal tissue, embryos and germ cells, or to tissue from deceased persons, even if scientific research is done with it. In the Netherlands, there is already specific legislation applying to the use of such material. The code emphasises that the interests of the donors need to be safeguarded. These interests are optimal privacy protection, dealing responsibly with any (incidental) ‘findings’ and a balanced system of consent (i.e., a system that gives donors adequate influence, without unduly restricting the availability of human tissue for scientific research). Further, the code argues for transparency and consent in the whole chain of scientific research, from taking the tissue sample to ultimately using it in scientific research.</p>
<p>Which life stage is addressed in the document?</p>	<p>X Adults Minors (excluding newborns) Newborns Prenatal</p>
<p>Which ethical challenges are addressed in the document? (you can see lists above for ideas of types)</p>	<p>Informed consent, privacy/anonymity, dealing with incidental findings, transparency towards donors, etc.</p>

<p>How are the ethical issues addressed? (practically, theoretically etc? Any comment on this is fine</p> <p>Are solutions offered? If so, which ones? (depending on the document, this may be redundant with above)</p>	<p>See above.</p>
<p>How is the document structured? e.g. Which format is used in the document (checklist, continuous text, other)?</p>	<p>Bulleted text.</p>
<p>Is the document clearly understandable?</p>	<p>Yes.</p>
<p>Why is the document important/useful for your country? why did you choose this doc? TELL US WHY IMPORTANT for country and why for SIENNA</p>	<p>It is the only real code of ethics/conduct by a professional organisation found in our search that deals with particular issues in genetics/genomics in any significant way.</p>
<p>Is the document useful for the development of the SIENNA codes and other ethical frameworks? If yes, please explain.</p>	<p>The code contains practical guidelines for biobanking that may be useful for SIENNA codes/frameworks.</p>

Netherlands

Document found via (national associations or google or another database)	Google
Title of document	Ingrijpen in het DNA van de mens (intervention in human DNA)
Scope/ main topic	5Bi-5Bvi, gene editing
Kind of document (PEC or NAEG or other)	NAEG
Document developed by whom (organisation, profession)?	Gezondheidsraad (Health Council) & COGEM (The Netherlands Commission on Genetic Modification)
Year the document was published	2017
Document saved in folder as	cogem-gezondheidsraad_kiembaanmodificatie_2017.pdf
Who is the stated audience, if none specified, write not stated (NS)	Government, general public
For Clinical or research or both or not specified	Not specified

<p>What level of guideline is provided?</p>	<p>Very broad: i.e.: on many aspects of genomics or genetics, including many issues all together e.g. X Very specific: only focuses on 1- 2 issues More Theoretical (values, principles) More practical (a practitioner could apply them)</p>
<p>What is the scope of the document, and what are the main recommendations?</p>	<p>The document is about human germline gene editing. It discusses the treatment of genetic diseases in this context, but also designer babies and human enhancement. The Health Council advises the Dutch government to remove a ban on the use of embryos in research that have been specially created for this purpose, thus allowing fundamental research into the use of CRISPR for human germline engineering. The Health Council finds that when used to prevent serious diseases, human germline gene editing does not harm human dignity.</p>
<p>Which life stage is addressed in the document?</p>	<p>Adults Minors (excluding newborns) Newborns X Prenatal</p>
<p>Which ethical challenges are addressed in the document? (you can see lists above for ideas of types)</p>	<p>Moral status of the embryo, harms to human dignity, uncertainty about the consequences of germ line gene editing, parents becoming their child's "makers" and the inability for the child to consent to having their genome edited (including a discussion about whether a child's "numerical identity" changes as a result of editing its genome), issues with regard to equality</p>

Netherlands

<p>How are the ethical issues addressed? (practically, theoretically etc? Any comment on this is fine</p> <p>Are solutions offered? If so, which ones? (depending on the document, this may be redundant with above)</p>	<p>The issues are addressed in a theoretical way and a case is made for the removal of a ban in the Netherlands on the use of embryos in research that have been specially created for this purpose. This would allow for fundamental research to be conducted into the use of CRISPR for human germline engineering in order prevent serious genetic diseases in new-borns.</p>
<p>How is the document structured? e.g. Which format is used in the document (checklist, continuous text, other)?</p>	<p>Continuous text.</p>
<p>Is the document clearly understandable?</p>	<p>Yes.</p>
<p>Why is the document important/useful for your country? why did you choose this doc? TELL US WHY IMPORTANT for country and why for SIENNA</p>	<p>It provides a Dutch perspective on a topic of significance (in terms of potential future consequences) within genetics/genomics.</p>
<p>Is the document useful for the development of the SIENNA codes and other ethical frameworks? If yes, please explain.</p>	<p>It provides an in-depth theoretical ethical discussion and offers only one major governance recommendation, and is therefore somewhat less relevant for SIENNA's codes.</p>

Netherlands

Document found via (national associations or google or another database)	Website of the Dutch Health Council
Title of document	Het 'duizend dollar genoom': een ethische verkenning (The "thousand dollar genome": An ethical exploration)
Scope/ main topic	5Bii genomic screening
Kind of document (PEC or NAEG or other)	NAEG
Document developed by whom (organisation, profession)?	Gezondheidsraad (Health Council)
Year the document was published	2010
Document saved in folder as	signalement-duizenddollargenoom.pdf
Who is the stated audience, if none specified, write not stated (NS)	Government, general public
For Clinical or research or both or not specified	Not specified

Netherlands

<p>What level of guideline is provided?</p>	<p>X Very broad: i.e.: on many aspects of genomics or genetics, including many issues all together e.g. Very specific: only focuses on 1- 2 issues More Theoretical (values, principles) More practical (a practitioner could apply them)</p>
<p>What is the scope of the document, and what are the main recommendations?</p>	<p>The document outlines the major ethical issues that exist in relation to whole genome sequencing and analysis (genomic testing and screening, prenatal screening, screening of embryos). The document does not provide many recommendations. It recommends having a stakeholder/public debate on the issues outlined, and stresses the importance of figuring out how to deal with accidental findings from genomic testing in a responsible way.</p>
<p>Which life stage is addressed in the document?</p>	<p>X Adults X Minors (excluding newborns) XNewborns X Prenatal</p>
<p>Which ethical challenges are addressed in the document? (you can see lists above for ideas of types)</p>	<p>Ethical issues in relation to informed consent (for adults undergoing screening and embryos), retention of the genomic testing/screening data, practitioner’s duties to inform patients/clients</p>

<p>How are the ethical issues addressed? (practically, theoretically etc? Any comment on this is fine</p> <p>Are solutions offered? If so, which ones? (depending on the document, this may be redundant with above)</p>	<p>The document mainly provides an exposition of ethical issues in genomic testing and screening. No solutions are offered.</p>
<p>How is the document structured? e.g. Which format is used in the document (checklist, continuous text, other)?</p>	<p>Continuous text.</p>
<p>Is the document clearly understandable?</p>	<p>Yes.</p>
<p>Why is the document important/useful for your country? why did you choose this doc? TELL US WHY IMPORTANT for country and why for SIENNA</p>	<p>It provides a Dutch perspective on a topic of significance within genetics/genomics.</p>
<p>Is the document useful for the development of the SIENNA codes and other ethical frameworks? If yes, please explain.</p>	<p>It provides an in-depth ethical discussion of genomic testing and screening and does not offer many practical recommendations, and is therefore somewhat less relevant for SIENNA's codes.</p>

Table 5C GDREC DOCUMENTS for HUMAN GENOMICS

Document found via (national associations or google or another database)	
Title of document	
Scope/ main topic	(choose from list above, 5Bi-5Bvi, write out the topic)
Kind of document (PEC or NAEG or other)	
Document developed by whom (organisation, profession)?	
Year the document was published	
Document saved in folder as	
Who is the stated audience, if none specified, write not stated (NS)	
For Clinical or research or both or not specified	

Netherlands

<p>What level of guideline is provided?</p>	<p>Very broad: i.e.: on many aspects of genomics or genetics, including many issues all together e.g. Very specific: only focuses on 1- 2 issues More Theoretical (values, principles) More practical (a practitioner could apply them)</p>
<p>What is the scope of the document, and what are the main recommendations?</p>	
<p>Which life stage is addressed in the document?</p>	<p>Adults Minors (excluding newborns) Newborns Prenatal</p>
<p>Which ethical challenges are addressed in the document? (you can see lists above for ideas of types)</p>	

<p>How are the ethical issues addressed? (practically, theoretically etc? Any comment on this is fine</p> <p>Are solutions offered? If so, which ones? (depending on the document, this may be redundant with above)</p>	
<p>How is the document structured? e.g. Which format is used in the document (checklist, continuous text, other)?</p>	
<p>Is the document clearly understandable?</p>	
<p>Why is the document important/useful for your country? why did you choose this doc? TELL US WHY IMPORTANT for country and why for SIENNA</p>	
<p>Is the document useful for the development of the SIENNA codes and other ethical frameworks? If yes, please explain.</p>	

7.8 Poland

TABLE 1: INDIVIDUAL AND COUNTRY INFORMATION

Your organisation	Helsinki Foundation for Human Rights
Your country	Poland
Search conducted in which language	Polish

TABLE 2: LIST OF ALL RELEVANT PROFESSIONAL ETHICS CODES

SIENNA area	Title of document (original + English translation)	URL	Year	Author/organisation	Stated audience	comments
HG HE (art 75, only in the context of sports and doping)	Kodeks Etyki Lekarskiej (The Code of Medical Ethics)	https://www.nil.org.pl/dokumenty/kodeks-etyki-lekarskiej	1991	Krajowy Zjazd Lekarzy (General Medical Assembly)	Medical profession	Art. 3 Art. 29 Art. 38.3 Chapter II b Human Genome Art. 75

Poland

<p>HG</p>	<p>Stanowisko Polskiego Towarzystwa Genetyki Człowieka (PTGC) oraz Polskiego Towarzystwa Ginekologów i Położników (PTGP) dotyczące zasad i warunków podejmowania indywidualnych decyzji prokreacyjnych w przypadku ryzyka wystąpienia wad wrodzonych lub wystąpienia zaburzeń rozwojowych u potomstwa</p> <p>Statement of the Polish Society of Human Genetics (PTGC) and the Polish Society of Gynecologists and Obstetricians (PTGP) regarding the principles and conditions for making individual reproductive decisions in the event of the risk of congenital malformations or developmental disorders in the offspring</p>	<p>http://www.imid.med.pl/files/imid/Aktualnosci/Aktualnosci/1_Stanowisko_PTGC_PTGP.pdf</p>	<p>2018</p>	<p>Polskie Towarzystwo Genetyki Człowieka (Polish Society of Human Genetics) and Polskiego Towarzystwa Ginekologów i Położników (Polish Society of Gynecologists and Obstetricians.)</p>	<p>NS</p>	
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Poland

HG	<p>Stanowisko Zarządu Polskiego Towarzystwa Genetyki Człowieka (PTGC) w sprawie testów genetycznych wykonywanych niezgodnie z obowiązującymi standardami oraz z naruszeniem zasad etycznych</p> <p>Statement of the Board of the Polish Society of Human Genetics (PTGC) on genetic tests performed in violation of applicable standards and in violation of ethical principles</p>	<p>http://ptgc.pl/wp-content/uploads/2016/06/Stanowisko_Zarz%C4%85du_PTGC_testy_genetyczne.pdf</p>	2016	Polskie Towarzystwo Genetyki Człowieka (Polish Society of Human Genetics)	NS	
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TABLE 3: LIST OF ALL RELEVANT DOCUMENTS FROM NATIONAL ADVISORY/ETHICS GROUPS

SIEN NA area	Title of document (original + English translation)	URL	Year	Author/organization	Stated audience	comments
HG	Stanowisko Komitetu Bioetyki przy Prezydium PAN nr 2/2012 z dnia 8 czerwca 2012 r. w sprawie preimplantacyjnej diagnostyki genetycznej (Statement no. 2/2012 by the Committee of Bioethics on the issue of preimplantation genetic diagnosis of 8 June 2012)	http://www.bioetyka.pan.pl/index.php/stanowiska-iopinie-komitetu/45-stanowiska-komitetu/100-stanowisko-komitetu-bioetyki-przy-prezydium-pan-nr-22012-z-dnia-6-czerwca-2012-r-w-sprawie-preimplantacyjnej-diagnostyki-genetycznej	2012	Komitet Bioetyki PAN (The Committee of Bioethics at the Polish Academy of Science)		

Poland

HG	<p>Stanowisko Komitetu Bioetyki przy Prezydium PAN nr 1/2012 z dnia 15 marca 2012 r. w sprawie etycznych problemów medycyny reprodukcyjnej i genetyki klinicznej oraz konieczności ich regulacji prawnej</p> <p>(Statement no. 1/2012 by the Committee of Bioethics on the ethical problems of reproductive medicine and clinical genetics, and the need for legal regulation of 15 March 2012)</p>	<p>http://www.bioetyka.pan.pl/index.php/stanowiska-iopinie-komitetu/45-stanowiska-komitetu/94-stanowisko-nr-12012-1</p>	2012	Komitet Bioetyki PAN (The Committee of Bioethics at the Polish Academy of Science)		
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Poland

HG	<p>Stanowisko Komitetu Bioetyki przy Prezydium PAN nr 3/2013 z dnia 25 marca 2013 r. w sprawie rynku prywatnych usług genetycznych</p> <p>(Statement 3/2013 by the Committee of Bioethics concerning the private market of genetic services of 25 March 2013)</p>	<p>http://www.bioetyka.pan.pl/index.php/standowiska-iopinie-komitetu/45-standowiska-komitetu/106-standowisko-komitetu-bioetyki-przy-prezydium-pan-nr-32013-z-dnia-25-marca-2013-r-w-sprawie-rynku-prywatnych-usug-genetycznych</p>	2013	Komitet Bioetyki PAN (The Committee of Bioethics at the Polish Academy of Science)	Public authorities	
HG	(A letter to the Minister of Health on the need to introduce a law on genetic tests)	<p>https://www.rpo.gov.pl/sites/default/files/Wyst%C4%85pienie%20RPO%20do%20Ministra%20Zdrowia%20ws.%20uregulowania%20test%C3%B3w%20genetycznych%20.pdf</p>	2018	Rzecznik Praw Obywatelskich (Office of the Commissioner for Human Rights)	Minister of Health	

Poland

HG	(A letter to the Minister of Health on the need to introduce a law on biobanks for research purposes)	https://www.rpo.gov.pl/sites/default/files/Do_MZ_ws._prawnego_uregulowania_dzialalnosci_biobankow.pdf	2015	Rzecznik Praw Obywatelskich (Office of the Commissioner for Human Rights)	Minister of Health	
HG	(A letter to the Minister of Health on genetic counselling)	https://www.rpo.gov.pl/sites/default/files/testy%20genetyczne%2C%20poradnictwo%20psychologiczne.pdf	2017	Rzecznik Praw Obywatelskich (Office of the Commissioner for Human Rights)	Minister of Health	

TABLE 4: LIST OF ALL RELEVANT GUIDANCE DOCUMENTS ON HOW TO WRITE RESEARCH ETHICS PROTOCOLS

Please note: these are examples of recommendations published by some of the local RECs. Not all 54 RECs have been looked at. Because the documents are not technology specific, it was not possible to carry out analysis referred to in Step 3, p. 7 of the Work plan.

Name of national REC	Title of document (original + English translation)	Ethical issues addressed in which SIENNA area (HG, HE, AI&R)?	URL	Stated audience	Comments

Poland

<p>Bioethics Committee by the Warsaw Chamber of Physicians</p>	<p>Regulamin Komisji Bioetycznej</p> <p>Rules of proceeding of a the bioethics committee (REC) by the Warsaw Chamber of Physicians</p>	<p>n/a (non-specific)</p>	<p>https://izba-lekarska.pl/wp-content/uploads/2015/03/Regulamin-Komisji-Bioetycznej.pdf</p>	<p>Researchers submitting the application.</p>	<p>Rules of proceeding the REC contain a form that has to be filled by the applicant. However there is no guidance on how to write a research ethics protocol</p>
<p>Bioethics Committee by the Warsaw Medical University</p>	<p>(Information on required documents)</p>	<p>n/a (non-specific)</p>	<p>https://komisja-bioetyczna.wum.edu.pl/content/szczeg%C3%B3%C5%82owe-informacje-oraz-wzory-dokument%C3%B3w</p>	<p>Researchers submitting the application.</p>	<p>REC provides information on what information should be given to participants and an example of an informed consent form.</p>

Poland

<p>Bioethics Committee by the Copernicus University in Toruń</p> <p>Uniwersytet Mikołaja Kopernika w Toruniu</p>	<p>(Bioethics Committee - Remarks on the most common formal mistakes made when filling in applications)</p>	<p>n/a</p>	<p>https://www.cm.umk.pl/aktualnosci-2/2-collegium-medicum/165-komisja-bioetyczna.html</p> <p>https://www.cm.umk.pl/aktualnosci-2/2-collegium-medicum/561-komisja-bioetyczna-uwagi-odnosnie-najczestszych-bledow-formalnych-popelnianych-przy-wypelnianiu-wnioskow.html</p>		<p>REC provides information on the „most common formal mistakes”, and are related to e.g. recruitment of participants, the use of medical data in research or on biological material.</p>
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Poland

Poland

TABLE 5A: General questions on your document search in Human Genomics

1- Did you find guidance documents specific to human genomics or genetics?	Yes
2a How many documents did you find from each of these sources that specifically address human genetics or genomics	PECs 4 (counting in the Code of Medical Ethics) NAEG 6 GDREC 0 Total: 10
2b Did you find more documents than you had the time to analyse in this task?	No

3- a) How many documents specifically ONLY address high throughput genomics? By high throughput genomics, we mean using sequencing of larger parts of the genome, or exome (including large gene panels).

There were no such documents found

3b Please list the year of publication of each documents here:

3c please specify the topics addressed explicitly, ideally indicate the number of documents that address each, place an X beside the topic and add the number of docs

Genomics used for diagnostics in the clinic (genomic testing)

Genomics used for research

Consent in genomics in the clinic

Consent in genomics in research

Return of results in genomics in the clinic

Return of results in genomics in research

Reinterpretation and recontact in genomics in the clinic

Reinterpretation and recontact in genomics in the clinic

Genomics for newborn screening

Genomics for prenatal screening or testing

Non-invasive prenatal testing/screening

Discrimination

Eugenics

Justice

Genetic exceptionalism

4 a How many documents only address genetics (so testing or screening but without a lot of sequencing)?

Please see the comment for point 5 below.

4b List the years of publications of each document here

PECs 3 (but see the problem with classification referred to in the tables below)

NAEG: 6

GDREC: 0

2012, 2012, 2013, 2015, 2016, 2017, 2018, 2018, 2018

4c please specify the topics addressed explicitly, ideally indicate the number of documents that address each, place an X beside the topic and add the number of docs

Genetics used for diagnostics in the clinic (genetic testing) ____

Genetics used for research ____

Consent in genetics in the clinic ____

Consent in genetics in research ____

Return of results in genetics in the clinic ____

Return of results in genetics in research ____

Reinterpretation and recontact in genetics in the clinic ____

*Reinterpretation and recontact in genetics in the research (only in the context of accidental findings)

newborn screening ____

prenatal screening or testing ____

*Non-invasive prenatal testing/screening (in some cases it is not specified if the testing is or is not invasive)

Discrimination ____

*Eugenics (*but not as public policy, but choice made by parents)

Justice ____

Genetic exceptionalism____

<p>5. How many documents address both genetics and genomics?</p>	<p>The documents refer to “genetics”, the word “genomics” is not used. It could be assumed, however, that the documents that speak of “genetics” in fact mean both genetics and genomics (e.g. the documents that refer to “genetic” testing refer also to “genomic” testing.) For this reason it was difficult to categorize the documents (whether field “4” – only genetics, or “5” – genetics and genomics is the right choice).</p>
<p>6 a) How many documents specifically address Genome Modification, gene editing, crispr, or gene therapy</p> <p>6b) do they address somatic or germline editing or both?</p>	<p>Only the Code of Medical Ethics refers to “changes made to the genome” – inheritable and not.</p>
<p>How many documents specifically address (so in the title of the document)</p>	<p>Genetic or genomic testing – 2 (one does not have a title though – the letter from the Commisioner)</p> <p>Genetic or genomic screening (pol. badania przesiewowe) – 0 (although screening is sometimes mentioned in the context of testing, e.g. in the case of the statement on preimplantation diagnosis, thus it is a bit unclear whether “testing” is not meant as an umbrella term for both)</p> <p>Biobanks or databases – 2 (Ombudsperson letter, the draft code of conduct)</p> <p>Pharmacogenomics or pharmacogenetics - 0</p> <p>Patents - 0</p>

Table 5B RELEVANT PEC or NAEG DOCUMENTS IN HUMAN GENOMICS, copy the table for as many documents as you have to describe.

5Bi: genomic testing (if none on genomics, use genetic testing)

5Bii genomic screening (if none on genomics, use genetic screening)

5Biii biobanks, databases or registries

5Biv pharmacogenomics or pharmacogenetics

5Bv patents

5Bvi gene editing, crispr, or gene therapy

Document found via (national associations or google or another database)	National association
Title of document	Position of the Polish Society of Human Genetics (PTGC) and the Polish Society of Gynecologists and Obstetricians (PTGP) regarding the principles and conditions for making individual reproductive decisions in the event of the risk of congenital malformations or developmental disorders in the offspring
Scope/ main topic	5bi, prenatal (genetic) diagnosis
Kind of document (PEC or NAEG or other)	PEC (classified as PEC because produced by a professional organisation, but in fact it has a character of advice or recommendation)
Document developed by whom (organisation, profession)?	Professional organisation
Year the document was published	2018
Document saved in folder as	1. Stanowisko_ryzyko wad wrodzonych+reproductive decision+PTGC+2018

Who is the stated audience, if none specified, write not stated (NS)	NS Presumably the general public, the authorities
For Clinical or research or both or not specified	Clinical
What level of guideline is provided?	<p>Very broad: i.e.: on many aspects of genomics or genetics, including many issues all together e.g.</p> <p>Very specific: only focuses on 1- 2 issues</p> <p>More Theoretical (values, principles)</p> <p>More practical (a practitioner could apply them)</p> <p>It refers to a general problem but some specific solutions are proposed (see below)</p>
What is the scope of the document, and what are the main recommendations?	<p>The statement underscores that the autonomy of the woman should be respected in all cases, and that the duty of the doctor (according to the Code of Medical Ethics) is to provide all necessary information about the pregnancy and potential risks in an unbiased way.</p> <p>The statement also refers to the negative experience and obstacles faced by people with disabilities and their parents, which impact the decisions about family planning.</p>
Which life stage is addressed in the document?	<p>Adults</p> <p>Minors (excluding newborns)</p> <p>Newborns</p> <p>Prenatal</p> <p>Mainly prenatal, mentions the issue of care for new-borns, pregnant women, as well as adult people with disabilities.</p>

<p>Which ethical challenges are addressed in the document? (you can see lists above for ideas of types)</p>	<p>Autonomy of a pregnant woman, genetic counselling, lack of adequate care for people with disabilities offered by the state</p> <p>genetics used for diagnostics in the clinic return of results in the clinic (new-born testing prenatal testing</p>
<p>How are the ethical issues addressed? (practically, theoretically etc? Any comment on this is fine)</p> <p>Are solutions offered? If so, which ones? (depending on the document, this may be redundant with above)</p>	<p>Practically, but with reference to values and general rules (e.g. the Oviedo Convention)</p>
<p>How is the document structured? e.g. Which format is used in the document (checklist, continuous text, other)?</p>	<p>Continuous text</p>
<p>Is the document clearly understandable?</p>	<p>Yes</p>

<p>Why is the document important/useful for your country? why did you choose this doc? TELL US WHY IMPORTANT for country and why for SIENNA</p>	<p>It addresses a pressing social problem, it is quite a rare example of a professional organisation being vocal about a current social problem, it seems to be a result of a broad consensus – it states: “the boards of PTGC and PTGP, despite the existing differences in opinions (...)”</p> <p>For SIENNA: it is an example of building consensus in a hotly debated issue</p>
<p>Is the document useful for the development of the SIENNA codes and other ethical frameworks? If yes, please explain.</p>	<p>it may be useful because it addresses the difficult issue of the intersection of politics and medicine</p>

<p>Document found via (national associations or google or another database)</p>	<p>National association</p>
<p>Title of document</p>	<p>Statement of the Board of the Polish Society of Human Genetics (PTGC) on genetic tests performed in violation of applicable standards and in violation of ethical principles</p>
<p>Scope/ main topic</p>	<p>5Bi</p>
<p>Kind of document (PEC or NAEG or other)</p>	<p>PEC (classified as PEC because produced by a professional organisation, but in fact it has a character of advice or recommendation)</p>
<p>Document developed by whom (organisation, profession)?</p>	<p>Professional organisation</p>

Year the document was published	2016
Document saved in folder as	2. Stanowisko_Zarządu_PTGC_testy_genetyczne+genetic tests+PTGC+2016
Who is the stated audience, if none specified, write not stated (NS)	<ul style="list-style-type: none"> • Patients, physicians, diagnosticians • Potential users of the genetic tests • Local bioethics commissions (RECs), heads of institutions involved in genetic research • Parties involved in the unethical genetic tests performed for non-medical purposes without consent (see below) • All interested parties: patients, their families, victims of unethical use of genetic testing
For Clinical or research or both or not specified	Tests for health purposes, research as well as other purposes (e.g. paternity)
What level of guideline is provided?	<p>Very broad: i.e.: on many aspects of genomics or genetics, including many issues all together e.g.</p> <p>Very specific: only focuses on 1- 2 issues</p> <p>More Theoretical (values, principles)</p> <p>More practical (a practitioner could apply them)</p> <p>It focuses on tests and provides some practical advice for different audiences.</p>

<p>What is the scope of the document, and what are the main recommendations?</p>	<ul style="list-style-type: none"> • Lack of relevant laws on genetic testing is a source of practices that are unethical and that violate standards; • There is a lack of political will to introduce the laws • Genetic tests are becoming increasingly available and are vehemently advertised • Genetic tests performed without oversight are dangerous for consumers • It is unethical to perform genetic tests for purposes other than medical (e.g. tests to dis/prove paternity) without consent from all parties and without court supervision <p>Recommendations:</p> <ul style="list-style-type: none"> • Potential users of the tests should be very careful and critical about the tests • RECs and heads of institutions involved in genetic research should have supervision over the process of return of results • All interested parties: patients, their families, victims of unethical use of genetic testing should report such cases (to PTGC) • There should be disciplinary penalties for people involved in the described unethical activities • Law on genetic tests should be adopted
<p>Which life stage is addressed in the document?</p>	<p>Adults Minors (excluding newborns) Newborns Prenatal</p> <p>Minors in the case of paternity tests.</p>

<p>Which ethical challenges are addressed in the document? (you can see lists above for ideas of types)</p>	<p>Protection of genetic data and national security (only briefly mentioned)</p> <p>Genetic testing Genetics used for research Lack of consent in the case of paternity tests Return of results in the clinic (i.e. lack of any genetic counselling) Return of results in research</p>
<p>How are the ethical issues addressed? (practically, theoretically etc? Any comment on this is fine)</p> <p>Are solutions offered? If so, which ones? (depending on the document, this may be redundant with above)</p>	<p>Practically, but on a general level</p>
<p>How is the document structured? e.g. Which format is used in the document (checklist, continuous text, other)?</p>	<p>Continuous text</p>
<p>Is the document clearly understandable?</p>	<p>Yes</p>

<p>Why is the document important/useful for your country? why did you choose this doc? TELL US WHY IMPORTANT for country and why for SIENNA</p>	<p>This issues has been debated for years, the need to adopt the law has been repeated by many intuitions, still no provisions have been introduced; the statement points out the lack of political will to address the issues relevant for SIENNA</p>
<p>Is the document useful for the development of the SIENNA codes and other ethical frameworks? If yes, please explain.</p>	

<p>Document found via (national associations or google or another database)</p>	<p>Advisory body</p>
<p>Title of document</p>	<p>(A letter to the Minister of Health on the need to introduce a law on genetic tests)</p>
<p>Scope/ main topic</p>	<p>(choose from list above, 5Bi-5Bvi, write out the topic)</p>
<p>Kind of document (PEC or NAEG or other)</p>	<p>NAEGs</p>
<p>Document developed by whom (organisation, profession)?</p>	<p>By a public body</p>

Year the document was published	2018
Document saved in folder as	3. Wystąpienie RPO+Ombudsperson+genetic tests+2018
Who is the stated audience, if none specified, write not stated (NS)	Minister of Health
For Clinical or research or both or not specified	Not specified, but judging on the context for clinical
What level of guideline is provided?	<p>Very broad: i.e.: on many aspects of genomics or genetics, including many issues all together e.g.</p> <p>Very specific: only focuses on 1- 2 issues</p> <p>More Theoretical (values, principles)</p> <p>More practical (a practitioner could apply them)</p> <p>It focuses on genetic tests.</p>
What is the scope of the document, and what are the main recommendations?	<p>The law on genetic tests needs to be introduced to ensure a proper protection of individual rights, such as the right to life and health, self-determination, privacy.</p> <p>Genetic tests performed without a proper oversight are a serious risk to patient's health and life.</p>

<p>Which life stage is addressed in the document?</p>	<p>Adults Minors (excluding newborns) Newborns Prenatal</p> <p>Not stated, but most probably adults and minors</p>
<p>Which ethical challenges are addressed in the document? (you can see lists above for ideas of types)</p>	<p>Genetic counselling Data protection</p> <p>Genetic testing Return of results in the clinic (plus informing members of family) Discrimination</p>
<p>How are the ethical issues addressed? (practically, theoretically etc? Any comment on this is fine)</p> <p>Are solutions offered? If so, which ones? (depending on the document, this may be redundant with above)</p>	<p>They are not addressed practically, but rather as issues that need regulation, without going into too much detail.</p>
<p>How is the document structured? e.g. Which format is used in the document (checklist, continuous text, other)?</p>	<p>Continuous text</p>

Is the document clearly understandable?	Yes
Why is the document important/useful for your country? why did you choose this doc? TELL US WHY IMPORTANT for country and why for SIENNA	It shows the intersections between ethics, human rights and new technologies.
Is the document useful for the development of the SIENNA codes and other ethical frameworks? If yes, please explain.	It addressed the issues from the point of view of human rights and places them in the context of human rights.

Document found via (national associations or google or another database)	Advisory Body
Title of document	Statement 3/2013 by the Committee of Bioethics concerning the private market of genetic services of 25 March 2013
Scope/ main topic	5Bi
Kind of document (PEC or NAEG or other)	NAEG

Poland

Document developed by whom (organisation, profession)?	By members of the Committee
Year the document was published	2013
Document saved in folder as	4. Stanowisko KB+Bioethics Committee+private market+2013
Who is the stated audience, if none specified, write not stated (NS)	Public authorities
For Clinical or research or both or not specified	Direct to consumer
What level of guideline is provided?	<p>Very broad: i.e.: on many aspects of genomics or genetics, including many issues all together e.g.</p> <p>Very specific: only focuses on 1- 2 issues</p> <p>More Theoretical (values, principles)</p> <p>More practical (a practitioner could apply them)</p>

<p>What is the scope of the document, and what are the main recommendations?</p>	<p>The statement concerns the issue of direct to consumer genetic testing for health purposes, and the need to formally regulate it. It lists the rights of patients that form the ethical basis for allowing DTC (see below), as well as specific features of genetic tests and the nature of genetic information that should be considered when introducing the law (e.g. the difficulties with correct interpretation of data, conflicts between members of the family, the need for confidentiality so that the information is not used against the patient in a discriminatory manner). In addition it lists ethical principles that have significant meaning in the discussed context (the list is non-exhaustive): justice, non-maleficence, beneficence, autonomy, prohibition of discrimination; it stresses the need for education and awareness raising activities.</p> <p>It refers to art. 12 of the Oviedo Convention</p> <p>Recommendations:</p> <ul style="list-style-type: none"> - Law on genetic tests should be adopted
<p>Which life stage is addressed in the document?</p>	<p>Adults Minors (excluding newborns) Newborns Prenatal</p> <p>Not stated</p>

<p>Which ethical challenges are addressed in the document? (you can see lists above for ideas of types)</p>	<p>Genetic counselling The right of patients to know their health status, the health situation of their children, the right to choose the treatment, the right to make informed reproductive decisions</p> <p>Genetic testing (specifically direct to consumer tests) Discrimination, Justice (as principles that need to be adhered to) Other ethical principles: non-maleficence, beneficence, autonomy</p>
<p>How are the ethical issues addressed? (practically, theoretically etc? Any comment on this is fine)</p> <p>Are solutions offered? If so, which ones? (depending on the document, this may be redundant with above)</p>	<p>Rather theoretically, some practical consequences of the lack of regulation are addressed (e.g. the lack of counselling)</p>
<p>How is the document structured? e.g. Which format is used in the document (checklist, continuous text, other)?</p>	<p>Continuous text with lists</p>
<p>Is the document clearly understandable?</p>	<p>Yes</p>

<p>Why is the document important/useful for your country? why did you choose this doc? TELL US WHY IMPORTANT for country and why for SIENNA</p>	<p>it situates the discussed issue (DTC) in the ethical context, it provides reasons why there is a need to regulate</p>
<p>Is the document useful for the development of the SIENNA codes and other ethical frameworks? If yes, please explain.</p>	<p>It lists some of the most relevant principles, as well as speaks of the right of patients</p>

<p>Document found via (national associations or google or another database)</p>	<p>National Association (General Medical Assembly)</p>
<p>Title of document</p>	<p>5. Code of Medical Ethics</p>
<p>Scope/ main topic</p>	<p>n/a (It has a general scope)</p>
<p>Kind of document (PEC or NAEG or other)</p>	<p>PEC</p>
<p>Document developed by whom (organisation, profession)?</p>	<p>Profession</p>
<p>Year the document was published</p>	<p>1991</p>

Document saved in folder as	5. Kodeks-Etyki-Lekarskiej+NIL+medical ethics+1991
Who is the stated audience, if none specified, write not stated (NS)	Medical professionals
For Clinical or research or both or not specified	Both
What level of guideline is provided?	Very broad: i.e.: on many aspects of genomics or genetics, including many issues all together e.g. Very specific: only focuses on 1- 2 issues More Theoretical (values, principles) More practical (a practitioner could apply them)

What is the scope of the document, and what are the main recommendations?

The scope is general.

Some aspects of genetics are mentioned in four provisions

Art. 3

The physician should fulfil their duties with respect for the patient regardless of their (...) genetic heritage (pl. wyposażenie genetyczne)

Art. 29

The doctor and people cooperating with them are obliged to protect the confidentiality of information contained in the genetic material of patients and their families.

Art. 38.3

The doctor is obliged to familiarize patients with the possibilities of modern medical genetics as well as diagnostics and pre-birth therapy. By providing the above information, the physician is required to inform about the risks associated with conducting pre-birth tests.

Chapter II b Human Genome

Art. 51h

1. A physician is not allowed to discriminate against people due to their genetic heritage.
2. A physician participating in procedures (badaniach) aimed at identifying whether a person is a carrier of a genetic disease or is susceptible to a genetic condition, may carry it out only for health purposes or scientific research related to them, after obtaining the consent of the patient and allowing them genetic consultation.
3. A doctor may intervene in the human genome only for preventive or therapeutic purposes in accordance with Article 46 of the Code of Medical Ethics.
4. A physician cannot participate in activities aimed at causing heritable genetic changes in humans.

As far as HE is concerned, Art. 75 concerns the issue of doping in sports

Art. 75

The doctor may not use doping products or methods for non-medical purposes. The use of products and

methods recognized as doping in people practicing sports is unethical.

Which life stage is addressed in the document?	Adults Minors (excluding newborns) Newborns Prenatal Not stated, presumably all
Which ethical challenges are addressed in the document? (you can see lists above for ideas of types)	Discrimination/ equal treatment Confidentiality of genetic information Risks associated with prenatal tests Consent in genetic research Consent in genetics in the clinic
How are the ethical issues addressed? (practically, theoretically etc? Any comment on this is fine) Are solutions offered? If so, which ones? (depending on the document, this may be redundant with above)	Rather practically (see the provisions cited above)
How is the document structured? e.g. Which format is used in the document (checklist, continuous text, other)?	It consists of 78 articles, preceded by a medical oath
Is the document clearly understandable?	Yes

<p>Why is the document important/useful for your country? why did you choose this doc? TELL US WHY IMPORTANT for country and why for SIENNA</p>	<p>It is the key document concerning medical ethics, there are no specific documents that would address the issues relevant for the project.</p>
<p>Is the document useful for the development of the SIENNA codes and other ethical frameworks? If yes, please explain.</p>	

<p>Document found via (national associations or google or another database)</p>	<p>Website of the national advisory body</p>
<p>Title of document</p>	<p>6. Statement no. 2/2012 by the Committee of Bioethics on the issue of preimplantation genetic diagnosis of 8 June 2012</p>
<p>Scope/ main topic</p>	<p>5Bi (preimplantation genetic diagnosis)</p>
<p>Kind of document (PEC or NAEG or other)</p>	<p>NAEG</p>
<p>Document developed by whom (organisation, profession)?</p>	<p>Advisory body</p>

Year the document was published	2012
Document saved in folder as	6. Stanowisk KB+Bioethics Committee+PGD+2012
Who is the stated audience, if none specified, write not stated (NS)	NS (presumably the authorities)
For Clinical or research or both or not specified	Clinical
What level of guideline is provided?	Very broad: i.e.: on many aspects of genomics or genetics, including many issues all together e.g. Very specific: only focuses on 1- 2 issues (PGD) More Theoretical (values, principles) More practical (a practitioner could apply them)
What is the scope of the document, and what are the main recommendations?	It discusses medical, ethical and legal aspects of PGD. Main recommendation is that PGD should be explicitly legalized by a law on in-vitro fertilization (such a laws was introduced in 2015)
Which life stage is addressed in the document?	Adults Minors (excluding newborns) Newborns Prenatal

<p>Which ethical challenges are addressed in the document? (you can see lists above for ideas of types)</p>	<p>Prenatal screening/testing</p>
<p>How are the ethical issues addressed? (practically, theoretically etc? Any comment on this is fine)</p> <p>Are solutions offered? If so, which ones? (depending on the document, this may be redundant with above)</p>	<p>Both practically and theoretically; specific solutions on what should and should not be allowed are provided.</p>
<p>How is the document structured? e.g. Which format is used in the document (checklist, continuous text, other)?</p>	<p>Introduction, then 15 elaborated points</p>
<p>Is the document clearly understandable?</p>	<p>Yes</p>
<p>Why is the document important/useful for your country? why did you choose this doc? TELL US WHY IMPORTANT for country and why for SIENNA</p>	<p>It is one of the few documents that address ethical issues related to genetics.</p>

Is the document useful for the development of the SIENNA codes and other ethical frameworks? If yes, please explain.	Potentially, it combines technical, ethical and legal perspectives.
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Document found via (national associations or google or another database)	Website of the national advisory body
Title of document	7. Statement no. 1/2012 by the Committee of Bioethics on the ethical problems of reproductive medicine and clinical genetics, and the need for legal regulation of 15 March 2012
Scope/ main topic	(very general, mentions different issues, see below)
Kind of document (PEC or NAEG or other)	NEAG
Document developed by whom (organisation, profession)?	National advisory body
Year the document was published	2012
Document saved in folder as	7. Stanowisko KB+Bioethics Committee+ reproductive medicine and clinical genetics+2012

Who is the stated audience, if none specified, write not stated (NS)	The authorities (the law makers)
For Clinical or research or both or not specified	Clinical
What level of guideline is provided?	<p>Very broad: i.e.: on many aspects of genomics or genetics, including many issues all together e.g.</p> <p>Very specific: only focuses on 1- 2 issues</p> <p>More Theoretical (values, principles)</p> <p>More practical (a practitioner could apply them)</p>
What is the scope of the document, and what are the main recommendations?	<p>The document is “an overview of the most important ethical problems related to reproductive medicine and genetics”.</p> <p>It expresses the need for the introduction of dedicated legislation that would address those problems, particularly in-vitro fertilization.</p> <p>In the field of genetics the Committee stated there is a need to introduce laws that would address the following issues:</p> <ul style="list-style-type: none"> - The status and the protection of human genome - Rights of patients who use genetic diagnosis and counselling - The standards of genetic diagnosis and counselling, pre-natal and pre-implantation tests, and screening - The rules on commercial genetic tests -

Which life stage is addressed in the document?	<p>Adults Minors (excluding newborns) Newborns Prenatal</p> <p>NS, but could be interpreted that all the above, due to the general character of the document.</p>
Which ethical challenges are addressed in the document? (you can see lists above for ideas of types)	<p>Genetic testing/screening Prenatal and preimplantation testing/screening</p> <p>The so-called “conscience clause” used by doctors to refuse to perform pre-natal diagnosis.</p>
<p>How are the ethical issues addressed? (practically, theoretically etc? Any comment on this is fine</p> <p>Are solutions offered? If so, which ones? (depending on the document, this may be redundant with above)</p>	<p>Both theoretically (in terms of ethical principles) and practically (in terms of the need to introduce specific laws).</p>
How is the document structured? e.g. Which format is used in the document (checklist, continuous text, other)?	<p>Short introduction an 5 points.</p>
Is the document clearly understandable?	<p>Yes</p>

<p>Why is the document important/useful for your country? why did you choose this doc? TELL US WHY IMPORTANT for country and why for SIENNA</p>	<p>It is one of the few documents that address ethical issues related to genetics.</p>
<p>Is the document useful for the development of the SIENNA codes and other ethical frameworks? If yes, please explain.</p>	<p>Potentially, it combines ethical and legal perspectives, is theoretically grounded (ethical principles) and offers concrete solutions (i.e. guidance on laws).</p>

<p>Document found via (national associations or google or another database)</p>	
<p>Title of document</p>	<p>8. A letter to the Minister of Health on the need to introduce a law on biobanks for research purposes</p>
<p>Scope/ main topic</p>	<p>5biii biobanks</p>
<p>Kind of document (PEC or NAEG or other)</p>	<p>NAEG</p>
<p>Document developed by whom (organisation, profession)?</p>	<p>The office of the Commissioner for Human Rights (the Ombudsperson)</p>
<p>Year the document was</p>	<p>2015</p>

published	
Document saved in folder as	8. Wystąpienie RPO+Ombudsperson+biobanks_2015
Who is the stated audience, if none specified, write not stated (NS)	The Minister of Health
For Clinical or research or both or not specified	Research
What level of guideline is provided?	<p>Very broad: i.e.: on many aspects of genomics or genetics, including many issues all together e.g.</p> <p>Very specific: only focuses on 1- 2 issues</p> <p>More Theoretical (values, principles)</p> <p>More practical (a practitioner could apply them)</p> <p>It focuses on the issue of biobanks.</p>
What is the scope of the document, and what are the main recommendations?	The letter addressed the need to introduce a national law on biobanking for research purposes.

Which life stage is addressed in the document?	Adults Minors (excluding newborns) Newborns Prenatal
Which ethical challenges are addressed in the document? (you can see lists above for ideas of types)	Consent in research Discrimination Return of results in research, Re-contacting (incidental findings) Data protection
How are the ethical issues addressed? (practically, theoretically etc? Any comment on this is fine) Are solutions offered? If so, which ones? (depending on the document, this may be redundant with above)	Rather theoretically, it signals problems (e.g. related to consent) but does not offer concrete solutions.
How is the document structured? e.g. Which format is used in the document (checklist, continuous text, other)?	Continuous text
Is the document clearly understandable?	Yes

<p>Why is the document important/useful for your country? why did you choose this doc?</p> <p>TELL US WHY IMPORTANT for country and why for SIENNA</p>	<p>It addresses a pressing need to introduce a specific law on biobanking for research purposes.</p>
<p>Is the document useful for the development of the SIENNA codes and other ethical frameworks? If yes, please explain.</p>	<p>n/a</p>

<p>Document found via (national associations or google or another database)</p>	
<p>Title of document</p>	<p>9. A letter to the Minister of Health on genetic counselling</p>
<p>Scope/ main topic</p>	<p>5bi genetic testing</p>
<p>Kind of document (PEC or NAEG or other)</p>	<p>NAEG</p>
<p>Document developed by whom (organisation, profession)?</p>	<p>The office of the Commissioner for Human Rights (the Ombudsperson)</p>

Year the document was published	2017
Document saved in folder as	9. Wystąpienie RPO+Ombudsperson+counselling+2017
Who is the stated audience, if none specified, write not stated (NS)	The Minister of Health
For Clinical or research or both or not specified	Clinical
What level of guideline is provided?	<p>Very broad: i.e.: on many aspects of genomics or genetics, including many issues all together e.g.</p> <p>Very specific: only focuses on 1- 2 issues</p> <p>More Theoretical (values, principles)</p> <p>More practical (a practitioner could apply them)</p> <p>Genetic counselling</p>
What is the scope of the document, and what are the main recommendations?	The document addressed the need to provide appropriate and comprehensive genetic counselling for patients (including by a psychologist, currently it is limited to consultation by a geneticist).
Which life stage is addressed in the document?	<p>Adults</p> <p>Minors (excluding newborns)</p> <p>Newborns</p> <p>Prenatal</p> <p>NS, presumably adults and minors</p>

<p>Which ethical challenges are addressed in the document? (you can see lists above for ideas of types)</p>	<p>Genetic counselling</p> <p>Problems related to the lack of appropriate information and counselling in the case of direct to consumer genetic tests.</p>
<p>How are the ethical issues addressed? (practically, theoretically etc? Any comment on this is fine)</p> <p>Are solutions offered? If so, which ones? (depending on the document, this may be redundant with above)</p>	<p>Rather theoretically, it diagnoses a problem and a need.</p>
<p>How is the document structured? e.g. Which format is used in the document (checklist, continuous text, other)?</p>	<p>Continuous text</p>
<p>Is the document clearly understandable?</p>	<p>Yes</p>
<p>Why is the document important/useful for your country? why did you choose this doc? TELL US WHY IMPORTANT for country and why for SIENNA</p>	<p>It shows how little has been done at national level to address the challenges related to genetics.</p>

<p>Is the document useful for the development of the SIENNA codes and other ethical frameworks? If yes, please explain.</p>	
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<p>Document found via (national associations or google or another database)</p>	
<p>Title of document</p>	<p>10. Code of conduct on processing of personal data for scientific purposes by biobanks in Poland</p>
<p>Scope/ main topic</p>	<p>5biii biobanks</p>
<p>Kind of document (PEC or NAEG or other)</p>	<p>PEC</p>
<p>Document developed by whom (organisation, profession)?</p>	<p>Consortium</p>
<p>Year the document was published</p>	<p>2018</p>
<p>Document saved in folder as</p>	<p>10. Kodeks postępowania BBMRIpl biobanking 2018</p>
<p>Who is the stated audience, if none specified, write not stated (NS)</p>	<p>Entities who are in charge of biobanks</p>

For Clinical or research or both or not specified	Research
What level of guideline is provided?	Very broad: i.e.: on many aspects of genomics or genetics, including many issues all together e.g. Very specific: only focuses on 1- 2 issues More Theoretical (values, principles) More practical (a practitioner could apply them)
What is the scope of the document, and what are the main recommendations?	It is a code of conduct adopted on the basis of art. 40 of GDPR according to which “Associations and other bodies representing categories of controllers or processors may prepare codes of conduct, or amend or extend such codes, for the purpose of specifying the application of this Regulation”.
Which life stage is addressed in the document?	Adults Minors (excluding newborns) Newborns Prenatal NS
Which ethical challenges are addressed in the document? (you can see lists above for ideas of types)	Ethical challenges related to the processing of personal data for research purposes, i.s. rights of individuals, particularly privacy

<p>How are the ethical issues addressed? (practically, theoretically etc? Any comment on this is fine</p> <p>Are solutions offered? If so, which ones? (depending on the document, this may be redundant with above)</p>	Practically
<p>How is the document structured? e.g. Which format is used in the document (checklist, continuous text, other)?</p>	<p>Introduction followed by 12 points aim, definitions (points 1-2) each of the further points consist of: a rule, explanations and recommendations</p>
<p>Is the document clearly understandable?</p>	Yes
<p>Why is the document important/useful for your country? why did you choose this doc? TELL US WHY IMPORTANT for country and why for SIENNA</p>	This is the one of the first codes of conducts adopted after the entry into force of the GDPR.
<p>Is the document useful for the development of the SIENNA codes and other ethical frameworks? If yes, please explain.</p>	The structure – a rule, explanations, recommendations seems useful.

7.9 South Africa

TABLE 1: INDIVIDUAL AND COUNTRY INFORMATION

Names and emails of persons who did the work (if different from above)	Jantina de Vries
Your organisation	University of Cape Town
Your country (again)	South Africa
Search conducted in which language	English (main official language of the country; all professional documents would be released in this language)
Acknowledgements (any researcher who helped you to complete this task)	None

TABLE 2: LIST OF ALL RELEVANT PROFESSIONAL ETHICS CODES

SIENNA area	Title of document (original + English translation)	URL	Year	Author/organisation	Stated audience	comments
HG	Ethics in Health Research	www.nhrec.org.za/index.php/grids-preview?download=10:doh-2015-ethics	2015	National Department of Health	All health researchers in the country	These are the South African national health research ethics guidelines which are legally binding for the country. They are enforced by the National Health Research Ethics Council. Whilst they could theoretically apply to all three topical areas, the guidelines are silent about HE and AI&R
HG (to some extent)	SA GCP Guidelines	http://www.hsrc.ac.za/uploads/pageContent/181/SAgoodclinicalpracticeguidelines.pdf	2006	National Department of Health	All those conducting clinical trials in the country	These are the South African adaptation of GCP guidelines that are also often used by researchers to inform ethical practice. Not specific to HG, AI/R or HE

All 3	HSRC Code of Research Ethics	http://www.hsrc.ac.za/en/about/research-ethics/code-of-research-ethics	2006	Human Sciences Research Council	All those conducting research with/through the HSRC	Whilst not specific to any of the topical areas, this code could inform the design of the SIENNA code in that it places important value on community engagement and social benefit of research as guiding values for research conducted in the country.
All 3	General Ethical Guidelines For Health Researchers	http://www.hpcsa.co.za/Uploads/editor/UserFiles/downloads/conduct_ethics/rules/generic_ethical_rules/booklet_6_gen_ethical_guidelines_for_researchers.pdf	2008	Health Professionals Council South Africa	All HPCSA registered professionals conducting health research in South Africa	These guidelines also emphasize the importance of research being of 'social benefit' and addressing the health needs of the country's poorest communities. These guidelines are very comprehensive and include ethical aspects of conducting research on communities, ethical aspects of using animals in research, and ethical aspects of doing research in general (e.g. do not falsify data; do not plagiarise)

<p>HG</p>	<p>Human Genetics Policy Guidelines for the Management And Prevention of Genetic Disorders, Birth Defects and Disabilities</p>	<p>http://www.geneticalliance.org.za/wp-content/uploads/2015/09/Policy-Guidelines-for-the-management-and-prevention-of-genetic-disorders-birth-defects-and-disabilities.pdf</p>	<p>2001</p>	<p>National Department of Health</p>	<p>Policy makers, health care providers etc</p>	<p>Guidelines include a section on ‘ethics guidelines’</p> <p>“Genetic counselling should be non-directive, supportive, responsive to patients’ needs, and should respect the choices of clients and families.</p> <p>Information conveyed to the patient and/or the family will be conducted in a language that is easily understood, enabling them to make informed, independent decisions. Informed consent is always to be obtained prior to investigation or treatment.”</p> <p>The rest of the document doesn’t really touch on ethics but rather explores the state of training and professional practice in medical genetics and genetic counselling in South Africa.</p>
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HG	Standards of Practice for Genetic Counsellors	http://sashg.org/wp-content/uploads/2016/09/standards_prac_genetic_counsellors_may2013.pdf	2013	Genetic Counsellors South Africa	Genetic Counsellors	<p>Document defines what genetic counsellors are and do, what training they should have, and how they should conduct themselves. It lightly touches upon ethics in the following ways:</p> <p>“Ethical Values: Can act in accordance with the ethical, legal, and philosophical principles and values of the profession; can recognize and respond to ethical and moral dilemmas arising in practice Can advocate for patients, recognize own limitations in knowledge and capabilities and seek consultation or refer and can show initiative for continued professional growth” (pg6)</p> <p>The document also outlines that genetic counsellors should ‘facilitate the informed consent process’.</p>
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HG	Code of Ethical Practice for Medical Biotechnology Research in South Africa	https://juta.co.za/support-material/resource/275/	2005	Health Professionals Council South Africa	Those conducting or regulating medical microbiotechnology research in South Africa	The document reiterates all the usual components of research ethics discussions – e.g. informed consent, benefit of the research to the SA population, integrity and competency of the researcher, privacy and confidentiality etc. The content of this document is not specific to genomics in South Africa really – it just reiterates ethics guidance for that audience.
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TABLE 5A: General questions on your document search in Human Genomics

<p>1- Did you find guidance documents specific to human genomics or genetics?</p>	<p>Not many. There was only one (from 2005) which was specific to ‘medical biotechnology’ which specifically included gene mapping, gene sequencing, development of medical genetic tests, genetic modification and cloning (but situated strongly in the kinds of discussions and concerns that prevailed in 2005). It speaks about the “Genetically Modified Organisms Act” which introduces an Executive Council that reviews anything to do with GMOs, but interestingly this Council is regulated by the Department of Agriculture and mostly regulates food (plants and animals). The University of Witwatersrand has a Biobanking Policy which I have included in the summaries below, but this is not really specific to genomics.</p>
<p>2a How many documents did you find from each of these sources that specifically address human genetics or genomics</p> <p>2b Did you find more documents than you had the time to analyse in this task?</p>	<p>PECs, NAEG, GDREC</p> <p>Total: 2 – there are no professional ethics codes or guidelines from ethics committees that are specific to genetics or genomics in South Africa. I looked up all the SOPs and Guidelines for the ethics committees of the six main universities in the country (UCT, Stellenbosch University, Pretoria, University of Johannesburg, Wits University and NorthWest University). However, I did find the two documents described in the box above, which are not specific to human genetics or genomics but do somewhat touch on these. In the summaries below I have also included the H3Africa Guidelines on Informed Consent and the Guidelines on Community Engagement, because some ethics committees (notably the UCT and Stellenbosch ctees) refer to these in their SOPs as setting the standard for consent for genomics research in the country. They are also echoed in the National Ethics Guidelines.</p> <p>No</p>
<p>3- a) How many documents specifically ONLY address high throughput genomics? By high throughput genomics, we mean using sequencing of larger parts of the genome, or exome (including large gene panels).</p> <p>3b Please list the year of publication of each documents here:</p>	<p>PECs NAEG GDREC None; see comment above.</p> <p>see table below</p>

3c please specify the topics addressed explicitly, ideally indicate the number of documents that address each, place an X beside the topic and add the number of docs

Genomics used for diagnostics in the clinic (genomic testing): the policy document that I found about genetic testing addresses this issue but does not really concern ethics. I've provided a summary in the table below

Genomics used for research: there are no South African policy or ethics documents that I could find that specifically address genomics used for research, although some consideration of genomics research is included in the National "Ethics in Health Research" guidelines that are legally binding in the country.

Consent in genomics in the clinic: the guidelines for the Genetic Counsellors that I referred to above do talk about 'consent' but just in one sentence. Other than that I have found no documents detailing how consent ought to be obtained

Consent in genomics in research: the ethics committees from Stellenbosch University and from UCT require researchers to seek separate consent (on a separate form) for genetic research when the primary research project is not a genomics one (e.g. if the primary project is a clinical trial but samples are collected for genetics research as well). Both those committees, and the National "Ethics in Research" guidelines reference the H3Africa Guidelines for Informed Consent, which are not specific to the South African context but very relevant because they have been endorsed at that level. They are included in the table below

Return of results in genomics in the clinic: no specific guidance found

Return of results in genomics in research: no specific guidance found

Reinterpretation and recontact in genomics in the clinic: no specific guidance found

Reinterpretation and recontact in genomics in the clinic: no specific guidance found

Genomics for newborn screening: the National Policy offers some insight but very summarily. Have included in the tables below. Note that South Africa does not have a newborn screening programme

Genomics for prenatal screening or testing: the National Policy offers some insight but very summarily. Have included in the tables below

Non-invasive prenatal testing/screening: the National Policy offers some insight but very summarily. Have included in the tables below

Discrimination: : no specific guidance found, but obviously an issue of huge concern in the SA context with its history of apartheid and ongoing efforts to create a fairer society where everyone has equal opportunities regardless of skin colour, sexual orientation, gender or other characteristics or traits. This concern permeates all ethics guidelines and broader policy discussions in the country and is obviously also a primary ethical concern for genomics.

Eugenics: : no specific guidance found

Justice: this is a very important concern in the SA context but again, there is no specific guidance on this. However, concerns of justice permeate all ethics documents and deliberations in SA and as such are key also to thinking about genomics.

	Genetic exceptionalism: no specific concern
<p>4 a How many documents only address genetics (so testing or screening but without a lot of sequencing)?</p>	<p>PECs: 1 (the genetic counsellors SOPs which is not really an ‘ethics code’) NAEG: 1 (the policy document which is not really an ‘ethics code’ though) GDREC</p>
<p>4b List the years of publications of each document here</p>	<p>See table above</p>
<p>3c please specify the topics addressed explicitly, ideally indicate the number of documents that address each, place an X beside the topic and add the number of docs</p>	<p>Genetics used for diagnostics in the clinic (genetic testing) 1 (the National policy document)____ Genetics used for research 1 (National Health Research Ethics Guideline, which is not specific to genomics though, but does talk about it explicitly)____ Consent in genetics in the clinic 1 (the GC code, but refers to consent only in 1 sentence)____ Consent in genetics in research 1 the National Health Research Ethics Guidelines)____ Return of results in genetics in the clinic____ Return of results in genetics in research____ Reinterpretation and recontact in genetics in the clinic____ Reinterpretation and recontact in genetics in the clinic____ newborn screening 1 (the national policy doc)____ prenatal screening or testing 1 (the national policy doc)____ Non-invasive prenatal testing/screening 1 (the national policy doc)____ Discrimination____ Eugenics____ Justice____ Genetic exceptionalism____</p>
<p>5. How many documents address both genetics and genomics?</p>	
<p>6 a) How many documents specifically address Genome Modification, gene editing, crispr, or gene therapy 6b) do they address somatic or germline</p>	<p>None that relate to humans; 2 if looking at agriculture (see comment below)</p>

editing or both?	
How many documents specifically address (so in the title of the document)	Biobanks or databases - the With University Biobanking Policy

Table 5B RELEVANT PEC or NAEG DOCUMENTS IN HUMAN GENOMICS

5Bi: genomic testing (if none on genomics, use genetic testing)

5Bii genomic screening (if none on genomics, use genetic screening)

5Biii biobanks, databases or registries

5Biv pharmacogenomics or pharmacogenetics

5Bv patents

5Bvi gene editing, crispr, or gene therapy

Document found via (national associations or google or another database)	Targeted search for University of Witwatersrand Ethics Committees
Title of document	The Human Research Ethics Committee Medical (Hrec) Principles and Policy on Biobanks University of the Witwatersrand, Johannesburg South Africa
Scope/ main topic	5Biii biobanks
Kind of document (PEC or NAEG or other)	University Policy

Document developed by whom (organisation, profession)?	By the Wits Biobanking Ethics Committee
Year the document was published	2015
Document saved in folder as	https://www.wits.ac.za/media/wits-university/research/documents/HREC%20principles%20and%20policy%20on%20biobanking.pdf
Who is the stated audience, if none specified, write not stated (NS)	
For Clinical or research or both or not specified	Research
What level of guideline is provided?	Very broad: i.e.: on many aspects of genomics or genetics, including many issues all together e.g. Very specific: only focuses on 1- 2 issues More Theoretical (values, principles) More practical (a practitioner could apply them)
What is the scope of the document, and what are the main recommendations?	The policy sets out the ethical requirements for the establishment, operation and regulation of biobanks at Wits University

<p>Which life stage is addressed in the document?</p>	<p>Adults Minors (excluding newborns) Newborns Prenatal</p> <p>Not specific to any of these</p>
<p>Which ethical challenges are addressed in the document? (you can see lists above for ideas of types)</p>	<p>Informed consent, community engagement, right to withdraw, importance of continuing to provide information post-enrollment, privacy and risk, benefits, governance and issues to do with closing down the biobank</p>
<p>How are the ethical issues addressed? (practically, theoretically etc? Any comment on this is fine</p> <p>Are solutions offered? If so, which ones? (depending on the document, this may be redundant with above)</p>	<p>As a policy document, the document is quite prescriptive and described the parameters of biobanking, e.g. people 'need to ensure some form of community engagement is conducted'. So I'd say it is quite prescriptive (even if unfortunately quite haphazard and inconsistent).</p>
<p>How is the document structured? e.g. Which format is used in the document (checklist, continuous text, other)?</p>	<p>Some continuous text and some checklists</p>
<p>Is the document clearly understandable?</p>	<p>Individual sentences are understandable; unfortunately the document is not entirely consistent and leaves lots of gaps in terms of the regulation of biobanks.</p>

<p>Why is the document important/useful for your country? why did you choose this doc? TELL US WHY IMPORTANT for country and why for SIENNA</p>	<p>It is the only such document that I am aware of in the country. This is also the only biobank-specific ethics committee – at the other universities, the main Health Research Ethics Committee includes considerations of biobanking. I know anecdotally that this committee is very actively engaged in research and often pushes back on proposals that it doesn't think match the ethical standards required. The committee strongly emphasises (in its practice if not in this document) social benefit and wants to ensure that the establishment of a biobank is not exploitative and leads to some kind of benefit for the participants and their communities. Considering all of that, I think this is an important document</p>
<p>Is the document useful for the development of the SIENNA codes and other ethical frameworks? If yes, please explain.</p>	<p>Sort of, except that this is not necessarily a very good or comprehensive document and so would be unlikely to really inform the Sienna developments. I don't think that this document says anything that other docs don't also so much more eloquently.</p>

<p>Document found via (national associations or google or another database)</p>	<p>Targeted search of the Health Professionals Council South Africa website</p>
<p>Title of document</p>	<p>Code of Ethical Practice for Medical Biotechnology Research in South Africa</p>
<p>Scope/ main topic</p>	
<p>Kind of document (PEC or NAEG or other)</p>	<p>PEC</p>

Document developed by whom (organisation, profession)?	Health Professionals Council South Africa
Year the document was published	2005
Document saved in folder as	https://juta.co.za/support-material/resource/275/
Who is the stated audience, if none specified, write not stated (NS)	NS
For Clinical or research or both or not specified	Research
What level of guideline is provided?	<p>Very broad: i.e.: on many aspects of genomics or genetics, including many issues all together e.g.</p> <p>Very specific: only focuses on 1- 2 issues</p> <p>More Theoretical (values, principles)</p> <p>More practical (a practitioner could apply them)</p>

What is the scope of the document, and what are the main recommendations?

The document is not really specific to any of the fields relevant to the SIENNA project but could be taken to inform on gene editing by proxy (bearing in mind that this document is 13 years old and responded to concerns pertinent at that time). Medical Biotechnology is defined as a “broad term for a wide range of technologies which use living organisms, biochemistries or synthetic DNA to make or modify products, improve plants or animals, or develop micro-organisms for specific uses.” In its specification of the kinds of research covered in this term are ‘gene mapping’ and ‘genetic modification’.

The document clearly positions considerations of justice and equity as central ethical concerns, describing how the dual features of a relatively strong research infrastructure (certainly when compared to other African countries) and the traditional features of a developing country (many poor people, low levels of health- and research literacy, many treatment-naïve people, high levels of infectious disease etc etc) both give great opportunities for research but also carry the risk for exploitation. This, btw, is an ethics concern that permeates all ethics docs and discussions in the country and should be central to the SIENNA framework.

Unfortunately, the document is not really specific to ‘medical microbiology’ but rather sets out conventional ethics principles that should be respected in the conduct of research. These include integrity, autonomy, beneficence, non-maleficence, justice and fairness. The ‘ethical duties’ identified include: respect for the law; relevance of the research; competence of the researchers; informed consent; privacy and confidentiality; and intellectual property and commercialisation. But all of these are extremely generic and not specific to genomics.

Which life stage is addressed in the document?	Adults Minors (excluding newborns) Newborns Prenatal Not specified
Which ethical challenges are addressed in the document? (you can see lists above for ideas of types)	See above
How are the ethical issues addressed? (practically, theoretically etc? Any comment on this is fine) Are solutions offered? If so, which ones? (depending on the document, this may be redundant with above)	Not really addressed
How is the document structured? e.g. Which format is used in the document (checklist, continuous text, other)?	Continuous text
Is the document clearly understandable?	Yes

<p>Why is the document important/useful for your country? why did you choose this doc? TELL US WHY IMPORTANT for country and why for SIENNA</p>	<p>Somewhat. On the one hand it is one of two documents specific to technologies involving genetics, on the other the text is so generic to research ethics that it doesn't really engage with the particular ethical issues raised by genomics.</p>
<p>Is the document useful for the development of the SIENNA codes and other ethical frameworks? If yes, please explain.</p>	<p>I don't think so, other than introducing the value of justice perhaps.</p>

<p>Document found via (national associations or google or another database)</p>	<p>Targeted search of the website of the South African Society for Human Genetics (SASHG) and conversation with genetic counselling colleagues at UCT</p>
<p>Title of document</p>	<p>Standards of Practice for Genetic Counsellors</p>
<p>Scope/ main topic</p>	<p>Genetic Counselling</p>
<p>Kind of document (PEC or NAEG or other)</p>	<p>other</p>
<p>Document developed by whom (organisation, profession)?</p>	<p>Genetic Counsellors South Africa</p>

Year the document was published	2013
Document saved in folder as	http://sashg.org/wp-content/uploads/2016/09/standards_prac_genetic_counsellors_may2013.pdf
Who is the stated audience, if none specified, write not stated (NS)	Genetic Counsellors
For Clinical or research or both or not specified	Clinic
What level of guideline is provided?	<p>Very broad: i.e.: on many aspects of genomics or genetics, including many issues all together e.g.</p> <p>Very specific: only focuses on 1- 2 issues</p> <p>More Theoretical (values, principles)</p> <p>More practical (a practitioner could apply them)</p>

<p>What is the scope of the document, and what are the main recommendations?</p>	<p>This is mostly describing the roles and duties of genetic counsellors in South Africa, including a detailed description of what GC training should involve in order for persons to be allowed to register with the HPCSA, what the parameters are for exchange with GCs from Australia and the UK etc. It only includes mention of ethics in two places, namely:</p> <p>“Ethical Values:</p> <ul style="list-style-type: none"> - Can act in accordance with the ethical, legal, and philosophical principles and values of the profession; can recognize and respond to ethical and moral dilemmas arising in practice - Can advocate for patients, recognize own limitations in knowledge and capabilities and seek consultation or refer and can show initiative for continued professional growth” (pg6) <p>The document also outlines that genetic counsellors should ‘facilitate the informed consent process’ but does not outline how they should go about this.</p>
<p>Which life stage is addressed in the document?</p>	<p>Adults Minors (excluding newborns) Newborns Prenatal</p> <p>Not specific to any of these</p>
<p>Which ethical challenges are addressed in the document? (you can see lists above for ideas of types)</p>	<p>None other than the three listed above (to act in accordance with ethics of the profession; to be allowed to advocate on behalf of patients, and to seek informed consent).</p>

<p>How are the ethical issues addressed? (practically, theoretically etc? Any comment on this is fine</p> <p>Are solutions offered? If so, which ones? (depending on the document, this may be redundant with above)</p>	Not addressed, just mentioned
<p>How is the document structured? e.g. Which format is used in the document (checklist, continuous text, other)?</p>	Continuous text
<p>Is the document clearly understandable?</p>	Yes
<p>Why is the document important/useful for your country? why did you choose this doc? TELL US WHY IMPORTANT for country and why for SIENNA</p>	Not really
<p>Is the document useful for the development of the SIENNA codes and other ethical frameworks? If yes, please explain.</p>	Not really

Document found via (national associations or google or another database)	Google
Title of document	Human Genetics Policy Guidelines for the Management And Prevention of Genetic Disorders, Birth Defects and Disabilities
Scope/ main topic	Diagnostic genetic testing
Kind of document (PEC or NAEG or other)	National Health Policy
Document developed by whom (organisation, profession)?	National Department of Health
Year the document was published	2001
Document saved in folder as	http://www.geneticalliance.org.za/wp-content/uploads/2015/09/Policy-Guidelines-for-the-management-and-prevention-of-genetic-disorders-birth-defects-and-disabilities.pdf
Who is the stated audience, if none specified, write not stated (NS)	NS (but as a health policy document it would likely be those involved in health resource allocation decisions, medical genetics professionals and the like).
For Clinical or research or both or not specified	Clinical

<p>What level of guideline is provided?</p>	<p>Very broad: i.e.: on many aspects of genomics or genetics, including many issues all together e.g. Very specific: only focuses on 1- 2 issues More Theoretical (values, principles) More practical (a practitioner could apply them)</p> <p>This guideline specifically talks about challenges and opportunities for offering diagnostic genetic testing services in the country</p>
<p>What is the scope of the document, and what are the main recommendations?</p>	<p>See above. It recommends that genetic testing services should be made more widely available in the country, and that there is an urgent need to train medical genetics professionals (both doctors and counsellors) to provide these services. It recommends that posts be made available for genetic counsellors.</p> <p>I don't think that any or many of these recommendations were followed up as the situation set out in the report is still pretty much the same today.</p>
<p>Which life stage is addressed in the document?</p>	<p>Adults Minors (excluding newborns) Newborns Prenatal</p> <p>All. It talks about prenatal, newborn, paediatric and adult genetic testing.</p>

<p>Which ethical challenges are addressed in the document? (you can see lists above for ideas of types)</p>	<p>The document includes the following paragraph on ethics: “The use of genetic information has major ethical implications for affected individuals and those related to them. Ethical guidelines adapted from the WHO document “Hereditary Disease Programme 1995” and other relevant publications, have been included in the document. Genetic counselling should be non-directive, supportive, responsive to patients’ needs, and should respect the choices of clients and families. Information conveyed to the patient and/or the family will be conducted in a language that is easily understood, enabling them to make informed, independent decisions. Informed consent is always to be obtained prior to investigation or treatment.”</p> <p>It recommends that the Medical Genetics Advisory Board includes one expert in ‘law/ethics’. I could find no information about this Board, including its membership and activities.</p>
<p>How are the ethical issues addressed? (practically, theoretically etc? Any comment on this is fine)</p> <p>Are solutions offered? If so, which ones? (depending on the document, this may be redundant with above)</p>	<p>Not addressed just mentioned</p>
<p>How is the document structured? e.g. Which format is used in the document (checklist, continuous text, other)?</p>	<p>Cts txt</p>

Is the document clearly understandable?	Yes
Why is the document important/useful for your country? why did you choose this doc? TELL US WHY IMPORTANT for country and why for SIENNA	Not really
Is the document useful for the development of the SIENNA codes and other ethical frameworks? If yes, please explain.	Not really

Document found via (national associations or google or another database)	I know this document as it is the national research ethics guideline for the country
Title of document	Ethics in Health Research
Scope/ main topic	Not specific; generic about research ethics (but includes some mention of genetics/genomics)
Kind of document (PEC or NAEG or other)	NAEG

Document developed by whom (organisation, profession)?	National Department of Health
Year the document was published	2015
Document saved in folder as	www.nhrec.org.za/index.php/grids-preview?download=10:doh-2015-ethics
Who is the stated audience, if none specified, write not stated (NS)	All health researchers in the country
For Clinical or research or both or not specified	Research
What level of guideline is provided?	<p>Very broad: i.e.: on many aspects of genomics or genetics, including many issues all together e.g.</p> <p>Very specific: only focuses on 1- 2 issues</p> <p>More Theoretical (values, principles)</p> <p>More practical (a practitioner could apply them)</p> <p>Not sure how to answer; document describes general health research in the country, with specific mention of genomics and biobanking.</p>
What is the scope of the document, and what are the main recommendations?	The Guidelines refer to the Nagoya Protocol on Access to Genetic Resources and the Fair and Equitable Sharing of Benefits, which was ratified by South Africa in 2011 and should therefore be taken to inform on issues relating to human genomics in South Africa.

Which life stage is addressed in the document?	Adults Minors (excluding newborns) Newborns Prenatal
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Which ethical challenges are addressed in the document? (you can see lists above for ideas of types)

The report draws attention to these ethical issues:
 Genetic privacy of children, with a recommendation that their interests ‘may be more important than those of adults who manifest a particular genetic condition’
 Risks to confidentiality, although the report describes that ‘genetic markers make it possible to identify groups rather than individuals’
 Consent (see below);
 Potential for harm e.g. through stigmatisation or discrimination
 Potential that genetics research may reveal (sensitive) information about group ancestry or about families;
 Potential for pertinent individual genetic research results

W.r.t. Consent the document describes that:

- “if samples or data are stored without explicit consent for re-use, then researchers need to seek permission from a REC for re-use. The REC may require researchers to obtain new consent for the proposed re-use”;
- “in the case of genetic or genomic research, information should be provided about the implications of genetic testing (e.g. paternity determinations, insurance risks, reproduction decisions) and associated confidentiality risks”
- “the purpose and nature of a repository, including the specifics for which consent is being sought, how a repository works and the types of research it supports” (NOTE the doc does not use the word ‘biobank’ but rather talks about ‘repository’)
- “the conditions and requirements under which data or material will be shared with other researchers”
- “the nature and extent of specific risks of harm related to use and storage of material or data, especially if identifiers are retained”

<p>How are the ethical issues addressed? (practically, theoretically etc? Any comment on this is fine</p> <p>Are solutions offered? If so, which ones? (depending on the document, this may be redundant with above)</p>	<p>Proposals submitted to RECs must include a plan for how information revealed by genetic research will be managed. This plan needs to be explained to participants.</p> <p>About individual research results: “Plans to share findings with participants must include opportunities for participants to choose whether they wish to receive the information personally, and whether the information may be shared with biological relatives. Genetic counselling must be available if findings will be disclosed to participants.”</p>
<p>How is the document structured? e.g. Which format is used in the document (checklist, continuous text, other)?</p>	<p>Ctd txt</p>
<p>Is the document clearly understandable?</p>	<p>Yes</p>
<p>Why is the document important/useful for your country? why did you choose this doc? TELL US WHY IMPORTANT for country and why for SIENNA</p>	<p>As the main ethics guideline for the country, which is legally binding, this document is important in the country. Also, its specific mention of genetics and genomics (albeit in short paragraphs) also make it relevant</p>

Is the document useful for the development of the SIENNA codes and other ethical frameworks? If yes, please explain.	Somewhat – but doesn't really raise ethical challenges that other documents do not also raise.
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Document found via (national associations or google or another database)	I was central to the development of this document
Title of document	Ethics and Governance Framework for Best Practice in Genomic Research and Biobanking in Africa
Scope/ main topic	(choose from list above, 5Bi-5Bvi, write out the topic)
Kind of document (PEC or NAEG or other)	Ethics framework (guideline)
Document developed by whom (organisation, profession)?	The H3Africa Working Group on Ethics, endorsed by the H3Africa Steering Committee
Year the document was published	2017
Document saved in folder as	https://h3africa.org/9-news/361-framework-for-african-genomics-and-biobanking

Who is the stated audience, if none specified, write not stated (NS)	stakeholders involved in the design, conduct, participation, regulation and ethical review of genomic research and biobanking across the African continent
For Clinical or research or both or not specified	Research
What level of guideline is provided?	Very broad: i.e.: on many aspects of genomics or genetics, including many issues all together e.g. Very specific: only focuses on 1- 2 issues More Theoretical (values, principles) More practical (a practitioner could apply them)

What is the scope of the document, and what are the main recommendations?	<p>“The purpose of this Framework is to provide a principled and practical approach to promote best practice for genomic research and biobanking in Africa. In recognising that the standards and principles outlined here may not be currently achievable in all countries and may need to be adapted to the local context, its primary goals are to:</p> <ul style="list-style-type: none">• Promote responsible conduct of genomic research and biobanking in Africa that fosters shared decision making, accountability, transparency and fairness;• Guide development of national regulation for genomic research and biobanking in African countries that will promote social value and maximize benefits to African scientists and nationals who engage in biobanking and genomics research;• Provide a framework for evaluating the ethical soundness of genomic research and biobanking by ethics review committees across the African continent.”
Which life stage is addressed in the document?	Adults Minors (excluding newborns) Newborns Prenatal

Which ethical challenges are addressed in the document? (you can see lists above for ideas of types)

The document outlines a number of key principles and ethics elements that should be considered when conducting genomics research and biobanking in the African continent. Primarily, the focus is on ensuring fair and equitable collaborations that lead to the development of sustainable and independent research capacity on the African continent. Importantly, the document recommends 'genuine African intellectual leadership' over projects to ensure that African researchers are not just sample collectors but lead research.

The document recommends that broad consent is a viable consent option for the African continent, but is only sustainable and ethical if accompanied by genuine community engagement and a governance framework that takes into account rights of participants and their needs.

The core principles of the Framework are:

“* Research should be sensitive to and respectful of African values and cultures;

- Research should be for the benefit of African people recognizing that it likely also yields benefits to the global population;
- Research and the dissemination of data in publications should take place with genuine and active intellectual participation of African investigators and other African stakeholders;
- Research should promote ways of relating typified by respect for individuals and communities, fairness, equity and reciprocity.”

The range of issues considered include:

Consent

Community Engagement

Ethics Review

Avoiding group harm or stigma

Benefit sharing

Capacity building

International collaboration and export of samples

Feedback of individual genetic research results

Good governance

<p>How are the ethical issues addressed? (practically, theoretically etc? Any comment on this is fine</p> <p>Are solutions offered? If so, which ones? (depending on the document, this may be redundant with above)</p>	<p>The framework blends practical recommendations with ethical or normative observations. I have already described some practical solutions (broad consent, African intellectual leadership, genuine CE). Other recommendations are that a project should be undertaken to explore challenges and opportunities for feeding back individual research results (now being done by IFGENERA, an H3Africa Project) etc etc. The document isn't very long and it makes no sense for me to copy and paste it here – may be good to consult the primary source</p>
<p>How is the document structured? e.g. Which format is used in the document (checklist, continuous text, other)?</p>	<p>Ctd txt</p>
<p>Is the document clearly understandable?</p>	<p>Yes</p>
<p>Why is the document important/useful for your country? why did you choose this doc? TELL US WHY IMPORTANT for country and why for SIENNA</p>	<p>Yes – although not relevant to the SA research context only, several ethics committee across the country refer to H3A and to the Framework for their ethical principles.</p>

Is the document useful for the development of the SIENNA codes and other ethical frameworks? If yes, please explain.	Yes, I'd say this is very relevant as it is the result of very extensive consultation with over 100 members of African ethics committees, national ethics councils and policymakers from close to 30 countries over three years. The framework also received some international coverage, for instance in Nature (https://www.nature.com/articles/d41586-018-04685-1 and https://www.nature.com/articles/d41586-018-04589-0) and from the Global Alliance for Genomics and Health (https://www.ga4gh.org/news/ffY_2As3Q-iv39gB2Un4Bw.article).
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Document found via (national associations or google or another database)	I know this doc as I wrote it
Title of document	H3Africa Guidelines for Informed Consent
Scope/ main topic	Genomics
Kind of document (PEC or NAEG or other)	Guidelines
Document developed by whom (organisation, profession)?	H3Africa (developing by the H3Africa Working Group on Ethics)
Year the document was published	2013
Document saved in folder as	https://h3africa.org/images/PDF/H3A%20WG%20Guidelines%20Informed%20Consent_FINAL_01082013.pdf

Who is the stated audience, if none specified, write not stated (NS)	All those involved in genomics research on the continent
For Clinical or research or both or not specified	research
What level of guideline is provided?	<p>Very broad: i.e.: on many aspects of genomics or genetics, including many issues all together e.g.</p> <p>Very specific: only focuses on 1- 2 issues</p> <p>More Theoretical (values, principles)</p> <p>More practical (a practitioner could apply them)</p>
What is the scope of the document, and what are the main recommendations?	The document offers guidance on how to seek informed consent for African genomics research and biobanking. Its main recommendation is that there is no a priori reason to assume that broad consent should not be used in the African context. It proposes that whilst tiered consent could be used, there are some concerns about its practical implementation.
Which life stage is addressed in the document?	<p>Adults</p> <p>Minors (excluding newborns)</p> <p>Newborns</p> <p>Prenatal</p> <p>Not specific</p>
Which ethical challenges are addressed in the document? (you can see lists above for ideas of types)	Informed consent and the issues associated with it.

<p>How are the ethical issues addressed? (practically, theoretically etc? Any comment on this is fine</p>	<p>The document offers practical guidance about how to seek informed consent for genomics research. The 2013 document was the first edition; the guidelines have since been adapted twice and the third edition is now available (but I see that the 1st edition is still on the H3A website, will seek to have that corrected).</p>
<p>Are solutions offered? If so, which ones? (depending on the document, this may be redundant with above)</p>	<p>It would go a bit too far to summarise the entire document here, but this is what I think the H3A recommendation for informed consent in Africa is: That there is no reason that broad consent shouldn't or couldn't work on the African continent, and that many of the known challenges relating to seeking informed consent are equally true regardless of the consent model used; That it is, however, imperative that broad consent is used in conjunction with a governance framework that protects the interests of research participants AND that ensures that re-use is fair, beneficial and equitable; It is also imperative that broad consent be used in conjunction with 'genuine' community engagement (where 'genuine' is used to distinguish it from 'tokenistic' engagement).</p> <p>It is the emphasis on the incorporation of ethical values in the governance of genomics research that led us to develop the Ethics and Governance Framework – in the H3A thinking, these two things need to go hand in hand.</p> <p>H3Africa also published a paper where it analysed practical examples of the way in which H3Africa researchers sought consent. It found that the majority of projects had in fact used broad consent and that only one of 16 projects included clear information about the return of individual genetic research results.</p>

<p>How is the document structured? e.g. Which format is used in the document (checklist, continuous text, other)?</p>	<p>Ctd txt</p>
<p>Is the document clearly understandable?</p>	<p>Yes</p>
<p>Why is the document important/useful for your country? why did you choose this doc? TELL US WHY IMPORTANT for country and why for SIENNA</p>	<p>Yes in the sense that both the National Ethics in Health Research guidelines make reference to these, and because at least 2 ethics committees in the country reference these guidelines as the 'gold standard' for genomics research.</p>
<p>Is the document useful for the development of the SIENNA codes and other ethical frameworks? If yes, please explain.</p>	<p>Somewhat useful perhaps – although I don't think that the guidelines say anything that other guidelines don't also say or recommend.</p>

Table 5C GDREC DOCUMENTS for HUMAN GENOMICS (Sorry I listed them all above here)

Document found via (national associations or google or another database)	
Title of document	
Scope/ main topic	(choose from list above, 5Bi-5Bvi, write out the topic)
Kind of document (PEC or NAEG or other)	
Document developed by whom (organisation, profession)?	
Year the document was published	
Document saved in folder as	
Who is the stated audience, if none specified, write not stated (NS)	
For Clinical or research or both or not specified	

What level of guideline is provided?	Very broad: i.e.: on many aspects of genomics or genetics, including many issues all together e.g. Very specific: only focuses on 1- 2 issues More Theoretical (values, principles) More practical (a practitioner could apply them)
What is the scope of the document, and what are the main recommendations?	
Which life stage is addressed in the document?	Adults Minors (excluding newborns) Newborns Prenatal
Which ethical challenges are addressed in the document? (you can see lists above for ideas of types)	

<p>How are the ethical issues addressed? (practically, theoretically etc? Any comment on this is fine</p> <p>Are solutions offered? If so, which ones? (depending on the document, this may be redundant with above)</p>	
<p>How is the document structured? e.g. Which format is used in the document (checklist, continuous text, other)?</p>	
<p>Is the document clearly understandable?</p>	
<p>Why is the document important/useful for your country? why did you choose this doc? TELL US WHY IMPORTANT for country and why for SIENNA</p>	
<p>Is the document useful for the development of the SIENNA codes and other ethical frameworks? If yes, please explain.</p>	

7.10 Spain

TABLE 1: INDIVIDUAL AND COUNTRY INFORMATION

Names and emails of persons who did the work (if different from above)	
Your organisation	University of Granada (Spain)
Your country (again)	Spain
Search conducted in which language	Spanish
Acknowledgements (any researcher who helped you to complete this task)	

TABLE 2: LIST OF ALL RELEVANT PROFESSIONAL ETHICS CODES

SIENNA area	Title of document (original + English translation)	URL	Year	Author/organisation	Stated audience	Comments
HG	CÓDIGO DE DEONTOLOGÍA MÉDICA-GUÍA DE ÉTICA MÉDICA (Medical Deontology Code-Guide to medical ethics)	https://www.cgcom.es/sites/default/files/codigo_deontologia_medica.pdf (cap. XIII).	2011	Consejo general de colegios oficiales de medicos	Dr. Serafín Romero	
HG	CÓDIGO DE BUENAS PRÁCTICAS EN INVESTIGACIÓN (Code of good research practices)	http://diposit.ub.edu/dspace/bitstream/2445/28543/1/codibonpractiques_spa.pdf		Universidad de Barcelona	Vicerrectorado de política científica	
HG	CÓDIGO DE BUENAS PRÁCTICAS CIENTÍFICAS (Code of good scientific practice)	http://www.prbb.org/system/uploads/attachment_data/file/3/es/CBPC_PRBB_CAT_CAST_ENG.PDF	2009	Parque de investigación biomédica de Barcelona		

HG	<p>PROCEDIMIENTO DE TRABAJO DEL COMITÉ DE ÉTICA PARA LA INVESTIGACIÓN CON AGENTES BIOLÓGICOS Y ORGANISMOS GENÉTICAMENTE MODIFICADOS (Working procedure of the Ethics Committee for Research with biological agents and genetically modified organisms)</p>	<p>https://www.ehu.eus/documentos/2458096/3362171/PNT_CEIA_B</p>	2017	Universidad del País Vasco	Vicerrectorado de Investigación	
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TABLE 3: LIST OF ALL RELEVANT DOCUMENTS FROM NATIONAL ADVISORY/ETHICS GROUPS

SIENNA area	Title of document (original + English translation)	URL	Year	Author/organization	Stated audience	comments
HG	DOCUMENTO SOBRE BIOÉTICA Y EDICIÓN GENÓMICA EN HUMANOS (Document on bioethics and gene editing in humans)	http://www.publicacions.ub.edu/refs/observatoriBioEticaDret/documents/08543.pdf	2016	Observatorio de Bioética y Derecho (Univeridad de Barcelona)	Dra. María Casado	
HG	CÓDIGO DE BUENAS PRÁCTICAS APLICABLES A BIOBANCOS DE INVESTIGACIÓN BIOMÉDICA EN ESPAÑA (Code of good practices applicable to biobanks of biomedical research in Spain)	http://imas12.es/wp-content/uploads/2015/documentacion/GBP%20Biobancos.pdf	2012	Instituto de salud Carlos III	Isabel Novoa / Manuel Morente	
HG	INFORME SOBRE EL CONSEJO GENÉTICO PRENATAL (Report on prenatal Genetic advice)	http://assets.comitedebioetica.es/files/documentacion/consejo-genetico-prenatal.pdf		Comité de bioética de España	M ^a Teresa López López	

HG	CLONACIÓN TERAPÉUTICA: PERCPECTIVAS ÉTICAS, LEGALES Y CIENTÍFICAS (Therapeutic cloning: ethical, legal and scientific perspectives)	https://www.fundaciogrifols.org/documentos/4662337/4689111/informe3.pdf/651acbf-2068-4a74-a37c-8d0b037728e8		Fundación Grífols i Lucas	Víctor	Gemma Marfany, Josep Egozcue y Victòria Camps	
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TABLE 4: LIST OF ALL RELEVANT GUIDANCE DOCUMENTS ON HOW TO WRITE RESEARCH ETHICS PROTOCOLS

Name of national REC	Title of document (original + English translation)	SIENNA area	URL	Stated audience	Comments
Comité de Bioética de España	RECOMENDACIONES DEL COMITÉ DE BIOÉTICA DE ESPAÑA CON RELACIÓN AL IMPULSO E IMPLANTACIÓN DE BUENAS PRÁCTICAS CIENTÍFICAS EN ESPAÑA (Recommendations of the bioethics committee of Spain in relation to the impulse and implantation of good scientific practices in Spain)	HG	http://assets.comitedebioetica.es/files/documentacion/buenas_practicas_cientificas_cbe_2011.pdf	Dra. Victoria Camps	

Please choose for every area the most relevant documents and fill out the TABLES below.

HUMAN GENOMICS

TABLE 5A: General questions on your document search in Human Genomics

1- Did you find guidance documents specific to human genomics or genetics?	Yes No
2a How many documents did you find from each of these sources that specifically address human genetics or genomics 2b Did you find more documents than you had the time to analyse in this task?	PECs 4 NAEG 4 GDREC 1 Total: 9 Yes, roughly how many more documents than those listed here did you find? _____ No

<p>3- a) How many documents specifically ONLY address high throughput genomics? By high throughput genomics, we mean using sequencing of larger parts of the genome, or exome (including large gene panels).</p> <p>3b Please list the year of publication of each documents here:</p> <p>3c please specify the topics addressed explicitly, ideally indicate the number of documents that address each, place an X beside the topic and add the number of docs</p>	<p>PECs NAEG GDREC</p> <p>Genomics used for diagnostics in the clinic (genomic testing) X2 Genomics used for research X2 Consent in genomics in the clinic Consent in genomics in research Return of results in genomics in the clinic Return of results in genomics in research Reinterpretation and recontact in genomics in the clinic Reinterpretation and recontact in genomics in the clinic Genomics for newborn screening Genomics for prenatal screening or testing x1 Non-invasive prenatal testing/screening Descrimination Eugenics Justice Genetic exceptionalism</p>
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<p>4 a How many documents only address genetics (so testing or screening but without a lot of sequencing)?</p> <p>4b List the years of publications of each document here</p> <p>3c please specify the topics addressed explicitly, ideally indicate the number of documents that address each, place an X beside the topic and add the number of docs</p>	<p>PECs 1 NAEG 4 GDREC</p> <p>2017, 2016, 2012</p> <p>Genetics used for diagnostics in the clinic (genetic testing) <u> X 2 </u> Genetics used for research <u> X 2 </u> Consent in genetics in the clinic <u> </u> Consent in genetics in research <u> </u> Return of results in genetics in the clinic <u> </u> Return of results in genetics in research <u> </u> Reinterpretation and recontact in genetics in the clinic <u> </u> Reinterpretation and recontact in genetics in the clinic <u> </u> newborn screening <u> </u> prenatal screening or testing <u> x1 </u> Non-invasive prenatal testing/screening <u> </u> Discrimination <u> </u> Eugenics <u> </u> Justice <u> </u> Genetic exceptionalism <u> </u></p>
<p>5. How many documents address both genetics and genomics?</p>	<p>Specify docs or not? 3--NO</p>

<p>6 a) How many documents specifically address Genome Modification, gene editing, crispr, or gene therapy</p> <p>6b) do they address somatic or germline editing or both?</p>	<p>3</p>
<p>How many documents specifically address (so in the title of the document)</p>	<p>Genetic or genomic testing 3</p> <p>Genetic or genomic screening</p> <p>Biobanks or databases 1</p> <p>Pharmacogenomics or pharmacogenetics</p> <p>Patents</p>

Table 5B RELEVANT PEC or NAEG DOCUMENTS IN HUMAN GENOMICS,

5Bi: genomic testing (if none on genomics, use genetic testing)

5Bii genomic screening (if none on genomics, use genetic screening)

5Biii biobanks, databases or registries

5Biv pharmacogenomics or pharmacogenetics

5Bv patents

5Bvi gene editing, crispr, or gene therapy

Document found via (national associations or google or another database)	national associations
Title of document	DOCUMENTO SOBRE BIOÉTICA Y EDICIÓN GENÓMICA EN HUMANOS (Document on bioethics and gene editing in humans)
Scope/ main topic	Gene editing
Kind of document (PEC or NAEG or other)	NAEG
Document developed by whom (organisation, profession)?	Observatorio de bioética y derecho
Year the document was published	2016

Document saved in folder as	“NAEG Edición genética” PDF
Who is the stated audience, if none specified, write not stated (NS)	Dra. María Casado
For Clinical or research or both or not specified	Research
What level of guideline is provided?	<p>Very broad: i.e.: on many aspects of genomics or genetics, including many issues all together e.g.</p> <p>Very specific: only focuses on 1- 2 issues</p> <p>More Theoretical (values, principles)</p> <p>More practical (a practitioner could apply them)</p>

What is the scope of the document, and what are the main recommendations?

SCOPE: The current development of biotechnology permits us to structurally alter the genetic background of living beings, humans included; this has generated expectations, fears and many questions. Although public policies, regulations and economic decisions have been adopted in Spain and the European Union that tend to promote biomedical research, the enormous potentiality of genome editing techniques, including the CRISPR technique, demand additional reflection and debate that will allow us to articulate an adequate ethical-legal framework.

RECOMMEDATIONS:

Within the gradualist position that we defend in our documents and a framework based on respect for the precautionary principle, we believe that genome editing techniques should proceed in phases

With the aim of making the previous recommendation feasible, it is necessary to analyse and revise current international, national and regional regulations, updating them for the possible use of gene editing techniques under certain requirements and with due guarantees. In Spain, this will require revising the penal code

Decisions on the development of gene editing research and its application in any field – and especially in human beings – cannot remain in the hands of economic and financial powers, but must be guided by the idea of the common good and improving quality of life.

Gene editing techniques make it necessary to have public policies to determine research priorities and resources, in addition to ensuring transparency, accountability and control.

The system for the evaluation and control of research must be truly effective; thus, ethics committees and other existing guarantees must really serve to evaluate the scientific and methodological implications of the research, as well as its ethical, legal and social implications.

The media and the public must be involved in an inclusive, forward-looking and informed social debate, which will foster public research policy based on a respect for human rights and oriented toward justice and equality.

<p>Which life stage is addressed in the document?</p>	<p>Adults Minors (excluding newborns) Newborns Prenatal</p>
<p>Which ethical challenges are addressed in the document? (you can see lists above for ideas of types)</p>	<p>The objective is to revive a debate that had been considered purely speculation or even science fiction, rather than a real scientific possibility. The challenge is referring to whether or not the path toward the genetic modification of the human embryonic germ line should be initiated, thus modifying the genetic characteristics of future generations.</p>
<p>How are the ethical issues addressed? (practically, theoretically etc? Any comment on this is fine)</p> <p>Are solutions offered? If so, which ones? (depending on the document, this may be redundant with above)</p>	<p>Both</p>
<p>How is the document structured? e.g. Which format is used in the document (checklist, continuous text, other)?</p>	<p>continuous text</p>
<p>Is the document clearly understandable?</p>	<p>Yes</p>

<p>Why is the document important/useful for your country? why did you choose this doc? TELL US WHY IMPORTANT for country and why for SIENNA</p>	<p>Because we found a good description of the state of the question in Spain, the characteristics of this technique are identified over previous techniques for genetic modification and give it a potential and interest unknown until now, and specific recommendations are presented.</p>
<p>Is the document useful for the development of the SIENNA codes and other ethical frameworks? If yes, please explain.</p>	<p>Probably, for all the above mentioned</p>

<p>Document found via (national associations or google or another database)</p>	<p>Google</p>
<p>Title of document</p>	<p>CÓDIGO DE BUENAS PRÁCTICAS APLICABLES A BIOBANCOS DE INVESTIGACIÓN BIOMÉDICA EN ESPAÑA (Code of good practices applicable to biobanks of biomedical research in Spain)</p>
<p>Scope/ main topic</p>	<p>Biobanks</p>
<p>Kind of document (PEC or NAEG or other)</p>	<p>NAEG</p>
<p>Document developed by whom (organisation, profession)?</p>	<p>Instituto de Salud Carlos III</p>
<p>Year the document was published</p>	<p>2012</p>

Document saved in folder as	"NAEG Biobancos" PDF
Who is the stated audience, if none specified, write not stated (NS)	Isabel Novoa / Manuel Morente
For Clinical or research or both or not specified	Both
What level of guideline is provided?	<p>Very broad: i.e.: on many aspects of genomics or genetics, including many issues all together e.g.</p> <p>Very specific: only focuses on 1- 2 issues</p> <p>More Theoretical (values, principles)</p> <p>More practical (a practitioner could apply them)</p>

<p>What is the scope of the document, and what are the main recommendations?</p>	<p>SCOPE: This Code of good practice aims to establish a consensus guide of procedures, based on experience and framed by the current ethical-legal norms, which marks the operating guidelines of biobanks for biomedical research. Therefore, this document is applicable to all biobanks staff of biomedical research as well as health institutions staff or of research centers that during the development of their professional activity carry out biomedical researches with biological material and/or data of human origin.</p> <p>RECOMMENDATIONS: The management of the centers with biomedical research biobanks belonging to the National network of Biobanks (see www.redbiobancos.es) should make possible the dissemination and application of this code of good practices among its staff. It is also recommended that a link to this code of good practice be included on the website of the biobanks to facilitate their availability and consultation.</p>
<p>Which life stage is addressed in the document?</p>	<p>Adults Minors (excluding newborns) Newborns Prenatal</p>
<p>Which ethical challenges are addressed in the document? (you can see lists above for ideas of types)</p>	<p>How to protect the rights of individuals whose biological samples are to be included in a collection. To do so, it is crucial to inform the donor adequately regarding the purpose and use of the biological samples ceded, as well as obtaining informed consent.</p>

<p>How are the ethical issues addressed? (practically, theoretically etc? Any comment on this is fine</p> <p>Are solutions offered? If so, which ones? (depending on the document, this may be redundant with above)</p>	<p>Practical issues</p>
<p>How is the document structured? e.g. Which format is used in the document (checklist, continuous text, other)?</p>	<p>continuous text</p>
<p>Is the document clearly understandable?</p>	<p>Yes</p>
<p>Why is the document important/useful for your country? why did you choose this doc? TELL US WHY IMPORTANT for country and why for SIENNA</p>	<p>Because the document provides an overview of Spanish codes on the issue. For the elaboration of this Code of good practice, the recommendations of the Bioethics Committee of Spain have been taken into account (see www.comitedebioetica.es/documentacion/index.php), and the following codes of good practice:</p> <ul style="list-style-type: none"> • Código de buenas prácticas científicas del Instituto de Salud Carlos III (see www.isciii.es/htdocs/terapia/terapia_comiteetica.jsp) • Documento de Guía de Buenas Prácticas de Biobancos de Investigación Biomédica de la Red de Entidades Gestoras de Investigación Clínica (REGIC, www.regic.org).

Is the document useful for the development of the SIENNA codes and other ethical frameworks? If yes, please explain.	Probably, for all the above mentioned.
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Document found via (national associations or google or another database)	national associations
Title of document	INFORME SOBRE EL CONSEJO GENÉTICO PRENATAL (Report on prenatal Genetic advice)
Scope/ main topic	Prenatal diagnosis
Kind of document (PEC or NAEG or other)	NAEG
Document developed by whom (organisation, profession)?	Comité de bioética de España
Year the document was published	
Document saved in folder as	NAEG consejo-genetico-prenatal
Who is the stated audience, if none specified, write not stated (NS)	M ^a Teresa López López

For Clinical or research or both or not specified	Clinical
What level of guideline is provided?	Very broad: i.e.: on many aspects of genomics or genetics, including many issues all together e.g. Very specific: only focuses on 1- 2 issues More Theoretical (values, principles) More practical (a practitioner could apply them)

What is the scope of the document, and what are the main recommendations?

SCOPE:

The volume of the analyzes and the lack of coordination of the activity in Spain demonstrate the urgent need to implement the specialty of Clinical Genetics as well as the harmonization of genetic counseling in Spain. Adequate training in genetic counseling according to the recommendations of the European Union, will promote the regular development of genetic diagnosis practices in our country.

RECOMMENDATIONS:

1. The tests of genetic screening should be carried out with criteria of relevance, quality, equity and accessibility.
2. Faced with the possibility of transmission of a genetic alteration to children, parents are advised to access counseling
3. Genetic counseling in no case should have a managerial nature
4. The performance of genetic diagnostic tests must have the mandatory informed consent in accordance with the Biomedical Research Law
5. Before the eventual detection during pregnancy of a genetic alteration, parents should be informed of the therapeutic, palliative, and legal possibilities. In addition, information should be given on how many benefits exist and on associations that help families and patients with congenital disabilities.
6. The protection of the rights of the persons consulted should be ensured as regards the decision they adopt and the processing of the genetic data.
7. It is recommended to promote adequate training in universities

<p>Which life stage is addressed in the document?</p>	<p>Adults Minors (excluding newborns) Newborns Prenatal</p>
<p>Which ethical challenges are addressed in the document? (you can see lists above for ideas of types)</p>	<p>The protection of human beings who find themselves in contexts and situations of vulnerability and vulnerability, and must always keep in mind the dignity of the people. It is necessary to take into account that counseling, in addition to influencing the individual that is the object of the analysis, will have an impact on the people who have responsibility for the subjects that are the objects of diagnosis</p>
<p>How are the ethical issues addressed? (practically, theoretically etc? Any comment on this is fine)</p> <p>Are solutions offered? If so, which ones? (depending on the document, this may be redundant with above)</p>	<p>Practically</p>
<p>How is the document structured? e.g. Which format is used in the document (checklist, continuous text, other)?</p>	<p>continuous text</p>
<p>Is the document clearly understandable?</p>	<p>Yes</p>

<p>Why is the document important/useful for your country? why did you choose this doc? TELL US WHY IMPORTANT for country and why for SIENNA</p>	<p>Because this report refers to the why and how genetic counseling is carried out in Spain, in relation to what is stated in the Biomedical Research Law. The need to regulate genetic counseling is judged according to the Interterritorial Council of the National Health System.</p>
<p>Is the document useful for the development of the SIENNA codes and other ethical frameworks? If yes, please explain.</p>	<p>Probably, for all the above mentioned.</p>

<p>Document found via (national associations or google or another database)</p>	<p>national associations</p>
<p>Title of document</p>	<p>CLONACIÓN TERAPÉUTICA: PERCPECTIVAS ÉTICAS, LEGALES Y CIENTÍFICAS (Therapeutic cloning: ethical, legal and scientific perspectives)</p>
<p>Scope/ main topic</p>	<p>therapeutic and reproductive cloning</p>
<p>Kind of document (PEC or NAEG or other)</p>	<p>NAEG</p>
<p>Document developed by whom (organisation, profession)?</p>	<p>Fundación Víctor Grífols i Lucas</p>

Year the document was published	
Document saved in folder as	NAEG Clonación terapéutica
Who is the stated audience, if none specified, write not stated (NS)	Gemma Marfany, Josep Egozcue y Victòria Camps
For Clinical or research or both or not specified	Both
What level of guideline is provided?	Very broad: i.e.: on many aspects of genomics or genetics, including many issues all together e.g. Very specific: only focuses on 1- 2 issues More Theoretical (values, principles) More practical (a practitioner could apply them)

What is the scope of the document, and what are the main recommendations?

SCOPE:

The aim of this document is to focus on our existing knowledge and on the future possibilities offered by research into therapeutic cloning – the use of somatic cell nuclear transfer to obtain cells whose genetic information is identical to that of the donor individual for use in cell therapy. The document want also clarify the differences between this and ‘reproductive cloning’. While based on the same techniques, reproductive cloning has a radically different aim: the creation of genetically identical individuals

RECOMMENDATIONS:

1. The Spanish government should use every means at its disposal to authorise therapeutic cloning as widely as possible within the legal context of the European Union, and to finance research using SCNT stem cells.
2. Each project should be evaluated on an individual basis and approved, where appropriate, by the competent body.
3. The donation of oocytes should be permitted within the specified legal context, and the signed informed consent of donors should cover this eventuality. Financial compensation should be the same as for the donation of oocytes for reproduction, and the anonymity of the donor should be guaranteed. In the same way, the donation of somatic cells so that their nucleus can be extracted and used in SCNT should be permitted for use in cell therapy. The signed informed consent of the donors should cover this eventuality.
4. The guidelines that regulate research with embryos should be applied to donated embryos, with the words ‘father’ and ‘mother’ being replaced by the terms ‘oocyte donor’ and ‘nucleus donor’.
5. The implantation in the uterus of the products of SCNT should be banned.
6. Reproductive cloning should be prohibited.

<p>Which life stage is addressed in the document?</p>	<p>Adults Minors (excluding newborns) Newborns Prenatal</p>
<p>Which ethical challenges are addressed in the document? (you can see lists above for ideas of types)</p>	<p>Why is society as a whole so concerned by the phenomenon of cloning? Why does the very word 'clone' spark fears of the possible manipulation and control of the human species?</p>
<p>How are the ethical issues addressed? (practically, theoretically etc? Any comment on this is fine)</p> <p>Are solutions offered? If so, which ones? (depending on the document, this may be redundant with above)</p>	<p>Practically and theoretically</p>
<p>How is the document structured? e.g. Which format is used in the document (checklist, continuous text, other)?</p>	<p>continuous text</p>
<p>Is the document clearly understandable?</p>	<p>Yes</p>

<p>Why is the document important/useful for your country? why did you choose this doc? TELL US WHY IMPORTANT for country and why for SIENNA</p>	<p>The document try to contribute to the need for a significant body of consistent, rigorous research, both cognitive and experimental, encompassing a range of perspectives including both the scientific and the social, in order to identify realistic prospects for the application of these new genetic techniques</p>
<p>Is the document useful for the development of the SIENNA codes and other ethical frameworks? If yes, please explain.</p>	<p>Probably, for all the above mentioned.</p>

Table 5C GDREC DOCUMENTS for HUMAN GENOMICS

<p>Document found via (national associations or google or another database)</p>	<p>national associations</p>
<p>Title of document</p>	<p>RECOMENDACIONES DEL COMITÉ DE BIOÉTICA DE ESPAÑA CON RELACIÓN AL IMPULSO E IMPLANTACIÓN DE BUENAS PRÁCTICAS CIENTÍFICAS EN ESPAÑA (Recommendations of the bioethics committee of Spain in relation to the impulse and implantation of good scientific practices in Spain)</p>
<p>Scope/ main topic</p>	<p>Scientific research in general</p>
<p>Kind of document (PEC or NAEG or other)</p>	<p>PEC</p>
<p>Document developed by whom (organisation, profession)?</p>	<p>Spain bioethics committee</p>

Year the document was published	
Document saved in folder as	PEC cod deont_med CAPXIII
Who is the stated audience, if none specified, write not stated (NS)	NS
For Clinical or research or both or not specified	Research
What level of guideline is provided?	<p>Very broad: i.e.: on many aspects of genomics or genetics, including many issues all together e.g.</p> <p>Very specific: only focuses on 1- 2 issues</p> <p>More Theoretical (values, principles)</p> <p>More practical (a practitioner could apply them)</p>

<p>What is the scope of the document, and what are the main recommendations?</p>	<p>SCOPE: Inspire and guide the doctors/researchers in the practical issues</p> <p>RECOMMENDATIONS (specific about genetic testing):</p> <p>1. The analysis of biological samples will only be carried out for the intended purpose and consented by the patient. 2. If consent has been obtained for the sample to be used in research work, the appropriate precautions will be taken to preserve the anonymity of the specimen. 3. The research must preserve the genetic data of the patients he or she attends. 4. The genetic data can never be used as a discriminatory element. 5. biological DNA samples used in people identification should be obtained from the most reliable genomic regions. 6. Only research tests should be performed</p>
<p>Which life stage is addressed in the document?</p>	<p>Adults Minors (excluding newborns) Newborns Prenatal</p>
<p>Which ethical challenges are addressed in the document? (you can see lists above for ideas of types)</p>	<p>General ethical challenge: the commitment of the medical profession to the society to which it provides its service, including the advancement of scientific-technical knowledge and the development of new rights and responsibilities</p>

<p>How are the ethical issues addressed? (practically, theoretically etc? Any comment on this is fine</p> <p>Are solutions offered? If so, which ones? (depending on the document, this may be redundant with above)</p>	<p>Practical Issues</p>
<p>How is the document structured? e.g. Which format is used in the document (checklist, continuous text, other)?</p>	<p>Checklist</p>
<p>Is the document clearly understandable?</p>	<p>Yes</p>
<p>Why is the document important/useful for your country? why did you choose this doc? TELL US WHY IMPORTANT for country and why for SIENNA</p>	<p>It has been developed by a very representative institution in the country</p>
<p>Is the document useful for the development of the SIENNA codes and other ethical frameworks? If yes, please explain.</p>	<p>Probably, for all the above mentioned.</p>

7.11 Sweden

TABLE 1: INDIVIDUAL AND COUNTRY INFORMATION

Names and emails of persons who did the work (if different from above)	HC Howard, Emilia Niemiec, Caroline Gallant, and Cornelia Tandre Heidi.howard@crb.uu.se
Your organisation	Uppsala University
Your country (again)	SWEDEN
Search conducted in which language	Swedish and English
Acknowledgements (any researcher who helped you to complete this task)	NA

TABLE 2: LIST OF ALL RELEVANT PROFESSIONAL ETHICS CODES

Area	Title of document	URL	Year	Author/organisation	Stated audience	Comments
HG	Yrkesetisk kod för biomedicinska analytiker (<i>Professional ethic code for biomedical scientists</i>)	http://ibl-inst.se/wp-content/uploads/2016/03/Yrkesetisk-kod-A6.pdf	2011	Vårdförbundet/I FBLS/ IBL	Biomedical scientists	Very general for biomedical sciences. Example of general/macro guideline
HG	Läkarförbundets etiska regler (<i>The doctor federation's ethical rules</i>)	https://www.slf.se/Lon--arbetsliv/Etikochansvar/Etik/Lakarforbundets-etiska-regler	2017	Sveriges Läkarförbund	Doctors	Meta level for doctors
HG	<i>This is more than one guideline</i>	http://sfmg.se/riktlinjer/		Svensk förening för Medicinsk Genetik och Genomik (SFMG)	Professionals working with medical or clinical genetics	Contains very general ethical guidance and recommendations for screening
HG	Yrkesetisk kod för biomedicinska analytiker (<i>Professional ethic code for biomedical scientists</i>)	http://ibl-inst.se/wp-content/uploads/2016/03/Yrkesetisk-kod-A6.pdf	2011	Vårdförbundet/I FBLS/ IBL	Biomedical scientists	

Deliverable report

HG	Yrkesetiska koder	https://www.vardforbundet.se/rad-och-stod/regelverket-i-varden/etik/yrkes-etiska-koder/	2016	Vårdförbundet		Vårdförbundet is a union for nurses, midwives, biomedical scientists and radiology nurses
HG	Plötslig död hos unga: Förslag till riktlinjer för postmortal genetisk analys	https://sfmg.se/	2013 (update d 2016)	SFMG	Clinicians	Guidelines regarding clinical and genetic screening of sudden cardiac death in young individuals and their first degree relatives. No discussion of consent related to genetic testing of the dead patient and/or patient's relatives.

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HG	Rekommendationer från Svensk förening för medicinsk genetik (SFMG) avseende hantering av "oväntade" genetiska fynd vid genomvida analyser	https://sfmg.se/	2016	SFMG	Clinicians	Recommendations regarding if and when to return secondary findings to patients when performing whole exome or genome sequencing.
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HG	<p>HEREDITÄRT FEOKROMOCYTOM/PARAGANGLIOM: - Nationella rekommendationer för genetisk utredning av paragangliom/feokromocytom samt kontrollprogram för friska anlagsbärare av ett anlag i SDHx-, TMEM127 och MAX-generna (National recommendations for genetic analysis of pheochromocytoma/paraganglioma and control program for healthy landscapers of a landfill in SDHx)</p>	<p>https://sfmg.se/ https://sfmg.se/download/riktlinjer/Cancergenetik/Arftligt-feokromocytom-paragangliom_170627.pdf</p>	2017	SFMG	Clinicians	<p>Recommendations regarding genetic testing (gene panels) of close relatives, including healthy children.</p>
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Deliverable report

HG	BILAGA 6: HANDLÄGGNING AV INDIVIDER INOM FAMILJER MED FAMILJÄRT MELANOM	https://sfmg.se/	2015	SFMG & Svenska nätverket för familjärt melanoma (SweFaM)	Clinicians	Recommendations on contacting relatives of index melanoma patient when familial disease is suspected (including consent form). Information regarding research of melanoma-associated genes in family members
HG	Ärftlig kolorektalcancer - utredning, uppföljning och omhändertagande one of cancers, potentially all yes	https://sfmg.se/	2012 (update d 2015)	SFMG	Clinicians	Guidance on genetic counselling of patients with inherited cancer susceptibility, including possible socio/psychological impact.

Deliverable report

HG	Utredning, uppföljning och omhändertagande av personer med misstänkt ärftlig ökad risk för tumörsjukdom	https://sfmg.se/	2007 (update d 2012)	SFMG	Clinicians	Discussion/commentary about whether it is ethical to screen individuals for increased risk of pancreatic cancer whereas currently no effective treatment program exists.
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Deliverable report

HG	Biobank Sweden all documents (general web site)	http://biobanksverige.se/alladokument/		Biobank Sverige	Everyone who are working/are in contact with biobanks	This is a site rich with all things related to biobanks, and in some ways they address ethical issues but only in a very specific ways within documents about general procedures, (i.e. in the form for researchers they address coding of samples; in the form for patients they address consent and withdrawal of consent) but these are still not the focus per se
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TABLE 3: LIST OF ALL RELEVANT DOCUMENTS FROM NATIONAL ADVISORY/ETHICS GROUPS

Area	Title of document	URL	Year	Author/organisation	Stated audience	Comments
HG	Tekniken CRISPR/Cas9 och möjligheten att redigera det mänskliga genomet <i>(the CRISPR technique and the possibility to edit the human genome)</i>	http://www.smer.se/wp-content/uploads/2015/07/Sm-er-kommenterar-CRISPR_Cas91.pdf	2015	Statens medicinska råd (SMER)	Not available	SMER comments on international and national reports on gene editing tools such as CRISPR-Cas9
HG	Individanpassad medicin – möjligheter och risker <i>(Personalized medicine. opportunities and risks)</i>	http://www.smer.se/wp-content/uploads/2013/12/Sm-er-kommenterar-individanpassad-medicin.pdf	2013	Statens medicinska råd (SMER)		SMER comments on international and national reports on personalised medicine

Deliverable report

HG	Tillsätt en parlamentarisk utredning för att se över lagstiftningen på genteknikområdet This is an attachment to the letter below. In this attachment the ideas contained in the letter are developed, I havent analysed it,.	http://www.smer.se/skrivelse/r/tillsatt-en-parlamentarisk-utredning-for-att-se-over-lagstiftningen-pa-genteknikområdet/	2018	Statens medicinska råd (SMER)		Motivation for the government to investigate the impact and regulation of genetics and genomics
HG	Letter connected To report <i>Tillsätt en parlamentarisk utredning för att se över lagstiftningen på genteknikområdet</i>	http://www.smer.se/wp-content/uploads/2018/06/Skrivelse-om-utredning-av-lagstiftning-f%C3%B6r-ny-genteknik.pdf	2018	Statens medicinska råd (SMER)		Motivation for the government to investigate the impact and regulation of genetics and genomics

TABLE 4: LIST OF ALL RELEVANT GUIDANCE DOCUMENTS ON HOW TO WRITE RESEARCH ETHICS PROTOCOLS

Area	Title of document	URL	Name of national REC	Stated audience	Comments
HG + All types with humans	Vägledning till ansökan <i>(Guidance for application)</i>	https://www.epn.se/media/2469/vaegledning-till-ansokan.docx https://www.epn.se/start/	Etikprövningsnämnderna	Researchers	For all research involving humans. There is one part called “Redogör för om insamlad biologiskt material kommer att förvaras i en biobank” so it should apply to HG
All	Vägledning till forskningsplan/forskningssprotokoll (program) <i>(Guidance for research plan/research protocol (program))</i>	https://www.epn.se/media/1103/vaegledning_till_forskningsplan.pdf https://www.epn.se/start/	Etikprövningsnämnderna	Researchers	Could be relevant, but very brief and mostly about the research protocol application, not the ethics per se
Potentially all	Vägledning till forskningspersonsinformation <i>(Guidance for research person information)</i>	https://www.epn.se/media/2573/vaegledning-till-forskningspersonsinformationgdp-r-med-korrigeringar.pdf https://www.epn.se/start/	Etikprövningsnämnderna	Researchers	Guide to research professionals information. Useful for information needed to recruit human subjects

Deliverable report

HG, biome dical profess ions	YrkesYrketiska koder	https://www.vardforbundet.se/rad-och-stod/regelverket-i-varden/etik/yrkesetiska-koder/	Vårdförbundet		Vårdförbundet is a union for nurses, midwives, biomedical scientists and radiology nurses. PECs for all the professions can be found at their websites.
ALL All	Nationella etiknätverket (KI)	https://ki.se/lime/etik-i-praktiken		Researchers and others stakeholders in research	They have some good links on their website, like the mapping of all regional ethics groups for example. They also make documents where they collect new articles, laws etc. about ethics (e.g. https://ki.se/sites/default/files/2017/11/02/omvarldsbevakning_varen_2017.pdf) No ethical guidelines.

TABLE 5A: General questions on your document search in Human Genomics

1- Did you find guidance documents specific to human genomics or genetics?	Yes
2a How many documents did you find from each of these sources that specifically address human genetics or genomics	PECs: 6 NAEG: 3 GDREC: 0 Total: 9
2b Did you find more documents than you had the time to analyse in this task?	No

Deliverable report

<p>3- a) How many documents specifically ONLY address high throughput genomics? By high throughput genomics, we mean using sequencing of larger parts of the genome, or exome (including large gene panels).</p>	<p>PECs: 2 NAEG: 0 GDREC:0</p>
<p>3b Please list the year of publication of each documents here:</p>	
<p>3c please specify the topics addressed explicitly, ideally indicate the number of documents that address each, place an X beside the topic and add the number of docs</p>	<p>Genomics used for diagnostics in the clinic (genomic testing) x Genomics used for research Consent in genomics in the clinic Consent in genomics in research Return of results in genomics in the clinic x Return of results in genomics in research Reinterpretation and recontact in genomics in the clinic Reinterpretation and recontact in genomics in the research Genomics for newborn screening Genomics for prenatal screening or testing Non-invasive prenatal testing/screening Discrimination Eugenics Justice Genetic exceptionalism</p>

Deliverable report

<p>4 a How many documents only address genetics (so testing or screening but without a lot of sequencing)?</p> <p>4b List the years of publications of each document here</p> <p>3c please specify the topics addressed explicitly, ideally indicate the number of documents that address each, place an X beside the topic and add the number of docs</p>	<p>PECs 1 NAEG GDREC</p> <p>Genetics used for diagnostics in the clinic (genetic testing) _1_ Genetics used for research____ Consent in genetics in the clinic____ Consent in genetics in research____ Return of results in genetics in the clinic____ Return of results in genetics in research____ Reinterpretation and recontact in genetics in the research____ Reinterpretation and recontact in genetics in the clinic____ newborn screening__, Genetic exceptionalism____</p>
<p>5. How many documents address both genetics and genomics?</p>	<p>5 PECs, 2 NAEG</p>

<p>6 a) How many documents specifically address Genome Modification, gene editing, crispr, or gene therapy</p> <p>6b) do they address somatic or germline editing or both?</p>	<p>3</p> <p>both</p>
<p>How many documents specifically address (so in the title of the document)</p>	<p>Genetic or genomic testing</p> <p>Genetic or genomic screening</p> <p>Biobanks or databases</p> <p>Pharmacogenomics or pharmacogenetics</p> <p>Patents</p>

Table 5B RELEVANT PEC or NAEG DOCUMENTS IN HUMAN GENOMICS,

5Bi: genomic testing (if none on genomics, use genetic testing)

5Bii genomic screening (if none on genomics, use genetic screening)

5Biii biobanks, databases or registries

5Biv pharmacogenomics or pharmacogenetics

5Bv patents

5Bvi gene editing, crispr, or gene therapy

Please note that the quotes included in these tables are based on translations obtained using Google Translate software, which were then refined by the authors. Yet, they may not always precisely reflect the content of the documents; they are rather indicative of their content.

Table 5B 1)

Deliverable report

Document found via (national associations or google or another database)	
Title of document	Tekniken CRISPR/Cas9 och möjligheten att redigera det mänskliga genomet (<i>the CRISPR technique and the possibility to edit the human genome</i>)
Scope/ main topic	(choose from list above, 5Bi-5Bvi, write out the topic CRISPR
Kind of document (PEC or NAEG or other)	NAEG
Document developed by whom (organisation, profession)?	Statens medicinsk-etiska råd (SMER) Swedish National Council on Medical Ethics
Year the document was published	2015
Document saved in folder as	Smer-kommenterar-CRISPR_Cas91.pdf
Who is the stated audience, if none specified, write not stated (NS)	NS
For Clinical or research or both or not specified	Both

<p>What level of guideline is provided?</p>	<p>X Very broad: i.e.: on many aspects of genomics or genetics, including many issues all together e.g. (it focuses on gene editing, but discusses its many aspects) Very specific: only focuses on 1- 2 issues More Theoretical (values, principles) More practical (a practitioner could apply them)</p>
<p>What is the scope of the document, and what are the main recommendations?</p>	<p>Introduction what gene editing is. Then it reports on the articles of Baltimore’15 (calling for moratorium and discussion), Lanphier’15 (which says that maybe we shouldn’t perform research on embryos involving CRISPR(?)) and Liang’15. Swedish perspective: Swedish legislation discussed. SMER (authors) comments ethical issues discussed. And SMER stance (see below)</p>
<p>Which life stage is addressed in the document?</p>	<p>Adults Minors (excluding newborns) Newborns Prenatal</p> <p>Germline and somatic gene editing</p>
<p>Which ethical challenges are addressed in the document? (you can see lists above for ideas of types)</p>	<p>Knowledge gaps and unknown risks and human life at stake; risks also in somatic GE, but less There are alternative methods like PGD. Potential negative perception in public given ethical sensitivity Potential uses to enhance non health-related traits, attempts to achieve perfect human; questions of human dignity</p>

Deliverable report

<p>How are the ethical issues addressed? (practically, theoretically etc? Any comment on this is fine</p> <p>Are solutions offered? If so, which ones? (depending on the document, this may be redundant with above)</p>	<p>“SMER looks positively at the opportunities that CRISPR can mean, especially applicable somatic gene therapy for severe diseases. Also research on fertilized eggs with CRISPR can provide valuable knowledge, for example infertility and stem cell therapy. SMER considers that the current Swedish regulation of genetic engineering research is well balanced. The research community should proceed carefully current research aimed at introducing induce hereditary changes in the nuclear DNA. It is far too early to perform clinical research efforts today, taking into account the moods about medical risks and others possible effects. In addition, one should weigh in that today there are other methods for avoid severe hereditary diseases in children. SMER agrees with article call for a broad social discussion about the possibility of performing hereditary genetic changes in humans” (p.4 and 5)</p>
<p>How is the document structured? e.g. Which format is used in the document (checklist, continuous text, other)?</p>	<p>Short paragraphs</p>
<p>Is the document clearly understandable?</p>	<p>yes</p>
<p>Why is the document important/useful for your country? why did you choose this doc? TELL US WHY IMPORTANT for country and why for SIENNA</p>	

<p>Is the document useful for the development of the SIENNA codes and other ethical frameworks? If yes, please explain.</p>	<p>Yes, gives the Swedish perspective on gene editing</p>
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Table 5B 2)

Document found via (national associations or google or another database)	
Title of document	Original title: Rekommendationer från Svensk förening för medicinsk genetik (SFMG) avseende hantering av "oväntade" gemetosla fynd vid genomvida analysen Recommendations from the Swedish Association for Medical Genetics (SFMG) regarding handling "unexpected" genetic findings during whole genome analysis
Scope/ main topic	choose from list above, 5Bi-5Bvi, write out the topic - genomic testing
Kind of document (PEC or NAEG or other)	PEG
Document developed by whom (organisation, profession)?	Swedish Society of Medical Genetics and Genomics
Year the document was published	2016
Document saved in folder as	SFMG-hantering-av-oväntade-genetiska-fynd_22-juni-2016.pdf
Who is the stated audience, if none specified, write not stated (NS)	Clinicians

Deliverable report

For Clinical or research or both or not specified	Clinical	
What level of guideline is provided?	Very broad: i.e.: on many aspects of genomics or genetics, including many issues all together e.g. x Very specific: only focuses on 1- 2 issues More Theoretical (values, principles) More practical (a practitioner could apply them)	
What is the scope of the document, and what are the main recommendations?	Unsolicited findings obtained in whole genome sequencing (WGS)	
Which life stage is addressed in the document?	X Adults Minors (excluding newborns) Newborns Prenatal	
Which ethical challenges are addressed in the document? (you can see lists above for ideas of types)	Please see below	

Deliverable report

<p>How are the ethical issues addressed? (practically, theoretically etc? Any comment on this is fine</p> <p>Are solutions offered? If so, which ones? (depending on the document, this may be redundant with above)</p>	<p>“In order to avoid detecting "unexpected" genetic findings, SFMG recommends that you as far as possible, genetic investigations make targeted gene analyzes in the form of either dedicated gene panels or <i>in silico gen</i> panels (ie where data is filtered and only predetermined genes are analyzed) once analyzes have been performed. About exome- or WGS sequencing is still up to date, for example in the case of unclear genetic conditions or there Information about structural variants is paramount, it is of great importance for each center has a clearly formulated policy for dealing with unexpected genetic findings, especially as regards reports of such findings.</p> <p>According to SFMG, the following points should be considered regarding handling and reporting of unexpected genetic findings:</p> <ol style="list-style-type: none"> 1. Before ordering exome or WGS, referring physicians should inform the patient that unexpected findings may be identified and to clinically relevant findings can be reported. 2. An in-depth family name should be taken before exoms or WGS are ordered to get indication of other hereditary conditions may exist in the family. 3. Clinically relevant unexpected findings in high-penetrant genes where treatment / prevention is possible or may affect the processing can be reported back. Currently defined no list of genes to be reported without this being discussed on a case-by-case basis. 4. Reports only pathogenic or likely pathogenic genetic variants (i.e. no VUS, likely benign or benign variants to be answered). Heterozygous support for recessive hereditary disease can be reported back in a clinically relevant situation 5. If an unexpected genetic finding is detected, the laboratory must first have a dialogue with in-hospital physician if this is a clinically relevant finding for the individual / family before patient is informed. If necessary, the finding can be discussed with a reference group within SFMG. <p>Further detailed guidelines on unexpected genetic findings and how these are handled within Medical care is likely to need to be established as knowledge and experience in this area increases”</p>
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Deliverable report

<p>How is the document structured? e.g. Which format is used in the document (checklist, continuous text, other)?</p>	<p>Short introduction, and points to consider</p>
<p>Is the document clearly understandable?</p>	<p>Yes</p>
<p>Why is the document important/useful for your country? why did you choose this doc? TELL US WHY IMPORTANT for country and why for SIENNA</p>	
<p>Is the document useful for the development of the SIENNA codes and other ethical frameworks? If yes, please explain.</p>	

Table 5B 3)

<p>Document found via (national associations or google or another database)</p>	
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Deliverable report

Title of document	Original title: Plötslig död hos unga: Förslag till riktlinjer för postmortal genetisk analys Sudden death in young people: Proposal for guidelines for post mortem genetic analysis
Scope/ main topic	(choose from list above, 5Bi-5Bvi, write out the topic: genetic testing
Kind of document (PEC or NAEG or other)	PEG
Document developed by whom (organisation, profession)?	Swedish Society for Medical Genetics and Genomics
Year the document was published	2010. Updated 2016.
Document saved in folder as	Plotslig-dod-hos-unga_Forslag-till-riktlinjer-for-postmortal-genetisk-analys_uppdatering-2016.pdf
Who is the stated audience, if none specified, write not stated (NS)	Clinicians
For Clinical or research or both or not specified	Clinical
What level of guideline is provided?	Very broad: i.e.: on many aspects of genomics or genetics, including many issues all together e.g. X Very specific: only focuses on 1- 2 issues More Theoretical (values, principles) X More practical (a practitioner could apply them)

Deliverable report

<p>What is the scope of the document, and what are the main recommendations?</p>	<p>Investigations after sudden deaths in people age 1-35 years old and potentially also in younger children. See below for recommendations.</p>	
<p>Which life stage is addressed in the document?</p>	<p>X Adults X Minors (excluding newborns) X potentially Newborns Prenatal</p>	
<p>Which ethical challenges are addressed in the document? (you can see lists above for ideas of types)</p>	<p>When and how relatives of a deceased person (sudden death) should be screened, responsibilities of the physicians</p>	

<p>How are the ethical issues addressed? (practically, theoretically etc? Any comment on this is fine</p> <p>Are solutions offered? If so, which ones? (depending on the document, this may be redundant with above)</p>	<p>Very practical, concise guideline, ethical issues are not explicitly discussed: “Clinical screening of pre-existing patients. If completed autopsy cannot detect a cause of death, pre-trial situations (parents, children and siblings of the deceased) should be carefully screened for clinical hereditary cardiovascular disease screening. Contact with clinical geneticists is recommended at this stage, for planning / initiating information and surveys of the relatives. If the autopsy shows signs of structural heart disease of a likely hereditary nature (hypertrophic cardiomyopathy or arrhythmia right ventricular diacomomyopathy), pre-trial patients in collaboration with clinical geneticists and after careful information will be offered a targeted clinical examination of the current heart disease. If the deceased is not currently treated by a cardiologist, the physician who handled the death establishes contact between first-degree and heart specialist who is responsible for the above-described examination of the pre-existing patients. If the deceased is taken directly to forensic autopsy, and relatives contact with it forensic department for reporting the cause of death, the person who communicates should autopsy result (i.e. no detectable cause of death, or structural heart disease of a likely hereditary nature) informs that clinical screening of pre-existing patients should be performed by a heart specialist in consultation with clinical geneticists. As a basic clinical screening, it is recommended: - Family history - EKG - ECG test - Bandspejar</p> <p>Depending on the outcome of these studies, additional diagnostic measures may be applicable. Based on the findings of the clinical screening, directed molecular genetic investigation can then be made on the archived sample from the deceased or from the initial pains where the heart disease could be established to confirm the diagnosis molecular genetically. If a disease-causing mutation can be identified, this finding can be used for presymptomatic testing in the current family. Such molecular genetic investigation is conducted in collaboration between cardiologist and avd. for 3 clinical genetics, responsible for genetic analyzes. Family members who prove to be a carrier will be offered prevention and treatment and included in a control program. If a hereditary arrhythmia disease is diagnosed during clinical screening the family should be disposed of in a clinical control program for the current disease, even if no cause-causing mutation is identified. If signs of hereditary heart disease in first-degree patients can not be detected during screening, molecular</p>
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genetic investigation on the archived sample from the deceased may still be considered ("molecular autopsy"). This is from up to date if there was an alarming family name in the clinical screening (syncope attacks, unexplained seizures, drowning of unclear cause, or other cases of sudden death). In that case, consideration should be given primarily to genetic analyzes for junior diseases that are not associated with structural heart disease, i.e. long QT syndrome, catecholaminergic polymorphic ventricular tachycardia and Brugada syndrome.

Deliverable report

<p>How is the document structured? e.g. Which format is used in the document (checklist, continuous text, other)?</p>	<p>Continuous, a graph</p>
<p>Is the document clearly understandable?</p>	<p>Yes</p>
<p>Why is the document important/useful for your country? why did you choose this doc? TELL US WHY IMPORTANT for country and why for SIENNA</p>	
<p>Is the document useful for the development of the SIENNA codes and other ethical frameworks? If yes, please explain.</p>	<p>It potentially shows a gap – consent to genetic screening of deceased person is not addressed.</p>

Table 5B 4)

<p>Document found via (national associations or google or another database)</p>	
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Deliverable report

Title of document	Letter on Legislation for New Genetic Engineering (Skrivelse om utredning av lagstiftning för ny genteknik)
Scope/ main topic	(choose from list above, 5Bi-5Bvi, write out the topic)
Kind of document (PEC or NAEG or other)	NAEG
Document developed by whom (organisation, profession)?	Statens medicinsk-etiska råd (SMER) Swedish National Council on Medical Ethics
Year the document was published	2018
Document saved in folder as	Skrivelse_om_utredning_av_lagstiftning_för_ny_genteknik.pdf Attachment: Bilaga_till_skrivelsen.pdf
Who is the stated audience, if none specified, write not stated (NS)	Legislator
For Clinical or research or both or not specified	Both
What level of guideline is provided?	x Very broad: i.e.: on many aspects of genomics or genetics, including many issues all together e.g. Very specific: only focuses on 1- 2 issues More Theoretical (values, principles) More practical (a practitioner could apply them)

Deliverable report

What is the scope of the document, and what are the main recommendations?	In the letter, the authors suggest to investigate and develop a strategy for genetic engineering and review the relevant legislation. The attachment supporting their statement discusses each genomic technology and some related ethical issues.	
Which life stage is addressed in the document?	x Adults x Minors (excluding newborns) x Newborns x Prenatal	

<p>Which ethical challenges are addressed in the document? (you can see lists above for ideas of types)</p>	<p>“Basic ethical issues related to human worth and human perception, safety and risks, genetic and personal integrity, information, consent as well as justice issues. For some applications we are faced same ethical questions and considerations as before, to others we are facing new ones. The development challenges our view of what it means to be person. It's about us people, about our society and what future we want as well as in terms of changes in the genome that can go in of responsibility for future generations. It is also about how we can recruit and take advantage of the genetic engineering progress which is ethically good- sustainable and sustainable ways contribute to human health, functional capacity and quality of life.” (p.3 and 4) Then the document mentions the need for social debate.</p> <p>“In many areas it becomes increasingly difficult for Sweden to only through its own Foundations regulate different parts of genetic engineering. This applies, for example, to sale of online genetic tests and other services in or through other countries. Gene changes can cause uncontrolled spread of contagious diseases or lead to resistance development, which has global consequences.</p> <p>Gene technology can also be used to produce biological weapons of war.</p> <p>Many consider that we need to take joint responsibility for that with respect for human dignity promote ethically sustainable and secure development. During the last decades, several international recommendations have admitted that there are dangers related to genetic engineering.”</p> <p>“An ethical analysis is needed based on an overall perspective of the new treatment methods of genetic engineering. One such</p>
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should include embrace how they New methods affect privacy, self-determination, informed consent, utility, e.g. of individualized prevention and treatment, as well privacy, worry and unwanted information. Effects on cards and long-term view of human and human values, opportunities and threats and risks need to be weighed against each other and discussed widely.”

These issues are further elaborated in the context of specific technologies in the attachment to the letter.

<p>How are the ethical issues addressed? (practically, theoretically etc? Any comment on this is fine</p> <p>Are solutions offered? If so, which ones? (depending on the document, this may be redundant with above)</p>	<ul style="list-style-type: none"> - they call for national strategy on genetic engineering - “SMER considers in accordance with the Council’s previous position that consideration should be given to to allow mitochondrial exchange in certain circumstances even in Sweden. (p3) - “SMER believes that the many new clinical applications that concern people together justify a review of the regulatory framework from an ethical perspective.” p 4 (Smer anser...) - “A survey of the use, focus and quality of genetic self-tests is needed. A review needs to be made of the need for national requirements or rules, not least because of the new opportunities available for extended landline testing. One must include whether and how to ensure that the submitted material comes from the submitter of the sample, the quality of the test, how information about the result is given and whether genetic data will be destroyed or stored and who can access this data” - “The law on genetic integrity also regulates the use of genetic information in the insurance field. In the case of a risk-assessed personal insurance, an insurance company may, under certain conditions, investigate or use genetic information in connection with an agreement. Smer considers that the rules for the use of genetic information in the insurance field should be reviewed, inter alia, ased on the Council of Europe recommendation on health information in insurance contexts. This recommendation means that only information relevant to the assessment is to be submitted and that an independent evaluation of which diseases are obtained, and not allowed, should be sought. It should also be noted that genetic data is considered sensitive personal data according to the EU's new data protection regulation.”
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- “According to SMER, it is necessary to take a holistic approach based on an ethical perspective on criteria for and use of genetic studies in the field of fertility and pregnancy. It is important to review the new possibilities for detailed mapping of the genetic engineering plans already when planning for pregnancy. Further, it is necessary to consider whether differences in principles and criteria for genetic diagnosis of genital cells, fertilized eggs, embryos, fetuses and newborns (PKU tests) are motivated or need to be changed. It needs to be weighed in not least on the basis of proposals in the prop. 2017/18: 155 to allow IVF with donated germ cells after examination of the Health and Care Inspectorate (IVO). It is likely to lead to several actors who perform PGD and IVF with their own or donated cells.”
- “SMER believes that several issues governed by the Genetic Integration Law regarding Germline Cells and Fertilized Eggs need to be reviewed. This includes allowed storage time for frozen germ cells and eggs, as well as the right of disposal of frozen eggs. International discussion on the so-called 14-day rule for embryo research is ongoing. An extension of the border could contribute to increasing knowledge about the development of human embryos, which is important in for the treatment of infertility and reproduction. However, it also means that new ethical issues arise.
- “Furthermore, there is a need to design an ethically sustainable regulatory framework when it comes to stem cell development, synthetic organisms, and human and animal mixture. As Smer pointed out in 2008, it is unclear whether certain research within the stem cell area is unlawful in Sweden and if so, what trials would have to be

done for it to be allowed. The regulatory framework is unclear as regards the new possibilities of producing sex cells and embryos using the new stem cell technology iPS as well as attempting to produce synthetic organisms. The question of artificial mix of animals - humans has not been considered in Swedish legislation, something that Smer also mentioned in 2008.”

- “Technologically independent bioethical legislation to regulate the rapid development
Everything speaks for a continued or accelerated development rate within the genetic area. (...) Such legislation could regulate the research, development and use of knowledge and technology in life sciences and biomedicine. It should be based on universal ethical principles and establish a system of responsible authorities and clear processes for following, resolving and regulating genetic engineering and control systems to comply with compliance. (p6)”

- Add an investigation
“The task of SMER is to be a mediatorial body between science, people's opposition and politically responsible. SMER can also submit proposals for investigative activities. SMER proposes that a parliamentary inquiry be appointed with the task of reviewing the Swedish regulations for human applications of genetic engineering based on current state of knowledge and ethical perspective. SMER gladly contributes actively to such work.
The investigation should be commissioned to initiate a **broad social debate on the social**, ethical and societal consequences of the new genetic engineering. Proposals

	<p>should be submitted to the appropriate legal and other conditions for responsible genetic engineering that promote human health, functional ability and quality of life, as well as an appropriate division of responsibilities between authorities.</p> <p>In addition, a proposal for Swedish strategy for development in genetic engineering should be developed, including how Sweden should contribute to and promote an ethically balanced international regulatory framework.”</p>
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Deliverable report

<p>How is the document structured? e.g. Which format is used in the document (checklist, continuous text, other)?</p>	<p>Continuous text divided in paragraphs</p>
<p>Is the document clearly understandable?</p>	<p>Yes</p>
<p>Why is the document important/useful for your country? why did you choose this doc? TELL US WHY IMPORTANT for country and why for SIENNA</p>	<p>Seems to be very important, it calls for concrete changes of the legislation.</p>
<p>Is the document useful for the development of the SIENNA codes and other ethical frameworks? If yes, please explain.</p>	<p>Potentially</p>

Table 5B) 5

<p>Document found via (national associations or google or another database)</p>	
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Deliverable report

Title of document	Original title: Individanpassad medicin – möjligheter och risker Personalized medicine. <i>opportunities and risks</i>
Scope/ main topic	(choose from list above, 5Bi-5Bvi, write out the topic: genetic testing, screening and pharmacogenomics)
Kind of document (PEC or NAEG or other)	NAEG
Document developed by whom (organisation, profession)?	Statens medicinsk-etiska råd (SMER) Swedish National Council on Medical Ethics
Year the document was published	2013
Document saved in folder as	Smer-kommenterar-individanpassad-medicin.pdf
Who is the stated audience, if none specified, write not stated (NS)	
For Clinical or research or both or not specified	Clinical
What level of guideline is provided?	x Very broad: i.e.: on many aspects of genomics or genetics, including many issues all together e.g. Very specific: only focuses on 1- 2 issues More Theoretical (values, principles) More practical (a practitioner could apply them)

Deliverable report

<p>What is the scope of the document, and what are the main recommendations?</p>	<p>Personalized medicine. The document comments on Personalized Medicine for the European Citizen - Towards more precise medicine for the diagnosis, treatment and prevention of disease (iPM). November 2012. Report from European Science Foundation (ESF). http://www.esf.org/index.php?id=7988</p> <p>SMER among other issues, points out that the claims expressed in this documents seem to be exaggerated, see below for their recommendations.</p>	
<p>Which life stage is addressed in the document?</p>	<p>x Adults x Minors (excluding newborns) x Newborns x Prenatal</p> <p>The document doesn't mention explicitly all these life stages, but implicitly encompass them.</p>	

<p>Which ethical challenges are addressed in the document? (you can see lists above for ideas of types)</p>	<p>“The risks are especially large in terms of biobanks samples which contains genetic information, not at least when it has been found possible in some cases identify individual research persons based on published "unidentified" genetic information”</p> <p>Exaggerated expectations related to personalized medicine – see below</p> <p>Just distribution of goods in healthcare system “One question is, for example, how the individual said medicine refers to more populations focused healthcare efforts- such as vaccinations, screening programs and guidelines on diet and living habits.</p> <p>Funding of new expensive treatment methods</p> <p>It is positive about the individualized medicine. The drug can lead to new treatment methods for rare diseases as it is missing today effective treatment for. These drugs can Be very expensive and maybe just current for a few patients. A challenge comes to be to determine to what extent these medicines should be subsidized by society, and what criteria should be included in this assessment”</p> <p>“A component that is part of the individual said the medicine is the possibility of information about an individual's risks for various diseases mar based on genetic tests (and tests of biomarkers). So far, these possibilities are limited, but if the predictive and preventive medicine develops actualized ethical issues. But the knowledge that a disease can breaking out can generate strong concern, especially when it concerns diseases that can not be build or cure. A development involving individuals' disease risk are mapped to a greater extent, in the long run, can affect the willingness to finance and the view of both public and private insurance systems. Will people come with them "Healthy" genes want to pay for the less fortunate? A community development with an increasing focus on genetics and disease risks have the potential to lead to segregation, discrimination and injustice.”</p>
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Deliverable report

<p>How are the ethical issues addressed? (practically, theoretically etc? Any comment on this is fine</p> <p>Are solutions offered? If so, which ones? (depending on the document, this may be redundant with above)</p>	<p>“SMER has previously emphasized that it is important for the register's future that public confidence in the registers be maintained by satisfying the business from an ethical point of view. Furthermore, the Council has suggested that an ethical review function should exist for population-based registries that are not covered by other ethics testing.”</p> <p>“Exaggerated expectations? The report describes comprehensive measures and efforts that should be made to achieve a healthcare system that is structurally changed. However, from an ethical point of view it is difficult to predict exactly what effects and gains that can be achieved through individual sat medication. It is likely that new and more individualized treatment methods more to develop in certain areas - but in what extent? According to SMER, the expectations are high developments reflected in the report as exaggerated. There may be other out- development lines in health care which is equally important. One question is, for example, how the individual said medicine refers to more populations focused healthcare efforts- such as vaccinations, screening programs and guidelines on diet and living habits.”</p> <p>“SMER's preliminary assessment The ESF report describes a development as has already begun and is likely will be intensified in the future. However, the perception is that the positive effects as described in the report appears to be exaggerated, and that this area must Drawn against other areas of health and medical care when deciding on investments. As at all development and introduction of new systems ethical values should be safeguarded, in particular integrity, solidarity and patient influence”.</p>
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Deliverable report

<p>How is the document structured? e.g. Which format is used in the document (checklist, continuous text, other)?</p>	
<p>Is the document clearly understandable?</p>	<p>Yes</p>
<p>Why is the document important/useful for your country? why did you choose this doc? TELL US WHY IMPORTANT for country and why for SIENNA</p>	<p>It shows sceptical perspective of SMER on some prognosis related to development and benefits of personalized medicine.</p>
<p>Is the document useful for the development of the SIENNA codes and other ethical frameworks? If yes, please explain.</p>	

7.12 UK

Intro: This document presents the results of the UK country research for human genomics professional ethics codes, guidance documents from national advisory/ethics groups. It has following tables: 1. Individual and country information, 2. List of relevant professional ethics codes (PECs), 3. List of all relevant documents from national advisory/ethics groups (NAEGs) 4. List of relevant guidance documents on how to write research ethics protocols (GDRECs), 5A. General questions on the document search in Human Genomics, 5B. Relevant PEC or NAEG documents in human genomics, 5c GDREC documents for human genomics. A summary follows.

TABLE 1: INDIVIDUAL AND COUNTRY INFORMATION

Names and emails of persons who did the work (if different from above)	Rowena Rodrigues rowena.rodrigues@trilateralresearch.com David Wright david.wright@trilateralresearch.com
Your organisation	Trilateral Research Ltd
Your country (again)	UK
Search conducted in which language	English
Acknowledgements (any researcher who helped you to complete this task)	Gerard Porter, University of Edinburgh (reviewer)

TABLE 2: LIST OF ALL RELEVANT PROFESSIONAL ETHICS CODES (GENOMICS)

SIENNA area	Title of document (original + English translation)	URL	Year	Author/organisation	Stated audience	comments
HG	Code of ethics	http://www.agnc.org.uk/about-us/agnc-documents/code-of-ethics/	Due for review 2020.	Association of Genetic Nurses and Counsellors (AGNC)	Genetic counsellors at all levels of training and professional registration working in the United Kingdom and Ireland	
HG	Code of Practice	https://www.hfea.gov.uk/media/2565/hfea-draft-code-of-practice-9th-edition-consultation-version.pdf	2018 (9 th edition)	Human Fertilisation and Embryology Authority (HFEA)	Licensed centres, clinics, (reference for patients, donors, donor- conceived people, researchers and those working in the fertility sector).	

Deliverable report

HG	Concordat moratorium on genetics insurance and on and	https://www.abi.org.uk/globalassets/sitecore/files/documents/publications/public/2014/genetics/concordat-and-moratorium-on-genetics-and-insurance.pdf	2014	Government and the Association of British Insurers (ABI)	Government and the ABI and its members	
HG	Ethical collaboration policy	https://www.geneticalliance.org.uk/ethical-collaboration-policy/	Dec 2013	Genetic Alliance	Genetic Alliance UK encompassing its associated names and brands including Rare Disease UK (RDUK) and SWAN UK (Syndromes Without a Name)	
HG	Solid Tissue Best Practice Guidelines	http://www.acgs.uk.com/media/765681/acc_solid.tissue_bp_no_v2010_1.00.pdf	2010	Association for Clinical Cytogenetics (now merged into Association for Clinical Genomic Science)	Cytogenetics laboratories	Covers consent

Deliverable report

HG	A common framework of principles for direct-to-consumer genetic testing services	https://www.cellmark.co.uk/pdfs/HGCprinciples.pdf	2010	Human Genetics Commission (HGC)	individual providers, professional organisations, regulatory bodies	
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Deliverable report

HG	UK Biobank Ethics And Governance Framework	https://www.ukbiobank.ac.uk/wp-content/uploads/2011/05/EGF20082.pdf	2007	UK Biobank	UK Biobank project	Sets standards so that all necessary safeguards are in place to ensure that the data and samples are only used for scientifically and ethically approved research.
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Deliverable report

HG	Postnatal best Practice Guidelines	http://www.acgs.uk.com/media/765641/acc_postnatal_bp_mar2007_1.01.pdf	2007	Association for Clinical Cytogenetics (now merged into Association for Clinical Genomic Science)	Clinical cytogenetics	
HG	General best practice guidelines	http://www.acgs.uk.com/media/765607/acc_general_bp_mar2007_1.04.pdf	2007	Association for Clinical Cytogenetics (now merged into Association for Clinical Genomic Science)	Clinical cytogenetics	
HG	Ethical guidance for doctors	https://www.gmc-uk.org/ethical-guidance	Living document	General Medical Council (GMC)	Doctors	

TABLE 3: LIST OF ALL RELEVANT DOCUMENTS FROM NATIONAL ADVISORY/ETHICS GROUPS

SIENNA area	Title of document (original + English translation)	URL	Year	Author/organization	Stated audience	comments

Deliverable report

HG	Sample handling guidance	https://www.genomicsengland.co.uk/wp-content/uploads/2018/02/Sample-Handling-Guidance-v4.0.pdf	2018	Genomics England	National Health Service (NHS) Genomic Medicine Centres (GMCs). It is intended for use by NHS GMC colleagues involved in any aspect of the sample collection and handling process: clinicians, laboratory staff, pathologists, informaticians and project managers.	Covers consent.
HG	Identification and genomic data	http://www.phgfoundation.org/report/identification-and-genomic-data	2018	Foundation for Genomics Population and Health (PHG)	Healthcare professionals, data-processors and policy-makers who are interested in the use of genomic data.	

Deliverable report

HG	Non-invasive prenatal testing: ethical issues	http://nuffieldbioethics.org/wp-content/uploads/NIPT-ethical-issues-full-report.pdf	2017	Nuffield Council on Bioethics	Those who have an interest in the increasing use of NIPT	Covers NIPT for whole genome and exome sequencing
HG	Regulating Advanced Therapies	http://researchbriefings.parliament.uk/ResearchBriefing/Summary/POST-PN-0567	2017	Parliamentary Office of Science and Technology (POST)	Parliamentarians	
HG	Genome Editing	http://researchbriefings.parliament.uk/ResearchBriefing/Summary/POST-PN-0541	2016	Parliamentary Office of Science and Technology (POST)	Parliamentarians	
HG	Genome editing: an ethical review	http://nuffieldbioethics.org/wp-content/uploads/Genome-editing-an-ethical-review.pdf	2016	Nuffield Council on Bioethics	The Council and other identified bodies	

Deliverable report

HG	The retention and storage of pathological records and specimens	https://www.rcpath.org/asset/049EA966-DF5C-4A9F-9353BA24A69BB808/	2015	The Royal College of Pathologists and the Institute of Biomedical Science	Pathologists	
HG	Recommendations: <i>Enabling consistent and responsible data sharing</i>	http://www.phgfoundation.org/documents/507_1450350029.pdf	2015	ACGS and PHG Foundation	NHS, responsible health organisations	Report: <i>Data sharing to support UK clinical genetics and genomics services</i>
HG	Clinical whole genome analysis: delivering the right diagnosis	http://www.phgfoundation.org/briefing/clinical-whole-genome-analysis-delivering-the-right-diagnosis	2014	PHG Foundation	Clinical implementators of genomic analysis	

Deliverable report

HG	Consent and confidentiality in clinical genetic practice: Guidance on genetic testing and sharing genetic information	http://www.bsgm.org.uk/media/678746/consent_and_confidentiality_2011.pdf	2011	Royal College of Physicians, Royal College of Pathologists and British Society for Human Genetics	Professionals within the specialty of clinical genetics and also relevant for all medical specialties	
HG	A guide to genetics	https://www.rcn.org.uk/professional-development/publications/pub-003010	2005	Royal College of Nursing	Non-specialist nurses, midwives, teachers, and patients.	Includes genetic testing, gene therapy, preimplantation genetic diagnosis, genetic testing in pregnancy, genetic counselling and ethical issues, plus a comprehensive glossary and details of support groups and genetic centres in the UK.

Deliverable report

HG	Pharmacogenetics ethical issues	http://nuffieldbioethics.org/wp-content/uploads/2014/07/Pharmacogenetics-Report.pdf	2003	Nuffield Council on Bioethics	Public	
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TABLE 4: LIST OF ALL RELEVANT GUIDANCE DOCUMENTS ON HOW TO WRITE RESEARCH ETHICS PROTOCOLS

Name of national REC	Title of document (original + English translation)	SIENNA area	URL	Stated audience	comments
<i>(**please note not all below listed are NRECs as defined)</i>					
National Health Research Authority	Protocol guidance and template for use in a Clinical Trial of an Investigational Medicinal Product (CTIMP)	HG	https://www.hra.nhs.uk/planning-and-improving-research/research-planning/protocol/	Clinical trial researchers	Includes ethical and regulatory considerations

Deliverable report

National Health Research Authority	Protocol guidance and template for use in qualitative research	HG	https://www.hra.nhs.uk/planning-and-improving-research/research-planning/protocol/	Researchers developing protocols	Includes ethical and regulatory considerations
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TABLE 5A: General questions on your document search in Human Genomics

1- Did you find guidance documents specific to human genomics or genetics?	Yes
2a How many documents did you find from each of these sources that specifically address human genetics or genomics	PECs - 10 NAEG - 12 GDREC – 2 Total: 24
2b Did you find more documents than you had the time to analyse in this task?	Yes, of the above 24, we only had time to analyse six documents. This was also not an exhaustive search (given time limitations and the agreed scope of the work).

Deliverable report

<p>3- a) How many documents specifically ONLY address high throughput genomics? By high throughput genomics, we mean using sequencing of larger parts of the genome, or exome (including large gene panels).</p> <p>3b Please list the year of publication of each documents here:</p> <p>3c please specify the topics addressed explicitly, ideally indicate the number of documents that address each, place an X beside the topic and add the number of docs</p>	<p>PECs – 0 (of those we found) NAEG- 1 (of those we found) GDREC - 0 (of those we found)</p> <p>2017</p> <p>Genomics used for diagnostics in the clinic (genomic testing) X Genomics used for research Consent in genomics in the clinic Consent in genomics in research Return of results in genomics in the clinic Return of results in genomics in research Reinterpretation and recontact in genomics in the clinic Reinterpretation and recontact in genomics in the clinic Genomics for newborn screening Genomics for prenatal screening or testing Non-invasive prenatal testing/screening Discrimination Eugenics Justice Genetic exceptionalism</p>
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Deliverable report

<p>4 a How many documents only address genetics (so testing or screening but without a lot of sequencing)?</p> <p>4b List the years of publications of each document here</p> <p>4c please specify the topics addressed explicitly, ideally indicate the number of documents that address each, place an X beside the topic and add the number of docs</p>	<p>PECs - 6 NAEG – 3 GDREC – 0</p> <p>PECs: 2007 (three docs), 2010, 2014, 2018 NAEGs: 2003, 2011, 2017</p> <p>Genetics used for diagnostics in the clinic (genetic testing) ____8 Genetics used for research____4 Consent in genetics in the clinic ____7 Consent in genetics in research____1 (biobanking), 3 Return of results in genetics in the clinic____2 Return of results in genetics in research____1 Reinterpretation and recontact in genetics in the clinic ____ Reinterpretation and recontact in genetics in research____1 Newborn screening____2 (one was postnatal guidelines) Prenatal screening or testing____1 (<i>prenatal diagnosis</i>) Non-invasive prenatal testing/screening____1 Discrimination____3 Eugenics____1 Justice____3 Genetic exceptionalism____</p> <p><i>(Note: some documents cover or explicitly mention one or more of the above listed topics and the info above takes this into account; the above does not indicate the main focus of the docs. We also only looked for terms listed above)</i></p>
<p>5. How many documents address both genetics and genomics?</p>	<p>1 PEC, 7 NAEG docs cover or mention both.</p>

Deliverable report

<p>6 a) How many documents specifically address Genome Modification, gene editing, crispr, or gene therapy?</p> <p>6b) do they address somatic or germline editing or both?</p>	<p>PECs – 0</p> <p>NAEGs – 1 mentions germline sampling, including somatic and germline editing, 1 mentions genome editing, 1 covers genome editing, 1 mentions genetic modification, gene therapy, somatic therapy and germline therapy, 2 docs mention all terms.</p> <p>Note: in this search, we used the exact terms mentioned in the left column.</p>
<p>How many documents specifically address (so in the title of the document)</p>	<p>Genetic or genomic testing – 1 + 1 (NIPT)</p> <p>Genetic or genomic screening - 0</p> <p>Biobanks or databases - 1</p> <p>Pharmacogenomics or pharmacogenetics - 1</p> <p>Patents - 0</p>

Table 5B RELEVANT PEC or NAEG DOCUMENTS IN HUMAN GENOMICS

<p>Document found via (national associations or google or another database)</p>	<p>HGC website</p>
<p>Title of document</p>	<p>A common framework of principles for direct-to-consumer genetic testing services</p>
<p>Scope/ main topic</p>	<p>Genetic testing</p>
<p>Kind of document (PEC or NAEG or other)</p>	<p>PEC</p>

Deliverable report

Document developed by whom (organisation, profession)?	Human Genetics Commission
Year the document was published	2010
Document saved in folder as	HGC_Principles for direct-to-consumer genetic testing services_2010
Who is the stated audience, if none specified, write not stated (NS)	Individual providers, professional organisations, regulatory bodies
For Clinical or research or both or not specified	Cover all situations in which it is possible for a private consumer to purchase a genetic test without prescription by a qualified medical professional, subject to statutory regulation. Note, do not cover genetic testing carried out purely for medical research purposes, approved by a Research Ethics Committee (REC) where the results of the genetic test are not disclosed to the consumer nor do they apply to genetic tests for forensic purposes.
What level of guideline is provided?	Very broad: i.e.: on many aspects of genomics or genetics, including many issues all together e.g. Very specific: <i>focuses on issues relating to the provision of genetic testing services directly to the consumer</i> More Theoretical (values, principles) More practical (a practitioner could apply them)

<p>What is the scope of the document, and what are the main recommendations?</p>	<p>The Principles cover all situations in which it is possible for a private consumer to purchase a genetic test without prescription by a qualified medical professional, subject to statutory regulation. The Principles address the situation where genetic tests are marketed directly to consumers rather than to qualified medical professionals. The Principles are intended to cover all aspects of direct-to-consumer genetic testing services, including the marketing and advertising of tests, the collection, analysis and storage of biological samples, the interpretation of results and the provision of results to the consumer. These Principles do not cover genetic testing carried out purely for medical research purposes, approved by a Research Ethics Committee (REC) where the results of the genetic test are not disclosed to the consumer nor do they apply to genetic tests for forensic purposes. The Principles also exclude whole genome sequencing, which is not widely commercially available at the time of writing, although this will be considered in the context of planned future revision.</p>
<p>Which life stage is addressed in the document?</p>	<p>Adults Minors (excluding newborns) Newborns Prenatal</p>
<p>Which ethical challenges are addressed in the document? (you can see lists above for ideas of types)</p>	<p>Accuracy in marketing and advertisement of genetic tests, accuracy and adequacy of information to consumers, counselling and support, consent, data protection, use, storage, transfer and disposal of biological samples, interpretation and provision of results, complaints</p>

Deliverable report

<p>How are the ethical issues addressed? (practically, theoretically etc? Any comment on this is fine</p> <p>Are solutions offered? If so, which ones? (depending on the document, this may be redundant with above)</p>	<p>The document outlines requirements relating to the above i.e., what should be done.</p>
<p>How is the document structured? e.g. Which format is used in the document (checklist, continuous text, other)?</p>	<p>Continuous (numbered) text</p>
<p>Is the document clearly understandable?</p>	<p>Yes</p>

Deliverable report

<p>Why is the document important/useful for your country? why did you choose this doc? TELL US WHY IMPORTANT for country and why for SIENNA</p>	<p>As described, the Principles were “developed by a collaborative working group comprising representatives from the genetic testing industry, experts in regulation, clinical and molecular genetics and genetic counselling, representatives from groups that support individuals with genetic conditions and the UK Department of Health. The group was convened and supported by the UK Human Genetics Commission (HGC), the UK Government’s advisory body on developments in human genetics and their ethical, legal, social and economic implications. The purpose of these Principles is to promote high standards and consistency in the provision of genetic tests amongst commercial providers at an international level in order to safeguard the interests of people seeking genetic testing and their families. The Principles identify areas where individual providers, professional organisations, regulatory bodies, and/or national jurisdictions should have defined measures in place, and the nature of those measures”.</p>
<p>Is the document useful for the development of the SIENNA codes and other ethical frameworks? If yes, please explain.</p>	<p>Potentially.</p>
<p>Document found via (national associations or google or another database)</p>	<p>Human Fertilisation and Embryology Authority website</p>
<p>Title of document</p>	<p>Code of Practice</p>

Deliverable report

Scope/ main topic	Human Fertilisation and Embryology, Preimplantation genetic screening (PGS), genetic testing
Kind of document (PEC or NAEG or other)	PEC
Document developed by whom (organisation, profession)?	Human Fertilisation and Embryology Authority
Year the document was published	2018 (edition 9.0)
Document saved in folder as	HFEA_Code of practice_2018
Who is the stated audience, if none specified, write not stated (NS)	Licensed centres, clinics, (reference for patients, donors, donor-conceived people, researchers and those working in the fertility sector).
For Clinical or research or both or not specified	Both
What level of guideline is provided?	Very broad: i.e.: on many aspects of genomics or genetics, including many issues all together e.g. Very specific: only focuses on 1- 2 issues More Theoretical (values, principles) More practical (a practitioner could apply them)

Deliverable report

<p>What is the scope of the document, and what are the main recommendations?</p>	<p>The Code of Practice contains regulatory principles for licensed centres, and guidance notes which provides guidance to help clinics deliver safe, effective and legally compliant treatment and research in compliance with the Human Fertilisation and Embryology Act 1990.</p>
<p>Which life stage is addressed in the document?</p>	<p>Adults Minors (excluding newborns) Newborns Prenatal</p>
<p>Which ethical challenges are addressed in the document? (you can see lists above for ideas of types)</p>	<p>Covers, for example, responsibilities, consent, traceability, fairness, confidentiality, privacy</p>
<p>How are the ethical issues addressed? (practically, theoretically etc? Any comment on this is fine)</p> <p>Are solutions offered? If so, which ones? (depending on the document, this may be redundant with above)</p>	<p>It provides guidance on the above aspects (including others).</p>

Deliverable report

How is the document structured? e.g. Which format is used in the document (checklist, continuous text, other)?	Continuous (numbered) text.
Is the document clearly understandable?	Yes
Why is the document important/useful for your country? why did you choose this doc? TELL US WHY IMPORTANT for country and why for SIENNA	The Code offers good guidance on complying with legal requirements. We chose this document as it is a good example of a Code that takes into account latest evidence (functions as a good up-to date reference) and had an inclusive and participative Code review consultation process (that included workshops and a web-based survey).
Is the document useful for the development of the SIENNA codes and other ethical frameworks? If yes, please explain.	Yes, for reasons cited above.

Document found via (national associations or google or another database)	Nuffield Council on Bioethics website
Title of document	Non-invasive prenatal testing: ethical issues
Scope/ main topic	Non-invasive prenatal testing(NIPT)

Deliverable report

Kind of document (PEC or NAEG or other)	NAEG
Document developed by whom (organisation, profession)?	Nuffield Council on Bioethics
Year the document was published	2017
Document saved in folder as	Nuffield_Non-invasive prenatal testing ethical issues_2017
Who is the stated audience, if none specified, write not stated (NS)	Those who have an interest in the increasing use of NIPT
For Clinical or research or both or not specified	Both
What level of guideline is provided?	Very broad: i.e.: on many aspects of genomics or genetics, including many issues all together e.g. Very specific: only focuses on 1- 2 issues More Theoretical (values, principles) More practical (a practitioner could apply them)

<p>What is the scope of the document, and what are the main recommendations?</p>	<p>This report considers how NIPT could change the way we view pregnancy, disability and difference, and what the wider consequences of its increasing use might be.</p> <p>Main recommendations: The Working Group suggests three general principles, that should always be considered together, to guide policy making in relation to NIPT:</p> <p>Principle 1. The wider societal environment in which NIPT is provided and developed should be considered when developing policy relating to NIPT.</p> <p>Principle 2. Pregnant women and couples should have access, where appropriate, to NIPT within an environment that enables them to make autonomous, informed choices.</p> <p>Principle 3. Efforts should be made to reduce any risks of significant harms posed by the growing use and development of NIPT.</p> <p>It presents some overarching conclusions and recommendations (see below).</p>
<p>Which life stage is addressed in the document?</p>	<p>Adults Minors (excluding newborns) Newborns Prenatal</p>
<p>Which ethical challenges are addressed in the document? (you can see lists above for ideas of types)</p>	<p>It introduces ethical rights and wrongs, and harms and benefits that are relevant to NIPT as understood in terms of the values of choice, autonomy and consent; avoidance of harm; and equality, inclusion and fairness.</p>

<p>How are the ethical issues addressed? (practically, theoretically etc? Any comment on this is fine</p> <p>Are solutions offered? If so, which ones? (depending on the document, this may be redundant with above)</p>	<p>It provides a detailed account of the above ethical issues raised by the increasing availability and use of NIPT, and presents a suggested approach for considering these issues in policy making contexts.</p> <p>The Working Group suggests three general principles, that should always be considered together, to guide policy making in relation to NIPT (see above). It presents some overarching conclusions and recommendations e.g., Women and couples should be able to access NIPT to enable them to find out whether their fetus has a significant medical condition or impairment that manifests at birth or in childhood; NIPT should only be offered if it provides an accurate prediction of whether the fetus has or does not have the condition being tested for; NIPT should not normally be used to test whether a fetus has a less significant medical condition or impairment or an adult onset condition; to find out whether the fetus is the carrier of a gene for any kind of medical condition or impairment; nor to reveal non-medical traits of the fetus, including sex; professional guidance for health and social care professionals on the availability and provision of all types of NIPT in the UK should be developed, and existing guidance on the continuation of pregnancy after diagnosis of a fetal anomaly should be updated and expanded. It also has specific recommendations outlined for NIPT in NHS screening for Down's, Edwards' and Patau's syndromes; NIPT for rare genetic diseases in the NHS, NIPT in the private sector .</p>
<p>How is the document structured? e.g. Which format is used in the document (checklist, continuous text, other)?</p>	<p>Continuous text.</p>

Deliverable report

Is the document clearly understandable?	Yes
Why is the document important/useful for your country? why did you choose this doc? TELL US WHY IMPORTANT for country and why for SIENNA	As there is no UK-specific professional guidance on NIPT, this document is useful one especially for ist coverage of ethical issues.
Is the document useful for the development of the SIENNA codes and other ethical frameworks? If yes, please explain.	Yes, if we are considering codes and frameworks for NIPT.

Document found via (national associations or google or another database)	Nuffield Council on Bioethics website
Title of document	Genome editing: an ethical review
Scope/ main topic	Genome editing
Kind of document (PEC or NAEG or other)	NAEG
Document developed by whom (organisation, profession)?	Nuffield Council on Bioethics

Deliverable report

Year the document was published	2016
Document saved in folder as	Nuffield_Genome editing ethical review_2016
Who is the stated audience, if none specified, write not stated (NS)	The Council and other identified bodies
For Clinical or research or both or not specified	Research
What level of guideline is provided?	Very broad: i.e.: on many aspects of genomics or genetics, including many issues all together e.g. Very specific: only focuses on 1- 2 issues More Theoretical (values, principles) More practical (a practitioner could apply them)

<p>What is the scope of the document, and what are the main recommendations?</p>	<p>The terms of reference were:</p> <p>To identify and define ethical questions relating to developments in genome editing research.</p> <p>To review institutional, national and international policies and provisions relevant to genome editing, and to assess their current and likely future significance.</p> <p>To deliberate and to draw conclusions, as appropriate, about the nature of the ethical questions raised and how they might most suitably be addressed.</p> <p>To report on these matters and to make recommendations, as appropriate, for further initiatives by the Council or by other identified bodies, or for the development or revision of policy or legislation.</p> <p>The report contains no explicit recommendations for action, although it does contain a number of judgements and conclusions.</p>
<p>Which life stage is addressed in the document?</p>	<p>Adults Minors (excluding newborns) Newborns Prenatal</p>
<p>Which ethical challenges are addressed in the document? (you can see lists above for ideas of types)</p>	<p>The document identified a number of key moral perspectives (derived from ist Call for evidence) on genome editing such as Science as a moral enterprise; Intervening in the genome, moral conservatism, normality, moral norms and human rights, welfare and harm, social justice and just society, governance and democracy. The document highlights a number of features of genome editing, especially CRISPR-Cas9 and analogues, have emerged from our inquiry as sources of issues that require further ethical consideration:</p> <p>Novel mode of action Accessibility Speed of use and uptake Multiplexing</p>

Deliverable report

<p>How are the ethical issues addressed? (practically, theoretically etc? Any comment on this is fine</p> <p>Are solutions offered? If so, which ones? (depending on the document, this may be redundant with above)</p>	<p>The document covers each of the moral perspectives in detail (theoretically). The report contains no explicit recommendations for action, although it does contain a number of judgements and conclusions.</p>
<p>How is the document structured? e.g. Which format is used in the document (checklist, continuous text, other)?</p>	<p>Continuous text (also available in online format)</p>
<p>Is the document clearly understandable?</p>	<p>Yes</p>
<p>Why is the document important/useful for your country? why did you choose this doc? TELL US WHY IMPORTANT for country and why for SIENNA</p>	<p>As noted by the Council, “the review was carried out by an interdisciplinary working group that included expertise in science, law, philosophy, ethics, sociology and industry. In coming to its conclusions, the Working Group invited contributions from a wide range of people, including through an open call for evidence that ran from November 2015 until February 2016”. This makes it an important document.</p>

Deliverable report

Is the document useful for the development of the SIENNA codes and other ethical frameworks? If yes, please explain.	It is an important UK document on genome editing (ethical issues).
Document found via (national associations or google or another database)	Royal College of Physicians website
Title of document	Consent and confidentiality in clinical genetic practice: Guidance on genetic testing and sharing genetic information
Scope/ main topic	Genetic testing
Kind of document (PEC or NAEG or other)	NAEG
Document developed by whom (organisation, profession)?	Royal College of Physicians, Royal College of Pathologists and British Society for Human Genetics
Year the document was published	2011
Document saved in folder as	RCP et al_Consent and confidentiality in clinical genetic practice_2011
Who is the stated audience, if none specified, write not stated (NS)	Professionals within the specialty of clinical genetics and also relevant for all medical specialties

Deliverable report

For Clinical or research or both or not specified	Clinical genetic practice
What level of guideline is provided?	Very broad: i.e.: on many aspects of genomics or genetics, including many issues all together e.g. Very specific: only focuses on 1- 2 issues More Theoretical (values, principles) More practical (a practitioner could apply them)

What is the scope of the document, and what are the main recommendations?

It aims to provide up-to-date guidance on issues of consent and confidentiality arising in clinical genetic practice. (clarifies that there is considerable overlap, and consequently no clear distinction from, genomic practice).

Summary of recommendations:

The Joint Committee on Medical Genetics makes the following recommendations for good clinical practice.

As with other types of medical information, genetic information will vary in how sensitive it is: information should not necessarily be viewed as being sensitive, just because it is genetic. Some genetic information is of potential interest not only to the patient, but to others (such as family members, health services or even employers and insurers) and issues of consent and confidentiality should therefore be tailored to the particular situation.

The following issues should usually be discussed as part of a consent process during clinical consultations where medical and family history information is sought and/or where genetic investigations are initiated: (i) knowledge of a family history of a condition or disease, or genetic test results, has a potential benefit to other family members (ii) the fact that communication of certain aspects of information to family members may therefore be recommended (iii) the means of contacting those at-risk family members where relevant (iv) the fact that a summary of relevant clinical and genetic information will usually be sent to other appropriate health professionals (v) the likely timescales for availability of test results (vi) the possibility of unexpected or incidental findings from genetic testing and how these might be managed (vii) the predictive nature of certain genetic tests (for example, indicating risks many years in the future rather than current risks) (viii) the routine practice of long-term storage of samples for possible future analysis and the patient's preferences regarding further testing if it becomes available (ix) the routine practice of using stored samples from one family member as quality assurance for clinical testing in another family member.

Individuals seeking genetic advice are often motivated to do so, at least in part, by feelings of altruism and solidarity towards family members. We support the good practice of facilitating appropriate (and where possible anonymised) use of one person's genetic information to benefit the clinical management of family members. It is helpful to document agreement to such sharing in the patient's notes so that their wishes are clear should other family members be seen.

Where family members are identified as being at risk of developing a genetic condition, attempts should be made to communicate this information, either via the consultant, or where possible and appropriate, via their general practitioner. Such communication can usually be done in a stepwise fashion with first degree relatives being the initial point of contact and subsequent dissemination to appropriate relatives. Details should be provided of how those at risk can seek a genetics referral for themselves.

Family history and clinical information should be shared with other health professionals (regardless of their geographical location) if the sharing of confidential information is necessary for the purposes of healthcare, and disclosure is between healthcare professionals who share in their duty of confidence.

Where genetic testing involves a person who is unable to consent, a decision about testing should be made considering why testing would be in the patient's best interest. This should be documented in the notes.

Access to medical information about deceased relatives can be very helpful in determining or refining risks of disease in persons seeking genetic advice. We recommend that the medical record departments of healthcare facilities should help in accessing medical information about a deceased person for such purposes. In order to develop better evidence-based care and manage services, we support the good practice of sending relevant information to national or international disease or mutation registers. Patients should be informed in general terms that their information may be used in this way.

Health professionals within clinical genetic services should take the lead in disseminating good practice regarding the predictive and familial issues which arise in relation to the testing and storage of genetic samples.

NHS IT strategy should take into account the necessity for long-term storage of diagnostic information for the potential benefit of relatives.

Deliverable report

Which life stage is addressed in the document?	Adults Minors (excluding newborns) Newborns Prenatal
Which ethical challenges are addressed in the document? (you can see lists above for ideas of types)	Issues related to consent and confidentiality in clinical genetic practice.
How are the ethical issues addressed? (practically, theoretically etc? Any comment on this is fine) Are solutions offered? If so, which ones? (depending on the document, this may be redundant with above)	<p>The main section of the document introduces general principles of consent and confidentiality in clinical genetic practice and expands on these using hypothetical (but based on real) clinical case scenarios as illustrations to highlight key points.</p> <p>A summary of recommendations is provided (see above).</p>
How is the document structured? e.g. Which format is used in the document (checklist, continuous text, other)?	Continuous text
Is the document clearly understandable?	Yes

Deliverable report

<p>Why is the document important/useful for your country? why did you choose this doc? TELL US WHY IMPORTANT for country and why for SIENNA</p>	<p>As Lucassen and Hall state, “the report aims to guide healthcare professionals through the complex web of legislation and professional guidance relating to the use of genetic data and samples. It recognises the growing uncertainty among healthcare professionals about the duties and responsibilities they may have towards relatives of a patient, including when it might be appropriate or legitimate to utilise genetic information from one person for the benefit of another in order to recommend appropriate interventions. At the same time it attempts to formalise procedures to recognise that many service users attend genetic clinics not just for themselves, but often (at least in part) in order to help their families.” Further, the guidance provides an applied analysis of relevant legislation. See: http://www.clinmed.rcpjournals.org/content/12/1/5.full</p>
<p>Is the document useful for the development of the SIENNA codes and other ethical frameworks? If yes, please explain.</p>	<p>Yes, for coverage of issues of consent and confidentiality in clinical genetic practice.</p>

<p>Document found via (national associations or google or another database)</p>	<p>Nuffield Council on Bioethics website</p>
<p>Title of document</p>	<p>Pharmacogenetics ethical issues</p>
<p>Scope/ main topic</p>	<p>Pharmacogenetics</p>
<p>Kind of document (PEC or NAEG or other)</p>	<p>NAEG</p>

Deliverable report

Document developed by whom (organisation, profession)?	Nuffield Council on Bioethics
Year the document was published	2003
Document saved in folder as	Nuffield_Pharmacogenetics ethical issues_2003
Who is the stated audience, if none specified, write not stated (NS)	Public
For Clinical or research or both or not specified	Both
What level of guideline is provided?	Very broad: i.e.: on many aspects of genomics or genetics, including many issues all together e.g. Very specific: only focuses on 1- 2 issues (<i>focus on pharmacogenetics</i>) More Theoretical (values, principles) More practical (a practitioner could apply them)

<p>What is the scope of the document, and what are the main recommendations?</p>	<p>The report aims to encourage discussion of the issues and makes recommendations for future policy and practice. Terms of reference:</p> <p>1 To explore what pharmacogenetics offers now and is likely to offer in the near future; In particular to examine the effect of pharmacogenetics on: a) the design of medicines, the promotion of efficacy and safety in the administration of medicines to individuals; b) the conduct of trials in the context of pharmaceutical research & development; c) clinical practice.</p> <p>2 To consider ethical issues specifically raised by pharmacogenetics; In particular to examine the areas of: a) consent, privacy and confidentiality; b) the management of information about response likelihood; c) the implications of differentiating individuals into groups based on response likelihood.</p> <p>3 To consider the implications for the provision of healthcare.</p> <p>Main recommendations: It includes a detailed summary and recommendations (40 in total) covering: The nature of pharmacogenetic information The development of new medicines Using pharmacogenetics to improve existing medicines The use of pharmacogenetic information collected in research The use of pharmacogenetic information collected in research Withdrawn medicines The allocation of resources Stratification and the development of new medicines Pharmacogenetics and racial groups Clinical judgement and patient choice Privacy and confidentiality of pharmacogenetic information</p>
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Deliverable report

<p>Which life stage is addressed in the document?</p>	<p>Adults Minors (excluding newborns) Newborns Prenatal (<i>mentioned once</i>)</p>
<p>Which ethical challenges are addressed in the document? (you can see lists above for ideas of types)</p>	<p>Consent Allocation of resources Pharmacogenetics and racial groups Clinical judgement and patient choice Information, training and education Responsibility for test and treatment Off-label use in developing countries Privacy and confidentiality of pharmacogenetic information Use by third parties</p>
<p>How are the ethical issues addressed? (practically, theoretically etc? Any comment on this is fine)</p> <p>Are solutions offered? If so, which ones? (depending on the document, this may be redundant with above)</p>	<p>There is some discussion of the issues and some recommendations.</p>
<p>How is the document structured? e.g. Which format is used in the document (checklist, continuous text, other)?</p>	<p>Continuous text</p>

Deliverable report

Is the document clearly understandable?	Yes
Why is the document important/useful for your country? why did you choose this doc? TELL US WHY IMPORTANT for country and why for SIENNA	As described by itself, the report “aims to contribute to that process of anticipating the proper structures of incentive and constraint to guide the development and use of pharmacogenetics, from an ethical perspective” and it “aims to help people with diverse backgrounds and interests to think productively about the difficult and important ethical questions raised by pharmacogenetics.”
Is the document useful for the development of the SIENNA codes and other ethical frameworks? If yes, please explain.	See above.

Table 5C GDREC DOCUMENTS for HUMAN GENOMICS

Document found via (national associations or google or another database)	NHS HRA website
Title of document	Protocol guidance and template for use in a Clinical Trial of an Investigational Medicinal Product (CTIMP)
Scope/ main topic	
Kind of document (PEC or NAEG or other)	GDREC

Deliverable report

Document developed by whom (organisation, profession)?	NHS Health Research Authority
Year the document was published	2016
Document saved in folder as	HRA_ctimp-protocol-development-tool_2016
Who is the stated audience, if none specified, write not stated (NS)	Clinical trial researchers
For Clinical or research or both or not specified	Clinical trial research
What level of guideline is provided?	Very broad: i.e.: on many aspects of genomics or genetics, including many issues all together e.g. Very specific: only focuses on 1- 2 issues More Theoretical (values, principles) More practical (a practitioner could apply them)
What is the scope of the document, and what are the main recommendations?	The document is a Protocol guidance and template for use in a Clinical Trial of an Investigational Medicinal Product (CTIMP).
Which life stage is addressed in the document?	Adults Minors (excluding newborns) Newborns Prenatal

Deliverable report

<p>Which ethical challenges are addressed in the document? (you can see lists above for ideas of types)</p>	<p>It covers a wide range of ethical issues related to clinical trials across the document.</p> <p>E.g., ethical issues related to trial procedures such as participant recruitment (inclusion and exclusion criteria, payments etc), consent, randomisation, blinding, emergency unblinding, etc</p>
<p>How are the ethical issues addressed? (practically, theoretically etc? Any comment on this is fine)</p> <p>Are solutions offered? If so, which ones? (depending on the document, this may be redundant with above)</p>	<p>It has guidance on ethical issues and a specific section on ethical and regulatory considerations.</p>
<p>How is the document structured? e.g. Which format is used in the document (checklist, continuous text, other)?</p>	<p>As a template with detailed guidance.</p>
<p>Is the document clearly understandable?</p>	<p>Yes.</p>

Deliverable report

<p>Why is the document important/useful for your country? why did you choose this doc? TELL US WHY IMPORTANT for country and why for SIENNA</p>	<p>Per the HRA, guidance and template aims to support researchers developing protocols where the sponsor does not already use a template, support sponsors wishing to develop template protocols in line with validated guidance and support sponsors to review their existing protocol template to assess whether it is in line with national guidance. https://www.hra.nhs.uk/about-us/consultations/closed-consultations/qualitative-protocol-guidance-and-template-consultation/</p>
<p>Is the document useful for the development of the SIENNA codes and other ethical frameworks? If yes, please explain.</p>	<p>Relevant for development of research ethics protocol.</p>

7.13 USA

Due to the fact that the American partner did not have budget assigned to this task, only basic summary of relevant documents is provided in the Section 3.1.2.

Annex 2: International search – detailed results

Year	Title	Organization	URL	I/R	R/C	Specific	Saved as
2017	Recommendation of the OECD Council on Health Data Governance	OECD	http://www.oecd.org/health/health-systems/Recommendation-of-OECD-Council-on-Health-Data-Governance-Booklet.pdf	I	R, C	no	Recommendation-of-OECD-Council-on-Health-Data-Governance-Booklet.pdf
2016	CIOMS International Ethical Guidelines for Health-related Research Involving Humans	Council for International Organization of Medical Sciences (CIOMS)	https://cioms.ch/wp-content/uploads/2017/01/WEB-CIOMS-EthicalGuidelines.pdf	I	R	no	WEB-CIOMS-EthicalGuidelines
2016	WMA Declaration of Taipei on Ethical Considerations regarding health databases and biobanks	World Medical Association	http://www.humgen.org/virtual_library/4830_1/web/images/4830.pdf	I	R	no	WMA'17...

2015	Amsterdam Declaration on Genetic and Health Data, Challenges for Tomorrow and Data Protection Oversight of Security and Intelligence: The Role of Data Protection Authorities in a Changing Society	International Conference of Data Protection and Privacy Commissioners	https://icdppc.org/wp-content/uploads/2015/02/Amsterdam-Declaration-.pdf	I	R, C	yes	Amsterdam-declaration.pdf
2015	Medical Ethics Manual	World Medical Association	https://www.wma.net/wp-content/uploads/2016/11/Ethics_manual_3rd_Nov2015_en.pdf	I	C	no	Ethics_manual_3 rd
2015	An Update to Returning Genetic Research Results to Individuals: Perspectives of the Industry Pharmacogenomics Working Group	Industry Pharmacogenomics Working Group	http://www.humgen.org/databas e-laws-policies-results#National	I	R	yes	Industry Pharmacogenomics

2015	Whole-genome sequencing in newborn screening? A Statement on the continued importance of targeted approaches in newborn screening programmes	European Society of Human Genetics (ESHG) Human Genome Organization (HUGO) PHG Foundation Public Population Project in Genomics and Society (P3G)	https://www.nature.com/articles/ejhg2014289	R	C	yes	Newborn HCH'15
2014	Framework for Responsible Sharing of Genomic and Health-Related Data	Global Alliance for Genomics & Health	https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4685158/pdf/11568_2014_Article_3.pdf	I	R, C	Yes, but relevant to others too	GA4GH'14

Table 3.2.1. List of documents providing guidance with regard to the activities of “exploring genome” as oppose to “modifying genome” which is address in Table 2. I/R - International/Regional; R/C - Research/Clinic; Specific - Specific to genetics/genomics or not.

Year	Title	Organization	URL	I/R	R/C	Specific	Saved as
2017	The application of Genome Editing in humans	Federation of European Academies of Medicine (FEAM)	https://www.feam.eu/wp-content/uploads/HumanGenomeEditingFEAMPositionPaper2017.pdf	R	R,C	yes	HumanGenomeEditingFEAMPositionPaper2017.pdf
2017	Ethical issues of CRISPR technology and gene editing through the lens of solidarity	Human Genome Organization (HUGO)	https://academic.oup.com/bmb/article/122/1/17/3045812	I	R, C	yes	HUGO'17
2017	Human Genome Editing: Science, Ethics, and Governance	National Academies of Sciences, Engineering, and Medicine National Academy of Sciences (NAS)	https://www.nap.edu/read/24623/chapter/1	I	R, C	yes	NAS'17

Year	Title	Organization	URL	I/R	R/C	Specific	Saved as
2016	Mitochondrial Replacement Techniques: Ethical, Social, and Policy Considerations	National Academies of Sciences, Engineering, and Medicine	http://www.humgen.org/virtual_library/4778_1/web/images/4778.pdf	I	R, C	Yes (precisely molecular biology...)	NASEM'16 mt
2015	On Human Gene Editing: International Summit Statement	National Academies of Sciences, Engineering, and Medicine	http://www.humgen.org/virtual_library/4779_1/web/images/4779.pdf	I	R, C	yes	NASEM'15
2015	Statement on Genome Editing Technologies and Human Germline Genetic Modification	Hinxton Group, an International Consortium on Stem Cells, Ethics and Law	http://www.hinxtongroup.org/hinxton2015_statement.pdf	I	R, C	yes	Hinxton_statement

Year	Title	Organization	URL	I/R	R/C	Specific	Saved as
2015	The Alliance for Regenerative Medicine's (ARM) Position Statement Regarding Human Embryo or Germline Genome Modification	Alliance for Regenerative Medicine (ARM)	https://alliancerm.org/sites/default/files/ARM%20Normative%20Stance_FINAL.pdf	I	R, C (?)	yes	ARM Normative Stance_FINAL
2015	The ISSCR Statement on Human Germline Genome Modification	International Society for Stem Cell Research (ISSCR)	http://www.isscr.org/docs/default-source/policy-documents/isscr-statement-on-human-germline-genome-modification.pdf?sfvrsn=0	I	R, C	yes	isscr-statement-on-human-germline-genome-modification

Table 3.2.2. List of documents providing guidance with regard to the activities “modifying genome”. I/R - International/Regional; R/C - Research/Clinic; Specific - Specific to genetics/genomics or not.

Year	Title	Organization	Url	I/R	R/C	Specific	Saved us
2015	Report of the IBC on Updating Its Reflection on the Human Genome and Human Rights	UNESCO International Bioethics Committee (IBC)	- http://unesdoc.unesco.org/images/0023/002332/233258E.pdf	I	R, C	yes	UNESCO genome'15.pdf
2015	Ethical Issues in Obstetrics and Gynecology	International Federation of Gynecology and Obstetrics (FIGO) - Committee for the Study of Ethical Aspects of Human Reproduction	https://www.figo.org/sites/default/files/uploads/wg-publications/ethics/FIGO%20Ethical%20Issues%202015.pdf4893.pdf	I	R, C	no	FIGO Ethical Issues 2015.pdf
2015	WMA Declaration of Lisbon on the Rights of the Patient	World Medical Association	https://www.wma.net/policies-post/wma-declaration-of-lisbon-on-the-rights-of-the-patient/	I	C	No	wma-declaration-of-lisbon-on-the-rights-of-the-patient

Year	Title	Organization	Url	I/R	R/C	Specific	Saved us
2015	Global Health Ethics: Key Issues	World Health Organization	http://apps.who.int/iris/bitstream/handle/10665/164576/9789240694033_eng.pdf?sequence=1	I	C, R	No	WHO health ethics'15
2015	The Hague Declaration on Knowledge Discovery in the Digital Age	Association of European Research Libraries	https://thehaguedeclaration.com/wp-content/uploads/sites/2/2015/04/Liber_DeclarationA4_2015.pdf	R	R	No	Liber_Declaration
2014	Code of Practice for the Operation of the European Human Pluripotent Stem Cell Registry	European Human Embryonic Stem Cell Registry	https://hpscereg.eu/docs/downloads/Code_of_Practice_hPSreg_v1_%202014_03_21.pdf	R	R	No	Code_of_Practice_hPSreg_v1_2014_03_21

Table 3.2.3. List of documents providing guidance with regard to the activities of both “exploring genome” and “modifying genome”. I/R - International/Regional; R/C - Research/Clinic; Specific - Specific to genetics/genomics or not.

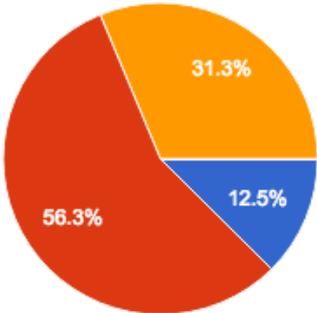
Annex 3: Online Survey – questions and answers

The online survey was developed for all three SIENNA areas, therefore questions are asked not only regarding AI&R, but also regarding Human Genomics and Human Enhancement. The responses were stripped from identifying information.

Questions and answers

With respect to Human Genomics, to what extent are you aware of technologies in this area?

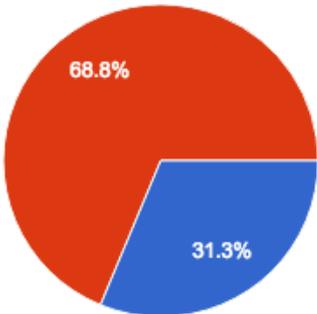
16 responses



- I am not aware of technologies in this area.
- I am slightly aware of technologies in this area.
- I am fully aware of technologies in this area.
- I am an expert in this technology area.
- I am not sure how to answer this question.

With respect to Human Enhancement, to what extent are you aware of technologies in this area?

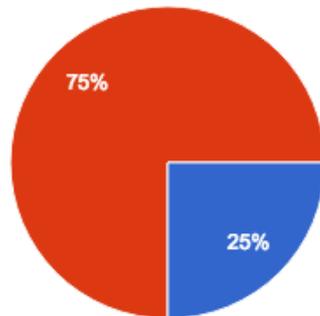
16 responses



- I am not aware of technologies in this area.
- I am slightly aware of technologies in this area.
- I am fully aware of technologies in this area.
- I am an expert in this technology area.
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With respect to Artificial Intelligence and Robotics to what extent are you aware of technologies in this area?

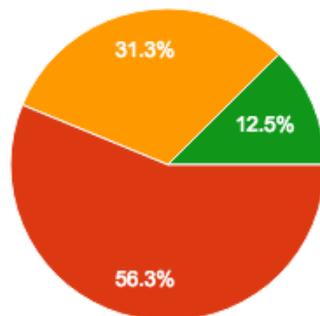
16 responses



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How aware are you of the ethical, legal and social issues relating to Human Genomics?

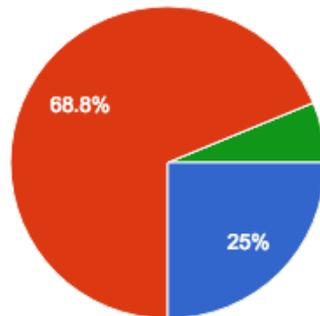
16 responses



- I am not aware of the ethical, legal or social issues.
- I am slightly aware of the ethical, legal or social issues.
- I am fully aware of the ethical, legal or social issues.
- I am an expert in the ethical, legal or social issues.
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How aware are you of the ethical, legal and social issues relating to Human Enhancement?

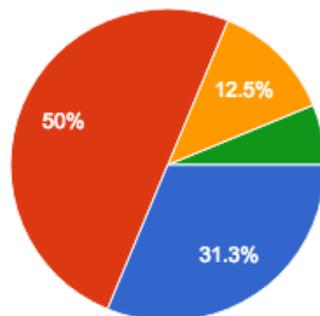
16 responses



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- I am an expert in the ethical, legal or social issues.
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How aware are you of the ethical, legal and social issues relating to Artificial Intelligence and Robotics?

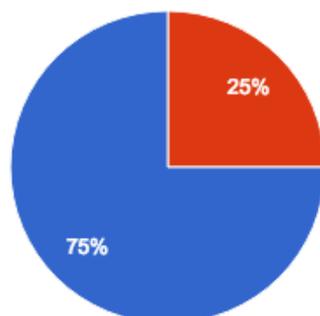
16 responses



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- I am an expert in the ethical, legal or social issues.
- I am not sure how to answer this question.

Does your REC address or offer any specific guidance for researchers working in Human Genomics?

16 responses



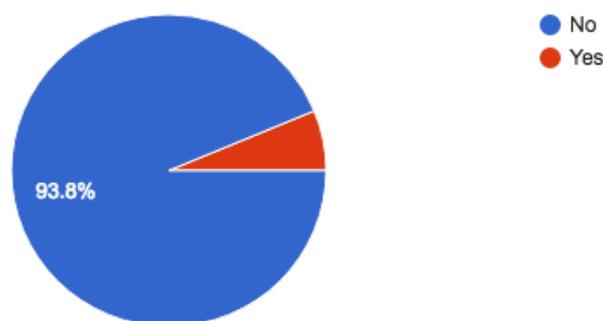
- No
- Yes

If yes, please provide links and/or information here: 4 responses

- The question is ambiguous as any REC has the responsibility to assess protocols on human genomics. Yet it does not necessarily require to draft specific guidance documents as there are already some available and the general principles of research ethics apply in any case which means that specific guidance documents may not be needed.
- we approve all medical research projects
- <http://www.nvk.dk/~media/NVK/Dokumenter/Guidelines-on-Genomics-Research.pdf?la=da>
- Management of Incidental Findings in projects involving whole-genome sequencing (<http://www.cner.lu/en-gb/procedures/incidentalfindings.aspx>)

Does your REC address or offer any specific guidance for researchers working in Human Enhancement?

16 responses

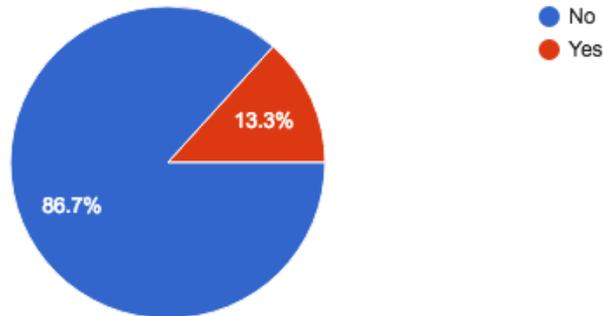


If yes, please provide links and/or information here: 2 responses

- The question is ambiguous as any REC has the responsibility to assess protocols on "human enhancement" (whether they are identified as such or not). Yet it does not necessarily require to draft specific guidance documents as there are already some available and the general principles of research ethics apply in any case which means that specific guidance documents may not be needed. In addition, human enhancement is as such a confusing concept as it implies that the research is actually enhancing human while some would argue that such technological enhancement may well be an impoverishment of mankind in a philosophical viewpoint. Innovation does not always equal progress.
- we have not received applications for this yet

Does your REC address or offer any specific guidance for researchers working in Artificial Intelligence and Robotics?

15 responses

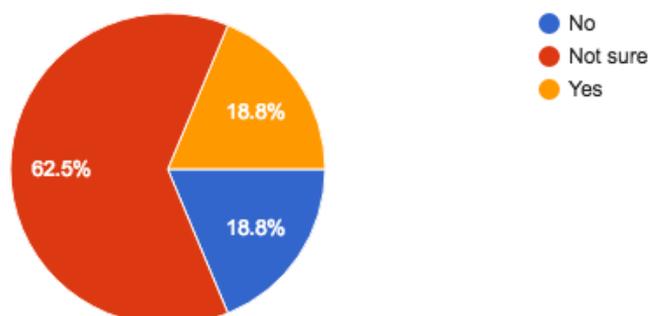


If yes, please provide links and/or information here: 2 responses

- The question is ambiguous as any REC has the responsibility to assess protocols on AI and Robotics. Yet it does not necessarily require to draft specific guidance documents as there are already some available and the general principles of research ethics apply in any case which means that specific guidance documents may not be needed.
- when the projects use real patient data

Does your REC have future plans to specifically deal with ethical, legal and social issues of Human Genomics?

16 responses



If yes, please specify here: 3 responses

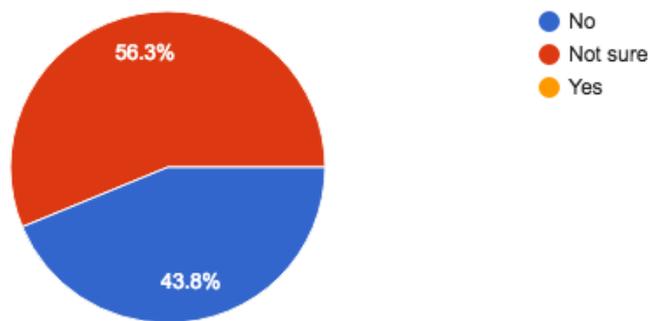
- RECs normally have limited control on the researches submitted to them. The region around the [X] presents itself as the health valley. This is therefore likely that there will be increased

research activities in this field. The REC will then adapt itself to this evolution (see remarks above).

- we have published guidelines
- We plan to help researchers to balance health needs and risks of high expectations, exploitation

Does your REC have future plans to specifically deal with ethical, legal and social issues of Human Enhancement?

16 responses

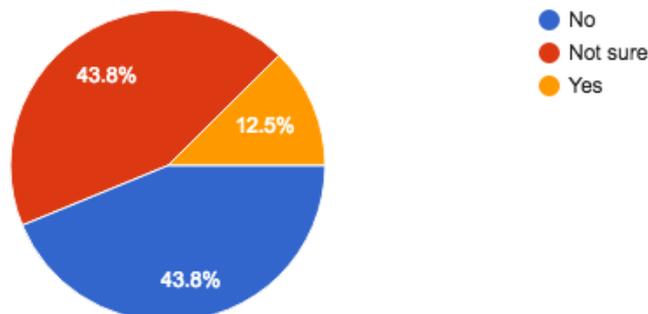


If yes, please specify here: 1 response

- It all depends on what is meant by human enhancement. Advance research is done on exoskeleton and repairing brain damages. There is also a lot of activities around doping. This is therefore likely that there will more activities in this field in the future (see remark on human genomics).

Does your REC have future plans to specifically deal with ethical, legal and social issues of Artificial Intelligence and Robotics?

16 responses

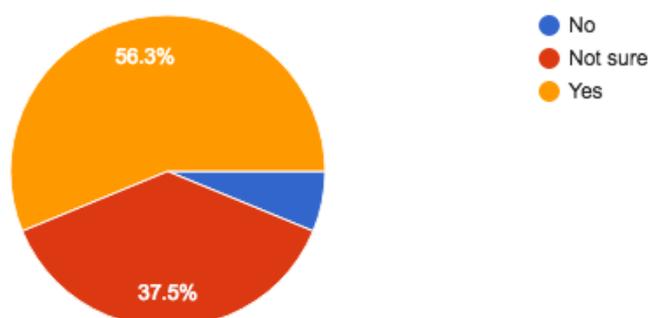


If yes, please specify here:3 responses

- RECs normally have limited control on the researches submitted to them. The region around the [X] presents itself as the health valley. This is therefore likely that there will be increased research activities in this field. The REC will then adapt itself to this evolution (see remarks above).
- [Z] is organising a symposium, specifically designed for members of the [Y] ethics committees, on ethical, legal and social issues of artificial intelligence, in [X] in 2018.
- Topics like data protection and validation of research are more important in big data;

Do you think there is a need to offer additional guidance to people doing research in Human Genomics?

16 responses

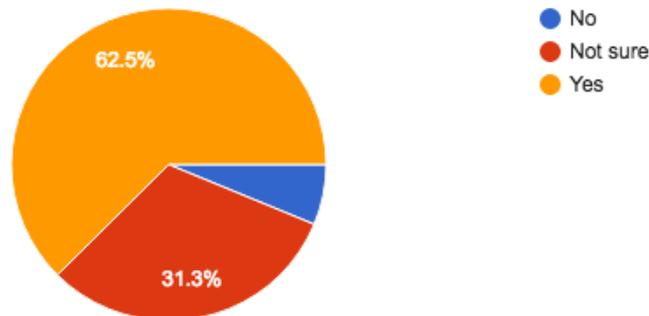


If yes, please specify here: 4 responses

- As recent (and past) history, most abuses do not happen due to a lack of norms but rather a lack of consideration for them and their underlying principles. Producing more norms has been a trend in research ethics and regulation since WWII. As Jay Katz said in 1969: "The proliferation of such codes testifies to the difficulty of promulgating a set of rules that does not immediately raise more questions than it answers. At this stage of our confusion, it is unlikely that codes will resolve many of the problems, though they may serve a useful function later. Even the much endorsed Declaration of Helsinki – praised, perhaps, because it is the newest and therefore the least examined – will create problems for those who wish to implement it". There has been limited progress in raising the ethical mentality within research institutions. Of course, this would be less lucrative for ethics centres as the industry and others are less likely to finance virtues behaviour rather than workshops and other publications.
- There is a need for the informed consent in this field
- risk management, realistic expectations
- Ethically difficult issue with rapid development

Do you think there is a need to offer additional guidance to people doing research in Human Enhancement?

16 responses

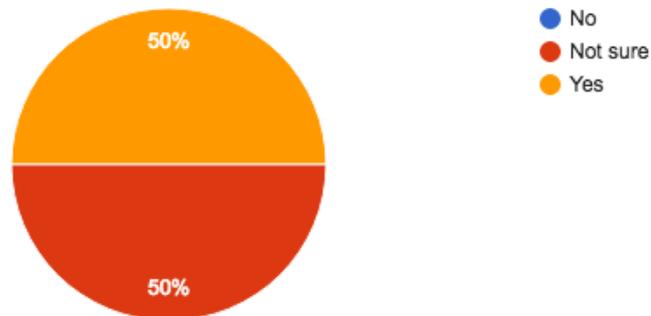


If yes, please specify here: 5 responses

- Not only the same remarks apply than for human genomics, but the very concept of "human enhancement" is at best confusing, at worst the entry door to totalitarianism. The very idea that humans need to be enhanced is worrying, especially if you refer to the previous time in history when similar proposals were formulated and, even worst, tested. As Hans Jonas said in 1969 (again), "Let us not forget that progress is an optional goal, not an unconditional commitment, and that its tempo in particular, compulsive as it may become, has nothing sacred about it". The best guidance in fact would be to explain to researchers why "human enhancement" should be banned as a concept.
- a guide for REC members regarding the ethical concerns such research projects may raise and possible approaches to deal with them could be useful
- The knowledge about these issues and their development is scarce. Identifying the ethical problems they pose is the first step
- risk Management, use and abuse,
- It is necessary to draw a line between enhancement and mere addiction to anything new

Do you think there is a need to offer additional guidance to people doing research in Artificial Intelligence and Robotics?

16 responses

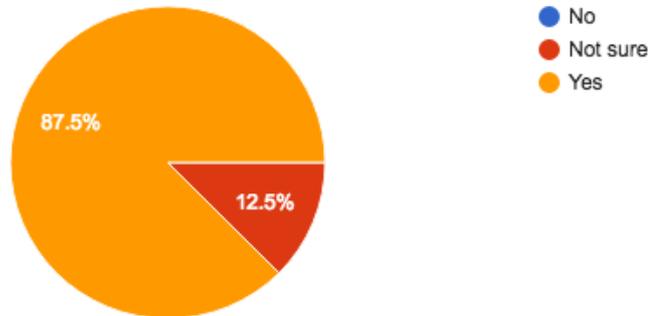


If yes, please specify here: 5 responses

- see remarks on human genomics
- a guide for REC members regarding the ethical concerns such research projects may raise and possible approaches to deal with them could be useful
- The knowledge about these issues and their development is scarce. Identifying the ethical problems they pose is the first step
- consequences of automated decision support
- Quite dangerous research with unpredictable progress

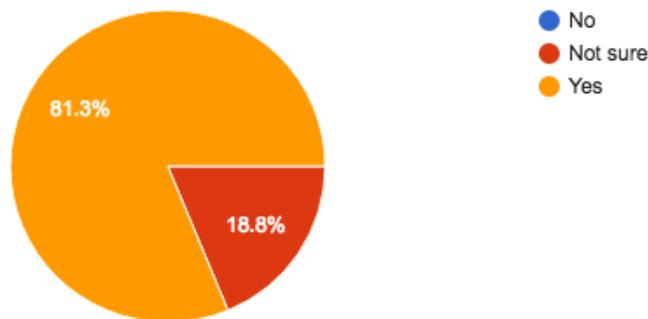
Do you think there is a need to offer additional education and training for REC members to learn more about ethical and social issues in Human Genomics?

16 responses



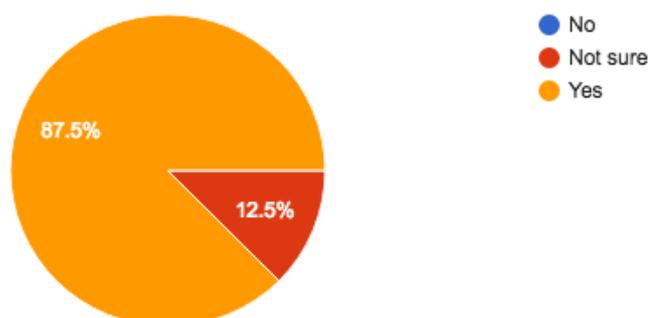
Do you think there is a need to offer additional education and training for REC members to learn more about ethical and social issues in Human Enhancement?

16 responses



Do you think there is a need to offer additional education and training for REC members to learn more about ethical and social issues in Artificial Intelligence and Robotics?

16 responses



What do you think are the most pressing needs/challenges facing RECs in Europe today? (open question) 12 responses

- As Adam Smith shrewdly pointed out in 1775: "A degree can pretend to give security for nothing but the science of the graduate; and even for that it can give but a very slender security. For his good sense and discretion, qualities not discoverable by an academical examination, it can give no security at all". promoting training of researchers and RECs' members is certainly a priority, but one can notice that such training does not offer much protection to research participants if its is not followed by a truly ethical evaluation of research with the aim to protect human participants before all other priorities. Instead of training, it seems time to educate all actors in the field that the rules they have learned actually apply to them and that they are morally and legally responsible to implement them.
- GDPR and open data
- In my opinion one of the major challenges is what happens in certain online communities of patients in which we assisted to two new phenomena: Lay crowdsourcing expertise and Patient Led Research. This bottom up kind of patient empowerment put in question the ethics regulation system.
- 1. lack of resources to adapt to new EU legal requirements; 2. fast advances in big data and genetic research; 3. lack of free international training offered to new REC members
- Do the RECs need to rethink the way they review the research projects to cope with the recent , and soon to come, changes in the EU legislations (CTR, MDR, IVDR)? Paediatric research, Data protection in an international research setting, New technologies: CAR-T cell therapy, CRISPR, AI and robots.
- Training
- The most pressing needs concern genomics and artificial intelligence
 - Training in emerging technologies and associated ethical issues
 - Resources for administration
 - Communication with RECs in other institutions and jurisdictions/regions.
- In my opinion, the major challenges for Italian RECs in this moment are to keep their independence and to assure reliable evaluations despite short timelines. Certainly, another important general need is to be updated with respect to new technologies and related ELSI
- there is no common legal base; there is no common social consent about "what is possible" and "what should be avoided"
- Lack of communication among RECs across Europe; theoretical background is gradually vanishing; education of members sharply differs in different countries.

If you have any other comments/suggestions/feedback which might help us, please specify here:4 responses

- The main difficulty in dealing with the latest innovation in biomedical progress is to confuse the technical enhancement they provide with a human one. Going back to the principles should always be the easiest solution. Yet, the scientific community and those expected to guide them in their action often prefer to create new rules to accommodate the so-called progress that industry, the market and the States are hoping for.
- I think that there are many new subjects that challenge the ethics regulation in force that's the need to reflect on these new issues is URGENT there not many expert that we have to set up meetings , workshops with the people that work in these domains
- n/a
- These methods will probably change the classical clinical trials, including biostatistical concepts, regulatory aspects, research methodology and social attitudes

8. Project officer request for revisions

8.1 Message from project officer to revise D2.3

"Request for revision of deliverable submission for the project "SIENNA (741716)".

On date 28/05/2019, the officer "Cristina MARCONE" has requested the revision of the submission for the deliverable with title "Survey of REC approaches and codes for genomics" and number "3" for the project "SIENNA (741716)".

Request for revision comment:

The deliverable provides an extremely detailed analysis (306 pages, including annexes) of existing ethical codes, protocols and REC approaches concerning human genomics. It presents the results of an extensive search for ethical documents (used by professional organisations, ethics advisory groups, and research ethics committees) which could provide normative guidance in the field. The results of the online survey among representatives of research ethics committees are included (bad timing of the survey resulted in poor response and diminished the relevance of the obtained data). The scope of the search and the amount of effort put into the activity should be commended, but the results are lacking a comprehensive summary and a specific conclusion (very general conclusions are provided in the deliverable)."

8.2 Answer from authors in revision of task 2.3

1- regarding the survey of RECs

It is important to note that a response rate of 43%, while not stellar is a fine rate for the time allotted and for our goals in this task and for the use of the results for SIENNA as a whole. Indeed academic journals often prefer a 60% or greater response rate when authors want to claim that the results are representative and generalizable. However, the lower response rate is less of an issue given that this survey was exploratory (i.e. not meant to test a hypothesis where an effect size would require a certain minimal sample size) nor is it a stand alone study where the results would be the end in and of themselves; rather, they provide complementary information to the the guideline search and both are meant as a resource to aid in future SIENNA tasks.

We have added the following to the text to make this clearer

"Sending the survey to EUREC members can be seen as a first round. For task 5.1 (the development of operational guidelines for RECs beyond biomedical research) EUREC will conduct semi-structured interviews with REC members, which focus also on other research fields then biomedical. EUREC will include the most important questions from the online survey in these semi-structured interviews. This shall lead to further insights."

2- Regarding the comprehensive summary and specific conclusion of the report

As stated above, task 2.3 results (guidelines and information from RECs) are meant to be a resource for future SIENNA work, such as 2.7, and much of WP5. As such, a descriptive approach was used to present the work. We highlight that the results section offers a summary of findings. We have nonetheless also added a conclusion specific to human genetics and genomics.

“In total, one hundred and sixty-four documents which specifically address human genetics and/or genomics were retrieved from 11 countries where country studies were specifically performed: Brazil, China, France, Germany, Greece, The Netherlands, Poland, South Africa, Spain, Sweden, and the United Kingdom. In addition, 22 documents from “international groups” like the OECD, or HUGO were also identified (between 2014-2017) (Annex 2); this includes documents from the USA since despite being issued at the national level, they often have worldwide recognition and/or impact. There was a large range of number of documents with the UK, France and Greece reporting mid to high twenties, and South Africa only reporting two documents addressing human genetics or genomics. Of the documents reported, most addressed genetic (testing or screening, excluding sequencing/genomics; rang 1-9), then a smaller portion addressed gene therapy or gene editing (range 0-6) and even less addressed high throughput genomics/sequencing (range 0-4) (table 4). This is not completely surprising since the latter two areas (re)emerged only recently in comparison to more traditional genetic testing and screening. That being said, having the concrete data to support our focus (in all of WP2 to date) on genomic sequencing and gene editing is valuable; it gives us evidence that we are working in areas where additional guidance is necessary. Indeed, this turns out to be somewhat different than what was initially described in the grant call (i.e. to focus on genetic testing and screening among other things). However, the way we have organized the tasks, we nevertheless still include these subjects in our work, yet give more focus on ELSI of genomic sequencing and gene editing.

Furthermore, the responses from the survey with REC members’ also indicate that addressing the ELSI of genomics is not only needed, but would be supported by the large majority of respondents in the context of research ethics. Respondents were in agreement that additional education on the ELSI of HG would be helpful.

In conclusion, this report provides information supporting our course of action in WP2 on ELSI of Human Genetics and Genomics, and it also offers a resource or tool to better inform the work required in task 2.7 (ethical framework in HG; e.g. what are the specific gaps? Are there documents we can use as a starting point?) and WP5 (consortium proposal for RECS and stakeholders).

9. References

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