Analysis of the legal and human rights requirements for genomics in and outside the EU

[WP2 – Human Genetics and Genomics, task 2.2]

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Executive Summary

The overall aim of this report is twofold. First, to examine how the law currently responds to challenges in the area of genetics and genomics, and identify what challenges, limitations and gaps emerge. Secondly, to identify key human rights norms and regulatory approaches that could be examined further for shaping legal responses to the new and emerging technology in the area with due regard to competences and authority of various actors regulating/contributing to shaping the regulatory environment in the area. The two aims are interrelated, so that aim two builds on the analysis of the current legal responses.

To achieve aims of SIENNA task 2.2, this report, through a literature review, identifies ethical concerns in the area of genomics (Chapter 3). Then it applies legal dogmatic and law-in-context methods and analyses the legal and human rights responses in the area of genomics at the international and regional human rights legal orders (Chapter 4), and the EU (Chapter 5). Furthermore, it carries out comparative analysis in selected EU Member States and non-EU countries and surveys the legal responses, academic legal discussions and legal developments in the areas of concern, and through a comparative review, it examines national comparative perspectives against the international and regional norms and human rights standards (Chapter 6). Finally, it synthesizes the findings and identifies key human rights and legal challenges that emerge concerning genetics and genomics and shows the convergences and distinctions in the regulation of genomics and the challenges this presents for future innovation (Chapter 7). Lastly, it offers concluding reflections on regulating genomics (Chapter 8).

The report shows overlaps and distinctions in regulating questions about genomics. While the UNESCO and CoE are considered as "frontrunners" in the area, many of the challenges can also be addressed in other legal orders of concern with due regard to the general human rights frameworks in place. Genomics touches upon a number of sensitivities where neither a straightforward human rights response exist, nor through the balancing act of competing rights and interests, the ultimate and right conclusion can be achieved. Although commonly reflections regarding the adequacy of the existing regulatory responses have been made about the surveyed national legal orders, a key question vis-a-vis SIENNA aims is whether given various sensitivities the area of genomics raises, a common approach could be found, and if so, what elements would constitute this approach. A common denominator might not necessarily be an adequate regulatory approach. Instead, in the subsequent tasks of SIENNA project account for tools that could help to find the balance might need to be given.

The juxtaposition of the identified challenges related to genetics and genomics with the mapped relevant international human rights norms proves that human rights framework may provide for an important point of reference for shaping legal responses. At the same time, the analysis suggests that in many aspects, the existing human right sources offer rather a starting point for further examinations and elaborations than a closing argument. In our view, the starting points should be grounded in the commonly shared civil and political rights and social, economic and cultural rights across the international human rights also enshrined in the CFREU. Simultaneously, due regard should be taken to the already found regional solutions; this, however, should not exclude the need to revisit them should that appear necessary. Although the EU could contribute to shaping the field of genomics within the EU and beyond, that has to be with due regard to the limits of competence, on the one hand, and aspirations for a social Europe on the other hand. The use of various regulatory tools, including soft measures, and different regulatory avenues should be further scrutinized in SIENNA task 4.2 to maximize the effects of any incentives EU could possibly take in the field.

Document history

Version	Date	Description	Reason for change	Distribution
V1.0	4 March 2019	First Draft	Reviewers' feedback, feedback from the national partners, further work towards finalizing the report.	Task contributors, External reviewers
V2.0	29 March 2019	Final Version		European Commission

Information in this report that may influence other SIENNA tasks

Linked task	Points of relevance
Task 4.2	This report will provide input to deliverable 4.2.

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Table 1 List of acronyms/abbreviations

Abbreviation	Explanation
APGT	Additional Protocol concerning Genetic Testing for Health Purposes
ART	Assisted reproductive technology
ASEAN	Association of South-East Asian Nations
AU	African Union
BMC	Convention on Human Rights and Biomedicine
CEDAW	Convention on the Elimination of All Forms of Discrimination against Women
CFREU	Charter of Fundamental Rights of the European Union
CJEU	Court of Justice of the European Union
CoE	Council of Europe
CRC	Convention on the Rights of the Child

CRPD	Convention on the Rights of Persons with Disabilities
CRISPR-Cas9	Clustered Regularly Interspaced Short Palindromic Repeats Cas-associated nuclease 9
DNA	Deoxyribonucleic acid
ECHR	European Court of Human Rights
ECtHR	European Court of Human Rights
ELSI	Ethical, legal and social issues
EU	European Union
FDA	Food and Drug Administration
GC	General Comment
GDPR	General Data Protection Regulation
HLA	Human leukocyte antigen
IBC	International Bioethics Committee
ICCPR	International Covenant on Civil and Political Rights
ICESCR	International Covenant on Economic, Social and Cultural Rights
IVDMD	*In vitro* diagnostic medical devices
IVF	*In vitro* fertilization
NIPT	Non – invasive prenatal testing advancement
OAS	Organization of American States
OECD-	Organisation for Economic Co-operation and Development
PKU	Phenylketonuria
PMs	Person months
PGD	Preimplantation genetic diagnosis
RNA	Ribonucleic acid
SWAFS	Science with and for Society
TFEU	Treaty on the Functioning of the European Union
UDHR	Universal Declaration of Human Rights
UN	United Nations
UNESCO	United Nations Educational, Scientific and Cultural Organization
WGS	Whole genome sequencing
WHO	World Health Organization

Table 2 Glossary of terms

Term	**Explanation**
Genetic Test	A genetic test: is an assay performed to obtain genetic information (directly on DNA or even on other molecules (RNA, proteins), which by extension could give us genetic information.
Genetic Testing	Genetic testing is usually offered to individual patients based on specific individual need on a one-on-one basis (diagnostic testing, prenatal testing, etc. https://ghr.nlm.nih.gov/primer/testing/uses). An assay (a genetic test) would be conducted after the clinician has decided to prescribe the test to the specific patient in question. Genetic testing is a type of medical test that identifies changes in chromosomes, genes, or proteins. The results of a genetic test can confirm

	or rule out a suspected genetic condition or help determine a person's chance of developing or passing on a genetic disorder.[1] Genetic testing could be accessed through a health care provider as part of clinical medical care, or as a service or product offered directly to consumers. Issues genetic testing raises relate to access rights, protection of integrity, medical oversight when these tests are being offered, as well as genetic counseling, and quality. Other questions that relate to genetic testing emerge in the area of advertising and the use of genetic information. In regard to advertising such issues as, for example, whether genetic testing providers are or are not allowed to advertise their offered genetic tests emerge (contrast with a distinction being made for advertising prescription and non-prescription drugs).[2] In regard to genetic information, such issues as who can access one's genetic information, and how this information could be used for different purposes, for example, life insurance. One of the key issues that emerges in this regard is discrimination relating to one's genome.
Genetic Screening	Genetic screening can have a few different meanings that differ based on subtle nuances and contexts, but in general, we contrast genetic screening from genetic testing because screening is offered to an entire group of people (not specific individuals). In particular screening cases, like new-born screening, for example, the screening programme follows public health frameworks (as opposed to genetic testing which is not a public health programme but within a clinical specialty; in countries where public health programmes do not differ so strongly from clinical practice, you may want to ignore this if it confuses you more. Differences between public health programmes and clinical (one patient at a time) programmes could mean differences in how we approach consent and the obligatory nature of any programme). Other than the "group/population" aim of screening programmes, screening programmes can vary hugely in different ways: population/group targeted (population-wide? Certain people/mothers at higher risks as a group?) etc.
Gene Editing/Genome Editing	Gene editing is a relatively recent term used for the description of modifying DNA (you may have previously heard in the past recombinant DNA technology or similar terms that all overlap in some fashion); the term gene editing has come along with the advent of particularly powerful and accurate tools like CRISPR-Cas9 (which is a tool that helps to change DNA). Gene editing can also be referred to as genome modification, which is more holistic, if you will as a concept, but addresses the same ideas; it basically includes not only changing individual DNA in one location but also many changes throughout the genome. One can edit genes in somatic cells (which are not inherited) or one could edit genes in gametes or germline

[1]U.S. National Library of Medicine, "What Is Genetic Testing?" (*Genetics Home Reference*). https://ghr.nlm.nih.gov/primer/testing/genetictesting.

[2] Kalokairinou, Louiza, Pascal Borry and Heidi Carmen Howard, "Regulating the Advertising of Genetic Tests in Europe: A Balancing Act", Journal of medical genetics, Vol. 54, 2017, p. 651.

	cells which are inherited and would pass down, in theory, DNA modifications/edits to future generations. The term human genome germline modification then includes the modification of 1 or many bits of DNA in an inheritable human cell. Many authors also include the replacement of mitochondria from one embryo to another as a type of germline modification, though this can be argued as (not) being very distinct from DNA modification, for the sake of comparison with other studies, we will include it for the purposes of this SIENNA study.

1. Introduction

1.1 Background and objectives

This report is the ultimate output for task 2.2 of the SIENNA project Work package 2. Work package 2 is devoted to the questions relating to genomics, and carries out ethical, legal and social analysis; task 2.2 specifically analyses the legal (including human rights) requirements relevant for genomics in and outside the EU. The output of this task will constitute an essential element of analysis carried out in task 4.2 of the project.

The overall aim of this report is twofold. First, to examine how the law currently responds to challenges in the area of genetics and genomics, and identify what challenges, limitations and gaps emerge. Secondly, to identify key human rights norms and regulatory approaches that could be examined further for shaping legal responses to the new and emerging technology in the area with due regard to competences and authority of various actors regulating/contributing to the shaping the regulatory environment in the area. The two aims are interrelated. The second aim builds on the analysis of the current legal responses. To achieve the aims, the report:

- analyses the ethical concerns, scrutinizes the legal and human rights responses in the area of genomics at the international and regional human rights legal orders, and the EU;
- carries out comparative analysis in selected EU Member States and non-EU countries, and surveys the legal responses, academic legal discussions and legal developments in the areas of concern;
- examines national comparative perspectives against the international and regional norms and human rights standards;
- identifies key human rights and legal challenges that emerge regarding genetics and genomics and shows the convergences and distinctions in the regulation of genomics and the challenges this presents for future innovation.

1.2 Scope and limitations

The scope of this report and limitations are defined by peculiarities relating to SIENNA consortium as well as topicalities in the area of genomics. Therefore, the study is focused on identifying challenges and responses, highlighting the commonalities and differences, rather than finding the ultimate responses to the questions that have been scrutinized.

With due regard to the indications in SWAFS-18-2016 and Description of Actions of the SIENNA project, the analysis focuses on genomics in the areas of a) human germline gene editing, b) genetic screening, c) genetic testing, and specific questions in the area of genetic testing and screening in these domains: prenatal and new-born testing/screening and direct-to-consumer advertising of genetic testing. Due to extensive considerations of such questions as biobanking in other places, including recently funded EU projects,[3] and ongoing scholarly work in the field,[4] they have not been of key concern when shaping questions for analysis. Nonetheless, they have been considered in so far as triggered by the substantive questions of concern[5] and highlighted in the national reports provided by SIENNA consortium members. The challenges that are covered in these domains focus on those concerns highlighted in the scholarly

[3] For example, H2020 B3Africa. See B3Africa Project (http://www.b3africa.org/), which has received funding under grant agreement nr 654404 from the EU's Horizon 2020 research and innovation programme.

[4] See, for example, Slokenberga, Santa, Olga Tzortzatou and Jane Reichel (eds), *Individual Rights, Public Interest and Biobank Research. Article 89 GDPR and European Legal Responses*, Springer, Forthcoming 2019.

[5] Questions can be found in Chapter 2.

analysis that is summarized in Chapter 3, as well as responses within the international, regional human rights, EU and national legal orders.

The six questions that are forming the basis for the national studies and this report that are introduced in Chapter 2.2 have been approached as *guiding questions*. Therefore, neither in national studies, nor international and regional human rights or EU law analysis, nor in the comparative study between the national legal orders these questions are responded in full. Instead of perceiving them as the ultimate answers, they should be approached as highlights of the regulatory trends. Furthermore, national partners have been enabled to provide additional considerations regarding other regulatory trends than *prima facie* covered within the six questions of concern. However, when carrying out comparative analysis, in so far as relevant and possible, these considerations have been integrated within analysis relating to the respective six questions.

Peculiarities relating to SIENNA consortium include such considerations as states and human rights legal orders represented within the consortium and different partner statuses (full partner or associate partner), accessibility of human rights legal orders, as well as considerable time constraints. In particular, the analysis focuses on those legal orders summarized in Table 4 below, covering UN, UNESCO, ASEAN, AU, CoE, OAS, the EU and the following national legal orders: Brazil, China, France, Germany, Greece, Japan, Netherlands, Poland, South Africa, Spain, Sweden, UK and USA. National reports annexed to this study also include considerations regarding, for example, the OECD and WHO and other beyond-the-state actors as has been relevant nationally; however, these have only sporadically been considered in this report.

Comparative analysis between the national legal orders is based on the reports provided by SIENNA partners and associate partners about their national legal orders; exceptionally, under the auspices of the Helsinki Foundation for Human Rights a draft report regarding the U.S. has been prepared. These national studies have been carried out within 0.6 PMs (12-13 working days) per country. Similarly, also reviews of the international, regional and EU legal orders have been carried out in a limited time, affording 0.2 PMs per legal order (approximately four working days). Consequently, this report is a selective overview of the regulatory responses, as well as legal developments and scholarly considerations among the surveyed states rather than a complete survey of an in-depth analysis of the legal situation in each of the legal orders concerned. Furthermore, even though the reports follow the same questionnaire and guidance provided by the task leaders, and all national partners have received at least one feedback and have been involved in joint discussion calls, the task leader has not had influence over the extent to which the guidance was followed. Consequently, national reports are of diverse quality. Key challenges include not fully responded questions, misplaced responses within the national reports, as well as an incomplete report of an associated partner (report for Japan); as well as an incomplete report regarding the US legal system, which is related to the limited time resources at the disposal of the author of the study. Given these limitations and with due regard to the different stages in which draft reports regarding Japan and the U.S. are, only the draft report regarding Japan is not annexed to this study. In attempt to mitigate challenges associated with interpreting the provided data in these reports, national partners have been asked to review the draft version of the report as of March 4, 2019. All feedback has been implemented.

This report has been reviewed by Dr Safia Mahomed and Professor Hermany Nys. Overall, the feedback of the reviewers has been followed. Exceptionally, due to the limited time resources a suggestion to complement Chapter 4 with tables to ensure a better oversight could not be accommodated. This is not perceived as a significant shortcoming, as the tables would be based on the existing analysis and would

not add any substantive changes. The reviewers have not had any control regarding the extent to which their suggestions were followed.

International and regional human rights organizations	European Union	National legal orders in Europe	National legal orders for comparative study
UN UNESCO ASEAN AU CoE OAS	EU	France, Germany, Greece, the Netherlands, Poland, Spain, Sweden, UK	Brazil, Japan, South Africa, the U.S.

Legal orders surveyed in the legal and human rights analysis (Table 3)

1.3 Structure of the report

This report is structured to effectively meet the aims of the study. Chapter 2 sets forth methodological foundations of the study and identifies six key questions that shape subsequent analysis. Chapter 3 expands on these questions in greater detail to scrutinize what challenges the scientific advances pose to the current legal and human rights frameworks that should be further scrutinized. Subsequent three chapters in the report follow this theme with analytical insights analysis in legal orders of interest to SIENNA. Chapter 4 examines relevant international and regional laws and human rights standards in light of the challenges identified as part of the literature review. Chapter 5 examines EU law, including its current responses and limitations, in the areas of concern. Chapter 6 carries out a comparative analysis to obtain insights in how different national legal orders respond to genomics and examines relevant national legal and human rights standards, surveying the current academic legal debates, regulatory developments and responses to the six questions of concern. After that, Chapter 7 examines the adequacy of existing international human rights standards vis-à-vis the ethical challenges, and Chapter 8 shows the convergences and distinctions on the regulation of genomics and reflects on challenges this presents for future innovation.

This report has three annexes. Annex 1 is a list of areas of inquiry, annex 2 is a list of human rights and legal concerns, annex 3 is a list of national reports annexed to this study provided as separates files.

2. Methodology and research questions

2.1 Methodology

For the purposes of this study, building on the guidance provided in SIENNA Handbook, a doctrinal method of law coupled with the functional and comparative method is applied. The doctrinal method is used to study applicable law and reach a systematic exposition of the principles, rules and concepts that are relevant to the area of genetics and genomics with a view to solving ambiguities and gaps in the existing law.[6] Functional method is used to examine "the way practical problems of solving conflicts of interest are dealt with in different societies according to different legal systems".[7] This method takes its expression in analysing legal responses to the six questions raised in different national and international legal orders. Thereafter, a comparative method is applied to examine various approaches these legal orders have taken

[6] See Smits, Jan M., *What Is Legal Doctrine? On the Aims and Methods of Legal-Dogmatic Research* in van Rob Gestel, Hans-W. Micklitz and Edward L. Robin (eds), CUP, 2017, p. 210.

[7] Van Hoecke, M., "Methodology of Comparative Legal Research", *Law and Method,* Vol.9, 2015, p.1.

in order to tackle the questions and identify the commonalities and differences.[8] In that way, comparative analysis is limited to selecting illustrative examples that allow identifying challenges and highlighting commonalities and differences. This analysis is carried out with due regard to the considerations on the competence and authority limitations of legal orders concerned, and as far as national legal orders are concerned – with due regard to their different external commitments.

The aims of this study are achieved using the sources of law that are relevant to each of the legal orders concerned; in so far as national comparative study is carried out, this report exclusively relies on the analysis carried out by SIENNA partners. Furthermore, to scrutinize the object, namely, the advance in genetics and genomics, as well as scientific challenge posed to law, relevant scientific literature is used, and including that which accounts for the ethical, legal and social issues of these advances, and helps to identify challenges human rights and EU law shall tackle in the area of genetics and genomics. Further considerations on approach and materials for the literature review, international and regional human rights, EU law and comparative national analysis can be found in Chapters 3, 4, 5, and 6 respectively.

2.2 Questions

In order to scrutinize legal (including human rights) challenges and responses to genomics, simultaneously also allowing national legal orders to report on other current challenges, we have pre-selected the below-listed questions.

Question 1: Human Germline Gene Editing
How, if at all, human germline gene editing is regulated? Please address the following:

- Regulation of basic research. In particular how, if at all, basic research is defined, whether basic research in human or embryos/gametes using germline modification is permitted, prohibited, or restricted.
- Regulation of pre-clinical research. In particular, whether pre-clinical research of germline modification technologies in non-humans is permitted, prohibited, or restricted (animal use for gene editing research purposes).
- Regulation of clinical research in humans. In particular, whether clinical research in humans using germline modification technologies is permitted, prohibited, or restricted.
- Regulation of clinical applications. In particular whether findings of research using germline modification technologies can be used in a clinical setting (i.e., to initiate a pregnancy with edited embryos or with edited gametes).

Question 2: Genetic screening in general
What are the legal rules applicable to/governing genetic screening as part of a public health measure? In particular:

- Conditions that are part of screening is defined or delineated in any way;
- Patient's rights in screening; in particular, type of consent and requirements for consent;
- Screening provider's responsibilities.

Question 3: Genetic testing in general
What are the legal rules applicable to/governing genetic testing?

Question 4: Prenatal testing/screening
How, if at all, is prenatal testing or screening regulated in the legal order of concern? If it is regulated:

[8] Ibid.

- Whether screening/testing is compulsory or an opt-out exists;
- What conditions are included in screening/testing?
- Is non-invasive pre-natal testing/screening mentioned?
- Whether abortion is available following the screening/testing; if so,
    - Until what week?
    - Whether any further conditions (rights, obligations) in relation to the abortion are relevant?

Question 5: New-born screening
How, if at all, is new-born screening regulated in the legal order of concern? If it is regulated:

- Whether testing is compulsory or an opt out exists;
- What type of consent is required?
- If also legislated:
    - how should it be obtained?
    - For the procedure?
    - For any secondary use of sample or data?
    - For how long the data/samples can be retained?
    - What can the data/samples be used for?
    - What conditions are included in screening?

Question 6: Advertising of genetic testing or screening

- Is the advertising of genetic tests/genetic screening for health purposes directly to consumers permitted? What, if any, special rules apply?
- Is the advertising of genetic tests/genetic screening for non-health purposes directly to consumers permitted? What, if any, special rules apply?

Question 7: Please report on other issues relevant to regulating genomics being at the forefront in the national legal order.

3. Legal issues and human rights challenges of genetics and genomics

3.1 Introduction, note on methodology

Each of the six guiding questions, outlined in Chapter 2.2., entails a number of legal issues, including human rights implications. This section presents a non-exhaustive overview of such issues identified through a literature review. The preliminary review was conducted using EBSCO Discovery Service database and limited to text labelled as legal articles published since 2012 (due to the dynamic technical developments in the field). In the second step, the review was extended also to older texts identified by using the Google Scholar database, the Google general search engine and via the references in the already reviewed articles. Since the six guiding questions partly overlap, the following overview is generally intended to highlight issues that are the most specific for each of the questions.

3.2 Human germline editing

Over the last decade, considerable advances in the area have occurred that, according to many, render human germline editing technically soon to be a feasible intervention. Before that can happen, and

despite the occurrences in late 2018,[9] there are considerable technical, as well as ethical and legal issues that need to be resolved.

In order to scrutinize the diversity of legal and human rights questions, the spectrum of concerns has been divided into two major groups: research and application. Moreover, each can be divided into further groups. In regard to research, the following three groups can be distinguished: basic research, pre-clinical research, and clinical research, though, elsewhere a distinction between pre-clinical and clinical research only is done.[10] In regard to application, a distinction can be drawn between health and non-health application, even though it can be challenging to make a clear distinction between the two. Although there are distinct legal questions that can be attributed to each of the groups, there are also legal concerns that transcend either some of these groups or all of them; for example, disability concerns and liability could be seen as relevant for all four of these groups, whereas, some are exclusively associated with application of human germline editing vis-à-vis humans, disregarding whether or not it is being done as part of clinical research or clinical care.

Basic research and human embryo involvement in research

As basic research on gene editing in germline and in embryos involve trials on human embryos, their moral and legal status is at the forefront of some of the discussions on gene editing. Apart from the more fundamental disputes on the embryo value, several areas of controversy emerge. First, there are concerns with how embryos are acquired for research (including e.g. questions whether embryos donors understand the information they are given about the research goals[11] and whether they are not pressured in any way,[12] what also relates to the particular risks of coercion and exploitation of women donating their eggs[13]). Second, there are debates surrounding the issue of handling of human embryos during the course of research (e.g. should a donor have any recourse when it is discovered that the embryos were used in a manner contrary to the donor's wishes?).[14] Thirdly, the ways in which embryos are destroyed after they have been used for scientific research.[15] In relation to the embryo status, also questions of potential duties vis-à-vis embryos have been raised, as well as legal liability for causing harm to embryos in the course of such treatment.[16] These questions require thoroughly examining the legal status of human embryo and protections set forth in that regard under such human rights, including principles, as dignity, right to life and right to private life.

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[9] In November 2018, Dr. He Jiankui from China announced that the first two children whose genome had been edited using CRISPR-CAS9 technology had been born, as well as another pregnancy with a foetus containing an edited human germline was initiated. These actions have generally been condemned by the scientific community and the Chinese government. See Slokenberga, Santa, and Heidi Carmen Howard, "The Right to Science and Human Germline Editing: Sweden, Its External Commitments and the Ambiguous National Responses under the Genetic Integrity Act", *Förvaltningsrättslig Tidskrift*, 2019 forthcoming.

[10] Evitt, Niklaus H., Shamik Mascharak and Russ B Altman, "Human Germline CRISPR-Cas Modification: Toward a Regulatory Framework", *The American Journal of Bioethics* Vol. 15, 2015, p.25.

[11] Tomlinson, T., "A CRISPR Future for Gene-Editing Regulation: A Proposal for an Updated Biotechnology Regulatory System in an Era of Human Genomic Editing", *Fordham Law Review,* Vol.87, 2018, p.437. Leach Scully, Jackie and others, "Donating Embryos to Stem Cell Research", *Journal of Bioethical Inquiry,* Vol. 9, 2012, p.19. Cogner, Christa, "New Approach to IVF Embryo Donations Lets People Weigh Decision", *Stanford Medicine News Center,* Vol. 2, 2019.

[12] Tomlinson, op. cit. 11.

[13] Ram, N., "Science as Speech", *Iowa Law Review,* Vol. 102, 2016, p.1187.

[14] Tomlinson, op. cit. 11.

[15] Ibid.

[16] Powell, G. Edward, "Embryos as Patients? Medical Provider Duties in the Age of CRISPR/Cas9", *Duke Law & Technology Review,* Vol. 15, 2017, p.344.

Animal involvement in pre-clinical research

Before clinical research is considered in an attempt to carry out gene editing that leads to human involvement, including implanting embryos and pregnancy thereafter, "preclinical studies should establish reliability, validity, safety, and efficacy" of the intended interventions.[17] However, if a country prohibits clinical trials but permits animal use for germline gene editing studies, one can question the consistency of the regulatory approach.[18] On the other hand, allowing animal research could be seen as an element furthering the right to science, as it allows to get information-data and question the current regulatory approaches.[19] A question of animal involvement in studies related to human germline editing falls within a broader category of animal rights and requires considering the value of these studies and their ultimate goal, especially, if clinical trials are often prohibited. Therefore, a question that should further be scrutinized is whether animals can be used in pre-clinical studies, even if a particular legal order does not permit the research to escalate to clinical phase.

Impact on future generations

The hereditary nature of germline gene editing generates discussions about its potential impacts on future generations. The debate is framed around such issues as lack of knowledge about possible risks and therefore, the difficulty to foresee the multigenerational consequences of altering genetic pool;[20] problem of consent of the affected persons in future generations,[21] including lack of consent for the probable long follow-up clinical trials of the descendants;[22] as well as threats of the so-called instrumentalization of genetically-modified children that relates to the impact of awareness of being "made" rather than having "grown".[23]

Some scholars claim that gene editing interventions – especially this type of germline editing that would be used for "artificial" shaping of specific traits – constitutes a transgression of some form of moral limits of human action. This concern is often expressed by such terms as "playing God" or "disrupting natural order", i.e. as ascribing to humans a role that should be left to other instances (God, nature etc.) – either due to moral or religious reasons or because we are unable to predict its consequences. [24] In a somewhat

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[17] Ormond, Kelly E., and others, "Human Germline Genome Editing", *The American Journal of Human Genetics,* Vol. 101, 2017, p.167.

[18] Slokenberga, S., and Heidi Carmen Howard, "The Regulation of Human Germline Genome Modification in Sweden" in Andrea Boggio, Cesare Romano and Jessica Almqvist (eds), *Human Germline Genome Modification and the Right to Science: A Comparative Study of National Laws and Policies*, Cambridge University Press, 2019.

[19] Slokenberga and Howard, op. cit. 9.

[20] Plummer, Kelly M., "Ending Parents' Unlimited Power to Choose: Legislation Is Necessary to Prohibit Parents' Selection of Their Children's Sex and Characteristics", *Saint Louis University Law Journal,* Vol. 47, 2003, p.517. Ossareh, Tandice, "Would You like Blue Eyes with That: A Fundamental Right to Genetic Modification of Embryos", *Columbia Law Review,* Vol. 117, 2017, p. 729.

[21] Collins, Francis S., and National Institutes of Health, "Statement on NIH Funding of Research Using Gene-Editing Technologies in Human Embryos". National Institutes of Health webpage. https://www.nih.gov/about-nih/who-we-are/nih-director/statements/statement-nih-funding-research-using-gene-editing-technologies-human-embryos. Gyngell, Christopher, Thomas Douglas and Julian Savulescu, "The Ethics of Germline Gene Editing", *Journal of applied philosophy*, Vol. 34, 2016, p. 498. Scott, Rosamund, and Stephen Wilkinson, "Germline Genetic Modification and Identity: The Mitochondrial and Nuclear Genomes", *Oxford journal of legal studies,* Vol 37, 2017, p.886.

[22] National Academies of Sciences "Human Genome Editing: Science, Ethics, and Governance". https://doi.org/10.17226/24623.

[23] Habermas, Jürgen, *The Future of Human Nature,* Polity, 2003. Plummer, op. cit. 20. Ossareh, op. cit. 20.

[24] Cole-Turner, Ronald, *The New Genesis: Theology and the Genetic Revolution*, Louisville, Kentucky, Westminster/John Knox Press, 1993, cited in: National Academies of Sciences, op. cit.22. Ram, op. cit.13. Ellison,

similar tone, it has been pointed out in early soft law instruments of the area that "the rights to life and to human dignity imply the right to inherit a genetic pattern which has not been artificially changed".[25] Critics of such view stress that it is based on very vague notions and notice that there is a wide acceptance of human interventions ("artificial changes") in other "natural" processes, e.g.in agriculture or medicine.[26] These concerns relate to re-assessing the adequacy of existing regulatory approaches and in that regard, a distinction between research and care needs to be particularly observed, and beneficiaries of human rights protection frameworks.

Scope of application and imperative to treat illnesses, right to health

One of the recurring themes relates to current and potential uses of gene editing for medical and non-medical purposes, though the distinction between the two might not always be easy to draw. In regards to medical purposes, it is being stressed – usually in terms of the individual's right to health or of a societal obligation to treat illnesses (or not to refuse treatment) – that any calls for banning, postponing or otherwise limiting clinical research of some forms of gene editing have to take into account the interests of those suffering illnesses that could be treated using this technology,[27] as well as benefits of potential general eradicating of some genetic diseases.[28] Hence, a key question to scrutinize is the scope and limits of the right to health and the appropriate responses through constraining self-determination. Moreover, these questions can also be approached in terms of benefiting from scientific advances and thus, be seen in the light of the right to science.

Safety considerations

Safety is identified as one of the key concerns by most of the participants of the scholar debate related to gene editing. Often are indicated risks such risks as off-target effects (unexpected, erroneous mutations) and mosaicism (the presence of two or more populations of cells with different genotypes in one individual).[29] Although safety concerns appear both in the context of somatic and germline gene editing the latter receives special attention in the context of human germline editing, and many authors indicate uncertainty about its near and long-term effects.[30] However, more recently scholars have pointed out that there are significant on-going technological developments and the unintended effects continue to be reduced.[31] Furthermore, due regard should also be paid to the fact that nothing is perfectly safe (and especially not the new technologies) – what should be considered is rather whether a given technology is

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Teddy, "Why Genetics Is CRISPR Than It Used to Be: Helping the Novice Understand Germ Line Modification and Its Serious Implications", *Southern California Interdisciplinary Law Journal,* Vol. 26, 2016, p. 595.

[25] CoE, Parliamentary Assembly, Recommendation 934, Genetic Engineering, 1982.

[26] National Academies of Sciences, op. cit. 22. Ellison, op. cit. 24.

[27] Miller, Henry I., "Germline Gene Therapy: Don't Let Good Intentions Spawn Bad Policy", *Issues in Science and Technology*, Vol. 32, 2016, p. 57. Nordberg, Anna, and others, "Cutting Edges and Weaving Threads in the Gene Editing (Я) Evolution: Reconciling Scientific Progress with Legal, Ethical, and Social Concerns", *Journal of Law and the Biosciences,* Vol. 5, 2018, p. 35.

[28] Mahoney, Julia D., and Gil Siegal, "Beyond Nature: Genomic Modification and the Future of Humanity", *Law & Contemp. Probs.,* Vol. 81, 2018, p. 195.

[29] Baltimore, David, and others, *On Human Gene Editing: International Summit Statement,* Washington DC: National Academy of Sciences, 2015. National Academies of Sciences, op. cit. 22. Santaló, J., and Casado M. Coords, "Document on Bioethics and Gene Editing in Humans" Barcelona University webpage., 2 March 2019. http://www.bioeticayderecho.ub.edu/en/document-bioethics-and-gene-editing-humans.

[30] See, for example, Howard, Heidi C., and others, "One Small Edit for Humans, One Giant Edit for Humankind? Points and Questions to Consider for a Responsible Way Forward for Gene Editing in Humans", *European Journal of Human Genetics*, Vol. 26, 2018, p.1. Ormond and others, op. cit. 17.

[31] National Academies of Sciences, op. cit. 22.

"safe enough", given the balance of risks and benefits[32] and, in particular, their possible risks should be analysed in the light of certain harms coming from illnesses.[33] In terms of human rights, this question triggers the standard of health, and in particular, it's quality dimension, including such angles as safety and efficacy. Moreover, it can also be conceptualized in terms of self-determination and integrity, individual freedom and state obligations to intervene and protect individuals from harm. It can also be seen in the light of the right to science, as a duty to protect from scientific harms.[34]

Inequality concerns

Gene editing technologies raise questions of equality, in particular, fair access to these technologies (especially, but not only, in the context of medical treatment).[35] This requires assessing such questions as whose illness is targeted, what are the research priorities and where should the funds be directed (e.g. whether to answer "that urgent needs of poor patients and overall public health" or "in favour of developing non-essential treatments for affluent patients"[36]).

Unequal access to the benefits of gene editing risks deepening existing inequalities in society. Some fear this would (especially in the context of germline editing and editing for non-therapeutic purposes) "make a culturally determined inequality into one that is biological", even up to the point of "parallel populations" of advantaged and disadvantaged[37] or of "genetics haves and haves notes".[38] Even though biological effects of social inequalities do already exist (e.g. as outcomes of better nutrition or use of vaccines), there are concerns that gene editing could largely contribute to development of this phenomenon.[39] These concerns, however, are not universal. Some have argued that "as with most technologies, costs will likely drop over time, closing the gap between those who have access to the technologies and those who do not"[40] and that "this inequality, however unfair it may feel, should not be found compelling enough to interfere with a parent's right to choose to have a healthy or otherwise modified child".[41] While enhancement and designer babies are a common concern in regard to the application of gene editing technology, and thus this would require examining limits of permissibility, others reject such an application of gene editing technology and the related risk of rising inequalities, because, as for today, there is no evidence that would suggest that the technology is capable of producing

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[32] Harris, John, "Germline Modification and the Burden of Human Existence", *Cambridge Quarterly of Healthcare Ethics,* Vol. 25, 2016, p. 6.

[33] Miller, op. cit. 27. Harris, op. cit. 32. Mahoney and Siegal, op. cit. 28.

[34] Slokenberga and Howard, op. cit. 9. For an overview of challenges see Eric S Lander and others, "Adopt a Moratorium on Heritable Genome Editing"(2019) 567 *Nature* , p.165.

[35] Jasanoff, Sheila, J. Benjamin Hurlbut and Krishanu Saha, "CRISPR Democracy: Gene Editing and the Need for Inclusive Deliberation", *Issues in Science and Technology,* Vol. 32, 2015, p. 37. Asbury, Bret D., "Counseling after CRISPR", *Stan. Tech. L. Rev.,* Vol 21, 2018, p. 1. Ruha, Benjamin, "Interrogating Equity: A Disability Justice Approach to Genetic Engineering", *Issues in Science and Technology,* Vol. 32, 2015, p. 51.

[36] Jasanoff, Hurlbut and Saha, op. cit. 35.

[37] Ibid.

[38] Mahoney and Siegal, op. cit. 28.
Center for Genetics and Society, *About Human Germline Gene Editing | Center for Genetics and Society.,* 2 March 2019. https://www.geneticsandsociety.org/internal-content/about-human-germline-gene-editing. The Guardian *Human Gene Editing Is a Social and Political Matter, Not Just a Scientific One.,* 2 March 2019. https://www.theguardian.com/science/2015/dec/04/human-gene-editing-is-a-social-and-political-matter-not-just-a-scientific-one.

[39] Mahoney and Siegal, op. cit. 28.

[40] Ossareh, op. cit. 20.

[41] Ibid.

such outcomes.[42] In terms of human rights, these questions relate to accessibility, usually expressed in terms of the right to health, which requires considering equal access to medical goods and services, as well as equal opportunities to benefit from scientific advances under the right to science.

Reproductive (procreative) autonomy

Decisions of using germline gene editing may fall within the sphere of reproductive (procreative) rights, as they are connected to parents' authority to decide "whether the characteristic in question is one that is central or material to a reproductive decision".[43] Similar arguments have been made regarding parental rights (autonomy), i.e. that the parents" liberty to make choices regarding the upbringing of their children includes the right to choose a future's child genetics.[44] Other scholars, however, remain more sceptical as to whether reproductive autonomy could include the right to a genetically related child.[45] The question of reproductive autonomy can be anchored in a more general right to privacy, as well as considered under the right to health where sexual and reproductive health is one of its dimensions.

Disability, equality and diversity

Some forms of research and clinical applications of gene editing may frame particular traits as "undesirable for future generations"[46] and as such lead to stigmatization of certain groups and reinforcement of existing prejudices.[47] This kind of practices and risk attached to it, especially when editing is used for enhancement, has been labelled by some scholars as a form of "neoeugenics".[48] This question is brought up perhaps most often in relation to people with disabilities and led to repeated calls for the inclusion of disability rights groups in the debates surrounding the developments of these technologies, in line with the slogan "nothing about us without us".[49] These questions trigger the considerations for persons with disabilities, and more generally, protection of the vulnerable societal groups; as well as stigmatization and considerations relating to (genetic) discrimination.

Liability and access to justice

Germline gene editing might pose several challenges for the existing framework of liability. For example, as germline gene editing may negatively affect several generations down the line, it is problematic to what extent a physician or researcher may be held liable for damages that potentially have no end[50] or who should be held liable when an unintended consequence of editing, hidden in the first generation, is

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[42] Enríquez, Paul, "Genome Editing and the Jurisprudence of Scientific Empiricism", *Vand. J. Ent. & Tech. L.,* Vol. 19, 2016, p. 603.

[43] Robertson, John A., "Genetic Selection of Offspring Characteristics", *BUL Rev.,* Vol. 76, 1996, p. 421 cited in Ossareh, op. cit. 20.

[44] Ibid.

[45] Haker, Hille, Germline gene editing of human embryos is wrong, response was submitted to the Call for Evidence held by the Nuffield Council on Bioethics on Genome editing between 15 May 2017 and 14 July 2017. http://nuffieldbioethics.org/wp-content/uploads/Hille-Haker-Chair-of-Catholic-Moral-Theology-Loyola-University-Chicago-USA.pdf.

[46] Lewis, Myrisha S., "How Subterranean Regulation Hinders Innovation in Assisted Reproductive Technology", *Cardozo L. Rev.,* Vol. 29, 2017, p. 1239.

more generally, see also Emens, Elizabeth F., "Framing Disability", *U. Ill. L. Rev.,* Vol. 5, 2012, p.1383.

[47] Oral statement cited in Benjamin, op. cit. 35.

ibid. Suter, Sonia M., "A Brave New World of Designer Babies", *Berkeley Tech. LJ,* Vol. 22, 2007, p.897. Jasanoff, Hurlbut and Saha, op. cit. 35.

[48] Suter, op. cit. 47.

[49] Benjamin, op.cit. 35. Baltimore and others, op. cit. 29.

[50] Marchant, Gary E., "Legal Risks and Liabilities of Human Gene Editing", *Scitech Lawyer,* Vol. 13, 2016, p.26.

detected in future generations several decades later.[51] Likewise, it raises questions regarding the protection of the rights and interests of those being affected by the application of gene editing technology to human germline. Different states may choose to protect human germline with different means, including envisaging liability for the violations of the statutory prohibitions. Therefore, questions of liability in terms of tri-partial obligations stemming from human rights (respect, protect, fulfil) and access to justice are of importance.

Summary of challenges, and human rights at stake and legal concerns regarding human germline gene editing (Table 4)

Challenges	**Human rights at stake and legal concerns**
Governance of human germline gene editing	• Who has the competence and authority to regulate human germline interventions
Human embryo involvement in research	• Legal status of the human embryo • Protections afforded to embryo under such human rights, including principles, as dignity, right to life and right to private life.
Animal involvement in pre-clinical research in light of the overall legal framework	• Legality of animal involvement and objective • The consistency of regulatory strategies • Right to science
Impact on future generations human germline editing could have	• The beneficiary of human rights protection • Right to science
Scope of application of human germline editing technologies	• Right to health • Scope and limits of self-determination
Safety considerations regarding of germline editing technologies	• Right to health, in particular, quality dimension, including such angles as safety and efficacy • Protection of integrity • Right to science
Access to scientific advances and inequality in that regard	• Right to health, which requires considering equal access to medical goods and services. • Right to science
Reproductive autonomy and freedom to decide of to have an offspring with genetic impairments	• Right to sexual and reproductive health • Right to private life/privacy
Elimination of certain features or diseases triggers disability, equality and diversity concerns	• Equality • Disability rights • Stigmatization • Genetic discrimination
Protection afforded to human genome	• Liability • Access to justice

3.3 Genetic screening

Genetic screening generally is offered to a particular group of people and not specific individuals, which is what differs this procedure from genetic testing.[52] Nonetheless, from the perspective of individual rights, genetic testing and genetic screening often overlap. As is surveyed in Chapter 6, it is also common

[51] Lovell, Kendall, "CRISPR/Cas-9 Technologies: A Call for a New Form of Tort", *San Diego Int'l LJ,* Vol. 19, 2017, p. 407. Powell, op. cit. 16.

[52] See more on genetic screening at Pinsky, Leonard, and Morris Kaufman, *Genetics of Steroid Receptors and Their Disorders* in Harry Harris and Kurt Hirschhorn (eds), *Advances in Human Genetics*, Springer, US, 1987, p.319.

that both terms, genetic screening and genetic testing are used interchangeably. Here below, we review the following areas of concern relating to legal, including human rights analysis, that have been highlighted in the literature specifically regarding genetic screening.

Availability, accessibility, acceptability, and quality of the screening programmes

One of the key issues related to genetic screening is its benefits for public health, which has been the main argument for introducing or expending of such programmes.[53] In organizing public health screening programs, such questions as existence of screening programmes is of particular importance, as well as its accessibility, acceptability, and quality. Not only is it important that various groups are targeted, including those located in the marginalized areas, for example, rural areas, but also the programmes are organized in a way that a particular societal group is singled out and could lead to group discrimination or marginalization/stigmatization.[54] While the latter concerns are part of organizing a public health measure, they can also be seen as distinct concerns that need further attention. Therefore, from a human rights perspective, the right to the highest attainable standard of health and public health shall be further analysed. Questions of discrimination and stigmatisation are considered below.

Discrimination and stigmatization

Stigmatization and different types of discrimination are a pertinent issue that emerges in relation to genetic screening. First, since certain genetic conditions are more common in people from particular ethnic groups, concerns have been expressed regarding the acceptability of screening programmes specifically targeting members of these sub-populations. It has been stressed that such an approach could lead to stigmatization of these groups[55] and to the reification of race categories[56] and advocated rather for more resource-intensive, but more equitable universal screening of specified genetic disorders.[57] Some calls for population-wide screening for the reasons related to racial discrimination have been also raised in the context of forensic DNA databases – as these data are gathered by arrests or convictions, the racial distribution of samples within the databases may reflect nation's overall racial disproportions in arrests and convictions.[58] Besides ethnic or racial context, inequality in relation to genetic screening has

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[53] Andermann, Anne, and Ingeborg Blancquaert, "Genetic Screening: A Primer for Primary Care", *Canadian Family Physician Medecin De Famille Canadien,* Vol. 56, 2010, p. 333. Burke, Wylie and others, "Genetic Screening", *Epidemiologic Reviews,* Vol. 33, 2011, p. 148. It has, however, also been noted that its beneficial effects should be measured taking into account more general economics of a public healthcare system: even though the test themselves may be inexpensive, they do require a costly infrastructure (counselling, storage, follow-up etc.). Andermann, Anne, and others, "Revisiting Wilson and Jungner in the Genomic Age: A Review of Screening Criteria over the Past 40 Years", *Bulletin of the World Health Organization,* Vol. 86, 2008, p. 317.

[54] Wauters, Annet, and Ine Van Hoyweghen, "Global Trends on Fears and Concerns of Genetic Discrimination: A Systematic Literature Review", *Journal of Human Genetics,* Vol. 61, 2016, p. 275.

[55] Nuffield Council on Bioethics. "Genetic screening: a supplement to the 1993 report by the Nuffield Council on Bioethics", London, Engl: Nuffield Council on Bioethics, 2006. http://nuffieldbioethics.org/wp-content/uploads/2014/07/Genetic-Screening-a-Supplement-to-the-1993-Report-2006.pdf.

Berghs, Maria, Simon M. Dyson and Karl Atkin, "Resignifying the Sickle Cell Gene: Narratives of Genetic Risk, Impairment and Repair", *Health,* Vol. 21, 2017, p. 171.

Karpin, Isabel Ann, "Protecting the Future Well: Access to Preconception Genetic Screening and Testing and the Right Not to Use It", *Griffith Law Review,* Vol. 25, 2016, p. 71.

[56] Sokhansanj, Bahrad A., "Beyond Protecting Genetic Privacy: Understanding Genetic Discrimination Through Its Disparate Impact on Racial Minorities", *Colum. J. Race & L.,* Vol. 2, 2012, p. 279.

[57] Cameron, Louise, and Hilary Burton "Genetic screening programmes: an international review of assessment criteria" PHG Foundation webpage. http://www.phgfoundation.org/documents/560_1470143671.pdf.

[58] Kaye, David H., and Michael E Smith, "DNA Identification Databases: Legality, Legitimacy, and the Case for Population-Wide Coverage", *Wis. L. Rev.,* 2003, p. 413.

been also discussed with reference to discrimination based on traits identified in genetic screening (genetic discrimination, e.g. in the sphere of employment or insurance).[59] Therefore, in genetic screening programmes, not only a question of addressing stigmatization and discrimination concerns in planning public health measures, but also how genetic data and information are being handled are of paramount particular importance. These shall be further analysed in terms of prohibition of discrimination, stigmatization, as well as equality requirements. Moreover, in so far as the use of data is concerned, also as part of data protection or private life protection. Lastly, this could also be seen as concern that relates to the human value and accordingly anchors in dignity.

Voluntariness of screening

In contemporary literature, there seems to be a general consensus that genetic screening should not be mandatory and it always requires informed consent of the affected persons (except for newborns screening, where the discussion continues).[60] In this context, screening for sickle cell disease among African American, that was obligatory in several states in the USA in the 1970s, is used as a negative point of reference.[61] However, it is also sometimes acknowledged that in case of wide, mass genetic screening programmes, it might be hard – due to practical constraints – to provide adequate counselling[62] and perhaps a more standardized form of counselling might be necessary.[63] In terms of human rights and legal frameworks, these questions may be addressed, for example, as part of the right to private life or privacy or liberty, as well specifically, a right to informed consent and counselling.

Reproductive freedom

Genetic screening relates to reproductive autonomy. It can be seen as enhancing it by offering more informed reproductive options, particularly to people belonging to population groups at the highest risks of carrying specific genetic disorders. For example, implementing genetic screening programmes by Ashkenazi Jewish communities, that have a high prevalence of the recessive Tay-Sachs trait, is one of the main examples.[64] Some scholars emphasize at the same time, that it would be unacceptable to preclude individuals who test positive as carriers of some genetic conditions from reproducing their own genetic materials if they wish so.[65] In this context, such questions as reproductive freedom are of particular importance and ways in which it is safeguarded/furthered. It includes questions of information and decision-making. In a broader perspective, it includes questions of non-stigmatization towards a particular group. They require further considerations in terms of reproductive freedom, as well as decision-making, which is addressed, for example, within the scope of privacy/right to private life.

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[59] Godard, Béatrice and others, "Population Genetic Screening Programmes: Principles, Techniques, Practices, and Policies", *European Journal of Human Genetics,* Vol. 11, 2004, p. 49. Prince, Anya E.R., "Tantamount to Fraud: Exploring Non-Disclosure of Genetic Information in Life Insurance Applications as Grounds for Policy Rescission", *Health Matrix,* Vol. 26, 2016, p. 255. Adjin-Tettey, Elizabeth, "Potential for Genetic Discrimination in Access to Insurance: Is There a Dark Side to Increased Availability of Genetic Information", *Alta. L. Rev.,* Vol. 50, 2012, p. 577.

[60] Andermann and others, op. cit. 53. Berghs et. al., op. cit. 55.

[61] Fulda, Kimberly G., and Kristine Lykens, "Ethical Issues in Predictive Genetic Testing: A Public Health Perspective", *Journal of medical ethics,* Vol. 32, 2006, p. 143. Sokhansanj, op. cit. 56.

[62] van El, Carla G., and others, "Whole-Genome Sequencing in Health Care", *European Journal of Human Genetics,* Vol. 21, 2013, p. 580.

[63] Australian Law Reform Commission, "Essentially Yours–The Protection of Human Genetic Information in Australia", Volume 1 and Volume 2 Report 96. https://www.alrc.gov.au/publications/report-96.

[64] Berghs et. al., op. cit. 55. The Human Genetics Commission, "Increasing options, informing choice: A report on preconception genetic testing and screening", https://tegalsi.hypotheses.org/files/2011/04/2011.HGC_Increasing-options-informing-choice-final1.pdf. van El and others, op. cit. 62.

[65] Karpin, op. cit. 55.

Data protection

Genetic screening could produce large aggregations of genetic information.[66] This raises questions of adequate protection of personal data and privacy and, consequently, of legal liability regimes for data breaches.[67] Protection of one's control over his or her stored DNA data is also connected with the topic of secondary uses of the gathered information (e.g. for research).[68] Some of these issues are further complicated in, for example, cases of long-term retention of the data (which highlights problems of correction or withdrawal of data, as well as questions of renewing the given consent over time).[69] Moreover, subsequent use of this information/samples is of concern, for example, for criminal justice purposes.[70] Therefore how, if at all, data protection in regard to genetic screening is handled is of particular importance; which can expressed in terms of data protection or the protection of private life or privacy.

Summary of challenges, and human rights at stake and legal concerns in genetic screening (Table 5)

Challenges	**Human rights at stake and legal concerns**
Availability, accessibility, acceptability, and quality of the screening programmes	• Public health • Right to the highest attainable standard of health • Stigmatization • Discrimination
Risks of singling out and marginalizing some societal groups	• Discrimination • Stigmatization • Equality • Data protection • Private life/privacy • Dignity
Voluntariness of screening and choice	• Right to private life/ privacy /liberty • Right to informed consent • Counselling
Reproductive freedom, ability to decide whether or not to proceed with conception following risk identification	• Reproductive decision-making Right to private life/privacy • Stigmatization
Storage and use of genetic data and information	• Data protection • Right to private life/ privacy

[66] Commission, op. cit. 63.

[67] Cameron, op. cit. 57.

Ajunwa, Ifeoma, "Genetic Testing Meets Big Data: Tort and Contract Law Issues", *Ohio St. LJ,* Vol. 75, 2014 p. 1225.

Pike, Elizabeth R., "Securing Sequences: Ensuring Adequate Protections for Genetic Samples in the Age of Big Data", *Cardozo L. Rev.*, Vol. 37, 2015 p. 1977.

[68] Gutmann Koch, Valerie, "PGTandMe: Social Networking-Based Genetic Testing and the Evolving Research Model", *Health Matrix,* Vol. 22, 2012, p. 33.

[69] Capps, B., and others, "Imagined Futures: Capturing the Benefits of Genome Sequencing for Society" Human Genome Organisation (HUGO) Report. DOI: 10.13140/RG.2.1.5153.5521.Cameron, op. cit. 57.

[70] On biobanking and criminal justice see Dranseika, Vilius, Jan Piasecki and Marcin Waligora, "Forensic Uses of Research Biobanks: Should Donors Be Informed?", *Medicine, Health Care and Philosophy,* Vol. 19, 2016, p. 141.

3.3 Genetic testing

Genetic tests (even though there is no universally accepted definition of what is a genetic test)[71] are usually defined to cover tests being provided to individuals usually after genetic counselling and aim to detect whether there is an existing genetic disorder (diagnostic purpose) or a risk of developing one (predictive purpose). They raise a number of profound ethical and legal questions, and these questions often differ deepening on type of test as well as intended application. However, some common concerns surveyed below also emerge.

Informed choice, counselling, return of results and informing family members

Due to the complicated nature of information related to genetics and serious implications that results of genetic testing may have for a tested person, it has been emphasized that genetic testing should be accompanied by an appropriate communication process (genetic counselling), that would enable patients to make informed choices.[72] In the context of the return of results, this respect for the autonomy of a tested person has been expressed by the concept known as the "right not to know", i.e. a patient's right to decide whether or not to be informed about the results of a genetic test and their consequences.[73] These questions shall be scrutinized under information about the results of the test and counselling requirement, as well as right to private life/privacy protection and the right not to know (whether a self-standing right or addressed as part of the right to private life/privacy or right to information).

Another considerably discussed issue connected to the return of results is the question of whether members of a family of a tested person should be informed without his or her consent of genetic risks identified by genetic testing that they may be sharing.[74] This has been described sometimes as a conflict between the principle of confidentiality of genetic information and privacy rights of a tested person on the one hand and his or her family's "right to know" about risks important for their health or reproductive choices on the other.[75] These issues shall be scrutinized further in light of the right to private/family life, data protection and right to know, as well as confidentiality, in particular, how the balance between competing rights and interests has been struck.

Furthermore, the New Generation Sequencing raised additional issues, including those related to the questions of informed choice, as these techniques are likely to produce secondary or incidental results (i.e. findings outside the primary or original purpose for which a test was conducted).[76] Therefore, handling of incidental findings is of a particular concern, which is reviewed further below.

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[71] Varga, Orsolya, and others, "Definitions of Genetic Testing in European Legal Documents", *Journal of Community Genetics,* Vol. 3, 2012, p. 125.

[72] Weil, Jon, "Genetic Counselling in the Era of Genomic Medicine: As We Move towards Personalised Medicine, It Becomes More Important to Help Patients Understand Genetic Tests and Make Complex Decisions about Their Health", *EMBO reports,* Vol. 3, 2002, p. 590. Heller, Karen, *Genetic Counseling: DNA Testing for the Patient*, *Baylor University Medical Center Proceedings*, Taylor & Francis, 2005.

[73] Berkman, Benjamin E., and Sara Chandros Hull, "The "Right Not to Know" in the Genomic Era: Time to Break from Tradition?", *The American Journal of Bioethics,* Vol. 14, 2014, p. 28. Borry, Pascal, Mahsa Shabani and Heidi Carmen Howard, "Is There a Right Time to Know? The Right Not to Know and Genetic Testing in Children", *The Journal of Law, Medicine & Ethics,* Vol. 42, 2014, p. 19.

[74] Black, Lee, and Kelly A. McClellan, "Familial Communication of Research Results: A Need to Know?", *The Journal of Law, Medicine & Ethics,* Vol. 39, 2011, p. 605.

[75] Ibid.

[76] Brown, Teneille R., "Needles, Haystacks and Next-Generation Genetic Sequencing", *Health Matrix,* Vol. 28, 2018, p. 217. Chico, Victoria, "Requiring Genetic Knowledge: A Principled Case for Support", *Legal Studies,* Vol. 35, 2015,

Gene patents and their impact on access to genetic tests

Access to genetic tests is one of the major issues in the debate on gene patents. Some fear that patenting DNA may have a negative impact on access to genetic test, either by directly hindering the access to the diagnostic products that use the protected material or by creating barriers to research that could lead to developing genetic tests. Others argue that gene patents are necessary for incentivizing innovation and, on contrary, by inciting research also expensive areas they rather increase the access to genetic testing.[77] Therefore, patentability of scientific advances in the area of genetics and genomics shall be considered as well as property rights and the right to science.

Secondary use of genetic information, data, and samples, genetic testing for non-medical purposes

Genetic discrimination – understood as an adverse treatment on the basis of genetic characteristics – is one of the issues that is at the forefront of legal discussions on genetic testing. It is not only a potential outcome of a genetic test, but fear of it has also been identified in empirical studies as one of the most common reasons for declining a recommended genetic test.[78] Genetic discrimination is often discussed in the context of employment and insurance,[79] but it also addressed in relation to forensic criminal investigation (especially in reference to DNA criminal databases and discrimination of minorities),[80] as well in connection to immigration policy.[81] In the latter case, genetic testing is used in among others in family reunification procedures for verifying family filiations, what may lead to an adverse treatment compared to native citizens in at least two aspects – first, it narrows a definition of family to a biological meaning and second, in many jurisdictions immigrants have no right to decide what happens later to their genetic information, and in some jurisdictions it may be used for criminal investigations.[82] In the context of genetic testing, of particular concern is whether and how this information can be further used, which can be scrutinized under the data protection as well as under the right to private and family life/privacy, how protection of genetic discrimination and equality is addressed. This question is concerned with the

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p. 532. Wolf, Susan M., "The Continuing Evolution of Ethical Standards for Genomic Sequencing in Clinical Care: Restoring Patient Choice", *The Journal of Law, Medicine & Ethics,* Vol. 45, 2017, p. 333.

[77] Vogel, Jennifer, "Patenting DNA: Balancing the Need to Incentivize Innovation in Biotechnology with the Need to Make High-Quality Genetic Testing Accessible to Patients", *U. Kan. L. Rev.,* Vol. 61, 2012, p. 257. Mueller, Janice M., "Facilitating Patient Access to Patent-Protected Genetic Testing", *J. Bus. & Tech. L.,* Vol. 6, 2011, p. 83. Hawkins, Naomi, "The Impact of Human Gene Patents on Genetic Testing in the United Kingdom", *Genetics in Medicine,* Vol. 13, 2011, p. 320. WHO Human Genetics Programme. "Genetics, genomics and the patenting of DNA: review of potential implications for health in developing countries." 2015. http://www.who.int/iris/handle/10665/43100.

[78] Wauters and Van Hoyweghen, op. cit. 54.

[79] Joly, Yann, and others, "Comparative Approaches to Genetic Discrimination: Chasing Shadows?", *Trends in Genetics,* Vol. 33, 2017, p. 299. De Paor, Aisling, and Charles O'mahony, "The Need to Protect Employees with Genetic Predisposition to Mental Illness? The UN Convention on the Rights of Persons with Disabilities and the Case for Regulation", *Industrial Law Journal,* Vol.45, 2016, p. 525. Davey, James, "Genetic Discrimination in Insurance: Lessons from Test Achats" in Aisling De Pao, Gerard Quinn and Peter Blanck (eds), *Genetic Discrimination - Transatlantic Perspectives on the Case for a European Level Legal Response*, Taylor & Francis, 2014.

[80] Kaye, David H., and Michael E Smith, "DNA Identification Databases: Legality, Legitimacy, and the Case for Population-Wide Coverage", *Wis. L. Rev.,* 2003, p. 413. Chow-White, Peter A., and Troy Duster, "Do Health and Forensic DNA Databases Increase Racial Disparities?", *PLoS medicine,* Vol. 8, 2011.

[81] M'charek, Amade, Katharina Schramm and David Skinner, "Topologies of Race: Doing Territory, Population and Identity in Europe", *Science, Technology, & Human Values,* Vol.39, 2014, p. 468. Joly, Yann, and others, "DNA Testing for Family Reunification in Canada: Points to Consider", *Journal of International Migration and Integration,* Vol. 18, 2017, p. 391.

[82] Heinemann, Torsten, and Thomas Lemke, "Biological Citizenship Reconsidered: The Use of DNA Analysis by Immigration Authorities in Germany", *Science, Technology, & Human Values,* Vol. 39, 2014, p. 488.

secondary use of genetic data/information. Moreover, the permissibility of using genetic testing for other purposes than strictly health-related should be scrutinized.

Genetic testing in minors

Genetic testing in minors – children and adolescents – is usually discussed as a separate category. Since they do not have the capacity to consent, the choice whether to perform a test on them has to be made others and, as it has been widely accepted, a test may be performed only when it is in the child best interest.[83] What shall be done, however, when parents and physicians disagree about what is in the child's best interest, continues to generate discussions.[84] Furthermore, there is much debate about a test that has no immediate benefit for a child's health, such as in particular predictive testing for late-onset conditions (discussion concern both performing of the test altogether, as well as disclosing such results that were obtained as incident findings).[85] It has been argued that these type of tests should be generally avoided, as they impose a heavy psychological burden, have a negative social impact (the child is treated as being sick even before symptoms occur) and, as some argue, they deprive the child of "an open future".[86] These issues shall be further scrutinized in light of the right to health right to private life/privacy, right to the highest attainable standard of health for minors, as well as specifically under access to genetic testing by minors.

Summary of challenges, and human rights at stake and legal concerns in genetic testing (Table 6)

Challenges	**Human rights at stake and legal concerns**
Are patients making an informed choice and get appropriate counselling? How, if at all, the return of results and informing family members, is balanced against the right not to know and confidentiality?	• Information about the results of the test Counselling • Data protection • Right to information • Right not to know • Right to private life/privacy • Family interests/ rights • Confidentiality • Incidental findings
Gene patents, impact on access to genetic tests	• Patenting • Property rights • Right to science
Secondary use of genetic information, data and samples, genetic testing for non-medical purposes	• Genetic discrimination • Right to private life/privacy • Data protection • Secondary use of genetic data/information

[83] Friedman Ross, Lainie, "Predictive Genetic Testing of Children and the Role of the Best Interest Standard", *The Journal of Law, Medicine & Ethics,* Vol. 41, 2013, p. 899.

[84] Ibid.

[85] Borry, Shabani and Howard, op. cit. 73. Wilfond, Benjamin S., Conrad V Fernandez and Robert C. Green, *Disclosing Secondary Findings from Pediatric Sequencing to Families: Considering the "Benefit to Families"*, SAGE Publications Sage CA: Los Angeles, CA, 2015. European Society of Human Genetics, "Genetic Testing in Asymptomatic Minors: Recommendations of the European Society of Human Genetics." *European journal of human genetics,* Vol. 17, 2009. Friedman Ross, op.cit. 83.

[86] Wilfond, Benjamin S., Conrad V. Fernandez and Robert C. Green, *Disclosing Secondary Findings from Pediatric Sequencing to Families: Considering the "Benefit to Families"*, SAGE Publications Sage CA: Los Angeles, CA, 2015.

Genetic testing in minors	• Right to the highest attainable standard of health for minors • Right to private life/privacy • Access to genetic testing by minors

3.4 Prenatal genetic testing

Part of the genetic counselling offered to pregnant women consists of questions relating to genetic testing of the foetus. The purpose of the genetic testing at this stage is to provide women the necessary information they need in order to enhance their reproductive choice. However, it is their personal ethical beliefs and moral values which should lead them to make one choice over the other in regards to the continuation of their pregnancy after receiving the results of the genetic tests.[87] Arguably, the most morally debated outcome of such testing is that early diagnosis of genetic disorders offers the possibility women have to legally request the termination of their pregnancy in case they do not wish to give birth to a child with medical needs.[88] Prenatal genetic screening has advanced during the last decades with the non – invasive prenatal testing advancement (NIPT), offering more possibilities to pregnant women to be tested with non-invasive methods for any genetic marker and therefore rendering these ethical dilemmas even more complicated as the volume of the produced data will be significantly increased, rendering decision taking a more difficult procedure. [89]

Reproductive choices

Prenatal genetic testing/screening is strongly related to the reproductive choices of the pregnant woman. More recently, NIPT, a method of determining the risk that the foetus will be born with certain genetic abnormalities, which involves analysis of small fragments of DNA that are circulating in a pregnant woman's blood,[90] is being increasingly used. This method allows the pregnant women to obtain genetic information about their foetus early in the pregnancy without the increased risk of miscarriage and potentially terminate an affected pregnancy within the legal gestational limit (for example, as commonly related to amniocentesis). This possibility has been considered to increase woman's reproductive autonomy. Still, some authors pointed out that the early timing of NIPT might unnecessarily increase the burden of choice on women since pregnancies involving foetus abnormalities sometimes end in spontaneous abortion.[91] In light of reproductive choices, considerations over the availability of prenatal testing/screening are of particular importance. On the other hand, caution regarding NIPT if advertised directly-to-consumers has also been expressed.[92] These concerns will be considered as part of the direct-to-consumer advertising of NIPT under Chapter 3.6.

[87] Garcia, Elisa, Danielle R.M. Timmermans, Evert van Leeuwen, "The impact of ethical beliefs on decisions about prenatal screening tests: Searching for justification", *Social Science and medicine,* Vol. 66, 2008, p. 753.

[88] Jones R.J., "Genetic counselling and prevention of birth defects", JAMA 1982: 248(2), p. 221-4.

[89] Dukhonvy, Stephanie, Mary E. Norton, "What are the goals of prenatal genetic testing", *Seminars in perinatology,* Vol.42, 2018, p. 270.

[90] U.S. National Library of Medicine, "What Is Noninvasive Prenatal Testing (NIPT) and What Disorders Can It Screen For?" (*Genetics Home Reference*), 2 March 2019. https://ghr.nlm.nih.gov/primer/testing/nipt.

[91] Haidar, Hazar, Charles Dupras and Vardit Ravitsky, "Non-Invasive Prenatal Testing: Review of Ethical, Legal and Social Implications", *Bioéthique Online,* Vol.5, 2016, p. 5.

[92] Farrell, Ruth M., and others, "Online Direct-to-Consumer Messages about Non-Invasive Prenatal Genetic Testing", *Reproductive Biomedicine & Society Online,* Vol. 1, 2015, p. 88.

Scholars discuss whether any and what kind of legal limitations should be placed on the use of prenatal testing.[93] Some commentators are concerned that the availability of NIPT may increase the number of sex-selective abortions. It has been raised, however, that the laws banning sex-selective abortion restrict women's reproductive rights.[94] Moreover, it has been argued that laws and policies that define as unacceptable selection based on specific traits (sex or race for example) send a message that some forms of equality and respect have priority over others.[95]

Another group of challenges related to reproductive choices discussed in literature concerns the need to provide women and their families with appropriate, accurate and patient-centred information and counselling based on up-to-date genetic knowledge, and accommodating informed patients' legal choices.[96] These questions shall be further scrutinized in light of regulating the termination of pregnancy, including considerations over reasons for termination of pregnancy, including, such practices as sex-selection.

The growth of pre-natal testing creates tension between reproductive rights and respect for persons with disabilities. Scholars look at the relationship between reproductive autonomy (and selection practices) and disability rights and interests,[97] and question how to reconcile disability equality with support for parental choice.[98] The debate focuses on how and based on which criteria to decide which conditions can and should be tested for before implantation and prenatally. According to some commentators,[99] law has a role to play in counteracting negative assumptions about disability that impact medical advice regarding prenatal selection by requiring medical professionals to provide up to date information about the condition and contact information for support groups. In light of these considerations, questions over respect for diversity and disability as grounds for terminating a pregnancy should specifically be considered.

Standard of care and liability

The expansion of NIPT raises concerns about legal liability, the scope of physicians' legal obligations towards the patient and the standard of care to which physicians will be legally held. Claims may be based on non-provision of appropriate testing options, inadequate disclosure or failed communication of pertinent genetic risk, wrong interpretation of test result or on a failure of informed consent by failing to

[93] Ouellette, Alicia, "Selection against Disability: Abortion, ART, and Access", *The Journal of Law, Medicine & Ethics: A Journal of the American Society of Law, Medicine & Ethics,* Vol. 43, 2015, p. 211.

[94] Citro, Brian, and others, *Replacing Myths with Facts: Sex-Selective Abortion Laws in the United States* Cornell Law Faculty Publications, 2014.

[95] Ouellette, op. cit. 93.

[96] de Jong, Antina, Idit Maya and Jan MM van Lith, "Prenatal Screening: Current Practice, New Developments, Ethical Challenges: Prenatal Screening: Current Practice, New Developments, Ethical Challenges", *Bioethics,* Vol. 29, 2015, p. 1. Henneman, Lidewij, and others, "Responsible Implementation of Expanded Carrier Screening", *European Journal of Human Genetics,* Vol. 24, 2016, p.1. Ioannides, Adonis S., "Preconception and Prenatal Genetic Counselling", *Best Practice & Research Clinical Obstetrics & Gynaecology,* Vol. 42, 2017, p. 2.

[97] De Jong, Antina, and others, "Non-Invasive Prenatal Testing: Ethical Issues Explored", *European Journal of Human Genetics*, Vol. 18, 2010, p. 272. de Jong, Antina, and Guido MWR de Wert, "Prenatal Screening: An Ethical Agenda for the Near Future", *Bioethics,* Vol.29, 2015, p. 46.

[98] Asch, Adrienne, "Disability Equality and Prenatal Testing: Contradictory or Compatible", *Fla. St. UL Rev.,* Vol. 30, 2002, p. 315.

[99] Crossley, Mary, "Normalizing Disability in Families", *The Journal of Law, Medicine & Ethics: A Journal of the American Society of Law, Medicine & Ethics,* Vol. 43, 2015, p. 224.

thoroughly describe the characteristics of tests.[100] Omission to offer a test may be related to conscientious objection to an outcome, e.g. abortion to which a test may lead.[101] Failures or omissions may lead to "wrongful birth" or "wrongful life" claims. In light of these challenges, regulation of the professional obligations primarily should be further scrutinized, considering conscientious objection as well as wrongful birth/life issues.

Access, voluntariness, and consent

It has been pointed out that NIPT is caught "between a public health paradigm (with a mission for collective health and well-being) and a paradigm of reproductive autonomy and patient-centred health care (with a prospectus for individual rights)."[102] Some commentators have warned that offering prenatal testing on a wide scale and its routinization might undermine informed consent[103] and the voluntary character of the test.[104] On the other hand questions about equity in access for those with financial limitations have been raised,[105] as well as direct access of these tests by consumers.[106] In light of these challenges, access to NIPT vis-à-vis the highest attainable standard of health should be scrutinized, as well as decision-making regarding the testing with a view to voluntariness and consent.

Right not to know of the future children

There are also concerns related to the right not to know – parents who obtain genetic information about their future children may deny them the right not to learn their medical prognoses based on genetic tests.[107] Therefore, profound questions on safeguarding the right not to know can be raised as protected under the current legal frameworks.

Summary of challenges, and human rights at stake and legal concerns in prenatal testing (Table 7)

Challenges	**Human rights at stake and legal concerns**
Reproductive choices	• Regulating the termination of pregnancy • Sex selection • Abortion and disability: Impact on people with disabilities

Impact on people with disabilities	• Respect for diversity
	• Disability as grounds for terminating pregnancy:
	  ○ Equality
	  ○ Non-Discrimination

[100] Toews, Maeghan, and Timothy Caulfield, "Physician Liability and Non-Invasive Prenatal Testing", *Journal of Obstetrics and Gynaecology Canada,* Vol. 36, 2014, p. 907.

[101] Dickens, Bernard M., "Ethical and Legal Aspects of Noninvasive Prenatal Genetic Diagnosis", *International Journal of Gynecology & Obstetrics,* Vol. 124, 2014, p. 181.

[102] Ravitsky, Vardit, "The Shifting Landscape of Prenatal Testing: Between Reproductive Autonomy and Public Health", *Hastings Center Report,* Vol. 47, 2017, p. S34.

[103] De Jong and others, op. cit. 97.

[104] M Farrell, Ruth M., "Women and Prenatal Genetic Testing in the 21st Century", *Health Matrix,* Vol. 23, 2013, p.1.

[105] Allyse, Megan, and others, "Non-Invasive Prenatal Testing: A Review of International Implementation and Challenges", *International journal of women's health,* Vol, 7, 2015, p. 113.

[106] Tsuge, Azumi, "Ethical and Social Implications of Current Prenatal Genetic Testing", *Journal of Mammalian Ova Research,* Vol. 33, 2016, p. 109.

[107] De Jong and others, op. cit. 97. Deans, Zuzana, Angus J. Clarke and Ainsley J. Newson, "For Your Interest? The Ethical Acceptability of Using Non-Invasive Prenatal Testing to Test "Purely for Information"", *Bioethics,* Vol. 29, 2015, p. 19.

Standard of care and liability	• Quality of NIPT • Wrongful birth/life considerations
Access, voluntariness, and consent	• Right to the highest attainable standard of health • Decision-making about NIPT • Informed consent • Voluntariness
Right not to know	• Right not to know • Right to privacy • Genetic privacy

3.5 Newborn screening

Newborn screening in general may be controversial as the uncertainty of the screening interact with issues of privacy, religious and cultural beliefs. Mandating only certain well-established screening tests but also requiring from families to be informed about all tests and offering them the right to "opt in" in case they choose to, is considered to be the way to proceed with the case of newborn screening tests.[108] While genetic screening of newborns can lead to health benefits, it comes with challenges. Harms of screening programs include false positives (causing additional parental stress) and false negatives (potentially causing a delay in diagnosis). Therefore, proposals for neonatal screening require careful scrutiny by policymakers because of the potential above mentioned harms.[109]

Decision-making about screening: voluntariness, informed consent, and best interests

One of the central issues in the debate around new-born screening is the question of parents' consent. Screening for certain diseases is often mandatory or performed without their explicit consent, as it is seen to be in the best interest of the child's health[110] and having high benefits and low risks.[111] It is argued that benefits for the child outweigh any claims of parental autonomy and the state should protect its vulnerable citizens from preventable harms.[112] Others, however, argue that the question is not whether informed consent should be required for newborn screening or not – as it is framed too generally – but rather "under what circumstances should consent for new-born screening be required, and what form should that consent process take?"[113] Proponents of such view advocate for a more nuanced approach, under which, e.g. the "opt-out" option would be sufficient for clearly beneficial tests, whereas less straightforward cases would require opt-in consent.[114]

[108] Kraszewski, Jennifer, Taylor Burkeand, Sara Rosenbaum, "Legal Issues in Newborn Screening: Implications for Public Health Practice and Policy", *Public Health Rep.,* 2006 Jan-Feb; 121(1), p. 92.

[109] Cornel, M. et.al. Newborn screening in Europe 10 Expert Opinion document, EU tender, "Evaluation of population newborn screening practices for rare disorders in Member 5 States of the European Union" 03/07/2011.

[110] Howard, Heidi Carmen, and others, "Whole-Genome Sequencing in Newborn Screening? A Statement on the Continued Importance of Targeted Approaches in Newborn Screening Programmes", *European journal of human genetics,* Vol. 23, 2015, p. 1593.

[111] Fost, Norman, "Informed Consent Should Be a Required Element for Newborn Screening, Even for Disorders with High Benefit-Risk Ratios", *The Journal of Law, Medicine & Ethics,* Vol. 44, 2016 p.241.

[112] Ibid.

[113] Ibid.

[114] Ibid.

The debate on consent relates not only to parents" choices whether to perform the test but also to the acceptance of retention of the genetic data (or samples) and of their secondary uses for research.[115] It has been noticed that storing and reusing the data or samples without the knowledge of the parents might undermine the public trust in newborn screening programmes.[116] In light of the foregoing, such questions as voluntariness of screening programs, as well as voluntariness vis-à-vis risks it brings along in light of the child's best interests should be considered. Moreover, research on data and samples associated with the new-born screening should be further scrutinized.

Whole-genome sequencing in new-born screening and conditions part of a genetic screening

There is much debate about the potential use of whole-genome sequencing in newborn screening programmes. Some claim that it would allow producing a person's genetic data once for a lifetime (to be used when needed throughout one's whole life),[117] that it would constitute an "inevitable end point in the development of personalized medicine",[118] as well as that parents have a right to know everything that is possibly knowable about their children.[119] It has been also noticed that inclusion of the whole genome sequencing in new-born screening programmes could also create practical problems with storage of massive amount information for many years (especially in a manner that would have adequate privacy safeguards and at the same time that would make the data accessible for authorized uses).[120] Furthermore, such development could bring challenges for clinicians to interpret much of the gathered data and complications regarding incidental findings.[121]

What disorders are tested in new-born screening programmes and how to choose them are other pertinent issues in the discussion around newborn screening. The initial programmes aimed for well-understood disorders for which only early detection and treatment could lead to elimination or reduction of associated harm for the affected child.[122] In the later years, the scope also expanded to disorders going beyond these classical criteria. Some claim that the expansion has been going too fast, without sufficient deliberations and adequate medical evidence and also to reach the point in which mandatory performance of the tests are no longer justified.[123] While WGS new-born sequencing in itself presents challenges that ought to be resolved, e.g., regarding utility,[124] it is foremost important to scrutinize how,

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[115] Huckaby Lewis, Michelle, and Aaron J Goldenberg, *Return of Results from Research Using Newborn Screening Dried Blood Samples*, SAGE Publications Sage CA: Los Angeles, CA, 2015. Baily, Mary Ann, and others, "Exploring Options for Expanded Newborn Screening", *The Journal of Law, Medicine & Ethics: A Journal of the American Society of Law, Medicine & Ethics,* Vol. 33, 2005, p. 46.

[116] Carnahan, Sandra, "Biobanking Newborn Bloodspots for Genetic Research Without Consent", *Journal of Health Care Law and Policy,* Vol. 14, 2011, p. 299.

[117] Goldenberg, Aaron J., and Richard R. Sharp, "The Ethical Hazards and Programmatic Challenges of Genomic Newborn Screening", *Jama,* Vol.307, 2012, p. 461.

[118] Collins, Francis S., *The Language of Life: DNA and the Revolution in Personalized Medicine.* Harper Perennial, 2010., cited in: Howard and others, op. cit. 110.

[119] Clayton, Ellen Wright, "State Run Newborn Screening in the Genomic Era, or How to Avoid Drowning When Drinking from a Fire Hose", *Journal of Law, Medicine & Ethics,* Vol. 38, 2010, p. 697.

[120] Howard and others, op. cit. 110. Goldenberg and Sharp, op. cit. 117.

[121] Ibid.

[122] Howard and others, op. cit. 110. Clayton, op. cit. 119.

[123] Howard and others, op. cit. 110. Goldenberg and Sharp, op. cit. 117. Clayton, op. cit. 119.

[124] Matthijs, Gert, and others, "Guidelines for Diagnostic Next-Generation Sequencing", *European Journal of Human Genetics,* Vol. 24, 2016, p. 2. Richards, Sue, and others, "Standards and Guidelines for the Interpretation of Sequence Variants: A Joint Consensus Recommendation of the American College of Medical Genetics and Genomics and the Association for Molecular Pathology", *Genetics in medicine,* Vol. 17, 2015, p. 405.

if at all, selecting conditions for newborn screening programs are being regulated and whether there are any prevailing human rights arguments for the permissibility of such an expansive genetic analysis.

Summary of challenges, and human rights at stake and legal concerns in new-born screening (Table 8)

Challenges	Human rights at stake and legal concerns
Decision-making about screening	• Voluntariness of screening programs Conditions included in the screening programs • Child's best interests
Whole-genome sequencing in new-born screening, conditions in newborn screening	• Privacy • Immediate and future health benefits as part of the right to health and right to science

3.6 Advertising of genetic testing or screening

Advertising directly to consumers is usually discussed as only one of the aspects of the broader issue of direct-to-consumer genetic testing and the literature on this specific topic remains limited.[125] However, more generally, there is a considerable amount literature that scrutinizes ELSI challenges associated with direct-to-consumer genetic testing.

The common concern is that advertisements of these products may be misleading – exaggerate the benefits, create false hopes and downplay the risks, resulting in consumer misunderstanding of the offered test.[126] Impacts of misleading advertisements may range from psychological harms (e.g. creating unnecessary anxiety already on people only targeted by the advertisement[127] or imposing a psychological burden of learning about being at high risk for developing a disease in absence of professional counselling[128]) up to direct physical health problems, when results of such a test constitute an inappropriate basis for subsequent medical decisions. Beside affecting an individual, misleading adverting of genetic testing or screening may have a broader effect of increased costs to a healthcare system by causing unnecessary medical visits by concerned consumers, increasing physicians' time spent correcting misconceptions,[129] as well as by overprescribing of drugs because of consumer pressure on health care providers.[130]

In order to avoid these risks, some legal solutions have been proposed, starting from using a general consumer protection measures and tackling only advertising that is fraudulent or misleading, up to a complete ban on all advertising directly to consumers of genetic testing.[131] Some also advocated for a more graduate stand. Since direct-to-consumer tests are not homogenous – e.g. genealogical test may

[125] Kalokairinou, Borry and Howard, op. cit. 2.

[126] Kishore, Deepthy, "Test at Your Own Risk: Your Genetic Report Card and the Direct-to-Consumer Duty to Secure Informed Consent", *Emory LJ,* Vol. 59, 2009, p.1553. Robertson, Andrew S., "Taking Responsibility: Regulations and Protections in Direct-to-Consumer Genetic Testing", *Berkeley Tech. LJ,* Vol.24, 2009, p. 213. Hull, Sara Chandros, and Kiran Prasad, "Reading between the Lines: Direct-to-Consumer Advertising of Genetic Testing", *Hastings Center Report,* Vol. 31, 2001, p.33.

[127] Hogarth, Stuart, Gail Javitt and David Melzer, "The Current Landscape for Direct-to-Consumer Genetic Testing: Legal, Ethical, and Policy Issues", *Annu. Rev. Genomics Hum. Genet.,* Vol. 9, 2008, p. 161.

[128] Kishore, op. cit. 126.

[129] Myers, Mellanie F., "Health Care Providers and Direct-to-Consumer Access and Advertising of Genetic Testing in the United States", *Genome medicine,* Vol. 3, 2011, p. 81.

[130] Ibid.

[131] Kalokairinou, Borry and Howard, op. cit. 2.

[131] Ibid.

pose different threats than medical ones, then legal responses should also be differentiated and proportionate to the harm that they may cause, possibly banning advertising of some of them and imposing only informational obligations on those less dangerous.[132] It has been also raised that that commercial speech is to certain extent protected as a form freedom of expression and therefore that any regulation cannot limit it disproportionately.[133] A proponent of genetic tests advertising also argues that preventing individuals from accessing information about their genome (and ways to test) is paternalistic, as well as that such advertisements answer a "public appetite for information about the fruits of the Human Genome Project".[134] In that regard, the permissibility of direct-to-consumer genetic testing is a key concern, further scrutinizing conditions that are set for advertising, and whether there is a difference in requirements between health and non-health advertising of genetic testing.

Summary of challenges, and human rights at stake and legal concerns in direct-to-consumer advertising of genetic testing (Table 9)

Challenges	Human rights at stake and legal concerns
Advertising of genetic testing or screening Informed choice	• Permissibility of direct-to-consumer advertising • Requirements for direct-to-consumer advertising

4. Analysis of relevant international and regional laws and human rights standards

4.1 Relevant organizations and sources of law

The sources of hard and soft law reviewed in this chapter stem from the legal orders of interest: UN, ASEAN, AU, CoE, as well as OAS. Predominantly, treaties that define the competence and authority of the legal orders of interest, as well as key treaties and declarations that define civil and political rights, as well as economic, social and cultural rights have been scrutinized (see table 10 below). When relevant also other hard and soft laws of the respective legal orders have been considered, as well as interpretations of the norms by authoritative bodies, such as courts or human rights bodies, as well as arguments by the scholars. In case of UNESCO, the UNESCO Constitution has been reviewed as well as the following three declarations have been of particular concern: Universal Declaration on the Human Genome and Human Rights, as well as International Declaration on Human Genetic Data of 2003, Universal Declaration on Bioethics and Human Rights 2005. Occasionally, also WHO recommendations and policy instruments have been accounted for.

The analysis, on the one hand, considerably differs between questions that are at the nucleus of Task 2.2 study; on the other hand, many of concerns identified as part of these questions relate to the same normative framework. Therefore, unless it has been deemed necessary, substantive questions only are discussed providing a reference to the previously outlined normative framework.

Key sources of law (Table 10)

UN	ASEAN	AU	CoE	OAS

[132] Ibid. Hogarth, Javitt and Melzer, op. cit. 127.

[133] Gniady, Jennifer A., "Regulating Direct-to-Consumer Genetic Testing: Protecting the Consumer without Quashing a Medical Revolution", *Fordham L. Rev.,* Vol. 76, 2007. p. 2429. Hogarth, Javitt and Melzer, op. cit. 127.

[134] Human Genetics Commission, "Genes Direct: Ensuring the Effective Oversight of Genetic Tests Supplied Directly to the Public". http://www.hgc.gov.uk/genesdirect/genesdirect_full.pdf cited in: Hogarth, Javitt and Melzer, op. cit. 127.

UN Charter of the United Nations UN UDHR UN ICCPR UN ICESCR	ASEAN Charter ASEAN Human Rights Declaration	Constitutive Act of the African Union African Charter on Human and Peoples" Rights	Statute of the CoE CoE ECHR CoE (revised) European Social Charter	Charter of the Organization of American States OAS American Declaration of the Rights and Duties of Man OAS American Convention on Human Rights OAS Additional Protocol to the American Convention on Human Rights in the Area of Economic, Social and Cultural Rights "Protocol of San Salvador"

4.2 Analysis and assessment of the existing standards

4.2.1 Germline gene editing

Who has the competence and authority to regulate human germline interventions?

Except for the CoE BMC[135] as part of hard law, and UNESCO Universal Declaration on the Human Genome and Human Rights,[136] as well as CoE Parliamentary Assembly Recommendation 2115 (2017),[137] human germline editing is not *expresis verbis* addressed in human rights instruments. Therefore, concerns scientific advances in gene editing technology and their application to human germline raise, are to a considerable degree to be assessed through interpretation of the human rights norms of general nature, as well as hard and soft laws that address issues related to genetics and genomics, and in so far animals are concerned, also through the animal rights protection hard and soft laws.

The overall *governance of human germline gene editing* is rather fragmented. Neither of the human rights organizations have taken the initiative to comprehensively respond to the challenges that scientific advances in the area of human germline gene editing poses. Instead, currently they are to be tackled through the interpretation and application of the existing norms, including those that have been adopted considerably before the human germline gene editing application on humans was feasible. Another question is, however, whether any of the existing human rights organizations could take the initiative in elaborating and adopting a hard or soft law measure in the area.

The legal and human rights questions that human germline editing raises relates to all four purposes of the UN as outlined in Article 1 of the Charter of the United Nations. It could also relate more specifically to the competences of the Economic and Social Council, which is tasked with making recommendations to promote respect for, and observance of, human rights and fundamental freedoms for all.[138] Moreover, it may make recommendations concerning international economic, social, cultural, and educational, health, and related matters to the General Assembly to the Members of the United Nations, and to the specialized agencies concerned, which could act following its competences set forth in Article 13 of the UN Charter.

Under Article 1.1 of the UNESCO Constitution its purpose "is to contribute to peace and security by promoting collaboration among the nations through education, science and culture in order to further

[135] CoE, Convention for the protection of Human Rights and Dignity of the Human Being with regard to the Application of Biology and Medicine: Convention on Human Rights and Biomedicine, ETS 164, Article 13.
[136] UN Educational, Scientific and Cultural Organisation (UNESCO), Universal Declaration on the Human Genome and Human Rights, 11 November 1997.
[137] CoE, Parliamentary Assembly, The use of new genetic technologies in human beings Recommendation 2115 (2017).
[138] UN, Charter of the United Nations, 24 October 1945, 1 UNTS XVI, Article 62.

universal respect for justice, for the rule of law and for the human rights and fundamental freedoms which are affirmed for the peoples of the world, without distinction of race, sex, language or religion, by the Charter of the United Nations." Article 27 of the UDHR is within the scope of UNESCO competences.[139] In Article 1.2.a of the UNESCO Constitution, UNESCO's authority to "[c]ollaborate in the work of advancing the mutual knowledge and understanding of peoples, through all means of mass communication and to that end recommend such international agreements as may be necessary to promote the free flow of ideas by word and image."[140]

Regionally, the ASEAN Charter Article 1.7 sets forth purpose of the organization to promote and protect human rights and fundamental freedoms. Under Article 3(h) of the Constitutive Act of the African Union, the AU is tasked to promote and protect human and peoples' rights by the African Charter on Human and Peoples' Rights and other relevant human rights instruments. Although Article 2 of the Charter of the Organization of American States does not expressly assign OAS purpose to act in the area of human rights, general competencies have been used to pursue human rights within the region. Under Article 1 of the Statute of the Council of Europe, its aim is to achieve a greater unity between its members for safeguarding and realising the ideals and principles which are their common heritage and facilitating their economic and social progress, and in that regard actions, including agreements for the maintenance and further realisation of human rights and fundamental freedoms can be taken.

As derives from the analysis above, all human rights legal orders could take the initiative to tackle human germline editing, however, the question is who would be best suited to do that, and if done regionally, whether differences between regions could emerge, and if done at the UN level or by UNESCO, what level of protection/approach could the states agree on. In responding to the challenges, in 2017 the CoE called upon its members, who have not ratified the CoE BMC, to ratify it "without further delay, or, as a minimum, to put in place a national ban on establishing a pregnancy with germ-line cells or human embryos having undergone intentional genome editing".[141] This call has not lead to any further ratifications of the CoE BMC.[142] Recently, WHO has initiated establishing an expert panel to develop global standards for governance and oversight of human genome editing.[143] Recently the panel has highlighted that it is currently irresponsible to proceed with clinical applications of human germline editing, although neither has it called for a global moratorium.[144]

Legality of animal involvement and objective and consistency of regulatory strategies

[139] UN Educational, Scientific and Cultural Organisation (UNESCO) webpage, UNESCO and the Declaration. http://www.unesco.org/new/en/social-and-human-sciences/themes/human-rights-based-approach/60th-anniversary-of-udhr/unesco-and-the-declaration/.

[140] UNESCO, Constitution of the United Nations Educational, Scientific and Cultural Organisation (UNESCO), 16 November 1945, Article 1.3.

[141] CoE, Parliamentary Assembly, The use of new genetic technologies in human beings Recommendation 2115 (2017), para. 5.1.

[142] CoE, Chart of signatures and ratifications of Treaty 164. https://www.coe.int/en/web/conventions/full-list/-/conventions/treaty/164/signatures?p_auth=rujxJRj4.

[143] World Health Organization. Human Genome editing. https://www.who.int/ethics/topics/human-genome-editing/en/.

[144] Sara Reardon, "World Health Organization Panel Weighs in on CRISPR-Babies Debate" [2019] Nature. ahttp://www.nature.com/articles/d41586-019-00942-z.

The *use of animals in research* is addressed within the CoE, under European Convention for the Protection of Vertebrate Animals used for Experimental and other Scientific Purposes,[145] as amended under Protocol of Amendment to the European Convention for the Protection of Vertebrate Animals used for Experimental and other Scientific Purposes.[146] This convention ”applies to any animal used or intended for use in any experimental or other scientific procedure where that procedure may cause pain, suffering, distress or lasting harm”,[147] and permits use of animals for various purposes, including scientific research, and ”procedures related to avoidance or prevention of disease, ill-health or other abnormality, or their effects, in man, vertebrate or invertebrate animals or plants, including the production and the quality, efficacy and safety testing of drugs, substances or products; as well as diagnosis or treatment of disease, ill-health or other abnormality, or their effects, in man, vertebrate or invertebrate animals or plants.”[148] It sets forth well-being requirements,[149] and mandate authorizations for use of animals,[150] but does not set any restrictions relating to delivering knowledge that could later not be operationalized. While animal rights have received the attention of other human rights organizations, for example, the AU,[151] relevant protection mechanisms have not followed.[152]

As derives from the analysis above, animals are not precluded from being used for research for the benefits of humans. This use, as further below is scrutinized, is related to protecting a human right to science.

The beneficiary of human rights protection

Human germline editing could lead to fundamental changes in humanity. Hence a question of *beneficiaries of the existing human rights frameworks emerges*, as well as what direction human rights should take regarding human germline editing.

UN UDHR in Article 1 addresses all human beings, UN ICCPR and UN ICESCR in Article 1.1 address ”all peoples”, whereas, for example, the preamble of the UN CRC is concerned with the need to protect a child before birth. UN CRPD, for example, explicitly refers to ”persons” in Article 1, but in the preamble emphasizes ”all members of the human family”, whereas the preamble of the UN CEDAW explicitly refers to ”men and women”. UNESCO Universal Declaration on the Human Genome and Human Rights refers to ”everyone”, UNESCO Universal Declaration on Bioethics and Human Rights under Article 1.1 is concerned with application of scientific advances to ”human beings”, whereas Article 1.a of International Declaration

[145] CoE, European Convention for the Protection of Vertebrate Animals used for Experimental and other Scientific Purposes, ETS 123.

[146] CoE, Protocol of Amendment to the European Convention for the Protection of Vertebrate Animals used for Experimental and other Scientific Purposes, ETS 170.

[147] CoE, European Convention for the Protection of Vertebrate Animals used for Experimental and other Scientific Purposes, ETS 123, Article 1.

[148] CoE, European Convention for the Protection of Vertebrate Animals used for Experimental and other Scientific Purposes, ETS 123, Articles 2a and 2 d.

[149] European Convention for the Protection of Vertebrate Animals used for Experimental and other Scientific Purposes, op.cit., Article 5.

[150] European Convention for the Protection of Vertebrate Animals used for Experimental and other Scientific Purposes, op.cit., Article 13.

[151] AU. Animal Welfare Strategy for Africa (AWSA). http://www.rr-africa.oie.int/docspdf/en/2017/AWSA_Executive_Summary_Layout_ENG_2017.pdf.

[152] For example, the Universal Declaration of Animal Rights (15 October 1978) is of relevance, however, status of this declaration has been subject to discussions as it does not seem to have formally been adopted by the UNESCO. Roeder, Larry Winter, and Albert Simard, “Security, Risk Analysis and Intelligence” in Jr Roeder Larry Winter and Albert Simard (eds), *Diplomacy and Negotiation for Humanitarian NGOs*, Springer, New York, 2013. p.149.

on Human Genetic Data refers to protecting ”human genetic data, human proteomic data and of the biological samples from which they are derived”.

ASEAN Human Rights Declaration Principle 1 addresses ”all persons”. AU African Charter on Human Rights and Peoples” Rights Article 2 addresses ”every individual”, AU Protocol to the African Charter on Human and Peoples' Rights on the Rights of Women in Africa refers to ”women” specifically in Article 2.1. CoE ECHR refers to ”everyone” in its Article 1, CoE BMC refers to ”human beings” and ”everyone” in Article 2, and CoE revised ESC relates to ”everyone” in its Article 1. However, as derives from early instruments, such as CoE, Parliamentary Assembly, Recommendation 934, Genetic Engineering, 1982, “the rights to life and to human dignity protected by Articles 2 and 3 of the European Convention on Human Rights imply the right to inherit a genetic pattern which has not been artificially changed”. While this is an interpretation only, made by the Parliamentary Assembly and not the ECtHR, it has informed regulatory framework in the area within the CoE. OAS American Declaration of the Rights and Duties of Man Article I addresses every human being, OAS American Convention on Human Rights Article 1 addresses ”all persons”, whereas OAS Additional Protocol to the American Convention on Human Rights in the Area of Economic, Social and Cultural Rights "Protocol of San Salvador" in its preamble relates to ”rights of man”.

Generally, the beneficiary of the human rights instruments is aimed at addressing human species. Neither of these instruments places any restrictions regarding protection of persons with potentially edited germline, and consequently they would be at risk of not being protected under the current human rights frameworks.

Legal status of the human embryo and its protections

Generally, human rights organizations have been rather reluctant to address the question of the *legal status of the human embryo*, and whether embryo could be seen as a human rights beneficiary. These questions have emerged, for example, as part of responses to draft general on Article 6 of ICCPR that protects right to life,[153] nonetheless the adopted version is silent on any status of an embryo, but requires measures for the termination of pregnancy, for example.[154] Similar questions have been raised before the ECtHR, which this far has followed the approach previously taken by the European Commission on Human Rights and has not afforded special protection to human embryos.[155] It can be argued that this question falls within the state’s margin of appreciation and they are free to provide special protection.[156]

OAS American Convention on Human Rights "Pact of San Jose" includes the right to life (“in general, from the moment of conception” – article 4), which has been interpreted not to cover the phase before an embryo is being transferred into a woman’s womb.[157]

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[153] Human Rights Committee, Draft General Comment on Article 6 of the International Covenant on Civil and Political Rights – Right to life. https://www.ohchr.org/en/hrbodies/ccpr/pages/gc36-article6righttolife.aspx.

[154] Human Rights Committee, General comment No. 36 (2018) on article 6 of the International Covenant on Civil and Political Rights, on the right to life, CCPR/C/GC/36, para. 8.

[155] ECtHR, X. v. United Kingdom (7215/75) 12 October 1978, E Eur Comm HR, Paton v. United Kingdom (8416/78) 13 May 1980. "[t]his would mean that the ‘unborn life’ of the foetus would be regarded as being of a higher value than the life of the pregnant woman", para. 20. Eur Comm HR, H. v. Norway (17004/90) 19 May 1992. ECtHR, Boso v. Italy (50490/99) 05 September 2002. ECtHR, Vo v. France (53924/00) 08 July 2004, paras. 9–14.

[156] On margin of appreciation see ECtHR, (GC), S.H. AND OTHERS v. AUSTRIA, (57813/00), 03 November 2011.

[157] In the Inter-American Court of Human Rights judgement of 28 November 2012 in the case of Artavia Murillo et al (“In Vitro Fertilization”) v. Costa Rica, the Court has concluded that “«conception» in the sense of Article 4(1) occurs

The question of the human embryo and scientific research is *expressis verbis* addressed within the CoE. Article 18.2 of the CoE BMC strictly prohibits the creation of human embryos for research purposes, but it does not prohibit the conduct of research on human embryos per se.[158] Further restrictions are elaborated in the CoE Recommendation 1046(1986), regarding the Use of human embryos and foetuses for diagnostic therapeutic, scientific, industrial and commercial purposes, including calling on the CoE members to forbid anything that could be considered as undesirable use or deviations of these techniques,[159] "including

- the creation of identical human beings by cloning or any other method, whether for selection purposes or not;
- the implantation of a human embryo in the uterus of an animal or the reverse;
- the fusion of human gametes with those of another animal (the hamster test for the study of male fertility could be regarded as an exception, under strict regulation);
- the creation of embryos from the sperm of different individuals;
- the fusion of embryos or any other operation which might produce chimeras;
- ectogenesis, or the production of an individual and autonomous human being outside the uterus of a female, that is, in a laboratory;
- the creation of children from people of the same sex;
- choice of sex by genetic manipulation for non-therapeutic purposes;
- the creation of identical twins;
- research on viable human embryos;
- experimentation on living human embryos, whether viable or not;
- the maintenance of embryos *in vitro* beyond the fourteenth day after fertilisation (having deducted any time necessary for freezing)."[160]

Within the AU, Resolution on Bioethics has called for "supervision of research facilities on embryos especially those produced as a result of medical procedures offering assistance towards procreation and the attendant application of such procedures, so as to obviate selective eugenic by-products particularly those relating to sex considerations".[161] Sex selection has also been addressed within the Council of Europe, which under Article 14 of the CoE BMC permits it only where a serious hereditary sex-related disease is to be avoided.[162]

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at the moment when the embryo becomes implanted in the uterus, which explains why, before this event, Article 4 of the Convention would not be applicable" (para. 264) and "the embryo, prior to implantation, is not covered by the terms of Article 4 of the Convention" (para. 315). Furthermore, the Court formulated a "principle of the gradual and incremental protection of prenatal life" (para. 315) – "it can be concluded from the words "in general" that the protection of the right to life under this provision is not absolute, but rather gradual and incremental according to its development, since it is not an absolute and unconditional obligation, but entails understanding that exceptions to the general rule are admissible" (para. 264).

[158] Ledford, Heidi, "The Landscape for Human Genome Editing", *Nature,* Vol. 526, 2015, p. 310.

[159] CoE, Parliamentary Assembly, Use of human embryos and foetuses for diagnostic, therapeutic, scientific, industrial and commercial purposes, Recommendation 1046 (1986).

[160] CoE, Parliamentary Assembly, Use of human embryos and foetuses for diagnostic, therapeutic, scientific, industrial and commercial purposes, Recommendation 1046 (1986), Section 14.1.4.

[161] Assembly of Heads of State and Government of the Organization of African Unity, Resolution on Bioethics, AHG/Res 254(XXXII), 10 July 1996.

[162] This prohibition is also endorsed in CoE, Parliamentary Assembly, Prenatal sex selection, Resolution 1829 (2011).

As derives from the above, the questions of the status of human embryo and permissibility either to use it in research or to create it for research are rather controversial. While the human embryo does not enjoy human rights, it could be subject of some protections, and depending on what protection a state has opted for, research could either be limited or furthered in that regard.

Right to health Right to health, in particular, quality dimension, including such angles as safety and efficacy

Right to health/the highest attainable standard of health is a common concern to the human rights actors. Some legal orders and their human rights instruments place considerable emphasis to protecting a particular societal group, for example, women, youth or children, or address childhood as a specific period in life within which particular attention to health should be given, others set forth more generic requirements for creating standard that is adequate for health or ensuring the highest (elsewhere as "best") attainable standard of health and removing ill-health risks (for example, UN UDHR Article 25, UN ICESCR Article 12, UN CEDAW Article 12, UN CRC Article 24, UN CRPD Article 25, UNESCO Universal Declaration on Bioethics and Human Rights Article 14, OAS American Declaration of the Rights and Duties of Man Article XI, OAS Additional Protocol to the American Convention on Human Rights in the Area of Economic, Social and Cultural Rights "Protocol of San Salvador" Article 10, AU African Charter on Human and Peoples' Rights Article 16, AU Protocol to the African Charter on Human and Peoples' Rights on the Rights of Women in Africa Article 14, AU Youth Charter Article 16, CoE (revised) European Social Charter Article 11.[163]

Neither of these instruments that protect the *right to health or highest attainable standard of health expressis verbis* require granting access to certain scientific advances. Moreover, commonly either expressly or implied these advances are subject to the maximum available of resources. Nonetheless, in order to rely on this right in application, it should first be further assessed whether, if at all, the right to health or highest attainable standard of health could be relied on to argue for access to certain interventions, such as human germline modifications. Second, it should be assessed whether there is a duty to revisit these bans, and if so, under what circumstances access should be given, and whether, for example, such commonly recognized principles as the *principle of equality and prohibition of* is observed.

Safety relating to germline editing technologies require accounting for the protection of inherent dignity afforded to people. ICCPR Article 7 prohibits subjecting persons to medical or scientific experimentation, except for their free consent. This prohibition, albeit in a less stringent form – under strict conditions allowing involving persons unable to consent – is also upheld and endorsed through regional instruments, for example, CoE Additional Protocol to the Convention on Human Rights and Biomedicine, concerning Biomedical Research.[164] In AU, Articles 4 (2)(h) of the AU Protocol on the Rights of Women in Africa of the African Charter on Human and Peoples" Rights specifically prohibits medical experimentation on women without their informed consent. Commonly, human rights instruments protect life, prohibit torture, inhumane or degrading treatment, and protect private life or, in the case of AU, specifically integrity. Through tripartite obligations to respect, protect and fulfil, the balance has to be struck between self-determination regarding use of these technologies, and the obligation to take measures to protect from harm associated with these technologies. Neither of the human rights instruments provide for a particular

[163] Although commonly the right to the highest attainable standard of health or the right to health belongs to socioeconomic rights, which are characterized by their lack of enforceability, through indivisibility doctrine of human rights, it is often also enforced through civil and political rights provisions.

[164] See CoE, Additional Protocol to the Convention on Human Rights and Biomedicine, concerning Biomedical Research, ETS 195, Articles 13-15.

stand on the issue; they protect rights that could be relied upon to support both of the positions. Hence, depending on a right at stake in a particular situation, a careful assessment would need to be made.

While ill-health is a commonly shared concern and prevention has a considerable role in addressing it, only some human rights instruments *expressis verbis* address preventive care (for example, Article 12.2.c of ICESCR). However, in so far as human rights instruments address public health or the right to the highest attainable standard of health, it could be argued that prevention should be seen as a constituting element of these rights.

Accessibility and acceptability are not commonly addressed as a distinct requirement, nonetheless certain elements are set forth in law (for example, CoE BMC Article 3). In those that address the right to the highest attainable standard of health, following analogy with Article 12 ICESCR accessibility and acceptability could be seen as a component of this right.[165] Moreover, accessibility could be seen as relevant to such core and commonly shared rights as the prohibition of discrimination. Similarly, acceptability could be seen as relating to stigmatization. Both of these aspects are addressed separately below.

Quality can be seen as an element of the right to the highest attainable standard of health,[166] and therefore could be argued to extend to germline interventions in so far as they fall under this right.

To conclude, legal orders of concern provide a normative framework for protecting the right to health. Realization of this protection remains primarily at the national level, and similarly to other questions, depends on the permissibility of germline editing technology per se.

Right to sexual and reproductive health

While *sexual and reproductive health* generally forms part of the right to health normative framework, which has been outlined above, none of the human rights instruments expressly afford specific entitlements regarding human germline. Sexual and reproductive health could also be seen as relating to other rights, for example, reproductive freedom, information, and decision-making in that regard, which is commonly protected under family planning rights or the right to private life/privacy. The former is addressed, for example, under UN UDHR Article 16 that protect the right to found a family. Access to health care services in family planning is a specific right in Article 14.2.b of UN CEDAW, while article 10.h of UN CEDAW protects also access to information and advice on family planning in education. Reproductive rights including family planning are outlined in Article 14 of Protocol to the African Charter on Human and Peoples' Rights on the Rights of Women in Africa. It is also recognized as part of private life under Article 8 CoE ECHR. While sexual and reproductive health is generally protected it is ambiguous what entitlements it covers, and whether germline editing technologies could be seen as falling in the scope of this right.

Scope and limits of self-determination, right to private life/privacy, protection of the integrity and the right to science; adequacy of the existing regulatory approaches

While the question of scope and limits of self-determination, right to private life/privacy, protection of integrity and the right to science are distinct human rights, in the context of human germline as an

[165] UN Committee on Economic, Social and Cultural Rights (CESCR), General Comment No. 14: The Right to the Highest Attainable Standard of Health (Art. 12 of the Covenant), 11 August 2000, E/C.12/2000/4.

[166] UN Committee on Economic, Social and Cultural Rights (CESCR), General Comment No. 14: The Right to the Highest Attainable Standard of Health (Art. 12 of the Covenant), 11 August 2000, E/C.12/2000/4.

intervention the fundamental question is whether the right to benefit from scientific advances includes benefiting from human germline editing, and if so, under what circumstances this could be applied. Therefore, the normative framework to self-determination, right to private life/privacy, protection of integrity becomes secondary vis-à-vis the right to science and is considered jointly with the right to science.

The right to science initially was included in Article 27.1 of the UDHR, then it was enshrined in Article 15 ICESCR, and it has subsequently been given expression in various area-specific instruments, for example, UNESCO Universal Declaration on Bioethics and Human Rights which is tasked with tackling ”ethical issues related to medicine, life sciences and associated technologies as applied to human beings, taking into account their social, legal and environmental dimensions”.[167]

A right to benefit from scientific advances has also been regionally protected. ASEAN Human Rights Declaration Principle 32 affirms the right to benefit from scientific advances. The AU Charter of the African Union Article II (2) identifies scientific and technical cooperation as essential for meeting its goals. OAS, Charter of the Organization of American States, Article 38 stipulates that States ”shall extend among themselves the benefits of science and technology by encouraging the exchange and utilization of scientific and technological knowledge”. Within the CoE, as case law of the ECtHR affirms, access to advances goes hand in hand with protection from risks these advances pose. The CoE BMC in its preamble affirms “the need for international cooperation so that all humanity may enjoy the benefits of biology and medicine”, and in Article 2 declares that the ”interests and welfare of the human being shall prevail over the sole interest of society or science”, and in Article 15 states that ”scientific research in the field of biology and medicine shall be carried out freely, subject to the provisions of this Convention and the other legal provisions ensuring the protection of the human being”.

Substantively, the UN Special Rapporteur in the field of cultural rights noted that ”[t]he terms “benefits” of science and “scientific progress” convey the idea of a positive impact on the well-being of people and the realization of their human rights. The “benefits” of science encompass not only scientific results and outcomes but also the scientific progress, its methodologies and tools.”[168] Yet, under Article 4 ICESCR right to science can be restricted to protect form arising harms. Although in the UDHR and ICESCR these issues are not *expressis verbis* addressed, it has been argued that the right to science provisions have been adopted under this background, and through the indivisibility doctrine of human rights, other rights shall be safeguarded. Therefore, in order to argue that human germline editing is permissible in the first place, a question that should further be analysed is how the benefits should be balanced against harms these advances could lead to (both, at the individual level and societal level).[169]

[167] UN Educational, Scientific and Cultural Organisation (UNESCO), Universal Declaration on Bioethics and Human Rights 19 October 2005, Article 1.1.

[168] UN General Assembly, Report of the Special Rapporteur in the field of cultural rights, Farida Shaheed, ‘The right to enjoy the benefits of scientific progress and its applications’, A/HRC/20/26 14 May 2012, https://www.ohchr.org/Documents/HRBodies/HRCouncil/RegularSession/Session20/A-HRC-20-26_en.pdf III.A.

[169] Ibid.. General Comment in the area has not been adopted yet. CESCR, ‘General discussion on a draft general comment on article 15 of the International Covenant on Economic, Social and Cultural Rights: on the right to enjoy the benefits of scientific progress and its applications and other provisions of article 15 on the relationship between science and economic, social and cultural rights’, https://www.ohchr.org/EN/HRBodies/CESCR/Pages/Discussion2018.aspx.

It has been argued that the right to science requires furthering research to continuously re-assess the balance between entitlements and protections.[170] Therefore, the question regarding consistency of regulatory strategies (allowing research on animals relating to human germline editing, but not allowing to proceed to clinical trials), which was raised as part of the literature review, becomes irrelevant since furthering the right to science could be difficult to achieve without using animals and in the absence of other feasible alternatives.

Disability rights

The questions of disability, vulnerability, discrimination, and stigmatization have commonly been at the forefront of discussions relating to the human genome. Most recently, disability protection is set forth in UN CRPD that prohibits discrimination, sets forth equality protection, and mandates awareness raising of disability, including awareness raising to nurture receptiveness to the rights of persons with disabilities.[171] More generally, the human rights protection relates to that discussed below regarding discrimination, genetic discrimination, equality and stigmatization, which is mapped out below. While not expressly stated in CRPD, practices aimed at combating disability, could be seen as contrary to the values the convention aspires to protect.

Human germline gene editing is not the only application of genetics and genomics that raises considerable concerns relating to disability rights that can also be seen as anchoring in human dignity. For example, the CoE BMC that *expressis verbis* addresses practices relating to human genome does not preclude using PGD as such but place limitations relating to the use of technology for sex-selection purposes. ECtHR has heard a case relating to PGD and desire to conceive a child without a genetic disability. The ECtHR has noted that "desire to conceive a child unaffected by the genetic disease of which they are healthy carriers and to use ART and PGD to this end attracts the protection of Article 8, as this choice is a form of expression of their private and family life."[172] Therefore, while the intervention might be "by the law" and pursue a legitimate aim, it also has to be necessary in a democratic society.[173] It is well-established that in matters that raise moral concerns states enjoy a wider margin of appreciation. However, given the functioning of the CoE system,[174] and in particular, that CoE BMC prohibits human germline interventions, it is difficult to see how states might have such a margin of appreciation that would afford them to accommodate practices of human germline gene editing.[175]

The normative framework relating to disability rights and specifically elimination of diseases is rather sporadic and leaves considerable room for further scrutinizing controversial questions. While persons with disability are equal to those not having a disability, as evidenced by the CoE framework and judicial responses, technological advances can be used for eliminating disability. If diversity is a value that shall be protected (UNESCO), should disability-related practices be regarded as undesirable? It remains here responded how other regional legal orders that have similar adjudication mechanisms as in the CoE (AU, OAS) approach scientific advances vis-à-vis human rights.

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[170] Slokenberga and Howard, op. cit. 9.

[171] UN General Assembly, Convention on the Rights of Persons with Disabilities : resolution / adopted by the General Assembly, 24 January 2007, A/RES/61/106, Articles 3, 5, 8.

[172] ECtHR, Costa and Pavan v. Italy, (54270/10), 28 August 2012, para. 57.

[173] Costa and Pavan v. Italy, op.cit., paras. 58-69.

[174] Slokenberga, Santa, *European Legal Perspectives on Health-Related Direct-to-Consumer Genetic Testing*, Jure, 2016, ch. 9.

[175] On margin of appreciation and scientific advances see ECtHR, (GC), S.H. and others v. Austria, (57813/00), 3 November 2011, paras. 91-97.

Discrimination, genetic discrimination, equality and stigmatization

Different human rights instruments address the question of discrimination and stigmatization differently. Generally, human rights instruments prohibit discrimination only, or address discrimination jointly with a requirement of equality, or recognize equality of all persons (for example, UN UDHR Article 7, UN ICCPR Article 26, UN ICESCR Article 2, UN CRC Article 2, ASEAN Human Rights Declaration Article 3, OAS Protocol of San Salvador Article 3, OAS American Declaration of the Rights and Duties of Man Article II, OAS American Convention on Human Rights Article 1 and 24, AU African Charter on Human and Peoples' Rights Articles 2 and 3, CoE ECHR Article 14).

Some human rights instruments set forth protection requirements for particular groups with a view to combat discrimination that relates to the group (for example, UN CEDAW, UN CRPD, AU Protocol to the African Charter on Human and Peoples" Rights on the Rights of Women in Africa). Some human rights legal orders address group-specific needs in general human rights instruments (for example, ASEAN Human Rights Declaration Article 4), or have adopted a generic instrument to combat discrimination and intolerance (for example, OAS Inter-American Convention Against All Forms of Discrimination and Intolerance).

While grounds for discrimination are commonly set in law (for example, UN ICCPR Article 26, UN ICESCR Article 2, UN CRC Article 2, UNESCO Universal Declaration on Bioethics and Human Rights Article 11, OAS Protocol of San Salvador Article 3, OAS American Declaration of the Rights and Duties of Man Article II, OAS American Convention on Human Rights Article 1,OAS Inter-American Convention Against All Forms of Discrimination and Intolerance Article 1.1, AU African Charter on Human and Peoples' Rights Article 2, AU Protocol to the African Charter on Human And Peoples" Rights on the Rights of Women in Africa Article 1b, CoE ECHR Article 14), only those hard and soft law instruments that specifically address genetic interventions commonly address explicitly discrimination and/or stigmatization based on human genome (for example, UNESCO International Declaration on Human Genetic Data Article 7, UNESCO Universal Declaration on the Human Genome and Human Rights Article 6, CoE APGT Article 4); also a biology and medicine specific human rights instrument, such as CoE BMC expressly set forth prohibition of discrimination based on genetic heritage in Article 11. CoE Recommendation (92) 3 expressly address equality in terms of access to testing/screening under Principle 4. Therefore, it can be concluded that legal orders of concern provide normative framework for tackling discrimination, genetic discrimination, equality and stigmatization. Realization of this protection remains primarily at the national level, and similarly to other questions, depends on the permissibility of germline editing technology per se.

Liability and access to justice

A common feature to human rights treaties and declarations, is that states are required to ensure that the rights are protected in their jurisdiction. This obligation can be seen in terms of the respect, protect, fulfil obligations, whereby states are required not to intervene with the rights afforded to individuals, take measures to protect these rights from interferences by others, as well as take measures to enjoy these rights. In so far as protected rights are violated effective remedy is triggered. A right to effective remedy is commonly protected in all legal orders of concern, though the wording of respective provisions differs, including under UN ICCPR Article 2.3.a and UN UDHR Article 8; ASEAN Human Rights Declaration Principle 5; Article 25 of the Protocol to the African Charter on Human and Peoples" Rights on the Rights of Women in Africa; CoE ECHR Article 13, CoE BMC Articles 23-25; OAS American Declaration of the Rights and Duties of Man Article XVIII as well as OAS American Convention on Human Rights "Pact of San Jose, Costa Rica" Article 25.1. Therefore, it can be concluded that legal orders of concern provide a normative framework for securing protection to the human genome. Realization of this protection remains primarily at the national level.

4.2.2 Genetic Screening

Public health and the right to the highest attainable standard of health

Health, whether addressed as a right to public health and/or the right to the highest attainable standard of health is a common concern to the human rights actors and in general terms it has already been reviewed above. Here suffices to notice that neither of the human rights instruments expressly require to include genetic screening as part of the public health measure, nor do they define in any way what conditions shall be part of the genetic screening. As derives from General Comment 14 adopted under UN ICESCR Article 12, availability requires that "functioning public health and health-care facilities, goods and services, as well as programmes, have to be available in sufficient quantity within the State party".[176] The precise nature of these measures may differ, depending on numerous of factors, including the state developmental level.[177] Nonetheless, WHO has drawn attention to particular genetic conditions and their public health importance:[178]

- Birth defects;[179]
- Thalassaemia and other haemoglobinopathies;[180]
- Sickle cell anaemia.[181]

Accessibility and acceptability has already been reviewed above and it suffices to reiterate that is not commonly addressed as a distinct requirement, nonetheless certain elements are set forth in law.

Quality is only expressly addressed in instruments that expressly address genetic screening (see, for example, UNESCO International Declaration on Human Genetic Data Article 15, CoE BMC Article 3, CoE APGT Article 5 and 19, CoE Recommendation Nr. (92)3 Principle 2). However, it can also be seen as an element of the right to the highest attainable standard of health,[182] and therefore could be argued to extend beyond ASEAN Agreement On Medical Device Directive, which sets standards for medical devices placed on the markets of the ASEAN Member States, aimed at safety and health of their users.[183] In AU, where genetic screening is or were to be used in medical context, AU Model Law for Medical Products Regulation would be relevant, which among others stipulates that, as a rule, all medical products must be registered and have a valid marketing authorization and certificate of conformity (article 13).[184] It should be emphasized, however, that the Model Law is not a prescriptive, directly applicable instrument and it requires domestic implementation by the AU Member States.[185]

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[176]UN Committee on Economic, Social and Cultural Rights (CESCR), General Comment No. 14: The Right to the Highest Attainable Standard of Health (Art. 12 of the Covenant), 11 August 2000, E/C.12/2000/4, §12a.

[177]UN Committee on Economic, Social and Cultural Rights (CESCR), General Comment No. 14: The Right to the Highest Attainable Standard of Health (Art. 12 of the Covenant), 11 August 2000, E/C.12/2000/4, §12a.

[178]World Health Assembly, "Resolutions on Human Genomics". https://www.who.int/genomics/WHAGenomics/en/.

[179] World Health Assembly, "Birth defects". http://apps.who.int/gb/ebwha/pdf_files/WHA63/A63_R17-en.pdf?ua=1.

[180]World Health Assembly, "Thalassaemia and other haemoglobinopathies". http://apps.who.int/gb/ebwha/pdf_files/EBSS-EB118-2006-REC1/english/Res/listing/b118_r1-en.pdf?ua=1.

[181]World Health Assembly, "Sickle-cell anaemia". http://apps.who.int/iris/bitstream/handle/10665/21447/A59_R20-en.pdf?sequence=1.

[182] See UN Committee on Economic, Social and Cultural Rights (CESCR), General Comment No. 14: The Right to the Highest Attainable Standard of Health (Art. 12 of the Covenant), 11 August 2000, E/C.12/2000/4.

[183] The Association of Southeast Asian Nations, Agreement on Medical Device Directive, September 2015.

[184] AU, Model Law for Medical Products Regulation, January 2016.

[185] The New Partnership for Africa's Development, "Issue Brief: AU Model Law for Medical Products Regulation: Increasing access to and delivery of new health technologies for patients in need".

Within Council of Europe, hard and soft law measures exist that set forth detailed requirements to genetic screening. Article 19 of APGT addresses specifically genetic screening programmes for health purposes. Whereas detailed principles and recommendations in genetic screening are elaborated by the CoE in R (92) 3 on genetic testing and screening for health care purposes.

Article 19 of APGT	CoE in R (92) 3 on genetic testing and screening for health care purposes sets forth the following principles:
”A health screening programme involving the use of genetic tests may only be implemented if it has been approved by the competent body. This approval may only be given after independent evaluation of its ethical acceptability and fulfilment of the following specific conditions: A the programme is recognised for its health relevance for the whole population or section of population concerned; B the scientific validity and effectiveness of the programme have been established; C appropriate preventive or treatment measures in respect of the disease or disorder which is the subject of the screening, are available to the persons concerned; D appropriate measures are provided to ensure equitable access to the programme; E the programme provides measures to adequately inform the population or section of population concerned of the existence, purposes and means of accessing the screening programme as well as the voluntary nature of participation in it.”	Principle 1 - Informing the public Principle 2 - Quality of genetic services Principle 3 - Counselling and support Principle 4 - Equality of access - non-discrimination Principle 5 - Self-determination Principle 6 - Non-compulsory nature of tests Principle 7 - Insurance Principle 8 - Data protection Principle 9 - Professional secrecy Principle 10 - Separate storage of genetic information Principle 11 - Unexpected findings Principle 12 - Supervision Principle 13 - Handling of data

Detailed requirements for genetic screening set forth in CoE (Table 11)

As derives from the analysis above, while health is of concern to all surveyed human rights legal orders, specific conditions are not addressed as part of the human rights measures. Of the surveyed human rights legal orders, only the CoE has a detailed framework (both, hard and soft law) for genetic screening and the protection of individual rights in that regard.

Discrimination and stigmatization

The normative framework relating to discrimination and stigmatization has already been reviewed above. Here suffices to reiterate that questions of discrimination and/or stigmatization are addressed in all legal orders of concern. In those instruments that specifically address genetic screening or questions relating to the human genome, protection against discrimination and/or stigmatization is also required.

Right to private life/privacy and right to informed consent

Questions that relate to non-coerced genetic screening can be addressed through various rights, for example, general integrity-protecting rights (for example, right to private life/privacy), as well as specifically right to informed consent.

https://www.nepad.org/publication/issue-brief-african-union-model-law-medical-products-regulation-increasing-access.

The right to private life/privacy is commonly protected in the legal orders of concern, except for AU. AU has opted for protecting the integrity of a person under Article 4 African Charter on Human and Peoples' Rights. In the legal orders that address the protection of right to private life/privacy, it is included in the human rights instruments of general nature as well as those specifically addressing particular groups (for example, UN UDHR Article 12, UN ICCPR Article 17, UN CRPD Article 22, ASEAN Human Rights Declaration Principle 21, OAS American Declaration of the Rights Article V, the OAS American Convention on Human Rights and Duties of Man Article 11, and CoE ECHR Article 8). Privacy is also protected regarding children under Article 16 CRC, but it does not necessarily mean that children are immediately given a right to consent.

Some human rights instruments address expressly consent requirements and set forth also requirements for the withdrawal of consent (for example, UNESCO International Declaration on Human Genetic Data Article 8 and 9; UNESCO Universal Declaration on the Human Genome and Human Rights Article 5, CoE BMC Article 5, CoE APGT Article 9) and protection of the vulnerable groups (for example, UNESCO International Declaration on Human Genetic Data Article 8, UNESCO Universal Declaration on Bioethics and Human Rights Articles 6 – 8, UNESCO Universal Declaration on the Human Genome and Human Rights Article 5, CoE BMC Articles 6-7, CoE APGT Article 10) and protect autonomy (for example, UNESCO Universal Declaration on Bioethics and Human Rights Article 5), or self-determination (for example, CoE Recommendation (92) 3 Principle 5). Some also emphasize the non-compulsory nature of screening (for example, CoE Recommendation (92) 3 Principle 6).

Some human rights instruments not only address the protection of privacy, but also specifically addresses informed consent (for example, UN CRPD Article 25.d, also UNESCO International Declaration on Human Genetic Data Articles 8 and 9, and 14; a similar approach is taken under the UNESCO Universal Declaration on Bioethics and Human Rights in Articles 6 and 9. However privacy is addressed together with confidentiality and seems directed towards informational privacy, the same can be said about CoE APGT Articles 9 and 16).

Those instruments that address informed consent also pay due regard to the rights and interests of the persons unable to consent (UNESCO International Declaration on Human Genetic Data Article 8, CoE APGT Articles 10-12, CoE Recommendation (92) 3 Principle 5), moreover, CoE APGT Article 19 e emphasizes the voluntary nature of participation in a screening program, and CoE Recommendation (92) 3 Principle 6 specifically addresses the question of non-compulsory nature of the tests, nonetheless permitting exceptions ”stifled by reasons of direct protection of the person concerned or of a third party and be directly related to the specific conditions of the activity”.

As derives from the above, protection of human integrity is granted, whether through general human rights provisions addressing protection of private life or integrity, or specific measures regarding consent requirements in genetic screening. Of those instruments that specifically address genetic screening, *expressis verbis* protection is afforded to persons unable to consent; moreover, voluntariness has been recognized as a principle that shall be observed in genetic screening, even though it is not absolute. Given that all legal orders protect similar rights, one could argue for similar regulatory responses therein.

Counselling

Counselling in genetic screening is crucial in understanding risk information.[186] As this is a rather genetics-specific requirement, it is common that only those human rights instruments that address questions relating to human genome also address counselling (for example, UNESCO International Declaration on Human Genetic Data Article 11 address genetic counselling, and in that regard Article 10 also allows the discretion to decide whether or not to be informed about the results of genetic investigation results; such a discretion is also set forth in UNESCO Universal Declaration on the Human Genome and Human Rights Article 5; CoE BMC Article 12 set forth appropriate counselling as a pre-condition for permissibility of predictive genetic analysis, and CoE APGT Articles 8 and 11 addresses genetic counselling and support; genetic counselling and support are also addressed under CoE Recommendation (92) 3 Principle 3).

Of the CoE instruments that specifically address counselling, particular requirements apply. As derives from Article 8.2 CoE APGT, ”[t]he forms and extent of this genetic counselling shall be defined according to the implications of the results of the test and their significance for the person or the members of his or her family, including possible implications concerning procreation choices.” Moreover, this ”counselling shall be given in a non-directive manner”. Following Article 11, in case a person unable to consent is undergoing genetic testing, information including counselling, should be provided to the person authorizing the intervention as well as to the person concerned with due regard to their capacity to understand the information. CoE Recommendation (92) 3 delineates that appropriate counselling, both before and after the screening procedure should be provided. This counselling as specified under Article 3 ”must be non-directive. The information to be given should include the pertinent medical facts, the results of tests, as well as the consequences and choices. It should explain the purpose and the nature of the tests and point out possible risks. It must be adapted to the circumstances in which individuals and families receive genetic information.” Moreover, should that be necessary, continuing support for the tested person needs to be provided.

Even though counselling is meant to enhance autonomy, as derives from the above, only instruments that specifically address questions relating to human genetics and genomics, include the requirement of counselling.

Dignity

Human dignity is a value commonly recognized in human rights instruments. Often it is included in the preamble of a particular human rights instrument (for example, UN UDHR, UN ICCPR, UN ICESCR, OAS Protocol of San Salvador, OAS American Declaration of the Rights and Duties of Man, OAS Inter-American Convention Against All Forms of Discrimination and Intolerance, AU African Charter on Human and Peoples” Rights, AU Protocol to the African Charter on Human and Peoples' Rights on the Rights of Women in Africa, CoE BMC, CoE APGT), but it is also common to address it in a specific article as a right afforded to individuals or value to protect (UN UDHR Article 1, ASEAN Human Rights Declaration Principle 1, AU African Charter on Human and Peoples” Rights Article 5, AU Protocol to the African Charter on Human and Peoples' Rights on the Rights of Women in Africa Article 3, CoE BMC Article 1).

Some human rights instruments set forth specific requirements and references to dignity in particular context, for example, with respect to living conditions (UN UDHR Article 25), social security (ASEAN Human Rights Declaration Principle 30), with respect to persons being deprived of their liberty (UN ICCPR Article

[186] Genetic counselling: A procedure to explain the possible implications of the findings of genetic testing or screening, its advantages and risks and where applicable to assist the individual in the long-term handling of the consequences; It takes place before and after genetic testing and screening; UN Educational, Scientific and Cultural Organisation (UNESCO), International Declaration on Human Genetic Data, 16 October 2003.

10), in regard to education (UN ICESCR Article 13, ASEAN Human Rights Declaration Principle 31, OAS Protocol of San Salvador Article 13), property (OAS American Declaration of the Rights and Duties of Man Article XXIII), in the context of prohibition of slavery (OAS American Convention on Human Rights Article 6), right to humane treatment (OAS American Convention on Human Rights Article 5), and right to privacy (OAS American Convention on Human Rights Article 11), elderly women (AU Protocol to the African Charter on Human and Peoples' Rights on the Rights of Women in Africa Article 22), women with disabilities (AU Protocol to the African Charter on Human and Peoples' Rights on the Rights of Women in Africa Article 23), women in distress (AU Protocol to the African Charter on Human and Peoples' Rights on the Rights of Women in Africa Article 24).

Some do not address it expressly, but their enforcing bodies have interpreted the instrument as being built on and aspiring to protect human dignity (for example, ECHR[187]). Some *expressis verbis* relate human dignity with discrimination and intolerance based on the human genome, for example, OAS Inter-American Convention against All Forms of Discrimination and Intolerance Article 4.

UNESCO Universal Declaration on Bioethics and Human Rights sets forth a requirement for the protection of human dignity in the area of scientific developments and their application (Article 3), and in a number of rights reference dignity (for example, regarding equality, justice and equity under Article 10; non-discrimination and non-stigmatization under Article 11, and respect for cultural diversity and pluralism under Article 12). UNESCO International Declaration on Human Genetic Data has been specifically adopted with a view to ensuring the respect of human dignity and protection of human rights and fundamental freedoms in relation to human genetic data, human proteomic data and of the biological samples from which they are derived (Article 1). UNESCO Universal Declaration on the Human Genome and Human Rights affirms that everyone has a right to respect for their dignity and for their rights regardless of their genetic characteristics. Moreover, it stipulates that dignity makes it imperative not to reduce individuals to their genetic characteristics and to respect their uniqueness and diversity (Article 2). Furthermore, it expressly relates dignity to certain prohibitions (for example, discrimination under Article 6, and making available benefits of advances in genetic under Article 12). Furthermore, under CoE BMC Article 1 Parties to this Convention shall protect the dignity and identity of all human beings and guarantee everyone, without discrimination, respect for their integrity and other rights and fundamental freedoms with regard to the application of biology and medicine. As derives from CoE APGT Article 1, parties to this protocol shall protect the dignity and identity of all human beings and guarantee everyone, without discrimination, respect for their integrity and other rights and fundamental freedoms with regard to the tests to which this protocol applies in accordance with Article 2. Common to these instruments is stipulation of the limits of permissibility in regard to the application of science to individuals or genetics more specifically.

Reproductive decision-making

Rights that are relevant to reproductive decision-making are commonly addressed in human rights instruments, for example, private life or privacy protection (see above), as well as a right to found a family (for example, UN ICCPR Article 23, ASEAN Human Rights Declaration Article 19, similarly OAS American Declaration of the Rights and Duties of Man Article VI, and OAS American Convention of Human Rights n Article 17, CoE ECHR Article 12), and afford the family a special status or protection (for example, UN ICCPR Article 23 recognizes that the family is the natural and fundamental group unit of society and is

[187] For example, under Article 3 ECtHR has highlighted that 'the prohibition of torture and inhuman or degrading treatment or punishment is a value of civilisation closely bound up with respect for human dignity.' ECtHR, (GC), BOUYID v. BELGIUM, (23380/09), 28 September 2015. para. 81.

entitled to protection by society and the State, AU African Charter on Human and Peoples' Rights Article 18 recognizes family as the natural unit and basis of society). However, it is rather uncommon that questions relating to reproductive choices are expressly addressed. Historically, reproductive choices have been located in the domain of women's rights protection and accordingly are anchored in the respective instruments (for example, UN CEDAW Article 14.2.b and Article 10.h. addresses family planning, similarly AU Protocol to the African Charter on Human and Peoples' Rights on the Rights of Women in Africa Article 14). Moreover, questions that relate to reproduction have also been recognized as part of the right to private life under ECHR.[188]

Neither of human rights instruments points in the direction of limiting reproductive choices relating to the human genome, but also do not prohibit them. UNESCO Universal Declaration on Bioethics and Human Rights Article 12 requires respect for cultural diversity and pluralism. Likewise instruments that prohibit discrimination and stigmatisation and protect dignity could be seen as those supporting choice and diversity. Human rights instruments that are oriented towards tackling disability, for example, UN CRPD, can be used as a means to further diversity of genetic makeup in the context of disability.

Data protection

The question of data protection has differently been handled in different legal orders. Some have addressed it as part of private life, some have addressed data protection explicitly, and some – have adopted specific instruments to tackle the data protection question in regard to medical data banks. Among the legal orders that have addressed data protection in treaties, are AU and the CoE. Whereas among soft law tools are UNESCO, ASEAN, as well as CoE. Here below only considerations that expressly relate to data protection are addressed, since considerations on the normative framework of the right to private life/privacy have already been provided previously. Nonetheless, one could highlight that it is ambiguous to what extent provisions that protect private life/privacy could subsume data protection considerations.

AU Convention on Cyber Security and Personal Data Protection, albeit not in force yet, explicitly mentions genetic data as a form of health data, which are a type of sensitive personal data (Article 1).[189] The Convention stipulates, among others, that processing personal data involving genetic information and health research is one of the actions that require prior authorization by the national protection authority (Article 10.4). Furthermore it provides an exhaustive list of conditions under which sensitive data (including *expressis verbis* data revealing genetic information) may be collected and processed, which includes among others, situations when "processing, particularly of genetic data, is required for the establishment, exercise or defense of legal claims", where "processing is necessary in the public interest, especially for historical, statistical or scientific purposes" or when the data subject gave his or her appropriate consent (Article 14). CoE (consolidated)[190] Data Protection Convention in Article 6 addresses genetic data as a special category of data and permits processing them "only be allowed where appropriate safeguards are enshrined in law, complementing those of this Convention". As noted in Article 6.2, "such safeguards shall guard against the risks that the processing of sensitive data may present for the interests, rights and fundamental freedoms of the data subject, notably a risk of discrimination".

[188] For example, ECtHR, VC v. Slovakia (18968/07), 8 November 2011.

[189] AU Convention on Cyber Security and Personal Data Protection, EX.CL/846(XXV), 27 June 2014.

[190] CoE, Convention for the Protection of Individuals with regard to Automatic Processing of Personal Data, ETS 108. Consolidated text of the Convention as it will be amended by the Protocol CETS No. 223 upon its entry into force.

Under the UNESCO International Declaration on Human Genetic Data, genetic data have been given a special status (Article 4). Genetic screening, defined as ”[l]arge-scale systematic genetic testing offered in a programme to a population or subsection thereof intended to detect genetic characteristics in asymptomatic people” is among purposes for which genetic data can be processed (Article 5(i)).

ASEAN Framework on Personal Data Protection sets forth principles for data protection, including conditions under which data can be processed, but does not set forth any special requirements for health or genetic data or genetic screening. CoE Recommendation No. R (92) 3 Principle 8 requires that the ”collection and storage of substances and of samples, and the processing of information derived therefrom, must be in conformity with the CoE Data Protection Convention and the relevant Recommendations of the Committee of Ministers in this field”; in so far as medical data are being processed, CoE Recommendation on the Protection of Medical Data is of particular relevance.[191]

A question to consider is whether the secondary use of the samples/data is permitted. UNESCO’s International Declaration on Human Genetic Data sets forth two alternative routes: the change of purpose, as well as the work with stored biological samples. In regard to the former, it requires that human genetic data, human proteomic data and the biological samples that are collected for a specific purpose, ”should not be used for a different purpose that is incompatible with the original consent, unless the prior, free, informed and express consent of the person concerned is obtained”. Whereas, Article 17 of the same declaration sets forth conditions for using stored biological samples for producing human genetic data or human proteomic data. Under the article, that is permissible ”with the prior, free, informed and express consent of the person concerned”, however, if matter concerns data that ”have significance for medical and scientific research purposes, e.g. epidemiological studies or public health purposes”, provided it is in accordance with national law and an ”independent, multidisciplinary and pluralist ethics committee” has been consulted. This means that under the UNESCO International Declaration on Human Genetic Data, it should only be permissible to further use the samples and generate data for research purposes, but not other purposes. Such an approach could be related to trust in scientific research and furthering scientific in order benefits from scientific advances can be gained.

Regionally, the ASEAN Framework on Personal Data Protection under Principle 6 does not preclude using samples/data for other purposes but requires that in such a case it is done in accordance with national law, or it is done in so far as a reasonable person would consider appropriate in the circumstances. CoE Recommendation No. R (92) 3 Principle 13 a emphasizes that ”[s]amples collected for a specific medical or scientific purpose may not, without permission of the persons concerned or the persons legally entitled to permit on their behalf, be used in ways which could be harmful to the persons concerned.” The CoE (consolidated) Data Protection Convention under Article 5.4.b envisages further processing of data which is compatible with the initial purposes the data and samples were collected for, if it is done in ”for archiving purposes in the public interest, scientific or historical research purposes or statistical purposes is, subject to appropriate safeguards, compatible with those purposes”. Therefore, purposes for which further processing is limited and does not include, for example, using the data in criminal justice systems. A similar approach to permitting further processing of data for historical, statistical or research purposes is also outlined in AU Convention on Cyber Security and Personal Data Protection Article 13, Principle 3 d.

[191] CoE, Committee of Ministers, Recommendation No. R (97) 5 on the Protection of Medical Data. Although work towards revising this recommendation has been conducted for some time, information about finalizing the recommendation is not available yet.

As derives from the above, data protection is of a concern to the surveyed legal orders, albeit as of now, addressed in significantly different ways. While the CoE has addressed the question under hard and soft laws, AU has opted for a hard law measure, which is not in force yet, whereas ASEAN has opted for a soft law measure. OAS has not taken specific steps in data protection, but the Article 13 of the American Convention on Human Rights, that protects the right to seek and receive information, has been understood to include the right to have access "to information about him or herself contained in public or private databases and to modify, remove or correct such information due to its sensitive, erroneous, biased, or discriminatory nature"[192] ("right to access to and control over personal information", known as the "*habeas data* writ"[193]). Of the surveyed regulatory responses, it is clear that data/samples collected for a screening purpose should be permitted for limited further use activities. While they include research, criminal justice or other purposes are not covered. Such an approach seems to go hand in hand with the health data protection purpose acknowledged by the CoE ECtHR that it is crucial for trust in the public health system.[194]

4.2.3 Genetic Testing

Information about the results of the test, counselling,[195] "right to know and not to know", incidental findings and right to private life/privacy

Right to information is a commonly shared right in the human rights catalogues of concern (for example, UN UDHR Article 19, UN ICCPR Article 19, ASEAN Human Rights Declaration Principle 23, AU African Charter on Human and Peoples" Rights Article 9, CoE ECHR Article 10, OAS American Declaration of the Rights and Duties of Man Article XXI, OAS American Convention on Human Rights Article 13). Although this protection is focused on freedom of thought and expression, Committee has pointed at the close link between UN ICCPR Article 17 and UN ICCPR Article 19 and that Article 19.2 includes protection of information held by public bodies, whereby these bodies are "[a]ll branches of the State (executive, legislative and judicial) and other public or governmental authorities, at whatever level – national, regional or local – are in a position to engage the responsibility of the State party."[196] Likewise, information is strongly related to decision-making and self-determination, normative background of which has already been reviewed previously. Following the general framework of protection, it could be regarded that information is not only crucial in obtaining consent, but also for furthering health of the person concerned. Therefore, receiving of the results could be argued to be falling within the scope of the protected rights.

The normative background of genetic counselling has already been highlighted under question 2. Here suffices to recall that it is common that only those human rights instruments that address questions relating to the human genome also address counselling. The exact requirements could differ, but the need for support as well as the importance of choice to know or not to know has commonly been emphasized. One can also note that, for example, UNESCO International Declaration on Human Genetic Data Article 11

[192] Inter-American Commission on Human Rights, Background and Interpretation of Declaration of Principles on Freedom of Expression, 19 October 2000.

[193] Ibid.

[194] ECtHR, I v. Finland (20511/03), 17 July 2008, ECtHR, (GC). ECtHR Z v. Finland, (22009/93), 25 February 1997.

[195] UN Educational, Scientific and Cultural Organisation (UNESCO), International Declaration on Human Genetic Data, Article 11 addresses genetic counselling and states that '[i]t is ethically imperative that when genetic testing that may have significant implications for a person's health is being considered, genetic counselling should be made available in an appropriate manner. Genetic counselling should be non-directive, culturally adapted and consistent with the best interest of the person concerned.

[196] Human Rights Committee, General comment No. 34, Article 19: Freedoms of opinion and expression, CCPR/C/GC/34, para. 7 in correlation with para. 18.

states that ”when genetic testing that may have significant implications for a person’s health is being considered, genetic counselling should be made available in an appropriate manner.” Moreover, this counselling ”should be non-directive, culturally adapted and consistent with the best interest of the person concerned.” It is, however, unaddressed what health-related genetic testing is such that has significant implications for a person’s health, and what genetic testing does not have significant implications; moreover, the subjective consideration over importance remains relevant.

Of the instruments that address the information about genetic testing results are UNESCO declarations and Council of Europe instruments. UNESCO Universal Declaration on the Human Genome and Human Rights Article 5(c) and UNESCO International Declaration on Human Genetic Data Article 10 addresses the right to decide whether to be informed about research results. From UNESCO International Declaration on Human Genetic Data Article 10 follows that in case of medical genetic testing ”the information provided at the time of consent should indicate that the person concerned has the right to decide whether or not to be informed of the results.” Therefore, although the declaration does not *expressis verbis* regulate the handling of incidental findings; it requires the choice to be left to the persons being tested.

Under UNESCO Universal Declaration on the Human Genome and Human Rights Article 5c and UNESCO International Declaration on Human Genetic Data Article 10, the right to decide whether or not to be informed about research results remains with the person being tested. Under Article 10 of the UNESCO International Declaration on Human Genetic Data, it is indicated that this information should be provided at the time of consent.

CoE BMC Article 10 sets forth protection for private life and the right to information. It grants to everyone ”the right to respect for private life in relation to information about his or her health”, and notes that ”[e]veryone is entitled to know any information collected about his or her health. However, the wishes of individuals not to be so informed shall be observed.” Only in exceptional cases, the right to know and not to know may be restricted by law in the interests of the patient concerned. Similar protection, albeit focused on genetic testing. Moreover, it adds in Article 16.2 that ”[t]he conclusions drawn from the test shall be accessible to the person concerned in a comprehensible form.”

It is unclear to what extent, for example, right not to now could be accommodated under the general human rights provisions as, on the one hand, they relate to a choice of the individual, on the other hand, there could be reasons, such as the individual”s health or someone else”s health as to why this right should be restricted.

Family interests/ rights

The protection of family interests/rights in knowing or not knowing about their risk of a genetic condition due to another relative’s test, is a question that has been expressly addressed in some human rights legal orders or at least express pointers are given at solving a conflict of rights, and remains unregulated in the others.

Among the legal orders that have expressly addressed the question or given pointers, are UNESCO and CoE. UNESCO International Declaration on Human Genetic Data in Article 10 indicates that the right to decide whether or not to be informed about also extends to the identified relatives who may be affected by the results of genetic testing. However, it does not elaborate how the delivery of this information should be organized in order to safeguard the right to know/not to know, and simultaneously safeguard

the rights and interests of the person being tested.[197] Although it is not expressly addressed in UNESCO Universal Declaration on the Human Genome and Human Rights, Article 5 c is worded in a rather neutral way stating that, ”[t]he right of each individual to decide whether or not to be informed of the results of genetic examination and the resulting consequences should be respected.” It could be interpreted broadly to apply also to persons being concerned by testing.

CoE BMC does not expressly address the rights of the family members in genetic testing. Nonetheless, from Article 26 follows that rights set forth in Article 10 can be restricted. Therefore, it would be for the national legislature to balance between the rights of the person’s tested and their family members.[198] CoE APGT Article 18, however, addresses information relevant to family members, and states ”where the results of a genetic test undertaken on a person can be relevant to the health of other family members, the person tested shall be informed.”

In other legal orders, the question relates to balancing the privacy of the person being tested, ensuring data protection and, depending on circumstances, confidentiality owed to that person, against potential benefits to the relative and public health benefits in that regard.

Data protection

The normative framework for data protection has already been introduced in the previous sections. Here suffices to further explore how data protection should be reconciled with the rights and interests of family members in obtaining genetic information. ASEAN Framework on Personal Data Protection Principle 6.b permits disclosing personal data about an individual only for purposes that a reasonable person would consider appropriate in the circumstances. AU Convention on Cyber Security and Personal Data Protection does not provide particular guidance in that regard. CoE (consolidated) Data Protection Convention under Articles 5 and 6 permits processing genetic data, providing legitimacy requirements are met and safeguards that guard against the risks that the processing of genetic data may present for the interests, rights and fundamental freedoms of the data subject, notably a risk of discrimination are safeguarded. One can, therefore, conclude that the right to data protection as regulated in these instruments, should not be seen as absolute, and measures that foresee processing could be in place, provided protections are warranted.

Confidentiality

[197] UNESCO, International Declaration on Human Genetic Data, Article 14 (a) requires that “States should endeavour to protect the privacy of individuals and the confidentiality of human genetic data linked to an identifiable person, family or, where appropriate, group, in accordance with domestic law consistent with the international law of human rights.” However, section b of the same article enables disclosing genetic information to the family “for an important public interest reason in cases restrictively provided for by domestic law consistent with the international law of human rights or where the prior, free, informed and express consent of the person concerned has been obtained provided that such consent is in accordance with domestic law and the international law of human rights.”

[198] Nonetheless in paragraph70 of the Explanatory report to the convention it is noted that ’[a]t the same time, certain facts concerning the health of a person who has expressed a wish not to be told about them may be of special interest to a third party, as in the case of a disease or a particular condition transmissible to others, for example. In such a case, the possibility for prevention of the risk to the third party might, on the basis of Article 26, warrant his or her right taking precedence over the patient's right to privacy, as laid down in paragraph 1, and as a result the right not to know, as laid down in paragraph 2. In any case, the right not to know of the person concerned may be opposed to the interest to be informed of another person and the interests of these two persons should be balanced by internal law.”

Confidentiality is, on the one hand, strongly related to the protection of private life/privacy and integrity of the patient as a data subject. On the other hand, it is also common that confidentiality, further to being an ethical requirement is also enshrined in hard and soft laws. UNESCO Universal Declaration on Bioethics and Human Rights Article 9 states that ”[t]he privacy of the persons concerned and the confidentiality of their personal information should be respected. To the greatest extent possible, such information should not be used or disclosed for purposes other than those for which it was collected or consented to, consistent with international law, in particular international human rights law.” UNESCO International Declaration on Human Genetic Data Article 14 (a) requires that ”[s]tates should endeavour to protect the privacy of individuals and the confidentiality of human genetic data linked to an identifiable person, family or, where appropriate, group, in accordance with domestic law consistent with the international law of human rights.” However, section b of the same article enables disclosing genetic information to the family ”for an important public interest reason in cases restrictively provided for by domestic law consistent with the international law of human rights or where the prior, free, informed and express consent of the person concerned has been obtained provided that such consent is in accordance with domestic law and the international law of human rights”.

While ASEAN Framework on Personal Data Protection addresses confidentiality under Article 11, it has a general nature and not medical one; it permits disclosing confidential information if authorized in writing. CoE Recommendation No. R (97)5 on the protection of medical data Article 3 requires confidentiality, nonetheless Article 4.3.b permits processing of medical data ”i. for preventive medical purposes or for diagnostic or for therapeutic purposes with regard to the data subject or a relative in the genetic line; or ii. to safeguard the vital interests of the data subject or of a third person”.[199]

Intellectual Property rights, Right to science

The normative framework of the right to science has already been introduced under question 1. Here suffices to reiterate that right to science as protected under UN UDHR Article 27 and UN ICESCR Article 15 has two dimensions – that relating to enjoying the scientific advances, and that relating to the protection of the moral and material interests resulting from the advance a person is an author of. Moreover, UN UDHR Article 17 protects the right to property. While CoE ECHR does not *expressis verbis* address the right to science, in its Protocol 1 Article 1, it protects a right to property, which as demonstrated by extensive case law can be subjected to considerable limitations. Furthermore, CoE BMC Article 15 addresses freedom of science and the protection to the persons concerned. Right to property is also addressed in, for example, ASEAN Human Rights Declaration Principle 17, AU African Charter on Human and Peoples” Rights Article 14, OAS American Declaration of the Rights and Duties of Man Article XXIII, and OAS American Convention on Human Rights Article 21.

UNESCO Universal Declaration on the Human Genome and Human Rights Article 12 does not expressly address the rights of the authors, but requires that ”[b]enefits from advances in biology, genetics, and medicine, concerning the human genome, shall be made available to all, with due regard for the dignity and human rights of each.”[200] UNESCO International Declaration on Human Genetic Data Article 19 addresses the sharing of benefits ”with the society as a whole and the international community”, and

[199] Council fo Europe, Committee of Ministers, of The Committee of Ministers to Member States on the Protection of Medical Data, Recommendation No. R (97) 5.

[200] (b) Freedom of research, which is necessary for the progress of knowledge, is part of freedom of thought. The applications of research, including applications in biology, genetics and medicine, concerning the human genome, shall seek to offer relief from suffering and improve the health of individuals and humankind as a whole.

permits restrictions to this obligation. UNESCO Universal Declaration on Bioethics and Human Rights under Article 15 enshrines a similar requirement but does not note permission for restrictions.

Neither of these instruments points exactly at how human rights respond to patenting in the area of genomics, and therefore, this question, from the perspective of human rights law remains subject of interpretation. In light of permissibility of patenting, one can question the meaning of and the scope of sharing benefits from science.

Genetic discrimination

Normative framework relevant to genetic discrimination has already been introduced previously. Here suffices to note that it indistinctly applies to genetic testing.[201]

Secondary use of genetic information, data, and samples, genetic testing for non-medical purposes

There is limited regulation for using genetic data/samples for other purposes than research (discussed under question 2). Unless specifically addressed, this question is to be resolved with due regard to the protection of privacy/personal data of the data subject, and competing rights and interests for which the data/samples/information can be used. UNESCO International Declaration on Human Genetic Data Article 14 (b) addresses the question expressly and states that "[h]uman genetic data, human proteomic data and biological samples linked to an identifiable person should not be disclosed or made accessible to third parties, in particular, employers, insurance companies, educational institutions and the family, except for an important public interest reason in cases restrictively provided for by domestic law consistent with the international law of human rights or where the prior, free, informed and express consent of the person concerned has been obtained provided that such consent is in accordance with domestic law and the international law of human rights. The privacy of an individual participating in a study using human genetic data, human proteomic data or biological samples should be protected and the data should be treated as confidential." Nonetheless, generally, UNESCO declarations permit the collection and processing of genetic data for, for example, court proceedings.

In other legal orders, for example, the Council of Europe, the question shall be approached from permissibility of genetic testing as such. CoE BMC under Article 12 permits predictive genetic testing for limited purposes and does not include, for example, insurance. This approach is also upheld under CoE Recommendation CM/Rec(2016)8 Principle 4, which, whilst does not permit genetic testing with a view to obtain information for insurance purposes (Paragraph 15), enables the use of "[e]existing predictive data resulting from genetic tests" (Paragraph 16) if so authorized by national law and following assessment of the following criteria (Paragraph 5) with regard to the type of test and risk for the person concerned:

- the processing purpose has been specified and the relevance of the data has been duly justified;
- the quality and validity of the data are in accordance with generally accepted scientific and clinical standards;
- data resulting from a predictive examination have a high positive predictive value; and
- processing is duly justified in accordance with the principle of proportionality in relation to the nature and importance of the risk in question."

[201] UN Educational, Scientific and Cultural Organisation (UNESCO), International Declaration on Human Genetic Data, Article 3 states that '[e]ach individual has a characteristic genetic make-up. Nevertheless, a person's identity should not be reduced to genetic characteristics, since it involves complex educational, environmental and personal factors and emotional, social, spiritual and cultural bonds with others and implies a dimension of freedom.'

It prohibits under Paragraph 17 using ”[e]xisting data from genetic tests from family members of the insured person (..) for insurance purposes.”

As derives from the above, it is not necessarily precluded to use genetic testing for another purpose than health, as well as to use genetic testing results/data/samples for other purposes. While, in light of what has been discussed in question 2, research is seen as a generally permissible purpose for further processing of the samples/data/information relating to genetic testing, further processing for other purposes can be seen as rather problematic. This could be related to public trust in scientific research. However, it is not precluded that genetic testing for other purposes can be used.

Right to the highest attainable standard of health for minors and consent

Further to the right to the highest attainable standard of health protection enshrined in human rights instruments outlined previously, UN CRC Article 24 specifically addresses the question of health for minors. Moreover, while it acknowledges the right to privacy under Article 16, it does not regulate the matter of consent.

ASEAN Human Rights Declaration recognises the rights of children (Principle 4). Furthermore, the Declaration imposes a duty on ASEAN Member States to provide in general special care for childhood (Principle 30.3) (the ASEAN Member States however are obliged to achieve these and other economic and social rights progressively, by taking steps to the maximum of their available resources – Principle 33). In AU, the African Youth Charter Article 16.2 obliges State Parties to take measures to e.g. make available equitable and ready access to medical assistance and health care, especially in rural and poor urban areas and institute programmes to address health pandemics in Africa.[202] In CoE, children as a specific group are addressed under the CoE (revised) European Social Charter Articles 7 and 17, requiring *inter alia* to ensure adequate social protection. OAS Additional Protocol to the American Convention on Human Rights in the Area of Economic, Social and Cultural Rights ("Protocol Of San Salvador") includes right to health (Article 10.1), which is related to such State Parties” obligations as “prevention and treatment of endemic, occupational and other diseases“ (Article 10.2.d) and “satisfaction of the health needs of the highest risk groups and of those whose poverty makes them the most vulnerable” (Article 10.2.f). It also embraces the right of every child to the protection that his status as a minor requires from his family, society and the State (Article 16). In all legal orders, children are beneficiaries of general human rights provisions that shape their rights and protections. However, neither these instruments nor the general human rights provisions provide concrete guidance in safeguarding the rights of children in genetic testing.

Genetic analysis on minors is expressly regulated in human rights instruments that specifically address genetics related questions. For example, UNESCO International Declaration on Human Genetic Data Article 8(b) “when, in accordance with domestic law, a (..) [minor] is incapable of giving informed consent, authorization should be obtained from the legal representative, in accordance with domestic law. The legal representative should have regard to the best interest of the person concerned.” It requires under section c of the same article that ”[t]he opinion of a minor should be taken into consideration as an increasingly determining factor in proportion to age and degree of maturity.” However, following section d of the same article, ”[i]n diagnosis and health care, genetic screening and testing of minors (..) not able to consent will normally only be ethically acceptable when they have important implications for the health of the person and have regard to his or her best interest.” UNESCO Universal Declaration on Bioethics and Human Rights enshrines more general protection of persons without the capacity to consent under Article 7, permitting interventions if authorization is provided ”in accordance with the best interest of the person

[202] AU, African Youth Charter, 2 July 2006.

concerned and in accordance with domestic law". Moreover, it requires that "the person concerned should be involved to the greatest extent possible in the decision-making process of consent, as well as that of withdrawing consent." Moreover, more generally respect for human vulnerability and personal integrity should be ensured.

CoE BMC Article 6 protects persons not able to consent and requires that "[w]here, according to law, a minor does not have the capacity to consent to an intervention, the intervention may only be carried out with the authorisation of his or her representative or an authority or a person or body provided for by law. The opinion of the minor shall be taken into consideration as an increasingly determining factor in proportion to his or her age and degree of maturity." Moreover, this intervention shall be carried out if for a direct benefit of the child concerned. This authorization may be withdrawn at any time in the best interests of the child concerned. CoE APGT Article 10 addresses protection of persons not able to consent. With regards to minors it requires that "[w]here, according to law, a minor does not have the capacity to consent, a genetic test on this person shall be deferred until attainment of such capacity unless that delay would be detrimental to his or her health or well-being."

As derives from above, the general human rights instruments protect the rights of minors but leave specific questions a balancing act between the competing rights and interests. Those instruments specifically aimed at addressing questions relating to genomics take a rather restrictive approach over genetic analysis on minors incapable of consent.

4.2.4 Prenatal Testing

Availability of prenatal screening/testing, and information and decision-making in that regard, right to know and not to know

The question of availability of genetic screening/testing anchorsin the highest attainable standard of health of the pregnant woman as well as the foetus, can also be seen as relating to a right to enjoy benefits form the scientific advances specifically addressing the pregnant woman's reproductive care, and thus, relating to such rights as equality and non-discrimination. Substantively, the issues of prenatal testing/screening not only raise questions about health entitlements and measures to further health of the pregnant woman, as well as health of the foetus (at the pre-birth stage) but relating to the termination of pregnancy. Therefore, questions on balancing the health protection and self-determination of the pregnant woman and her integrity vis-à-vis interests in protecting the foetus emerges. This therefore excludes considerations regarding situations where a pregnant woman's health necessitates termination of pregnancy.

Normative background of the right to the highest attainable standard of health and of equality and non-discrimination protection has been reviewed previously. Here suffices to note that prenatal care is directly related to the obligations stemming from UN ICESCR Article 12.2(a), which focuses on the provision for the reduction of the stillbirth-rate and of infant mortality, and can also directly be anchored in the pregnant woman's protected rights under Article 12.1. Therefore, in the context of reproductive rights, availability and accessibility of such a screening relate to the woman's right to have control over and to decide freely on matters related to their sexuality, including reproductive health.[203] As to the obligations

[203] UN Member States at the United Nations Fourth World Conference of Women in Beijing in 1995 reaffirm the Cairo Programme's definition of reproductive health: The human rights of women include their right to have control over and decide freely on matters related to their sexuality, including sexual and reproductive health, free of coercion, discrimination and violence. See Toebes, Brigit, "Sex Selection under International Human Rights Law", *Medical Law International,* Vol. 9, 2008, p. 197. See also UN Committee on the Elimination of Discrimination Against

related to prenatal care in regional orders, the Protocol to the African Charter on Human and Peoples' Rights on the Rights of Women in Africa specifies the State Parties are obliged by the Protocol to take all appropriate measures to establish and strengthen existing pre-natal health services for women during pregnancy (Article 14.2.b) In a similar tone, the African Youth Charter obliges State Parties to take measures to provide access to youth-friendly reproductive health services including antenatal services (article 16.2), whereas the African Charter on the Rights and Welfare of the Child obliges the State Parties to take measures to ensure appropriate health care for expectant mothers (Article 14.2.5). Furthermore, the ASEAN human rights declaration imposes a duty on ASEAN Member States to accord special protection to mothers before childbirth (Article 30.2), while the OAS Additional Protocol to the American Convention on Human Rights in the Area Of Economic, Social And Cultural Rights ("Protocol Of San Salvador") imposes upon a state obligation to provide special care and assistance to mothers during a reasonable period before childbirth (Article 15.3.a).

More directly, of the regional legal orders of concern, only CoE has adopted a measure, CoE Recommendation No. R (90) 13, which contains recommendations regarding practices for prenatal screening. This recommendation does not expressly address whether or not such a screening should be offered, but sets forth recommendations regarding situations when genetic testing/screening is being offered.

Although WHO has adopted recommendations on antenatal care for a positive pregnancy experience, it has noted that "[s]pecific genetic tests for detection of inherited conditions were considered beyond the scope of this guideline."[204] UNESCO Report of the IBC on Updating its Reflection on the Human Genome and Human Rights, has noted that NIPT is being used as a second screening test to women already diagnosed with elevated risk due to a positive combined test. This has as a result that invasive genetic tests follow only if an unfavourable positive NIPT result occurs and consequently leads to a decrease of the number of the invasive genetic test.[205] Therefore, it has pointed out that it is "important to develop a framework that on the one hand acknowledges the right of an individual to make autonomous choices, and on the other hand ensures what is enshrined in articles 6 and 2 of the UDHGHR: that no one shall be subjected to discrimination based on genetic characteristics and that individuals should be respected in their uniqueness and diversity."[206] Further recommendations on balancing these competing rights and interests have not been given.

Normative basis for safeguarding the right to information and decision-making regarding genetic testing has already been reviewed previously. Likewise, considerations over the right to know and not to know have been made. While prenatal testing/screening can be seen as an intervention that requires to the previously considered requirements for genetic testing/screening, also within the CoE specific requirements applicable to prenatal testing/screening have been adopted. It recommends *inter alia* that counselling is provided prior and after the intervention (Principle 1), which shall be non-directive (Principle 4), and encouraged for both partners as prospective parents (Principle 4). It is undertaken for the purpose

Women (CEDAW), CEDAW General Recommendation No. 24: Article 12 of the Convention (Women and Health), 1999, A/54/38/Rev.1, chap. I.

[204] WHO, "WHO recommendations on antenatal care for a positive pregnancy experience". https://www.who.int/reproductivehealth/publications/maternal_perinatal_health/anc-positive-pregnancy-experience/en/ para. 5.

[205] IBC, Report of the IBC on updating its reflection on the Human Genome and Human Rights, SHS/YES/IBC-22/15/2 REV.2 para. 83-93.

[206] IBC, op.cit., para. 93.

of identifying risk to the health of an unborn child should be aimed only at detecting a serious risk to the health of the child (Principle 2), following informed consent and, if relevant, specifically protecting the incapacitated undergoing genetic testing/screening (Principle 6). In order to protect the woman's freedom of choice, she should not be compelled by the requirements of national law or administrative practice to accept or refuse screening or diagnosis. In particular, any entitlement to medical insurance or social allowance should not be dependent on undergoing these tests (Principle 9).

Termination of pregnancy, including sex selection and disability considerations

Termination of pregnancy is a question that is rarely *expressis verbis* regulated in the hard law human rights instruments. Nonetheless, a number of rights commonly protected in the human rights frameworks have been used to address the question of abortion and shape the regulatory responses at the national level.

At its core, regulation of abortion relates to the limits of self-determination and protection of integrity against the state's interest in protecting the foetus. As follows from General Comment No. 36 on article 6 of the UN ICCPR on the right to life that has been adopted in 2018, "[a]lthough States parties may adopt measures designed to regulate voluntary terminations of pregnancy, such measures must not result in violation of the right to life of a pregnant woman or girl, or her other rights under the Covenant. Thus, restrictions on the ability of women or girls to seek an abortion must not, *inter alia*, jeopardize their lives, subject them to physical or mental pain or suffering which violates article 7 [ICCPR, torture or to cruel, inhuman or degrading treatment], discriminate against them or arbitrarily interfere with their privacy. States parties must provide safe, legal and effective access to abortion where the life and health of the pregnant woman or girl is at risk, or where carrying a pregnancy to term would cause the pregnant woman or girl substantial pain or suffering, most notably where the pregnancy is the result of rape or incest or is not viable. (..) In addition, States parties may not regulate pregnancy or abortion in all other cases in a manner that runs contrary to their duty to ensure that women and girls do not have to undertake unsafe abortions, and they should revise their abortion laws accordingly."[207]

Under UN CEDAW, the Committee on the Elimination of Discrimination against Women has noted that "it is discriminatory for a State party to refuse to legally provide for the performance of certain reproductive health services for women."[208] It has also stated that "laws that criminalize medical procedures only needed by women and that punish women who undergo those procedures" are a barrier to women's access to health care,[209] and that punitive measures for women who undergo an abortion should be removed.[210] Although the exact approach remains to be taken by individual states, from these UN instruments and their interpretation can be derived that abortion shall be ensured at the circumstances when the woman's life is at risk, the continuation of pregnancy risks amounting to severe suffering. Following the mentioned, at the extreme, states are free to regulate abortion, including limiting it, constraining rights stemming from the pregnant woman's right to privacy. However, in such a case privacy would include situations where the woman's health is at risk or carrying a pregnancy to term would mean

[207] UN Human Rights Commitee, General comment No. 36 (2018) on article 6 of the International Covenant on Civil and Political Rights, on the right to life, CCPR/C/GC/36, para. 8.

[208] UN Committee on the Elimination of Discrimination Against Women (CEDAW), CEDAW General Recommendation No. 24: Article 12 of the Convention (Women and Health), 1999, A/54/38/Rev.1, chap. I, para. 11.

[209] UN Committee on the Elimination of Discrimination Against Women (CEDAW), CEDAW General Recommendation No. 24: Article 12 of the Convention (Women and Health), 1999, A/54/38/Rev.1, chap. I, para. 14.

[210] Concluding Observations on Peru, CEDAW/C/PER/CO/7-8 (2014), para. 36. Statement on sexual and reproductive health and rights: Beyond 2014 ICPD Review (2014).

substantial pain or suffering to the woman concerned, situations where pregnancy is a result of rape or incest. Moreover, as pregnancy or abortion should not be regulated in a way that runs contrary to the duty to ensure access to safe abortions, it could be argued that states are obliged to adopt mechanisms to enable safe abortions even when the above-outlined risks do not emerge, but a pregnant woman has chosen not to continue carrying pregnancy to the term. This means that the regulatory discretion remaining with the states should be located in defining circumstances for access to abortion.

Regionally, in AU Protocol to the African Charter on Human and Peoples' Rights on the Rights of Women in Africa Article 14.2 *expressis verbis* requires that States Parties shall take all appropriate measures to ”protect the reproductive rights of women by authorising medical abortion in cases of sexual assault, rape, incest, and where the continued pregnancy endangers the mental and physical health of the mother or the life of the mother or the foetus.” It does not address abortion for purposes beyond the listed ones. ASEAN Human Rights Declaration protects *inter alia* reproductive health under Principle 29, as well as privacy under Principle 21, whereas the right to life under Principle 11 and prohibition of torture or cruel, inhuman or degrading treatment under Principle 14, however, how these principles and rights are balanced regarding abortion remains undiscussed here.

OAS American Convention on Human Rights protects the right to life (”in general, from the moment of conception” – article 4). However, it has been acknowledged ”that the protection of the right to life under this provision is not absolute, but rather gradual and incremental according to its development, since it is not an absolute and unconditional obligation, but entails understanding that exceptions to the general rule are admissible”.[211] Furthermore, the OAS Committee of Experts of the Follow-up Mechanism to the Inter-American Convention on the Prevention, Punishment and Eradication of Violence against Women, adopted a Declaration on Violence against Women, Girls and Adolescents and their Sexual and Reproductive Rights, that stated ”there are still laws that perpetuate the exercise of violence against women, girls, and adolescents, that re-victimize them by violating their sexual and reproductive rights, and that violate the prohibition of torture and mistreatment, such as: maintaining restrictions on access to safe abortions and absolute prohibitions of abortions, or the denial of access to post-abortion care that contravenes de prohibition of torture and ill-treatment”.[212]

In CoE, abortion is not regulated under the ECHR. However, some rights protected under the ECHR have been relied upon regarding questions that concern abortion. In the case of A, B and C v Ireland the ECtHR recalled that ”interruption of pregnancy touches upon the sphere of the private life of the woman”, Therefore, ”[t]he woman’s right to respect for her private life must be weighed against other competing rights and freedoms invoked including those of the unborn child”.[213] The ECtHR found that the prohibition of the termination of pregnancies sought for reasons of health and/or well-being amounted to an

[211] See the Inter-American Court of Human Rights judgement of 28 November 2012 in the case of Artavia Murillo et. al. (“*In Vitro* Fertilization”) v Costa Rica, para. 264.

[212] Committee of Experts of the Follow-up Mechanism to the Inter-American Convention on the Prevention, Punishment and Eradication of Violence against Women, Declaration on Violence against Women, Girls and Adolescents and their Sexual and Reproductive Rights, September 18th and 19th 2014 Montevideo, Uruguay OEA/Ser.L/II.7.10 MESECVI/CEVI/DEC.4/14.

[213] ECtHR (GC), A., B. and C. v. Ireland, (25579/05), 16 December 2010, para. 214. “While Article 8 cannot, accordingly, be interpreted as conferring a right to abortion, the Court finds that the prohibition in Ireland of abortion where sought for reasons of health and/or well-being about which the first and second applicants complained, and the third applicant’s alleged inability to establish her eligibility for a lawful abortion in Ireland, come within the scope of their right to respect for their private lives and accordingly Article 8.”

interference with their right to respect for their private lives[214] and has assessed whether it can be justified, namely, whether the interference is "in accordance with the law" and "necessary in a democratic society" for one of the "legitimate aims" specified in Article 8 of the Convention.[215] It noted that "[b]y reason of their "direct and continuous contact with the vital forces of their countries", State authorities are in principle in a better position than the international judge to give an opinion on the "exact content of the requirements of morals [of the right to life of the unborn was one aspect]"[216] in their country, as well as on the necessity of a restriction intended to meet them".[217] The ECtHR noted that in assessing whether a fair balance between compeating rights and interests has been struck, the margin of appreciation is of particular importance.[218] Even though abortion for health-related reasons could, at the time of judgement, be accessed in most ECHR Contracting States, due "to the acute sensitivity of the moral and ethical issues raised by the question of abortion or as to the importance of the public interest at stake",[219] Ireland was afforded broad margin of appreciation. Therefore, "having regard to the right to travel abroad lawfully for an abortion with access to appropriate information and medical care in Ireland (..) the Court finds that the impugned prohibition in Ireland struck a fair balance between the right of the first and second applicants to respect for their private lives and the rights invoked on behalf of the unborn".[220] As derives from this case regarding two of the three applicants the case related to, which is adjudicated well-before the adoption of General Comment No. 36 on Article 6 of the UN ICCPR on the right to life, within the limits of privacy protection states enjoy a considerable margin of appreciation. When the continuation of pregnancy amounts to interventions with rights protected under Article 3 (prohibition of torture, inhumane or degrading treatment) ECHR or Article 2 (right to life) ECHR, a different assessment applies.

The normative framework on disability considerations in terms of non-discrimination and equality have already been reviewed in the previous sections. Here suffices to note that regarding the UN CRPD and UN CEDAW, in 2018 United Nations Committee on the Elimination of Discrimination against Women and the Committee on the Rights of Persons with Disabilities issues a joint statement regarding abortion and disability. It has been noted that "access to safe and legal abortion, as well as related services and information, are essential aspects of women's reproductive health. Access to such services is a prerequisite for safeguarding women's human rights to life, health, equality before the law and equal protection of the law, non-discrimination, information, privacy, bodily integrity and freedom from torture and ill treatment (..)."[221] Chairperson of the Committee on the Rights of Persons with Disabilities stated that she is "very concerned that opponents of reproductive rights and autonomy often actively and deliberately refer to disability rights in an effort to restrict or prohibit women's access to safe abortion

[214] ECtHR, (GC), A., B. and C. v. Ireland, (25579/05), 16 December 2010, para. 216.
[215] A., B. and C. v. Ireland, op.cit., para. 217.
[216] A., B. and C. v. Ireland, op.cit., para. 227.
[217] A., B. and C. v. Ireland, op.cit., para. 223.
[218] "Accordingly, and as underlined at para.raph 213 above, in the present cases the Court must examine whether the prohibition of abortion in Ireland for health and/or well-being reasons struck a fair balance between, on the one hand, the first and second applicants' right to respect for their private lives under Article 8 and, on the other, profound moral values of the Irish people as to the nature of life and consequently as to the need to protect the life of the unborn." =A., B. and C. v. Ireland, op.cit., para. 230.
[219] A., B. and C. v. Ireland, op.cit., para. 233.
[220] A., B. and C. v. Ireland, op.cit., para. 241.
[221] The Office of the High Commissioner for Human Rights (UN Human Rights), "Stop regression on sexual and reproductive rights of women and girls, UN experts urge". https://www.ohchr.org/EN/NewsEvents/Pages/DisplayNews.aspx?NewsID=23503&LangID=E.

(..)."[222] "This constitutes a misinterpretation of the Convention on the Rights of Persons with Disabilities."[223] According to her, "disability rights and gender equality are two components of the same human rights standard that should not be construed as conflicting. Chairperson of the United Nations Committee on the Elimination of Discrimination against Women noted that "[s]tates should also acknowledge that women's decisions about their own bodies are personal and private, and place the autonomy of the woman at the centre of policy and law-making related to sexual and reproductive health services, including abortion care."[224]

These above-quoted statements, however, are of a rather policy-shaping nature; it remains to be seen how they will be accommodated in subsequent work of the two respective committees. Nonetheless, from this follows that at least as regards the two UN conventions, one could argue that they should be interpreted as CRPD does not preclude exercising rights stemming from CEDAW. Although protections for reproductive rights and gender equality, as well as equality and non-discrimination relating to disability, can commonly be found in the legal orders of concern, one should note that the regional legal orders are the competent ones to interpret their sources of law. Although tendencies at the UN level may affect them, the ultimate say remains at the regional level. However, one can raise a question on the meaning of genetic diversity as affirmed in Article 1 of the UNESCO Universal Declaration on the Human Genome and Human Rights.[225]

Of the regional legal instruments that regulate prenatal screening/testing in a greater detail, of particular relevance are CoE BMC, Article 12 of which permits "[t]ests which are predictive of genetic diseases or which serve either to identify the subject as a carrier of a gene responsible for a disease or to detect a genetic predisposition or susceptibility to a disease". Similarly, also the CoE Recommendation No. R (90) 13 under Principle 2 specifically addresses screening for diseases which pose "serious risk to the health of the child" as permissible.

Sex selection

Sex-selective practices are generally seen in the light of gender inequality and as discriminatory towards women.[226] The United Nations Committee on the Elimination of Discrimination against Women has expressed concern regarding sex-selective abortion,[227] and it has called upon states parties to implement

[222] The Office of the High Commissioner for Human Rights (UN Human Rights), "Stop regression on sexual and reproductive rights of women and girls, UN experts urge". https://www.ohchr.org/EN/NewsEvents/Pages/DisplayNews.aspx?NewsID=23503&LangID=E.

[223] Ibid.

[224] Ibid.

[225] Due to the limitations of this report, the question remains unanswered here.

[226] 1994 Programme of Action of the International Conference on Population and Development (ICPD). "As part of this undertaking States agreed to:. .. eliminate all forms of discrimination against the girl child and the root causes of son preference, which result in harmful and unethical practices regarding female infanticide and prenatal sex selection." United Nations, Programme of Action of the International Conference on Population and Development (ICPD) (1994), paragraph 4.16. See also Toebes, Brigit, "Sex Selection under International Human Rights Law", *Medical Law International,* Vol. 9, 2008, p. 197.

[227] See, e.g., UN, Concluding comments of the Committee on the Elimination of Discrimination against Women: China, 17, 21, U.N. Doc. CEDAW/C/CHN/CO/6 (2006); United Nations, Concluding comments of the Committee on the Elimination of Discrimination against Women: India 38, U.N. Doc. CEDAW/C/IND/CO/3 (2007). See Center for Reproductive Rights, Abortion and Human Rights, Government Duties to Ease Restrictions and Ensure Access to Safe Services,

a comprehensive strategy to overcome traditional stereotypes regarding men's and women's roles in society, which underlie the practice.[228] Along similar lines, the Committee has recommended that a state party monitor implementation of national legislation prohibiting sex-selective abortion, which includes safeguards to prevent criminalization of women who are pressured to obtain the procedure.[229] Of the regional legal orders, CoE BMC addresses sex determination before embryo implantation and under Article 14 states that "use of techniques of medically assisted procreation shall not be allowed for the purpose of choosing a future child's sex, except where serious hereditary sex-related disease is to be avoided" but, as criticized by Toebes, fails to regulate sex-selective abortion.[230] The question of sex-selective abortion has relatively recently been addressed by CoE in 2011, under the CoE Parliamentary Assembly Resolution 1829 (2011) regarding Prenatal sex selection with which it called on the CoE Member States to take specific measures to tackle sex-selective practices.[231]

Conscientious Objection

Protection of conscience is strongly related to the freedom of thought, conscience and religion, which is a right commonly shared in the reviewed human rights legal orders (for example, UN UDHR Article 18, UN ICCPR Article 18, ASEAN Human Rights Declaration Principle 22, CoE ECHR Article 9, AU African Charter on Human and Peoples Rights Article 8, OAS American Declaration of the Rights and Duties of Man Article III, and OAS American Convention on Human Rights Article 12). However, how exactly a manifestation of conscience in healthcare is balanced against other human rights is not *expressis verbis* addressed in these treaties and declarations. Within the Council of Europe, however, Parliamentary Assembly has adopted a resolution on the right to conscientious objection in lawful medical care, and has invited the Council of Europe Member States to: a) guarantee the right to conscientious objection in relation to participation in the medical procedure in question; b) ensure that patients are informed of any conscientious objection in a timely manner and referred to another health-care provider; and c) ensure that patients receive appropriate treatment, in particular in cases of emergency.[232]

The CEDAW Committee has expressed concern over the lack of access to abortion services due to laws permitting conscientious objection by hospital personnel. The Committee has recommended that states parties ensure access to abortion in public health services.[233]

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https://www.reproductiverights.org/sites/crr.civicactions.net/files/documents/BRB_abortion_hr_revised_3.09_WEB.PDF.

[228] See, e.g., UN, Concluding comments of the Committee on the Elimination of Discrimination against Women: China, 17–18, U.N.Doc.CEDAW/C/CHN/CO/6 (2006); United Nations, Concluding comments of the Committee on the Elimination of Discrimination against Women: India 39, U.N. Doc. CEDAW/C/IND/CO/3 (2007). See Center for Reproductive Rights, Abortion and Human Rights, Government Duties to Ease Restrictions and Ensure Access to Safe Services,
https://www.reproductiverights.org/sites/crr.civicactions.net/files/documents/BRB_abortion_hr_revised_3.09_WEB.PDF.

[229] See UN, Concluding comments of the Committee on the Elimination of Discrimination against Women: India 39, U.N. Doc. CEDAW/C/IND/CO/3 (2007). See Center for Reproductive Rights, Abortion and Human Rights, Government Duties to Ease Restrictions and Ensure Access to Safe Services, https://www.reproductiverights.org/sites/crr.civicactions.net/files/documents/BRB_abortion_hr_revised_3.09_WEB.PDF.

[230] Toebes, op. cit.

[231] CoE, Parliamentary Assembly, Prenatal sex selection, Resolution 1829 (2011).

[232]CoE, Parliamentary Assembly,The right to conscientious objection in lawful medical care, Resolution 1763 (2010), para. 4.

[233] Center for Reproductive Rights, Abortion and Human Rights, Government Duties to Ease Restrictions and Ensure Access to Safe Services,

Standard of care, quality of NIPT, wrongful birth and wrongful life

Quality of prenatal genetic testing/screening, and in particular more recently NIPT, is a concern. Under the highest attainable standard of health (the normative framework of which has been reviewed previously) require that medical care is of appropriate quality. The human rights frameworks do not regulate how exactly care shall be delivered but protect the rights that have to be observed when care is being provided. Moreover, generally, for failures to ensure the protection of rights (as has been previously reviewed), one can request a remedy. The scope of his remedy in light of such claims as wrongful life and wrongful birth remains to be addressed at each regional legal order.

Genetic privacy, the right not to know

The protection of privacy and personal data has been reviewed previously. Here suffices to highlight concern over the extent to which the existing protections address interests of the children to be born or foetuses at the time of carrying out genetic screening /testing. With the advances in technology, the scope of protection of privacy and personal data needs to be reconsidered in terms of safeguarding privacy and the right to know and not to know;[234] moreover, questions over the considerations of permissibility of intrusions can be raised. As has been above reviewed, when prenatal testing/screening is regulated, its scope is constrained to serious health conditions. However, one can question how it is being implemented nationally, namely, whether all other applications are prohibited, or only application for serious health conditions is regulated, and others are not, and therefore possible.

4.2.5 New-born Screening

Decision-making about new-born genetic screening and conditions included in the screening programs

The normative basis for questions relating to decision-making about genetic screening has already been introduced previously. Here particular attention needs to be given to the child's perspective, and in particular, whether the screening should be a discretionary choice of the parents or there are sufficient grounds for the state to initiate compulsory genetic screening programmes.

Right to health is a common concern of the reviewed legal orders. Likewise, some of the legal orders envisage specific protection for children. UN CRC protects the child's privacy, requires that best interest is safeguarded, and protects the child's right to the highest attainable standard of health. Similar protection for children can be found in some of the regional legal orders. For example, in ASEAN children are recognized as a vulnerable group under Principle 4 of the ASEAN Human Rights Declaration; moreover, under Principle 30.3 childhood is a period of special assistance, and enjoyment to the highest attainable standard of health shall be granted under Principle 29.1. In AU, the African Charter on the Rights and Welfare of the Child address specifically rights and welfare of the child, and include protection for best interests (Article 4), obligation to ensure, to the maximum extent possible, the survival, protection and development of the child (Article 5.2), protection of privacy (Article 10),as well as grants the right to enjoy the best attainable state of physical, mental and spiritual health (Article 14.1), and lists particular measures that shall be taken (Article 14.2). In OAS and in the CoE extensive child's rights specific protection framework has not been adopted, nonetheless, child's rights and interests are protected through the civil and political rights, as well as socioeconomic rights protection frameworks; however, as

https://www.reproductiverights.org/sites/crr.civicactions.net/files/documents/BRB_abortion_hr_revised_3.09_WEB.PDF.

[234] Zillén, Kavot , Jameson Garland and Santa Slokenberga, "The Rights of Children in Biomedicine: Challenges posed by scientific advances and uncertainties". https://rm.coe.int/16806d8e2f, p.35.

elaborated below CoE has specifically addressed the rights and interests of minors in genetic interventions.

From UNESCO International Declaration on Human Genetic Data Article 8 (d) derives that genetic screening of minors not able to consent will normally only be ethically acceptable when they have important implications for the health of the person and have regard to his or her best interest. Article 5(e) of the UNESCO Universal Declaration on the Human Genome and Human Rights emphasizes a direct health benefit for the person concerned. CoE APGT specifically addresses genetic screening question and emphasizes the voluntary nature. However, while it regulates genetic testing and screening (to some degree) distinctly, these concepts have not been defined. Therefore, for genetic screening of minors also requirements set forth in Article 10, which addresses the protection of persons not able to consent, are of relevance. Regarding minors, it states that "[w]here, according to law, a minor does not have the capacity to consent, a genetic test on this person shall be deferred until attainment of such capacity unless that delay would be detrimental to his or her health or well-being." As derives from Explanatory Report to CoE APGT, not only conditions that "would allow appropriate therapeutic measures to be taken for a disease or disorder from which the subject is suffering"[235] fall within this exception, but also "situations where the genetic test would provide predictive information allowing timely preventive measures to be taken. In particular, this applies to tests for diseases which might develop before the subject has attained legal capacity."[236]

Following CoE APGT one could argue that for childhood-onset diseases genetic screening could be envisaged. Nonetheless, the principles of voluntariness apply. For the other regional legal order orders of concern, as well as for states that have not acceded to the CoE APGT, the question would need to be resolved with due regard to the generic human rights provisions, including finding a balance between an intervention of potential individual immediate or future value, as well as limits of parental rights regarding their children. It cannot be precluded that this assessment is also subject to technological development, and considerations over benefits and challenges can be shaped by progress in medicine, for example, availability of cures, preventive measures etc. Moreover, it should also not be excluded that there can be situations when screening is in the best interests of children, and mandatory policies could be implemented.

Research on samples collected in relation to the new-born genetic screening

The normative framework for research on residual samples/data has already been reviewed previously. Therefore, here we describe only considerations regarding those instruments that specifically consider research on samples stemming from minors.

Under UNESCO Universal Declaration on the Human Genome and Human Rights Article 5 (e) "[r]esearch which does not have an expected direct health benefit may only be undertaken by way of exception, with the utmost restraint, exposing the person only to a minimal risk and minimal burden and if the research is intended to contribute to the health benefit of other persons in the same age category or with the same genetic condition, subject to the conditions prescribed by law, and provided such research is compatible with the protection of the individual's human rights." A similar approach is echoed in CoE Recommendation CM/Rec(2016)6 of the Committee of Ministers to member States on research on biological materials of human origin Article 12.1. Furthermore, Article 12.5 of the same recommendation

[235] CoE, Explanatory Report to the Additional Protocol to the Convention on Human Rights and Biomedicine concerning Genetic Testing for Health Purposes, ETS 203, para. 90.
[236] Explanatory Report to the APGT, op.cit., para. 91.

emphasizes that "[w]here a person not able to consent, whose biological materials have been stored for future research, attains or regains the capacity to consent, reasonable efforts should be made to seek the consent of that person for continued storage and research use of his or her biological materials." Moreover, alteration of the scope of consent, as well as withdrawal, is of importance, and such an option should be provided.

While in Europe tendencies for regulating research on samples is evident, it is unclear whether the same can be said about other regional legal orders. For example, more generally, scholars have raised concerns over biobank governance in other regions, including adequacy of data protection in some African states.[237]

4.2.6 Direct-to-consumer advertising of genetic testing

Permissibility of direct-to-consumer advertising and difference between health and non-health testing

Regulating of direct-to-consumer advertising, and permissibility of direct-to-consumer advertising, in terms of human rights raise questions on the enjoyment of benefits scientific advances bring along with a view to furthering such rights as the right to the highest attainable standard of health, as well as need to protect from these enjoyments. Normative frameworks of these rights have already been introduced previously. Therefore, a question to scrutinize is whether a state has a duty to protect from challenges, in the alternative, whether individuals have freedom from the state's intervention with enjoying the advances. A response to this question considerably relates to the approach to be taken for protecting human rights, and these questions are beyond the scope of this report. However, from the tri-partial obligation to respect, protect, fulfil, one could argue that a state could take measures to protect from risks related to scientific advances, even if this includes limiting access to some of the benefits. However, then in light of the nature of these tests, a question to consider is appropriate oversight and enforcement of restrictions if, for example, the testing is advertised and sold over the internet.

Neither of the regional legal orders *expressis verbis* address permissibility of direct-to-consumer genetic testing. However, they set forth protection for the rights that can be affected through advertising of, and consequently often access to direct-to-consumer genetic testing. For example, in the AU, under the AU Model Law for Medical Products Regulation, all promotion and advertisement of medical products shall be approved by a National Regulatory Agency/Authority and the National Regulatory Agency/Authority shall issue guidelines relating to the promotion and advertising of medical products and an enforceable Code of Marketing Practice (Article 19).[238] The Model Law, however, is not a prescriptive, directly applicable instrument and it requires domestic implementation by the AU Member States.[239] In the OAS, consumers protection has been addressed only in general terms, by setting up the Network For Consumer Safety And Health,[240] designed as a platform for exchanging knowledge and experience in this field and

[237] CoE, The Committee of Ministers of the CoE adopts Recommendation CM/Rec(2016)6 of the Committee of Ministers to member States on research on biological materials of human origin https://www.coe.int/en/web/bioethics/biobanks , and also CoE, recommendation No. R (81) 1 of the committee of ministers to member States on regulations for automated medical data banks https://rm.coe.int/CoERMPublicCommonSearchServices/DisplayDCTMContent?documentId=09000016804eee77.

[238] AU, Model Law for Medical Products Regulation, January 2016.

[239] The New Partnership for Africa's Development, "Issue Brief: AU Model Law for Medical Products Regulation: Increasing access to and delivery of new health technologies for patients in need". https://www.nepad.org/publication/issue-brief-african-union-model-law-medical-products-regulation-increasing-access.

[240] OAS General Assembly, Resolution AG/RES. 2549 (XL-O/10) on "Consumer Protection: Network for Consumers Safety and Health in the Americas", 8 June 2010.

by creating an Inter-American Rapid Product-Safety Warning System, which is aimed at "rapid detection and coordinated action to prevent the entry of unsafe consumer goods into markets in the Americas".[241] Thus, it would be rather the OAS human rights framework that might be relevant. In CoE, provisions that are of relevance to direct-to-consumer advertising of genetic testing are set forth in APGT, which in Article 8.1 requires that "[w]hen a genetic test is envisaged, the person concerned shall be provided with prior appropriate information in particular on the purpose and the nature of the test, as well as the implications of its results." Moreover, under Article 8.2 it requires that consent for tests predictive of a monogenic disease; tests serving to detect a genetic predisposition or genetic susceptibility to a disease; as well as tests serving to identify the subject as a healthy carrier of a gene responsible for a disease is documented, whereas for other tests merely informed consent requirement applies. Therefore, advertising and acquiring of genetic testing requires that the informed consent criteria are met. Advertising could be seen as an element of informed consent process as it informes a consumer about the testing, and therefore, would require that in so far as information about the purpose and the nature of the test, as well as the implications of its results is provided, it is accurate.

5 Analysis of relevant EU laws

5.1 The extent to which addressing the identified legal issues including human rights challenges in genetics and genomics lie within the competences of the EU and sources of law

The questions of human germline gene editing, including research and application, as well genetic testing/screening, including various angles therein, triggers several areas of law in which EU enjoys either shared competence or competence to support the Member State actions.

Positive and negative integration are mutually exclusive in the EU legal order. Negative integration (removing barriers through adjudication) is relevant only to the extent that an EU secondary law measure does not harmonize particular requirements. While negative integration relates to particular occurrences, and in particular in the context of genomics, removing barriers to the free movement within the internal market, positive integration (adoption of regulatory measures; legislation) relates to EU-wide measures. Depending on a particular situation, these positive integration measures could also beyond the limits of EU territory, for example, through externalizing the internal market via agreements, or requiring compliance with EU law in order goods can be placed on the market or EU data subjects' data can be processed. As concern here is with the current regulatory responses, future challenges and possible ways forward, of key relevance are considerations on how the EU addresses the situation now, in particular, through positive integration measures.

Human germline editing

Questions relating to human germline editing within the EU are addressed through various avenues. Firstly, through clinical trials legal framework, which includes the Clinical Trials Directive which has been transposed in national laws in the EU Member State and the recently adopted Clinical Trials Regulation.[242] The Clinical Trials Regulation has been adopted and has entered into force but is not being applied yet. As specified in Article 99.2 of the Clinical Trials Regulation, its application depends on functionality of the EU portal (a single entry point for the submission of data and information relating to clinical trials in

[241] OAS General Assembly, Resolution AG/RES. 2769 (XLIII-O/13) on "Network for Consumer Safety and Health and Inter-American Rapid Product-Safety Warning System", 5 June, 2013.

[242] European Parliament and the Council, Regulation No 536/2014 of 16 April 2014 on clinical trials on medicinal products for human use, and repealing Directive 2001/20/EC. Slokenberga and Howard, op. cit. 18.

accordance with the regulation)[243] and EU database (an EU level database for the purposes of containing data and information that is submitted in accordance with the regulation).[244] Once the EU portal and the EU database have achieved full functionality, the European Commission shall publish a notice in the Official Journal of the European Union;[245] six months after this notice, the Regulation will be applicable.[246] It is expected to happen sometime in 2020.[247] Once applicable, the regulation will apply directly throughout the EU. At that point, the Clinical Trials Directive will be repealed; however, during a transition period, both, rules of the Clinical Trials Directive and the Clinical Trials Regulation will apply.[248]

Secondly, through animal protection framework relevant for animals used to test gene editing that can be used then in humans. Generally, in the area of research EU competence is limited to defining and implementing programmes and the exercise of that competence shall not result in the Member States being prevented from exercising theirs.[249]

Thirdly, through allocating funding for research. Substantively, the governance of research falls under Title XIX of TFEU, Article 179 TFEU of which emphasized the link between research and competitiveness of the EU. However, that is not only to be exercised in line with the applicable laws shaping research at the EU and national level, but also with due regard to the respect for "fundamental orientations and choices of the research policies of the Member States".[250]

Fourthly, it relates to the questions of patenting as addressed under the Biotech Directive [251] and touches upon matters addressed under the Human Tissue and Cell Directive.

Regulating genetic testing and screening

Questions relating to genetic testing and screening, including prenatal testing and newborn screening, trigger predominantly the applicability of *in vitro* diagnostic medical devices framework, which comprises IVDMD Directive that was transposed in the national laws of the EU Member States, which is being repealed by IVDMD Regulation. It also triggers data protection. It could also relate to other areas, such as cross border healthcare, however potentially of less significance. Therefore, the two areas of law are predominantly reviewed.

Data protection regulatory framework in the EU legal order is multi-layered. Further to data protection is a part of privacy – which is protected under Article 8 ECHR and, therefore, constitutes a general principle of EU law – it is distinctly protected under the CFREU, and detailed norms about the protection of the

[243] Clinical Trials Regulation, op.cit., Art. 80.
[244] Clinical Trials Regulation, op.cit., Art. 81.
[245] Clinical Trials Regulation, op.cit., Art. 82.3.
[246] Clinical Trials Regulation, op.cit., Art. 99.
[247] See European Commission, Medicinal products. https://ec.europa.eu/health/human-use/clinical-trials/regulation_en.
[248] Clinical Trials Regulation, op.cit.,,Art. 98.
[249] Article 4.3 TFEU Consolidated version of the Treaty on the Functioning of the European Union OJ C 326, 26.10.2012, p. 47–390.
[250] Protocol 34. Declaration on Article 179 of the Treaty on the Functioning of the European Union
The Conference agrees that the Union's action in the area of research and technological development will pay due respect to the fundamental orientations and choices of the research policies of the Member States.
[251] European Parliament and the Council, Directive 98/44/EC of 6 July 1998 on the legal protection of biotechnological inventions. OJ L 213, 30.7.1998, p. 13–21.

personal data and free movement of such data are outlined in GDPR.[252] The CFREU distinguishes the right to protect private life and data protection, assigning it the status of a new, autonomous right in the EU's fundamental rights catalogue. Such advancement can be said to be "the final point of a long evaluation, separating privacy and data protection".[253] Article 7 CFREU, which is modelled after Article 8 ECHR, seeks to protect privacy. Furthermore, with the entry into force of Lisbon Treaty, Article 16.1 TFEU recognizes a right to data protection and serves as a legal basis for adopting secondary law measures in securing the protection of personal data in the EU legal order, specifically the GDPR.

Direct-to-consumer advertising of genetic testing

Direct-to-consumer advertising of genetic testing triggers applicability of the above introduced *in vitro* diagnostic medical devices framework, but also the advertising framework. In particular, it comprises Unfair Commercial Practices Directive and Misleading and Comparative Advertising Directive.

Fundamental rights

Fundamental rights in the EU legal order are addressed through the general principles of EU law,[254] as well as through the CFREU. With the entry into force of the Lisbon Treaty, the CFREU has become a source of primary law in the EU legal order,[255] which content-wise overlaps with the ECHR. Unlike the Treaties, the CFREU as such is not a self-standing legal instrument vis-à-vis the Member States. As stipulated in the CFREU, it applies to the Member States when they are implementing the Union law, and the application of the CFREU shall not in any way extend the competencies of the Union as defined in the Treaties.[256]

5.2 Human germline gene editing

5.2.1 Basic research

The EU does not set requirements for basic research in human or embryos/gametes using germline modification in basic research. As basic research does not relate to human applications, it falls beyond the scope of Human Tissue and Cell Directive,[257] as well as the EU Clinical trials Framework.[258] Moreover, the

[252] Slokenberga, op.cit. 173, ch. 6.5.

[253] Rodotà, Stefano, "Data Protection as a Fundamental Right" in Serge Gutwirth and others (eds), *Reinventing Data Protection?*, Springer, Netherlands, 2009 p.79. In similar way de Hert and Gutwirth have claimed that the recognition of data protection as a fundamental right in the EU legal order has balanced out a discrepancy between the fundamental rights and economic objectives that was created as a result of the Data Protection Directive. De Hert, Paul, and Serge Gutwirth, "Data Protection in the Case Law of Strasbourg and Luxemburg: Constitutionalisation in Action", *Reinventing data protection?*, Springer, 2009, p. 9. See Slokenberga, op.cit. 252, 6.5.

[254] Article 6.3 TEU stipulates that the general principles of EU law are fundamental rights that stem from the ECHR. European Union, Consolidated version of the Treaty on European Union, 13 December 2007, 2008/C 115/01.There is a considerable and well-documented history for developing general principles in the EU legal order. See, for example, Gutiérrez-Fons, Jose, and Koen Lenaerts, "The Constitutional Allocation of Powers and General Principles of EU Law", *Common Market Law Review,* Vol. 47, 2010, p. 1629. De Witte, Bruno, "Interpreting the EC Treaty like a Constitution: The Role of the European Court of Justice in Comparative Perspective", *Judicial Control: Comparative Essays on Judicial Review,* 1995, p. 149.

[255] TEU, op.cit. Article 6.1.

[256] CFREU, European Union, Charter of fundamental rights of the European Union, 18 December 2000, 2000/C 364/01, Article 51.2.

[257] European Parliament and the Council, Directive 2004/23/EC of 31 March 2004 on setting standards of quality and safety for the donation, procurement, testing, processing, preservation, storage and distribution of human tissues and cells, OJ L 102, 7.4.2004, p. 48–58, Article 1 and 2.1.

[258] Clinical Trials Regulation *expressis verbis* excludes non-interventional studies from its scope. See Article 1. Non-interventional studies under Article 2.2.4 of the Regulation are defined as 'a clinical study other than a clinical trial'. The Clinical Trials Directive in Article 1.1 specifies its applicability to clinical trials that following Article 2.a focus on

Human Tissue and Cell Directive also does not ”interfere with decisions made by the Member States concerning the use or non-use of any specific type of human cells, including germ cells and embryonic stem cells.” [259] Likewise, it does not interfere with provisions of Member States defining the legal term “person” or “individual”,[260] which could be relevant in addressing the status of the human embryo.

5.2.2 Animals: pre-clinical research

EU and the Member States are under a duty to pay full regard to the welfare requirements of animals in the area of research.[261] Detailed requirements relating to animal use in scientific research care are set forth in the Animal Protection Directive.[262] The directive is a minimum harmonization measure that establishes measures for the protection of animals used for scientific or educational purposes.[263] The directive refers to 3R policies (*replacement, reduction, and refinement*) already in its recitals stating “the care and use of live animals for scientific purposes is governed by internationally established principles of replacement, reduction and refinement”.[264] These principles to shape animal protection are also enshrined in Article 13 of the directive. The directive does not *expressis verbis* refer to the use of animals for germline modification experiments *per se*. However the above mentioned three principles shall apply in so far as animals are being used for research. The directive sets forth special protection to non-human primates,[265] and in exceptional cases permit the use of great apes for research.[266] These exceptions include the avoidance, prevention, diagnosis or treatment of disease, ill-health or other abnormality or their effects in human beings, animals or plants, as well as manufacture or testing of the quality, effectiveness and safety of drugs, foodstuffs and feed-stuffs and other substances or products for purposes set forth in the Directive.[267]

5.2.3 Clinical research

The EU clinical trials regulatory framework comprises the Clinical Trials Directive, which has been transposed in national laws in the EU Member States, and the recently adopted Clinical Trials Regulation. Once applicable, the Regulation will apply directly throughout the EU. Both, the Clinical Trials Directive and the Clinical Trials Regulation expressly preclude clinical trials that result in modifications to the subject’s germ line genetic identity.[268] According Article 90 of the Clinical Trials Regulation, ”no gene

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the applicability of investigation in human subjects. See Directive 2001/20/EC of the European Parliament and of the Council of 4 April 2001 on the approximation of the laws, regulations and administrative provisions of the Member States relating to the implementation of good clinical practice in the conduct of clinical trials on medicinal products for human use.

[259] European Parliament and the Council, Directive 2004/23/EC of 31 March 2004 on setting standards of quality and safety for the donation, procurement, testing, processing, preservation, storage and distribution of human tissues and cells, OJ L 102, 7.4.2004, p. 48–58, Recital 12.

[260] Ibid.

[261] Article 13 TFEU Consolidated version of the Treaty on the Functioning of the European Union OJ C 326, 26.10.2012, p. 47–390.

[262] European Parliament and the Council, Directive 2010/63/EU of 22 September 2010 on the protection of animals used for scientific purposes Text with EEA relevance, OJ L 276, 20.10.2010, p. 33–79.

[263] Animal Protection Directive, op.cit., Article 1.1.

[264] Animal Protection Directive, op.cit., Recital 13.

[265] Animal Protection Directive, op.cit., Article 8.

[266] Animal Protection Directive, op.cit., Article 55.2., Article 8.3 and Recital 18.

[267] Animal Protection Directive, op.cit., Article 5 (b)(i), (c).

[268] Clinical Trials Directive, op.cit. Art 9.6. Clinical Trials Regulation, op.cit., Recital 75 and Article 90.

Article 2 (a) of the Clinical Trials Directive states that “”clinical trial”: any investigation in human subjects intended to discover or verify the clinical, pharmacological and/or other pharmacodynamic effects of one or more

therapy clinical trials may be carried out which result in modifications to the subject's germ line genetic identity". Mutatis mutandis, Clinical Trials Directive 2001/20/EC, Article 9.6 states that "[n]o gene therapy trials may be carried out which result in modifications to the subject's germ line genetic identity". This prohibition is also endorsed through Advanced Therapy Medicinal Products Regulation, which aims to address the complexity of combined advanced therapy medicinal products containing viable cells or tissues requires a specific approach as *lex specialis*, which introduces additional provisions to those laid down in Directive 2001/83/EC.[269]

The Clinical Trials Regulation,[270] as well as the Clinical Trials Directive, focuses on a medicinal product,[271] and Advanced Medicinal Products Directive addresses a gene therapy medicinal product,[272] which relates to disease preventing (prophylactic), diagnostic or therapeutic gene. This means that under the EU law health-related application/ disease preventing (prophylactic) and diagnostic interventions are regulated, whereas non-health related application falls outside the scope of the current harmonization measures. One could, however, examine further in SIENNA task 4.2, whether and how EU competences, for example, in the area of public health and internal market, could be used to legislate on non-health related human germline editing interventions.

5.2.4 Clinical care

Clinical care relating to human germline editing is not expressly regulated; arguably, relating to the limits of EU competences. However, because clinical trials are not permitted, it follows that these interventions shall not take place.

5.2.5 Funding

Advances in human germline editing are strongly related to the available funding for research activities. Article 19 of EU Horizon 2020 Regulation, laying down the rules for participation and knowledge

investigational medicinal product(s), and/or to identify any adverse reactions to one or more investigational medicinal product(s) and/or to study absorption, distribution, metabolism and excretion of one or more investigational medicinal product(s) with the object of ascertaining its (their) safety and/or efficacy."

Article 2.2 of the Clinical Trials Regulation states that ""Clinical trial" means a clinical study which fulfils any of the following conditions:

(a) the assignment of the subject to a particular therapeutic strategy is decided in advance and does not fall within normal clinical practice of the Member State concerned;

(b) the decision to prescribe the investigational medicinal products is taken together with the decision to include the subject in the clinical study; or

(c) diagnostic or monitoring procedures in addition to normal clinical practice are applied to the subjects."

European Parliament and the Council, Directive 2001/83/EC of 6 November 2001 on the Community code relating to medicinal products for human use [2001] OJ L 311/ 67, see M9.

[269] See Article 4.1 of the regulation that states "the rules set out in Article 6(7) and Article 9(4) and (6) of Directive 2001/20/EC in respect of gene therapy and somatic cell therapy medicinal products shall apply to tissue engineered products' European Parliament and the Council, Regulation No 1394/2007 of 13 November 2007 on advanced therapy medicinal products and amending Directive 2001/83/EC and Regulation (EC) No 726/2004.

[270] See Clinical Trials Regulation, op.cit., Article 2.2.1.

[271] See Community Code Directive, op.cit. Article 2.a.

[272] "For the purposes of this Annex, gene therapy medicinal product shall mean a product obtained through a set of manufacturing processes aimed at the transfer, to be performed either in vivo or ex vivo, of a prophylactic, diagnostic or therapeutic gene (i.e. a piece of nucleic acid), to human/animal cells and its subsequent expression in vivo. The gene transfer involves an expression system contained in a delivery system known as a vector, which can be of viral, as well as non-viral origin. The vector can also be included in a human or animal cell." Community Code Directive, op.cit.

dissemination, in Horizon 2020, places a general obligation on the funding seekers to comply *among other things* with the ethical principles or any applicable legislation. Recital 29 of the Regulation emphasizes that these are "fundamental ethical principles". It is not, however, further delineated what these principles are or how to identify what would be regarded as an ethical principle or a fundamental ethical principle under this regulation. The question of the notion of applicable legislation is not less ambiguous. Any applicable legislation under Article 19 means "relevant national, Union and international legislation, including the Charter of Fundamental Rights of the European Union and the European Convention on Human Rights and its Supplementary Protocols". This means that the EU law needs not to apply to the subject matter; it could also be other laws attributable to the issue in some way. As far as the EU law regulates human germline modification, the rules become clearer and more straightforward. For example, the EU clinical trials legislation prohibits gene therapy trials that result in modifications to the subject's germ line genetic identity.[273] This prohibition is also echoed in the Horizon 2020 ethics self-assessment document, which states that "research activity intended to modify the genetic make-up of human beings that could make such changes heritable (apart from research relating to cancer treatment of the gonads, which may be financed)" are not eligible for funding.[274] A proposal that fails to meet the applicable requirements "may be excluded from the evaluation, selection and award procedures at any time."[275] However, in light of the limited scope of the EU Clinical Trials framework, it could be argued that the prohibitions in research are rather more comprehensive. Thus, while the EU does not regulate the non-clinical application of germline gene editing, the funding for this purpose may not be allocated.

5.2.6 Patenting

The question of patents is regulated under the Biotech Directive. For the directive, "new inventions, which involve an inventive step and which are susceptible of industrial application shall be patentable even if they concern a product consisting of or containing biological material or a process by means of which biological material is produced, processed or used."[276] Furthermore, "Biological material which is isolated from its natural environment or produced by means of a technical process may be the subject of an invention even if it previously occurred in nature."[277]

Patents relating to the human body are specifically addressed under Article 5 of the Biotech Directive. Article 5.1 states that "[t]he human body, at the various stages of its formation and development, and the simple discovery of one of its elements, including the sequence or partial sequence of a gene, cannot constitute patentable inventions." However, "[a]n element isolated from the human body or otherwise produced by means of a technical process, including the sequence or partial sequence of a gene, may constitute a patentable invention, even if the structure of that element is identical to that of a natural element." As specified further, in such a case, "[t]he industrial application of a sequence or a partial sequence of a gene must be disclosed in the patent application."[278] This is related to the intended limitation that "a mere DNA sequence without indication of a function does not contain any technical

[273] See Clinical Trials Regulation, op.cit., Article 90. Clinical Trials Directive, op.cit., Article 9.4.
[274] European Commission, Directorate-General for Research & Innovation, Horizon 2020 Programme Guidance How to complete your ethics self-assessment, Version 5.3 of 21 February 2018.
[275] Horizon 2020 Regulation, op.cit., Article 13.4.
[276] Biotech Directive, op.cit., Article 3.1.
[277] Biological material under Article 2.1.a is defined as "any material containing genetic information and capable of reproducing itself or being reproduced in a biological system." European Parliament and the Council, Directive 98/44/EC of 6 July 1998 on the legal protection of biotechnological inventions, OJ L 213, 30.7.1998, p. 13–21.
[278] Biotech Directive, op.cit., Article 5.3.

information and is therefore not a patentable invention."[279] Such an approach is upheld in the CJEU case law, where the Court has also pointed out that the absence of the industrial application would mean merely a discovery of a DNA sequence, which is not patentable.[280]

5.2.7 Fundamental rights considerations

Human germline gene editing could touch upon a number of fundamental rights protected under the CFREU. For example, the regulatory approach towards not permitting human germline interventions could be strongly related to such fundamental rights as the protection of dignity (Article 1), as well as protection of integrity (Article 3), life (Article 3), ensuring humane treatment (Article 4), protecting private life (Article 7), as well as health (Article 35), and regulating the freedom of arts and sciences (Article 13).

5.3 Genetic screening and genetic testing

5.3.1 Genetic screening and testing in terms of public health

5.3.2 Data protection perspectives

GDPR applies *inter alia* "to the processing of personal data wholly or partly by automated means".[281] Personal data within the meaning of GDPR are "any information relating to a data subject", whereby the data subject is "an identified natural person or a natural person, who can be identified, directly or indirectly, by means reasonably likely to be used by the controller or by any other natural or legal person, in particular by reference to an identification number, location data, online identifier or to one or more factors specific to the physical, psychological, genetic, mental, economic, cultural or social identity of that person."[282] Therefore, in processing biological samples in genetic screening and testing, as well as other data relating to an individual so that screening can be provided, the applicability of GDPR is triggered.

The GDPR provides specific protection not only to medical data but also to genetic data and prohibits processing them under Article 9.1 GDPR unless special requirements set forth in Article 9.2 GDPR are met, or further national regulations under Article 9.4 GDPR have been adopted. The requirements differ for genetic testing/screening and scientific research. Furthermore, as below is shown, a distinction can also be drawn between genetic testing/screening for health-related purposes and other purposes. Furthermore, substantively it sets forth a number of controller's and processor's obligations and data subject's rights which ought to be ensured. Here below, only those GDPR requirements that directly relate to the lawfulness of genetic testing/screening and research will be reviewed; only exceptionally, other requirements will be reviewed.

Lawfulness for genetic testing/screening

Lawfulness of processing goes hand in hand with the requirements set forth for the processing of genetic data. Following Article 6.1 and 9.2 GDPR requirements, a data subject may consent to the processing of their personal data either for genetic testing/screening or research under Articles 6.1.a and 9.2.a. However, other lawfulness grounds could be relevant as well. For example, Article 6.1.d enables processing of personal data in order to protect vital interests of the data subject or another person. Healthcare purposes are echoed in Article 9.2.h of GDPR and 9.3 permitting processing based on the

[279] Biotech Directive, op.cit., Recital 23.

[280] CJEU, Case C-377/98, *Kingdom of the Netherlands v European Parliament and Council of the European Union,* 09 October 2001.

[281] European Parliament and the Council, Regulation 2016/679 of 27 April 2016 on the protection of natural persons with regard to the processing of personal data and on the free movement of such data, and repealing Directive 95/46/EC, OJ L 119, 4.5.2016, p. 1–88, Article 2.1.

[282] GDPR, op.cit., Article 4.2.

existence of an obligation of professional secrecy ”established by national competent bodies or by another person also subject to an obligation of secrecy under Union or Member State law or rules established by national competent bodies.” While it could be seen how genetic testing for some health-related purposes could fall within this category, it is rather difficult to see the lifestyle and fun-trait testing as being related to vital interests.

Article 6.1.e also permits processing in the public interest and Article 6.1.c permits processing that is necessary for compliance with a legal obligation to which the controller is subject. In these, both cases under Article 6.2 and 6.3, national law or EU law shall be in place. This public interest protection is echoed in *inter alia* Article 9.2.g that permits processing of genetic and health data if ”processing is necessary for reasons of substantial public interest” and conditions set forth in the article are met. However, one can question whether genetic screening and genetic testing could be part of public interest.

Furthermore under Article 9.4 GDPR Member States are allowed to ”maintain or introduce further conditions, including limitations, with regard to the processing of genetic data, biometric data or data concerning health”. Therefore, it could be that other conditions are set forth for the processing of genetic data in genetic screening or testing at the national level, thus creating a diverse genetic data protection framework at the national level.

Specific considerations for further processing for research involving genetic and health data, and other purposes

GDPR enables further use of genetic data for other purposes than they have been previously collected for. Further processing for archiving purposes in the public interest, scientific or historical research purposes or statistical purposes is distinctly regulated under Article 89 GDPR and is generally regarded to be compatible with the initial purposes the data were collected for, and are subject to derogations relating to individual rights, both, by directly applying GDPR provisions and through national implementing measures. The detailed rules, however, differ, whether the processing relates to personal data processed for scientific or historical research purposes or statistical purposes, or for archiving purposes in the public interest.[283] These exceptions also are echoed in Article 9.2.j GDPR that facilitate special category data processing for archiving purposes in the public interest, scientific or historical research purposes or statistical purposes. Furthermore, also Member States could provide further specifications under Article 9.4 GDPR. While it is rather easy to carry out further processing of genetic data relating to the purposes *expressis verbis* listed in the GDPR (scientific or historical research purposes or statistical purposes), for other purposes lawfulness grounds jointly with special requirements for processing special categories of data under Article 9.2 GDPR would need to be met, which might appear to be a rather challenging task. With respect to such purposes as a criminal investigation, GDPR permits a number of restrictions for the GDPR requirements under national law that follows guidance under Article 23 GDPR, following also requirements of Article 10 GDPR. In the context of use genetic data for such purposes as genetic investigation, national laws should be also in accordance with the so called ”Police Directive”, which ”lays down the rules relating to the protection of natural persons with regard to the processing of personal data by competent authorities for the purposes of the prevention, investigation, detection or prosecution of criminal offences or the execution of criminal penalties, including the safeguarding against and the prevention of threats to public security (Article 1) and explicitly covers genetic data as one of the special categories of data (Article 10).[284]

[283] See in particular Articles 89.2 and 89.3 GDPR, op.cit.

[284] European Parliament and of the Council, Directive 2016/680 of the of 27 April 2016 on the protection of natural persons with regard to the processing of personal data by competent authorities for the purposes of the prevention,

Data subject's rights

When data are being lawfully processed within genetic screening or testing, also other requirements set forth in the GDPR need to be observed. They relate to such considerations as fairness and transparency, purpose specification, data minimisation, accuracy, storage limitation, integrity, and confidentiality. The controller (and in the case of joint controllers or in the case of processor also they) are responsible for ensuring compliance with the GDPR under accountability principle.[285] These activities are subject to oversight and violations can be remedied with actions, including taken by the data subjects against the person having committed the breach, as well as sanctions imposed by the data protection authority.[286] In genetic screening and testing, all data subject rights apply, which means that the data subjects have such rights as the right to information,[287] access rights,[288] right to rectification,[289] right to erasure,[290] right to the restriction of processing,[291] right to data portability,[292] right to object.[293] Additional protective measures include, for example, a notification entitlement, providing the data subject has triggered it.[294] Further to these rights and protection measures, at the data subject's disposal is access to justice, and access to remedies, as well as trigger liability and penalties regarding violations of their rights protected under the GDPR. In research, however, depending on circumstances, and in particular, whether national law is in place following Article 89.2 GDPR derogations or through directly applying GDPR provisions regarding individual rights in individual research situations, a number of rights can be derogated from. There could be situations when the data subject is left with means only to protect their rights and without substantive protections of their rights.[295]

5.3.3 *In vitro* diagnostic medical devices and genetic screening

5.3.3.1 Requirements stemming from IVDMD Directive

The scope of application of IVDMD Directive

The IVDMD Directive lays down rules for *in vitro* diagnostic medical devices so that they can be placed on the market or put into service. It applies only to that genetic analysis within genetic screening that fall within the scope of the IVDMD Directive and do not fall under the in-house genetic test exemption under Article 1.5 of the IVDMD Directive.[296] The Member States, however, retain the right to subject in house testing to appropriate protection requirements. This means, as long as the genetic analysis is being

investigation, detection or prosecution of criminal offences or the execution of criminal penalties, and on the free movement of such data, and repealing Council Framework Decision 2008/977/JHA, OJ L 119, 4.5.2016, p. 89–131.

[285] GDPR, op.cit., Article 5.

[286] See GDPR, op.cit., Chapter VIII.

[287] GDPR, op.cit., Articles 12-14.

[288] GDPR, op.cit., Article 15.

[289] GDPR, op.cit., Article 16.

[290] GDPR, op.cit., Article 17.

[291] GDPR, op.cit., Article 18.

[292] GDPR, op.cit., Article 20.

[293] GDPR, op.cit., Article 21.

[294] GDPR, op.cit., Article 19.

[295] Slokenberga, Santa, "Direct-to-Consumer Genetic Testing: Changes in the EU Regulatory Landscape", *European Journal of Health Law,* Vol. 22, 2015, p. 463.

[296] It states 'This Directive shall not apply to devices manufactured and used only within the same health institution and on the premises of their manufacture or used on premises in the immediate vicinity without having been transferred to another legal entity. This does not affect the right of Member State to subject such activities to appropriate protection requirements.' European Parliament and the Council, Directive 98/79/EC of 27 October 1998 on *in vitro* diagnostic medical devices OJ L 331, 7.12.1998, p.1.

provided by the same entity, requirements from IVDMD Directive do not apply. Due to the nature of genetic screening as a population-based measure as opposed to individual genetic analysis, it is unlikely that genetic analysis forming part of genetic screening are an in-house test, however that cannot be excluded in its entirety. It is, however, a common approach to genetic testing, and has been a key loophole in the law for the vagueness of EU law regarding direct-to-consumer genetic testing,[297] and consequently problems with tackling these tests also nationally.

The IVDMD Directive applies to those tests that involve *in vitro* examination of a specimen derived from the human body for solely or principally providing information about a physiological or pathological state or congenital anomality, determining safety and compatibility with potential recipients, or monitoring therapeutic measures.[298] It does not, therefore, apply to non-health related genetic analysis. Other types of genetic analysis remain specifically unregulated and could be addressed through the general consumer protection legal requirements, except for treating specimen receptacles and accessories, e.g., equipment to remove and store cellular samples, as *in vitro* diagnostic medical devices.

Performance

Those genetic analysis in genetic screening that fall within the scope of the IVDMD Directive and do not fall under the in-house test exemption ought to comply with the harmonized requirements enshrined in the IVDMD Directive;[299] these requirements are transposed at the national level. They must correspond to the series of essential requirements set out in the IVDMD Directive.[300] The essential requirements mandate that the genetic analysis does not compromise the clinical condition or safety of the patients, users and other persons.[301] Further, they require that the tests are suitable for their intended purpose, as specified by the manufacturer, taking into account the generally acknowledged state of the art. The analysis must be able to achieve the performances stated by the manufacturer, in particular, analytical sensitivity, diagnostic sensitivity, analytical specificity, and diagnostic specificity.[302] As has been highlighted by scholars, despite the common usage of analytical and diagnostic sensitivity and analytical and diagnostic specificity, European Commission has defined them in a way, which renders all four regulated criteria to ensure the analytical validity of the regulated devices, including genetic analysis constituting genetic screening.[303] That is, the IVDMD Directive explicitly requires demonstrating the genetic analysis" probability of the results correlation with the targeted sequence. Potentially, clinical validity can be related to the test's ability to meet the requirements by the manufacturer, but it is not explicitly further addressed in the IVDMD Directive. The same can be said about the utility of the genetic analysis.[304] Therefore, it could be argued that it is rather ambiguous how effectively the IVDMD Directive addresses clinical validity and utility of the genetic analysis.

The IVDMD Directive is designed to ensure that genetic analysis, which is subjected to its requirements, fulfils the analytical capabilities assigned to it by the manufacturer.[305] However, by way of derogation from the general rule, European Commission is entitled to adopt common technical specifications to ensure

[297] Slokenberga, op. cit. 295.
[298] IVDMD Directive , op.cit., Article1.2 (b).
[299] IVDMD Directive, op.cit., Article 2.
[300] IVDMD Directive, op.cit., Article 3, Annex I, section A 3.
[301] IVDMD Directive, op.cit., Annex I, section A 1.
[302] IVDMD Directive, op.cit., Annex I, section A 3.
[303] Hogarth, Stuart, David Barton and David Melzer, "The European IVD Directive and Genetic Testing", in Ulf Kristoffersson,Jörg Schmidtke,J. J. Cassiman, *Quality Issues in Clinical Genetic Services*, Springer, 2010.
[304] This section is based on Slokenberga, op. cit.295.
[305] IVDMD Directive, op.cit., Annex I A.

conformity with the essential requirements (of analytical and diagnostic sensitivity) for those genetic analyses that are listed in List A and List B of Annex II of the IVDMD Directive. If genetic analysis is capable of meeting the relevant requirements, it can be deemed to comply with the specified essential requirements of the IVDMD Directive.[306]

Further to the general requirements, the genetic analysis that is subjected to the IVDMD Directive requirements needs to undergo the conformity assessment procedure to ensure that the respective devices meet the provisions of the IVDMD Directive, which apply to them. The requirements of the conformity assessment depend on the type of genetic analysis in question. For the genetic analysis, which is not enclosed in Annex II, the conformity assessment procedure is carried out by the manufacturer and is not controlled by a notified body. It involves certifying that a genetic analysis in question meets the essential requirements and requires the manufacturer or their authorized representative filling out the EC conformity declaration.[307] For the genetic analysis falling in Annex II of the IVDMD Directive the notified body needs to be involved to ensure that the test meets the relevant requirements of the directive, thus, analytical validity and, arguably, clinical validity.[308] As has been argued elsewhere, these assessments, however, are limited to determining whether the genetic analysis in question meets the requirements outlined in the IVDMD Directive. Thus, while analytical validity is controlled by the involvement of a notified body, it is questionable whether clinical validity and utility are subjected to the notified body's scrutiny. Once the scrutiny is completed, the CE mark can be affixed which makes the genetic analysis lawful in the market.[309] In practice, however, this means that a limited range of genetic analysis is subjected to higher scrutiny, which could have implications for quality.

Application of genetic analysis

The IVDMD Directive in its recital 33 makes a reference to the CoE BMC and requires that "in view of the need to protect the integrity of the human person during the sampling, collection and use of substances derived from the human body (..) whereas, furthermore, national regulations relating to ethics continue to apply." As has been analysed above in chapter 4, under the CoE BMC informed consent is a requirement, whereas in the case of a person being unable to consent, authorization by the competent person. Moreover, genetic counselling is being required.[310] The necessity for informed consent also stems from Article 3 CFREU. Article 3.1 CFREU sets forth a rather general statement that affords to everyone the right to respect for their physical and mental integrity. Article 3.2 CFREU specifically stipulates that "[i]n the fields of medicine and biology (..) the free and informed consent of the person concerned, according to the procedures laid down by law" shall be respected.[311] Thus, in so far as genetic screening relates to genetic analysis regulates under the IVDMD Directive, respective CoE BMC should be followed, which is to be done disregarding whether a particular EU Member State has ratified it.

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[306] See IVDMD Directive, op.cit., Article 5.3 and Recital 17.

[307] See IVDMD Directive, op.cit., Article 9.1 and Annex III.

[308] See IVDMD Directive, op.cit., Article 9 and 9.4

[309] IVDMD Directive, op.cit., Article 9.

[310] See Article 12 of CoE, Convention for the protection of Human Rights and Dignity of the Human Being with regard to the Application of Biology and Medicine: Convention on Human Rights and Biomedicine, ETS 164, that states "[t]ests which are predictive of genetic diseases or which serve either to identify the subject as a carrier of a gene responsible for a disease or to detect a genetic predisposition or susceptibility to a disease may be performed only for health purposes or for scientific research linked to health purposes, and subject to appropriate genetic counselling."

[311] Respect for consent has been endorsed by the CJEU before CFREU entered into force; see, for example, CJEU, Case C-377/98, *Kingdom of the Netherlands v European Parliament and Council of the European Union,* 09 October 2001.

5.3.3.2 Considerations regarding IVDMD Regulation

The scope of application of IVDMD Regulation

IVDMD Regulation applies to *in vitro* medical devices and their accessories. Under the IVDMD Regulation genetic analysis that is covered are those solely or principally for the purpose of providing information: concerning a physiological or pathological state or concerning a congenital abnormality, or to determine the safety and compatibility with potential recipients, or to monitor therapeutic measures.[312] Following Article 5.4, a genetic analysis that is manufactured and used within health institutions, except devices for performance studies, shall be considered as having been put into service and therefore subject to the requirements of the IVDMD Regulation. Nonetheless, based on Article 5.5 exceptional requirements to in house genetic analysis can be applied.[313] In house, genetic analysis requirements are not intended to apply to genetic analysis that is manufactured on an industrial scale.

Performance

In relation to quality of genetic analysis in the course of genetic screening, the IVDMD Regulation maintains the approach that the IVDMD Directive had established, but sets forth more stringent quality requirements. In order that a genetic analysis may be placed on the market or put into service, it shall comply with the requirements of the IVDMD Regulation.[314] As derives from Article 5.2 and 5.3 IVDMD Regulation, these requirements include the applicable general safety and performance requirements as outlined in Annex I of the IVDMD Regulation, demonstrated by a clinical evidence and performance evaluation report. Further, technical documentation needs to be drafted to demonstrate the conformity with the requirements of the IVDMD Regulation and an EU declaration of conformity needs to take place. Once that is completed, the device can be made available to consumers.

Under the IVDMD Regulation, a general principle with regard to the general safety and performance requirements of genetic analysis is that they are selected by the manufacturer and ensure that the device meets its intended purpose.[315] However, harmonized standards can be adopted. If the genetic analysis in question is manufactured in line with the harmonized standards, it is presumed to meet the relevant general safety and performance requirements.[316] Where the harmonized standards do not exist, or they are not sufficient – or where it is necessary to address public health concerns – the European Commission is empowered to set forth the common specifications,[317] therefore standardizing requirements for particular genetic analysis across the internal market. If the European Commission adopts common specifications, the manufacturers have to comply with them unless they can duly justify the decision not to comply with these specifications.[318] To the extent an analysis meets these requirements, it is presumed to comply with the general safety and performance requirements mandated by the IVDMD Regulation;[319] consequently, it can benefit from free movement within the internal market.

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[312] European Parliament and of the Council, Regulation 2017/746 of 5 April 2017 on *in vitro* diagnostic medical devices and repealing Directive 98/79/EC and Commission Decision 2010/227/EU OJ L 117, 5.5.2017, p. 176–332, Article 1.1 and 2(2).

[313] IVDMD Regulation, op.cit., Article 5.5.

[314] IVDMD Regulation, op.cit., Article 5.1.

[315] IVDMD Regulation, op.cit., Annex I 1 .

[316] IVDMD Regulation, op.cit., Article 8.1 .

[317] IVDMD Regulation, op.cit., Article 9.

[318] IVDMD Regulation, op.cit., Article 9.3.

[319] IVDMD Regulation, op.cit., Article 9.2.

Once the requirements are met, the manufacturer must undertake a performance evaluation and conformity assessment procedure that seeks to ensure that the genetic analysis conforms to the requirements that the IVDMD Regulation provides.[320] Generally, the IVDMD Regulation sets forth alternative quality verification mechanisms, which entail an assessment performed by a notified body of a sample of the device and its technical documentation before the tests are sold to consumers.[321] Among requirements that are assessed are the general safety and performance requirements that require support from relevant scientific validity and analytical and clinical performance data of the genetic analysis in question.[322] While the IVDMD Regulation does not set a particular threshold to these criteria, it requires that the characteristics assigned to genetic analysis are duly motivated, and thus transparent. Therefore, while a certain level of quality is not guaranteed, transparency is being required. Having undergone the conformity assessment, the manufacturer may draft its EU declaration of conformity and affix the CE mark to the testing.[323] Consequently, the test can benefit from free movement within the internal market, such that the Member States under Article 21 of the IVDMD Regulation are prohibited from putting obstacles to the free movement of the genetic analysis.

Application of genetic analysis

The IVDMD Regulation takes a different approach with regards to protecting fundamental rights in the application of IVDMD Regulation, including carrying out genetic screening activities. In line with the evolution of EU law, instead of CoE instruments, a reference to the CFREU is made. Recital 89 indicates that ”[t]his Regulation respects the fundamental rights and observes the principles recognised in particular by the Charter and in particular human dignity, the integrity of the person, the protection of personal data, the freedom of art and science, the freedom to conduct business and the right to property.” However, simultaneously, following Article 1.9 the IVDMD Regulation is not intended to ”affect national law concerning the organisation, delivery or financing of health services and medical care, such as the requirement that certain devices may only be supplied on a medical prescription, the requirement that only certain health professionals or health care institutions may dispense or use certain devices or that their use be accompanied by specific professional counselling.”

Article 4 IVDMD Regulation addresses genetic information, counselling and informed consent. Article 4.1 requires that ”Member States shall ensure that where a genetic test is used on individuals, in the context of healthcare as defined in point (a) of Article 3 of Directive 2011/24/EU of the European Parliament and of the Council (1) and for the medical purposes of diagnostics, improvement of treatment, predictive or prenatal testing, the individual being tested or, where applicable, his or her legally designated representative is provided with relevant information on the nature, the significance and the implications of the genetic test, as appropriate.”

Furthermore, except for the cases when a patient is already informed of the genetic condition, genetic counselling is required under Article 4.2 IVDMD Regulation.[324] The Member States, however, are not precluded ”from adopting or maintaining measures at national level which are more protective of

[320] Article 10.2-10.5, detailed requirements for performance evaluation can be found in Article 56.

[321] Slokenberga, op. cit. 294.

[322] See ANNEX II and Annex XIII 1.1. For genetic testing, following Annex VIII, 2.3 Rule 3 (i) requirements “class C” applies. The same requirements apply, for example, for screening for cognitional disorders in the embryo. Following Article 9.3 IVDMD Regulation, scientific validity, analytical performance and clinical performance shall be demonstrated.

[323] IVDMD Regulation, op.cit., Article 18.

[324] See IVDMD Regulation, op.cit., Article 4.3 IVDMD.

patients, more specific or which deal with informed consent".[325] This means, when organizing genetic screening with genetic analysis regulated under IVDMD Regulation, not only consent is crucial, but also appropriate counselling.

As derives from the above, the IVDMD Regulation will alter the situation for genetic testing, setting forth more stringent requirements. These requirements, however, apply to a limited range of genetic analysis, that which is health-related, leaving others beyond the scope of the harmonized requirements. Substantively, however, these requirements relate to the oversight and transparency, rather than performance thresholds, which is in line with the EU approach for regulating products within the internal market. One could further examine in SIENNA task 4.2 whether and how the EU could further regulate the area, addressing, for example, quality of non-health related genetic testing in a more comprehensive manner.

5.3.5 Genetic analysis and cross border healthcare

Cross-border Healthcare Directive provides rules for facilitating the access to safe and high-quality cross-border healthcare and promotes cooperation on healthcare between the Member States, in full respect of national competencies in organising and delivering healthcare.[326] While public vaccination programmes are exempted from the scope of the directive, genetic screening and testing are not. Therefore, in so far as genetic screening and testing can be defined as healthcare under the directive,[327] they can be subject to cross-border healthcare. This means that, unless a Member State of affiliation applies exceptions set forth in Article 7.9 of the directive, a patient may choose to receive the screening in a different EU country than the one organizing it. Once could, however, question why genetic screening is being treated differently than vaccination. Furthermore, they can also be subject of benefits in kind under the Social Security Coordination Regulation.

5.3.6 Fundamental rights considerations

In terms of fundamental rights as protected under CFREU, genetic screening and testing predominantly could trigger the protection of the right to private life (Article 7), data protection (Article 8), integrity (Article 3), as well as health (Article 35). A number of other rights, depending on circumstances, could also be relevant, including freedom of sciences (Article 12), and freedom of thought, conscience, and religion (Article 10).

5.4 Specific considerations regarding prenatal screening and relevant fundamental rights considerations

Further to the review of IVDMD Directive and IVDMD Regulation above, it shall be noted that under the former increased requirements apply to evaluate the risk of trisomy as it is listed in Annex II, List B.[328] Under the IVDMD Regulation, devices for screening for congenital disorders in the embryo or foetus are subject to class C requirements under Rule 3 (l), and follow overall the same requirements examined above.

[325]IVDMD Regulation, op.cit., Article 4.4 IVDMD.

[326] European Parliament and the Council, Directive 2011/24/EU of 9 March 2011 on the application of patients' rights in cross-border healthcare, OJ L 88, 4.4.2011, p. 45–65, Article 1.1.

[327] Under Article 3.a ""healthcare" means health services provided by health professionals to patients to assess, maintain or restore their state of health, including the prescription, dispen- sation and provision of medicinal products and medical devices."

[328] IVDMD Directive, op.cit, Annex II, List B.

GDPR applies to the protection of personal data of natural persons as specified in Article 4.1 GDPR and does not in any specific way expressly address protection of the embryo's or foetus"s data. However, it simultaneously fails to explain what a natural person within the meaning of GDPR is.[329] As noted by Pormeister and Drozdzowski, previously under the Data Protection Directive Article 29 WP had adopted opinion to clearly exclude the unborn from the scope of the Directive. However, a similar approach vis-à-vis GDPR has not been taken yet. According to them, also the definition of a child as provided in Article 4(18) GDPR and relates to Recital 29, does not bring in much clarity. Therefore, this question is a risk where divergent national laws could apply.[330]

In terms of fundamental rights as protected under CFREU, prenatal screening could predominantly trigger the protection of right to private life (Article 7), data protection (Article 8), integrity (Article 3), life (Article 2), prohibition of torture and inhuman or degrading treatment or punishment (Article 4), as well as health (Article 35) of the pregnant woman. A number of other rights, depending on circumstances, could also be relevant, including freedom of sciences (Article 12), and freedom of thought, conscience, and religion (Article 10). The CFREU does not specifically address the protection before birth, however, in so far as it could be applicable, a number of protections could potentially apply.

5.5 Specific considerations on new-born screening and relevant fundamental rights considerations

Under the IVDMD Regulation, devices for screening for congenital disorders in new-born babies where failure to detect and treat such situations could lead to life-threatening situations or severe disabilities are subject to class C requirements under Rule 3 (m) and follow overall the same requirements examined above. Furthermore, an implementation report on the EU Commission Communication on Rare Diseases states that "a report on the practices of new-born screening for rare disorders implemented in all the EU Member States including the number of centres, an estimation of the number of infants screened and the number of disorders included in the new-born screening as well as reasons for the selection of these disorders (..). On the basis of this report, the EU Committee of experts on rare diseases adopted an opinion on potential areas of European collaboration in the field of new-born screening", emphasizing the need of a targeted policy on new-born screening across EU Member States.[331]

On an EU level, the GDPR, specifically refers to genetic data of individuals (this includes new-borns as there is no limitation to the age of the data subject) as a special category of data which needs to be processed under state-of-the-art technical measures and safeguards.

In terms of fundamental rights as protected under CFREU, new-born screening could trigger the protection of the right to private life (Article 7), data protection (Article 8), integrity (Article 3), as well as health (Article 35). A number of other rights, depending on circumstances, could also be relevant.

5.6 Direct-to-consumer advertising of genetic testing and relevant fundamental rights considerations

Neither the IVDMD Directive nor IVDMD Regulation specifically addresses direct-to-consumer advertising of genetic testing. However, for example, the IVDMD Regulation leaves the discretion with the Member

[329] It does not apply to the protection of data of legal persons, GDPR, op.cit., recital 14.

[330] Kärt Pormeister, Łukasz Drożdżowski, Protecting the Genetic Data of Unborn Children: A Critical Analysis, European Data Protection Law Review, 4 (2018) 1, p.53.

[331] European Commission, Implementation report on the Commission Communication on Rare Diseases: Europe's challenges [COM(2008) 679 final] and Council Recommendation of 8 June 2009 on an action in the field of rare diseases (2009/C 151/02), 5.9.2014, COM(2014) 548 final.

States regarding prescription of certain *in vitro* diagnostic medical devices.[332] Furthermore, both, the IVDMD Directive and the IVDMD Regulation address labelling requirements.

Unfair Commercial Practices Directive prohibits misleading actions in advertising [333] and sets forth general rules that shall be observed, however, does not restrict direct-to-consumer advertising of genetic testing as such.

In terms of fundamental rights as protected under CFREU, direct-to-consumer advertising of genetic testing could trigger the protection of the right to private life (Article 7), integrity (Article 3), as well as health (Article 35). A number of other rights, depending on circumstances, could also be relevant.

6. Analysis of relevant national laws and human rights standards

6.1 Introduction, note on methodology and scope of the analysis

This section surveys the national legal responses in the area of genomics. It relates to the six guiding questions provided in Chapter 2.2, and additionally it considers information that the partners have provided as being current in their national legal order. With due regard to limitations outlined in Chapter 1.2, this Chapter takes a comparative approach and highlights issues that are at the forefront of the national legal academic debates, as well as legal developments, and marks commonalities and differences in how the law currently responds to the issues of concern. This consists of drawing attention to a particular approach a particular state has taken to regulate a specific concern in the area of genomics, without implying any meaning for the other legal orders that are reviewed.

As this section is based on national reports that are provided by SIENNA partners annexed to this report, when a particular state is mentioned in this Chapter, the respective national report should be consulted for obtaining supporting information for those statements. Therefore, in so far as possible, further references in this Chapter are omitted. The exceptions include situations when: a) it is believed that a reference facilitates analysis; b) additional complementary information is given; c) information is included in this study after the completion of the national report.

6.2 Summary of current legal academic debates in the area of genetics and genomics

6.2.1 Human germline gene editing

In most of the surveyed states (for example, Germany, France, the Netherlands, Poland, Spain, Sweden and the UK, South Africa, China, the US, and Brazil), the question of germline gene editing has received some attention in the academic legal debates, with the exception being Greece. The issues that are at the forefront in several states concern an adequate regulatory mechanism for human germline gene editing and the most appropriate ways to address it. In this regard, for example, Chinese scholars have discussed genetic self-determination, which in China has legal status and violation of human rights of future

[332] IVDMD Regulation, op.cit., Article 1.9.

[333] European Parliament and the Council, Directive 2005/29/EC of 11 May 2005 concerning unfair business-to-consumer commercial practices in the internal market and amending Council Directive 84/450/EEC, Directives 97/7/EC, 98/27/EC and 2002/65/EC of the European Parliament and of the Council and Regulation (EC) No 2006/2004 of the European Parliament and of the Council, OJ L 149, 11.6.2005, p. 22–39, Article 6. See also Kalokairinou et al., op.cit. 2.

children. The Brazilian scholars have considered more generally what challenges CRISPR-Cas9 poses,[334] and also focused on the enhancement concerns.[335]

In Germany, Poland, Sweden, France, and South Africa scholars have discussed the ambiguity and effectiveness of the existing legal frameworks. For example, in France the specific concern is whether the application of gene editing technology on embryos for research purposes is covered under the current bans relevant for France and whether the prohibitions are effectively secured through sanctions. In Sweden the scope of the existing legal framework has appeared to be a key concern as there is a risk that some human germline editing technologies could fall outside the scope of application of the key act regulating the field.

In countries that take a somewhat restrictive approach to embryo involvement in research, discussions regarding the necessity and appropriateness to revisit the framework have been had. In Germany, for example, calls to enable research have been made, in the Netherlands, discussions have clustered regarding the moral difference between creating embryos for research and using surplus embryos for research, and whether the prohibitions for the former should not be lifted to enable research. In France, discussions have emerged with a view to permitting creating transgenic embryos for research purposes. In the UK, however, the 14-day upper limit for using embryos for research has been debated, in particular, arguments regarding the arbitrarily drawn lines between the permissible and the forbidden have been made. Arguably, it has been done with a view to reviewing and extending it.

In terms of specific regulatory responses, scholars in Germany have called for a moratorium for germline gene editing experiments in humans. Such calls have also been made in France with a view to evaluating the technology and consider the societal consequences, disregarding that the use of germline gene editing technologies is already prohibited in France.

Among the countries that raise concerns over the appropriateness of the current regulatory approach towards human germline gene editing in humans, opinions split, whether it should be entirely prohibited, or exceptions could be made. While in France support has been expressed for Article 13 of the CoE BMC in its entirety, it has also been suggested that case-by-case exceptions could be supported for a restricted number of genetic diseases and that a European steering committee should be put in place in that regard. In Germany, scholars have argued that once the technology is deemed to be safe to be applied to humans, a distinction regarding therapeutic and enhancement purposes needs to be made. Disregarding the current bans in Poland, some voices have been raised that the Polish Constitution could be permissible, provided that the intervention has a therapeutic purpose.

Scholars in Spain have identified a diversity of regulatory responses as a key governance challenge and have considered the need for harmonization on this question. Scholars in Germany have suggested that the use of model laws could be a way forward.

6.2.2 Genetic screening

Most of the national legal orders, including Brazil, France, Greece, Germany, Sweden, and the UK have reported that questions related to genetic screening are not at the forefront of the scholarly discussions.

[334] Marcelo de Araujo, "Brasil e o genoma humano, discussões sobre o CRISPR-Cas9", *São Leopoldo*, 2016, 489, p.13.

[335] Marcelo de Araujo, "Editing the Genome of Human Beings: CRISPR-Cas9 and the Ethics of Genetic Enhancement", *Journal of Evolution and Technology*, Vol. 27, 1, 2017 , p. 24.

Regarding Poland, it has been noted that genetic screening and genetic testing are often addressed interchangeably, and scholars have emphasised the need for specific regulations in these areas. In South Africa, a discussion of "genomic sovereignty" has emerged, and concerns over exploitation of the South African population in the absence of a clear regulatory framework for screening activities and benefit-sharing requirements have been raised. In China, at the forefront are discussions relating to genetic discrimination and protection in that regard.

In the Netherlands, the medical community has scrutinized the ability of the existing laws to manage or prevent undesirable developments in the field without unduly blocking innovation. The boundaries between the prevention of undesirable developments and the facilitation of innovation in genetic screening and diagnosis are becoming increasingly blurred. Scholars in Spain have been concerned about the use of genetic screening for other purposes than healthcare, for example, in the workplace or a legal trial.

6.3.3 Genetic testing

In different states, genetic testing has been subject to academic legal scrutiny to differing degrees. In South Africa, scholars have raised concerns over the extent to which genetic testing infringes the common law and constitutional law rights to privacy. Genetic tests can also have implications for other rights, including, dignity and bodily integrity. Currently, there is little consensus on how these rights could affect the availability and use of genetic testing. Similarly, privacy along with scientific advances in the area of genetic testing has also been subject to discussions in China. The right not to know has been highlighted as yet fully un-discussed dimension of privacy in the Chinese context; this situation, however, is gradually improving. Spanish scholars have debated the impact of genetic testing on constitutional rights; however, with a particular focus on genetic tests offered directly to consumers.

Brazil has considered genetic testing in light of eugenics. The word "eugenics" (*Port.: eugenia*) has a derogatory connotation in Portuguese as Brazil pursued "eugenic policies" in the first half of the twentieth century in the attempt to increase the number of white people in the population.[336] For this reason, the word "eugenics" is still associated with the deliberate attempt to change the phenotypic profile of the population so as to favour human features that some people (out of prejudice) consider better than other features (having white skin rather than black skin, having blue eyes rather than dark eyes, etc.).

Poland has raised a concern about testing without appropriate information and genetic counselling. Questions of regulating secondary findings have been of particular interest. In France, scholars have highlighted concern over the limited rights given to family members concerning genetic testing carried out for their relatives; in particular, it can be said that their right to know could be perceived as the duty to know as the family members do not have a right to refuse the information. In that way, French scholars point out the lack of balance between the rights of the tested persons and the rights of their family members. In the Netherlands, discussions about the familial nature of genetic information have been raised concerning the duty of the caregiver, and confidentiality obligation. Confidentiality and data protection have also been concerns that have been discussed in Sweden, in particular, considering whether genetic information should merit special protection.

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[336] Stepan, Nancy L., "Eugenics in Brazil, 1917-1940" in Mark, B. Adams, *The wellborn science. Eugenics in Germany, France, Brazil, and Russia,* Oxford U. Press, New York/Oxford, pp.110–152. See also Hochman, Gilberto, Nísia Trindade Lima and Marcos Chor Maio, "The Path of Eugenics in Brazil: Dilemmas of Miscegenation", *The Oxford handbook of the history of eugenics,* 2010, pp. 493. Walsh, Sarah, "The Executioner's Shadow: Coerced Sterilization and the Creation of "Latin" Eugenics in Chile", *History of Science,* 2018. 0073275318755533.

France has also raised concerns over the application of the law in light of advances in genetics and genomics. According to the French national law, the person must be informed of the risk of identifying genetic characteristics unrelated to the prescription. It is unclear what is to be done with data that have an uncertain meaning at the time of the diagnostic intervention, but that may make sense later as the science develops. Moreover, the scope of information has been of concern concerning an informed decision to exercise the right to know or not to know. Although the Netherlands has taken a different approach on the matter, in the literature it is stated that it is rightly assumed to be reluctant towards a specific warning obligation, because these obligations generally put pressure on caregivers. Currently, in the Netherlands, there is some ambiguity regarding the patient's secret, on the one hand, and sharing information about genetic test results with family members, on the other hand.

In Germany, discussions regarding revisions of the national law to respond to the advances in the area of genetic testing have been at the forefront. The Netherlands and Poland are discussing the need for specific legislation that would address genetic testing. In France, in light of the current national prohibitions in the area of genomics, discussions have been raised regarding recognizing the right of each person to have access to the knowledge of his/her DNA. It has also been highlighted that the current criminal prohibition against conducting genetic testing beyond any medical context should be lifted. Also, Spanish scholars have discussed that the national legal framework for accessing genetic testing is somewhat restrictive, as genetic tests have to be done for a medical aim, which precludes obtaining genetic testing for curiosity only. However, contrary to the French scholars, the Spanish scholars note a reluctance to liberalize their frameworks. Some other countries, for example, Germany discuss the use of DNA samples in criminal investigations and revisiting the national laws in that regard.

6.3.4 Prenatal testing

Neither in Brazil nor the UK and Spain, have questions regarding prenatal testing/screening been at the forefront of legal scholarly discussions.

In Poland, discussions have emerged about the cases in the area of reproductive rights that have been brought before the ECtHR. In addition to these discussions, it has been discussed that the opportunity to choose an embryo without a severe genetic defect allows the avoidance of mental, physical, medical and moral issues regarding the termination of a pregnancy after prenatal diagnosis. In Poland, it is believed that PGD should not be permitted for reasons other than medical ones. Therefore, PGD to determine gender or other physical traits; testing for HLA with a view to creating a saviour sibling or testing for the diagnosis of a disease that appears later in life (like Alzheimer's) should be precluded. More generally, discussions concerning granting access to the achievements of modern reproductive medicine and a high level of medical care, health safety, and effective medically-assisted procreation treatments have emerged. PGD has also been topical in Greece. Scholars have noted the fact that more stringent conditions have been placed for accessing PDG than for obtaining the legal termination of pregnancy.

Scholars in France have raised concerns that the generalization of high throughput sequencing leads the society closer to the possibility for parents to choose babies of the highest "quality" possible. In South Africa, an important area of debate concerns the extent to which the constitutional right to health care services includes the right to prenatal testing/ screening. Implicit in the view that such testing/ screening is indeed covered under the constitutional law.

In China, as well as in Sweden, discussions have focused on NIPT. While in China it can be seen as a controversy, in Sweden NIPT is being applied in practice, and scholars are concerned with equal access to

the technology in different regions in Sweden due to the peculiarities of how healthcare is organized nationally.

In Greece and South Africa, questions of wrongful life and wrongful birth have been of concern. The Constitutional Court of South Africa in the case of *H v. Fetal Assesment Centre*[337] faced a question regarding whether a child can claim damages in a so-called "wrongful life" situations. South African law recognizes that a parent can claim damages from a doctor when the doctor performs prenatal testing negligently, consequently fails to inform the parents that their child will be born with a particular condition and because of this misinformation the parents choose to carry the child, and s/he is born with the impugned condition. However, at issue in *H v. Fetal Assesment Centre* was whether the child has any claim in such circumstances. Until *H* the position in South African law was that the child had no such claim. However, in *H,* the Court held that in such circumstances, and where the parents fail to bring a claim against the doctor, the child has prima facie claim against the doctor for damages.[338] Additionally, the question of child's best interests has been debated widely and is at the forefront of the academic debates. This principle extends to newborn screening, and therefore, permits it only if the intended screening meets the best interests requirements.

In Greece, however, particular concern has been whether the purported father is entitled to sue for wrongful birth, along with the mother or not have been at the forefront of the scholarly discussions. The prevailing legal theory along with jurisprudence accepts the purported father's right to sue, based on the violation also of his personality, when the defective prenatal testing also disallowed him, along with the mother, from making an informed decision relevant to the continuation of pregnancy. The opposite view sees the right to informed consent in prenatal screening and testing as only belonging to the mother, as well as the liberty to terminate a pregnancy under the national law, and therefore, fails to see any legal interest of the purported father being involved in the law of prenatal screening/testing. Scholars highlight that some courts have deduced the purported father's right from an article of the Greek Civil Code mandating that all family decisions should be taken by both married spouses, disregarding that for a legal termination of pregnancy, only the consent of the pregnant woman is necessary, therefore the decision to terminate is seen as a common decision, of which both were deprived by "defective" prenatal testing/screening.

6.3.5 Newborn screening

In some states (for example, France, Brazil, Greece, South Africa, the Netherlands, and Spain) new-born screening has not been at the forefront of the academic legal debates.

In Poland, scholars have emphasized the need for genetic screening to detect rare diseases, which should be introduced as a priority. In Sweden, scholars have raised legal issues relating to research regarding the newborn data and samples.

[337] H. v. Fetal Assesment Centre 2015 (2) BCLR 127 (CC).

[338] The basis of the ruling was that where the parents fail to bring a claim, it is in the best interests of the child to allow the child to bring such a claim. The Court explained that in such circumstances the child was harmed not by being born but by the parent's failure to bring a claim. Since the impugned doctor is a factual cause of the parent's failure to bring a claim the Court was able to hold that, prima facie, the child has a claim against the doctor. Of course, since the Court was only required to decide whether the child had a prima facie claim, it did not have to resolve whether the doctor could appropriately be regarded as the legal cause of the harm sustained by the child.

In Germany, scholars criticise the legal framework for newborn screening. According to the current law, a blood sample from a newborn can be taken only after parents give their informed consent, which means after receiving genetic counselling. However, paediatric nurses and midwives, who usually took the blood samples from newborns, are not allowed to provide genetic counselling. Therefore, they are strictly speaking not allowed to take the blood sample. However, there are indications that this approach is leading to the new-born screening not being carried out for some new-born babies, which can have severe consequences, in particular, life-long disability, which could have been avoided with early diagnosis and appropriate treatment.

In the UK, there is a debate about whether the genetic profiling of babies at birth should be permitted in the public health context. Although it may have significant benefits for health and research, there are several legal issues to be examined, such as privacy concerns and sharing genetic information, the storage and legal ownership of genetic material, patent issues, consent and reporting requirements, and safeguarding the autonomy of the future adult. It has been suggested that new-born genetic screening should not apply generally, but it should be available and accessible based on its clinical utility and cost-effectiveness and the best interests of children.

6.3.6 Direct-to-consumer advertising of genetic testing

Some countries, for example, Brazil, South Africa, Netherlands, and Spain, do not have considerable legal debates regarding advertising of genetic testing. On the other hand, other countries, for example, the UK, Germany, Greece, Poland, South Africa, and China have debated questions that relate to direct-to-consumer advertising of genetic testing.

Specifically, regarding China, it has been noted among scholars that many people are moved by the promise in some advertisements that genetic testing can “predict the future” or even “cure all diseases.” There is awareness, however, that the commercial gene testing on the market has many problems, such as confusion of qualifications, false propaganda, and no fee standard. In the view of the Chinese scholars, a gap between expectations and current regulatory requirements has emerged.

Scholars in France and Poland have raised general concerns about the accessibility of genetic testing over the internet and difficult to regulate them; scholars in Poland have highlighted the genetic testing practices advertised directly-to-consumers that are contrary to the applicable standards and ethical requirements. Practicing lawyers in Sweden have published advice on advertising medical devices, which also includes genetic testing. German scholars have compared direct-to-consumer advertising with the regulation of prescription drugs and have argued that there are good reasons for some prohibitions in the area. Scholars in Greece have highlighted a need for a coherent regulatory framework. Scholars in China have pointed at the legal controversy of gene sequencing technology, which mainly includes the technical norms of risks and uncertainties brought by the technology itself, as well as legal issues such as informed consent, privacy and patent rights of genes. Scholars in Poland have considered ratification of the CoE BMC and CoE APTG as means to resolve challenges relating to direct-to-consumer advertising.

The UK scholars have raised concerns over the terms of service of direct-to-consumer genetic tests, in particular, that they are lengthy, they use obscure and vague terminology, which is difficult to understand for the average service-user. It has also been regarded as problematic that these tests are offered in a take-it-or-leave-it form, and they fail to provide adequate information in clear and understandable language to enable consumers to make an informed choice. However, even if the necessary information is disseminated, it is usually in vast amount, vague, obscure and complex. Scholars in the UK agree that direct-to-consumer advertising needs to be regulated, focusing on the need for informing about the

potential risks and limitations of genetic testing, the utility, and credibility of genetic testing as well as an agreement on restricting this advertising when directed to children.

6.3 Comparative analysis of legal developments in the area of genetics and genomics

6.3.1 Human germline gene editing

In most of the surveyed EU Member States, similarly to non-EU states that have been surveyed, there are legal developments in the field. However, in most cases, these developments are constrained to legal and policy discussions on the existing legal frameworks and challenges. The exceptions are China, Greece, Poland, and Spain, which do not have significant legal developments in the field. Nonetheless, the EU countries uphold bans stemming from the EU law, and Greece and Spain – also those stemming from the CoE BMC, whereas China revisited its law in 2017 to regulate experimental clinical medical treatment. In Brazil, advances have focused on responding to CRISPR challenges, though in the field of agriculture and livestock raising;[339] it remains to be seen whether the legislature will initiate any works regarding human germline gene editing.[340]

In Germany, some attention to the challenges in the existing laws has been given, in particular, that the law does not explicitly prohibit gene therapy or gene editing on an embryo that aims to enable its survival or serves for its health. Likewise, ambiguities have emerged regarding the permissibility of therapeutic cloning – whether it is prohibited or not under the national laws. Also, the scope of application of sanctions for violating prohibitions outlined in the German law is considered, specifically, regarding research on arrested embryos.

In France, considerable work is being done to revise the existing national legal framework, which is also a statutory requirement. The emergence of new technologies, including CRISPR-Cas9, is expected to be accounted for in these revisions. Furthermore, the transfer of mitochondrial DNA could also be expected to be considered in this revision.

In the Netherlands, a temporary prohibition on the creation of embryos for research is in place. Previously, considerations had been made to enable the creation of embryos for research under strict accounts laid down in law. These were not passed as a law before the ministerial change, and the minister who entered in office in 2017 withdrew the proposal. Instead, alternative possibilities for research in gene editing should be considered, for example, the use of (induced) pluripotent stem cells.

In South Africa, questions relating to human germline are mainly addressed through soft law measures, which are expected to guide the regulations of the human germline editing.

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[339] Ledford, Heidi, "Gene-edited animal creators look beyond US market", Nature, Vol. 566, 2019, p. 433.
de Almeida Regitano, Luciana Correia, "Cattle genes", Pesquisa, Vol. 254, 2017.

[340] National Technical Commission of Biosafety – CTNBio of Brazil, Normative Resolution No. 16, of January 15, 2018, http://ctnbio.mcti.gov.br/resolucoes-normativas/-/asset_publisher/OgW431Rs9dQ6/content/resolucao-normativa-n%C2%BA-16-de-15-de-janeiro-de-2018;jsessionid=0DC3D2823FBBA6DE845927FE0B754BDD.rima?redirect=http%3A%2F%2Fctnbio.mcti.gov.br%2Fresolucoes-normativas%3Bjsessionid%3D0DC3D2823FBBA6DE845927FE0B754BDD.rima%3Fp_p_id%3D101_INSTANCE_OgW431Rs9dQ6%26p_p_lifecycle%3D0%26p_p_state%3Dnormal%26p_p_mode%3Dview%26p_p_col_id%3Dcolumn-2%26p_p_col_count%3D3.

In Sweden, various authorities have actively been debating the issue of human germline gene editing, technological advances, and whether and how the national legal framework can accommodate them. Furthermore, it has been discussed whether the national law still sets an adequate balance between various rights, interests, and values at stake in human germline editing. Specific considerations to mitochondrial transfer for serious inheritable diseases and the gene editing question vis-à-vis PGD have been made.

In the UK, potential obstacles for innovation, including genome editing, and possible legislative amendments are at the forefront. While some have argued that law strikes a good balance in regulating gene editing technology, others have pointed out that gene editing technologies may have considerable potential to be applied in a clinical context. Recently, the Nuffield Council on Bioethics has stated that human germline modification could be ethically acceptable in some circumstances, namely under the guiding principles of solidarity, social justice, and welfare, so that the person born under this procedure will not suffer from negative consequences such as discrimination. Nonetheless, any legislative change regarding heritable genome editing interventions is not currently urgent, and any amendment should follow public engagement and dialogue and the adoption of monitoring and safety mechanisms. Nonetheless, recently, the UK Government has clarified that there are no plans to amend the law to allow germline modification.

6.3.2 Genetic screening

Most of the reviewed national legal orders, including Brazil, France, Greece, Germany, Sweden, and the UK have reported that questions related to genetic screening are not at the forefront of the scholarly discussions.

In Poland, genetic screening and genetic testing are often addressed interchangeably, and scholars have emphasised the need for specific regulations in these areas. In South Africa, the above noted discussion of “genomic sovereignty” has also appeared to emerge in the context of genetic screening, and concerns over exploitation of the South African population in the absence of a clear regulatory framework for screening activities and benefit-sharing requirements. In China, at the forefront are discussions relating to genetic discrimination and protection in that regard. In the Netherlands, the medical community has scrutinized the ability of the existing laws to manage or prevent undesirable developments in the field without unduly blocking innovation in genetic screening and diagnostics. As already noted regarding genetic testing, the boundaries between the prevention of undesirable developments and the facilitation by the legislation of innovation in genetic screening and diagnosis are becoming increasingly blurred. Scholars in Spain have been concerned about using genetic screening for other purposes than healthcare, for example, in the workplace or a legal trial.

6.3.3 Genetic testing

Generally, there are relatively limited regulatory developments in the area of genetic screening. While some states are considering which conditions (tests) should be part of the publicly funded healthcare interventions (for example, UK), others are focusing on expanding a PKU biobank to include in the programs also those children that are not born within the country (for example, Sweden), some others report the previously adopted laws as most recent developments in the field (for example, Brazil, South Africa). China, on the other hand, has considered regulating the area of genetic screening, considering addressing such questions as supervision, protection of genetic privacy.

6.3.4 Prenatal testing

In Germany, Spain and the Netherlands, there are no particular recent legal developments in the field.

In France, discussions address over extending prenatal screening from the analysis of foetal DNA circulating in the maternal blood for the screening of other forms of aneuploidy than Down Syndrome. Likewise, considerations regarding revisiting regulation regarding screening before conception have emerged. At the moment in France, this screening only concerns couples with particular risks of transmitting monogenic heritable illnesses. It is conducted once a case has been identified in the family. Such a test could be extended to all couples, even those for whom no particular risk has been identified. Furthermore, it has also been considered to permit PGD for illnesses caused by genetic or metabolic characteristics for high-risk groups, or possibilities to preserve oocytes.

In Greece and Germany, at the forefront are discussions over the terms used in the law which affect the scope of application of prenatal screening/PGD.

In China, important regulatory steps have been taken to ensure the quality of genetic testing in foetal free DNA in a pregnant woman, including ensuring professional and ethical care and continuously improve the standard.

In Poland, questions relating to prenatal genetic testing/screening have been subject of judicial scrutiny nationally, as well as before the ECtHR. The Polish Supreme Court found that the right to prenatal testing is a derivative of women's rights to be informed about the health state of the foetus and the right to plan a family, which are considered personal interests protected by law. Preventing prospective parents from exercising the right to prenatal testing, leading to the birth of a child with a disability, against the will of parents, creates an obligation on a liable entity to pay adequate compensation for suffered harm as a result of a personal rights violation. The cases against Poland relating to reproductive rights have been brought before the ECtHR. In 2011, the ECtHR ruled that the failure to refer a patient for prenatal examination is a violation of Article 3 (prohibition of torture) and Article 8 (right to respect for private and family life) of ECHR.[341] Another important ECtHR judgment was Tysiąc v. Poland.[342] The ECtHR recalled the Polish Supreme Court's judgment of 13 October 2005[343] where the Polish Supreme Court "expressed the view that a refusal of prenatal tests in circumstances where it could be reasonably surmised that a pregnant woman ran a risk of giving birth to a severely and irreversibly damaged child, namely in circumstances set out by section 4a(1)2 of the 1993 Act,[344] gave rise to a compensation claim."[345] Lastly, in Poland, a draft law concerning the protection of human genome and human embryo was proposed by a group of Parliament Members on June 22, 2012. However, the legislative process ended April 9, 2015 and the proposal was rejected.

In South Africa, guidelines issued by the State indicate a desire to incorporate the provision of prenatal testing into South Africa's public healthcare system. As yet, however, it is unclear whether the right of

[341] ECtHR, Case of R.R. v. Poland, (2761/04), 26 May 2011.
Council of Europe, European Convention for the Protection of Human Rights and Fundamental Freedoms, as amended by Protocols Nos. 11 and 14, 4 November 1950.
[342] ECtHR, Case of Tysiąc v. Poland, (5410/03), 20 March 2007.
[343] SN IV CK 161/05, 13 October 2005, https://sip.lex.pl/#/jurisprudence/520275079/1?directHit=true&directHitQuery=IV%20CK%20161~2F05.
[344] Poland, Ustawa o planowaniu rodziny, ochronie płodu ludzkiego i warunkach dopuszczalności przerywania ciąży (Act on family planning, protection of the human fetus and conditions of acceptability of termination of pregnancy).
[345] Tysiąc v. Poland, op.cit.

access to healthcare requires such a provision. A further development in this area is a provision of the Regulations Relating to the use of Human Biological Material in terms of which "[p]re-implantation and prenatal testing for selecting the sex of a child is prohibited except in the case of serious sex-linked or sex-limited genetic conditions".[346] Furthermore, has already been discussed above, the Constitutional Court in South Africa has decided that a child might be able to claim delictual damages when, as a result of a negligently performed prenatal test and subsequent failure to abort, the child is born with a specific condition.

In Sweden, a national authority has particularly considered preimplantation genetic screening, as well as foetal genetic diagnostics, and ethical issues these practices raise in light of the scarcity of national regulatory responses. It has supported the use of NIPT; albeit has cautioned for ethical concerns that should be addressed. Also, previously, the national authority has considered ethical issues relating to foetal diagnostics. There have been considerations regarding how the current prenatal screening practices, in particular, those relating to disability, are compatible with the protection of persons with disabilities. The motions for the Parliament's support in this regard have been rejected. Recently, also a motion for revisiting the abortion legal framework has been rejected.

In Brazil, there is a government bill that proposes to introduce pre-nuptial genetic testing.[347] This law is aimed at couples who intend to initiate a pregnancy and want to know in advance if a child genetically related to them would be likely to have some genetic disorder. This law would not apply in the case of an ongoing pregnancy. As of September 2018, this bill has not been enacted into law.

6.3.5 New born screening

In some states, legal developments in the area of new-born genetic screening have not been identified (for example, Germany, South Africa, and Spain). In the states that have legal developments in the area, different issues have been put at the forefront. For example, in Greece, concerns over financial constraints that have made it challenging to realize genetic screening have been expressed. In France, one of the questions raised as part of the 2018 national consultation for the revision of the bioethics law is whether screening of newborn should be authorised for known causal mutations of genetic diseases, or even, the sequencing of the genome.

The UK, there is a debate about whether the genetic profiling of the newborn should be permitted in the public health context. It is discussed whether genome sequencing should be employed to expand NHS new-born screening to include additional specific genetic conditions. Moreover, recently, it has been reported that the National Health Service NHS has promised genomic tests for all children with cancer.[348] In Poland, the most recent legal development is defining the conditions that are part of the screening. Under the Newborn Screening Program for 2019-2022, it includes such conditions as congenital hypothyroidism, phenylketonuria, cystic fibrosis, rare defects in metabolism by MS method, congenital adrenal hyperplasia and biotinides deficiency. Similarly to Poland, also in the Netherlands, some developments are going on in relation to the extension of the number of conditions that are part of the screening. The minister of Public Health decided to extend the list of conditions in the upcoming years

[346] National Health Act, 2003: Republic of South Africa. Regulations Relating to the Use of Human Biological Material. Government Gazette 35099, 2 March 2012, regulation 2.
[347] Brasil, "Projeto de Lei No. 1971", 17 September 2007. http://www.camara.gov.br/proposicoesWeb/fichadetramitacao?idProposicao=366398.
[348] Grace O'Regan, "NHS plan promises genomic tests for all children with cancer", 14 January 2019, https://www.bionews.org.uk/page_140794.

with additional 12 diseases/conditions. The current list of conditions contains 21, and the other conditions are going to be included over the years 2018-2022.[349] Similarly also in Brazil, recently law entered into force that expands the scope of newborn screening.

In Sweden, questions of correct classification of PKU testing have emerged, and in particular, whether and to what extent such testing is genetic. Furthermore, the competent national authority has suggested to include severe combined immunodeficiency as part of PKU screening.

6.3.6 Direct-to-consumer advertising of genetic testing

Some States, including France, Germany, Poland, South Africa, Spain, UK, and Sweden have reported no or limited legal developments in the area of direct-to-consumer advertising of genetic testing.

Some EU Member States have pointed out that once the EU IVDMD Regulation becomes applicable, its labelling requirements will need to be followed. In the Netherlands, the Royal Dutch Medical Association and the Forum of biotechnology and genetics have acted in the area that concerns of direct-to-consumer advertising of genetic testing. The former has withdrawn their support for the only existing guideline they developed to regulate direct-to-consumer tests as the guideline was not effective, and recommended to establish law and regulation to protect citizens against risks of preventive medical testing. The latter has made a starting point for future policy by handing out a summary, which gives guidelines of key elements of which a supplier should be bond to in regard of consent, quality, follow up care and the right to not know.

6.4 Comparative analysis of specific legal considerations on human genetics and genomics

6.4.1 Human germline gene editing

Overall, the survey of national laws on regulating questions about human germline gene editing shows different regulatory approaches, though some patterns emerge.

Governance

The question of governance of human germline gene editing has been at the forefront in some countries (Germany, Spain, Sweden, and South Africa). The debates include considerations over coherence of the laws, the scope of application and whether they serve for the intended purpose. In light of the external commitments states have, the question of who has the competence and authority to address human germline gene editing needs to be scrutinized further. An area of specific concern is those EU Member States that have implemented EU law in the area and are not bound to the provisions in CoE BMC. This relates to the uncertainty of non-health related human germline editing not being regulated under the EU law.[350] In contrast, one can compare with the US regulatory approach, where regulating human germline gene editing falls under the comprehensive regulation of the FDA, which maintains pre-market control over such products based on their efficacy and safety.

Basic research, embryos

[349] National Institute for Public Health and the Environment Ministry of Health, Welfare and Sport, "Expansion of the heel prick screening: state of affairs". https://www.rivm.nl/hielprik/uitbreiding-van-hielprikscreening.
[350] A detailed account on how the EU law addresses human germline editing can be found in Chapter 5.

Generally, basic research is not *expressis verbis* addressed in the reviewed national legal orders. The regulations regarding human embryo involvement in research differ among the surveyed states. Some countries, for example, Germany, Greece, the Netherlands, and Spain prohibit creating embryos for research but enable the use of surplus embryos. Among non-EU states that allow research on stem cells or zygotes that are less than 14 days old, are also South Africa. Research on embryos is also possible in China.

Further regulation, e.g., specific conditions that are related to the permissibility of research differ in these states. In France, considerable ambiguities have emerged regarding the use of embryos for research. Though it is clear that such heritable genetic modification is forbidden in the clinical context; it is unclear whether this interdiction also applies to the field of research. Greece permits the use of embryos for research and requires that sufficient protection of the embryos is ensured.

In Sweden, it is possible to create and use fertilised eggs for research purposes. Although the existing national legal framework is surrounded by a degree of ambiguity, it is clear that research on fertilized eggs can be carried out up to the 14th day, and after that, it should be destroyed. The UK allows using embryos for research and creating human admixed embryos for this purpose. However, on the surface, the UK law seems more restrictive, as it takes the approach that it is illegal to conduct any research on human or admixed embryos unless specific exceptions apply. The general restrictions include the following: 14-day time limit for research and the intention is research for one of the specified purposes in law. These fourteen days is generally shared among the countries that permit the use of embryos for research, including, Greece, and the Netherlands.

Animal involvement in pre-clinical research

Among the surveyed EU Member States and non-EU states, generally, the involvement of animals in research is permitted and regulated (France, Brazil, and South Africa). As a matter of principle, for example, German law sets "a good purpose condition", namely, that "[n]o one may cause an animal pain, suffering or harm without good reason." None of the states have set any bans on animal use for germline gene editing pre-clinical studies. Nonetheless, it is common that restrictions apply, for example, by limiting what animals can be used for research (i.e., laboratory animals). Generally, following the EU law obligations, particular consideration is given to primates. The Netherlands, however, has relied on the discretion afforded under the EU law and placed a ban on the use of most great ape species (chimpanzees, bonobos, orangutans, and gorillas) for experiments on animals. Sweden is a representative of those states that have set forth criminal sanctions for the violation of the animal protection framework. In South Africa, there is a more general duty to obtain data from pre-clinical studies and/or clinical trials that support human exposure to the intended interventions.

Clinical research

Generally, among the surveyed EU Member States and non-EU states, clinical research on human germline gene modification is prohibited. There are differences in how the prohibition is enshrined in law. Some have pointed at transposing EU law measures (France, Spain), or CoE BMC (Greece), others have pointed out more general prohibitions (Brazil, Greece, the Netherlands, Sweden, and the UK). In South Africa, the law is reported to be ambiguous. In China, arguably, clinical trials are possible. However, when done, the rights of patients must be protected and ethical norms and relevant legal provisions promulgated in China must be abided by. Further to clinical trial bans, some states, for example, Sweden, have more general prohibitions against using gametes or fertilized eggs that have been subject to research, for implementation into a woman's body. This prohibition, however, potentially covers only those interventions that involve *in vitro* fertilization technology.

Clinical care

Some of the surveyed states, *expressis verbis* prohibit germline interventions as part of clinical care (the Netherlands, Sweden, UK), whereas for others this prohibition seems to relate to the clinical trials prohibition. In South Africa, the framework remains ambiguous and only arguably not permitted. In China, human embryo gene editing cannot be generalized. It should be analyzed according to its research stage and service purpose. Through reflecting on the four dimensions of security, human dignity, rights and responsibilities, and justice, and through careful reasoning, repeated adjustment and revision, the rational orientation of human embryonic gene editing was finally established. Human embryo gene editing must meet the conditions of a reasonable location; if not satisfy this condition, gene editing is unreasonable and non-conforming. However, non-health applications are arguably not permitted. The UK, however, has enabled mitochondrial donation, which is something not allowed in other countries.

Sanctions

Some countries, for example, Germany, Greece, the Netherlands, and Sweden set forth sanctions for violating the prohibitions outlined in the law. The amount and type of sanctions differ; they could be either a fine or imprisonment.

6.4.2 Genetic screening

Genetic screening is not commonly defined in law. In preparatory works to Swedish law, it is pinpointed that although genetic screening is difficult to define, it is intended to apply to such screening that includes "group of people is offered an investigation aimed at preventing disease, detecting an early stage of disease or finding out if the individual is at risk of having a child with hereditary disease." In the Netherlands, the authority to define genetic screening is delegated to a competent authority (the Dutch Health Council), which has stated that genetic screening is "systematic research in groups of people into hereditary phenomena which contain sickness or predisposition to themselves or their offspring."

Different states address the question of genetic screening in different ways. While some have more general population screening regulatory frameworks that also relate to genetics as part of the population screening (for example, the Netherlands), others specifically address genetic screening (for example, Sweden). There are also states that do not specifically regulate screening but apply other laws to handle issues in the field (South Africa). Others, for example, Brazil, address it under the screening policies, including soft law tools; China has adopted a series of normative documents to support the development and application of genetic testing/screening technology. Also, differences emerge in terms of conditions that are included within the genetic screening, as well as rights and obligations in genetic screening.

Genetic screening is overseen differently. Some counties have a specific authority that is responsible for appraising proposals for screening programmes, examine the evidence for screening programmes and implement and monitor the impact of approved programmes (for example, the UK, National Screening Committee was founded in 1996). In other states, the legal obligations that relate to screening are carried out by a competent authority in the field of healthcare administration (for example, in Sweden; the National Board of Health and Welfare).

Some states expressly set forth conditions under which screening is permissible. In aspiration to protect individuals against harmful genetic screening (in terms of examination and outcome about the physical and mental health of the examined individuals), the Netherlands has set up a licencing system and requires obtaining a particular population screening permission. Similarly, for most of the genetic screening interventions, a system of authorization exists also in Sweden.

Some states set forth conditions under which screening can be carried out. In Germany, genetic screening may be carried out for preventable or treatable conditions. In Spain, genetic screening can be carried out to predict genetic illness, identify a carrier of a gene that can be responsible for the illness.

Some states set forth a rationale for permitting a particular screening. For example, in the Netherlands, an important general criterion in screening is that the benefits must outweigh any harms for the participant. In that regard, the following criteria to assess the intended screening apply:

- it must concern a significant health problem;
- screening must have meaningful outcomes (health benefit or options for action);
- there must be a reliable and valid screening method with safe-guarded quality;
- participation in the screening is based on a voluntary, informed choice;
- screening must make efficient use of resources (including cost-effectiveness fairness and accessibility).

In Germany, for example, criteria for new target diseases as well as organizational requirements for genetic screenings are regulated with soft law adopted by Genetic Diagnostics Commission.

Some countries regulate genetic screening vis-à-vis a particular population group. In Poland, the conditions for screening in the case of children that are between one week old and 19 years old are prescribed by law. The Netherlands addresses conditions screened for within prenatal screening and newborn screening. In the UK, while there are screening programmes for children, there are no population genetic screening programmes for adults. In Sweden, apart from screening carried out relating to PKU biobanks, the law does not set forth a particular group that should be targeted. Generally, the screening could be offered as part of a legal requirement and as part of the recommendations given by the competent authority. In the former category falls, for example, prenatal genetic diagnosis and preimplantation genetic diagnosis and in the latter category falls conditions recommended by the competent national authority. Currently, there are nine screening programs, six recommended to offer to specific groups of population and three recommended not to provide to specific groups of the population. In Brazil, there are screening programmes. However, there are no official figures about the number or types of genetic tests performed as part of the screening programmes. In the US, apart from new-born screening programmes, the U.S. Preventative Services Task Force, which is convened each year by the U.S. Agency for Healthcare Research and Quality, "recommends that primary care providers screen women who have family members with breast, ovarian, tubal, or peritoneal cancer with 1 of several screening tools designed to identify a family history that may be associated with an increased risk for potentially harmful mutations in breast cancer susceptibility genes (*BRCA1* or *BRCA2*). Women with positive screening results should receive genetic counselling and if indicated after counselling, BRCA testing."[351] Though this recommendation comes from a federal task force, the practice of any such screening would be performed by physicians and is therefore also regulated at the state level.

Some states do not set forth particular rights or protections to persons undergoing a screening that are distinct from those applicable in a clinical setting (the Netherlands). However, other states either address

[351] U.S. Preventative Services Task Force, "BRCA-Related Cancer: Risk Assessment, Genetic Counseling, and Genetic Testing", December 2013. https://www.uspreventiveservicestaskforce.org/Page/Document/UpdateSummaryFinal/brca-related-cancer-risk-assessment-genetic-counseling-and-genetic-testing.

the protection of individuals through general national legal provisions or those that specifically relate to genetic screening. For example, Spanish law regulates the basic principles of genetic screening and in what cases can be used (basically to avoid a genetic illness) the kind of information that has to be given to the patient and how he/she will be consent. The basic principles are accessibility and equality; data protection; free of charge; consent; and use of data. Similarly, South Africa addresses the issues under the data protection laws and health sector regulations. Sweden requires that consent to genetic screening is given in writing. A similar requirement is outlined in Spanish law.

Some states set forth the responsibilities of the genetic screening providers. In the Netherlands, regarding population screening an increased degree for the utmost care in relation to safety, efficiency and effectiveness are required, which are stricter requirements compared to general medical treatment. In Sweden, a distinct requirement in genetic screening is that authorisation of the competent authority needs to be obtained.

Some states also address liability or the violation of rights and obligations in genetic screening. For example, such a law exists in Spain, as well as in Spanish Autonomous communities, for instance, Andalusia. Under the Spanish law, administrative sanctions are from 600€ up to 1.000.000€ depending on a particular violation.

6.4.3 Genetic testing

Genetic testing: law

The approaches national laws have taken for regulating genetic testing differ considerably, though common features emerge. First, a distinction can be drawn between states that regulate genetic testing specifically (for example Germany, France, South Africa) and those who have not adopted specific laws to tackle genetic testing (for example, Poland, Spain, and Sweden). Germany has noted that genetic testing is regulated to ensure the state's commitment to safeguarding human dignity and the right to informational self-determination, and France has highlighted the aim of the legal framework to ensure that the individual remains at the centre of concern about genetic testing.

Access to testing

In France, the law regulates situations in which genetic testing may be prescribed, the way it shall be conducted, as well as the delivery of genetic testing results. In the Netherlands, it is emphasized that when genetic testing is provided in a clinical setting, patients cannot request it; the caregiver decides about the necessity of a particular intervention, including testing. Germany has introduced a special authorization system in order for genetic testing can be provided. Moreover, Germany also regulates explicitly some types of testing. Besides the use of genetic testing for medical purposes, the national law governs the use of genetic testing in the field of insurance and working life. In Brazil, genetic testing is offered both by the public and by the private sector. Moreover, the government also offers (or intends to offer, as the case may be) a range of optional genetic tests for individuals under certain circumstances. The private sector offers a broader range of genetic tests and is subjected to legal requirements stemming from a range of laws.

South Africa and Germany exemplify that it can be regulated who is allowed to be involved in genetic testing. In Germany, genetic testing for medical purposes may only be carried out by a physician, whereas a general practitioner can conduct diagnostic testing, but a medical specialist must perform predictive testing. South Africa permits the removal of biological material in an authorised institution, prescribed institution and research institution for ancestry analysis. When genetic testing is carried out for health

purposes, South African law requires that it is done by a person trained and registered under the national law.

Quality

Quality of genetic testing is addressed in different ways. In the Netherlands, in case genetic testing is provided as part of healthcare, the healthcare provider needs to make sure that the genetic tests used by the healthcare provider will deliver good quality of the testing results. This is necessary in order for the caregiver, and the patient can rely on the results for follow up treatment. Similarly, in Sweden, care that is given to a patient, shall be of high quality and provided by science and proven experience. The concept of 'science and proven experience' however is surrounded by a considerable ambiguity, and it remains to be ascertained how this concept relates to regulating *in vitro* diagnostic medical devices. Although a national authority in Sweden is entitled to adopt rules to specify requirements for genetic testing in healthcare that has not been done yet.

Nonetheless, in so far as genetic testing is an *in vitro* diagnostic device, measures implementing EU law apply. A similar approach is also adopted in the UK, where medical genetic tests fall under the broader regulatory framework associated with medical devices.

In China, physicians should conduct clinical laboratory treatment and should be approved by the hospital. In Poland, there specific laboratory requirements outlined in national law that constitute the quality regulatory framework for genetic testing. Specifically, the law introduces quality standards in the field of laboratory activities of medical genetics, assessment of their quality and diagnostic value, as well as laboratory interpretation of test results. In the Netherlands, laboratory requirements co-exist with other requirements that shape the quality of texting. In Greece, all laboratories offering diagnostic testing must obtain ISO 9001:2008 from a national or international accreditation body, must conduct international external quality controls and all laboratories are analyzing biological samples for other health care entities, must obtain ISO 15189: 2007 from the national accreditation authority.

In some states, a distinction can be drawn between tests offered in a clinic and direct-to-consumer genetic testing. For example, in the UK, medical devices should be approved by a specific body. However, in direct-to-consumer genetic testing, the approval mechanism applies to the test kits sent to the customer to produce the saliva sample, but not to the tests themselves or the interpretation of the results. Consequently, there is no quality assurance for these tests in place. This approach can be attributed to implementing EU law in the area. Also, other EU Member States note the transposition of EU law measures to regulate genetic testing, and their limited reach vis-à-vis scientific advances in genetic testing (for example, Sweden, the Netherlands, Poland, and Spain). Consequently, for example, in Sweden, the applicable requirements differ, depending on whether the test is health care measure, a medical device, or a service, namely, whether the laws relating to healthcare, second, those relating to medical products, and third, those relating to consumer rights are applicable. On the contrary, although in Greece also questions over adequate regulation of direct-to-consumer genetic testing emerged, these concerns have been tackled.

Information and consent

Generally, states regulate in some way consent requirements. Either specific requirements are set forth for genetic testing, the general provisions regulating health care apply. Germany requires that before conducting genetic testing, informed consent of the person involved has to be obtained. France requires a free and informed consent in writing of the person and specifies that this consent may be withdrawn at any point in time. The Netherlands indicates that consent to genetic testing cannot be presumed, so it

needs to be explicated due to the big impact of the possible outcomes of genetic testing. In South Africa, it is prohibited to remove biological material for genetic testing by a competent person unless written informed consent of the person from whom such biological material is removed is obtained. In emergencies, however, exceptional rules apply.

In South Africa, specific requirements are set forth for carrying out genetic testing on a "mentally ill" person, requiring that written informed consent is obtained from the person concerned, but if the person is not capable of giving consent, a curator appointed by the court, a spouse, next of kin, a parent or guardian, major child, brother or sister, parent or associate may consent to genetic testing. In the case of emergency, consent can also be obtained from the head of the health establishment. Germany regulates genetic testing of persons unable to give consent, which also includes minors, and permits it only under strict conditions. In the Netherlands, genetic testing for children should be approached with great reluctance when the benefits in terms of treatment of prevention are not so clear. Greece permits genetic testing on minors, setting forth an obligation to obtain consent from a minor via her legal representative. In South Africa, Regulation 3 of the Regulations relating to the use of Human Biological Material prohibits the removal of biological material for genetic testing from a person younger than 18 years, unless written informed consent by a child over the age of 12, provided the child is of sufficient maturity and has the mental capacity to understand the benefits, risks, social and implications of the procedure, written informed consent of a parent, guardian or caregiver where the child is younger than 12 years or the child is over 12 years but has no sufficient maturity and the mental capacity to understand the benefits, risks, social and implications of the procedure. In emergencies, exceptional rules apply.

Only South Africa has reported that the national law specifically regulates genetic testing on the deceased. In particular, if is intended to use tissue from a deceased person for purposes of genetic testing where no consent has been given by the deceased person before her death and where there is no evidence that the removal of the tissue or cells would be contrary to a direction given by the deceased before her death, steps must be taken to locate the spouse, partner, major child, parent, guardian, major brother or major sister of the deceased person in order to obtain consent.

Some states regulate modalities regarding information that should be provided to the persons being tested; others address information as one of the informed consent elements. For example, in France, right to information is a distinct right. It includes information about the person's state of health, the proposed treatments, their usefulness and necessity, and the risks involved. In Germany, before giving consent, the person concerned shall be informed of the nature, significance, and consequences of the genetic test to be performed. In the Netherlands, physicians are under a duty to provide relevant information to patients so that they can make a decision about testing and exercise their right to self-determination. Physicians are also under a duty to provide relevant information to patients so that they can decide on testing. In the Netherlands, the right of self-determination has been recognized as an independent right. In China, a right to informed consent is a distinct right. Under the national law, medical institutions and their medical personnel shall truthfully inform patients of their illness, medical measures, medical risks, etc., and promptly answer their inquiries; however, adverse consequences for patients should be avoided. In Poland, the national law obliges a doctor to give accessible information to a patient (or their statutory representative) about the patient's state of health, proposed and possible diagnostic and therapeutic methods, foreseeable consequences of their use or omission, the results of the treatment and the prognosis.

Counselling

In Germany, genetic counselling is specifically regulated. In the case of predictive genetic testing, the person concerned shall receive genetic counselling from a doctor with specific qualifications before the test is performed and once the results are on hand unless this person - after having received written information on the contents of the counselling - has waived their right to genetic counselling in writing. After counselling, the person concerned shall be allowed adequate time for consideration before undergoing the test. In the Netherlands, in regards to identifying incurable diseases, it has been suggested that an agreement needs to be made whether or not the person tested should be informed about this disease. In the case of genetic testing with newborn or children, the parents or the caregiver should act in the child's interest. In China, physicians should truthfully introduce the condition to patients or their families, and they should take care to avoid adverse consequences for patients. Similarly, also in France delivery of results of genetic testing has been of regulatory concern.

Family interests/ rights

Some states have expressly addressed the question of family member interests in genetic testing; others respond to this challenge through the general data protection/privacy/confidentiality legal framework. For example, in Poland, a doctor-patient confidentiality privilege exists. Nevertheless, there is an exception set forth in the national law, which allows the disclosure of test results without a patient's consent. In South Africa, while confidentiality is paramount and extends to the disclosure of genetic information that would impact family member's interests, it is not an absolute right. In accordance with section 14(2) of the National Health Act, no person may disclose any information unless the user (patient) consents to that disclosure in writing; a court order or any law requires that disclosure; or non-disclosure of the information represents a serious threat to public health. If any of these requirements for disclosure are relevant, then confidentiality may be waived regarding the health status of the patient.

The Dutch legislation and case law does not place a "warning obligation" on caregivers to justify a breach of the secret of the patient for the benefit of the family members. In China, there is a prevailing belief that, unlike other citizens' privacy, genetic privacy is extremely special, which concerns the health rights of individuals and families. In this regard, the Chinese legislature attaches great importance to the protection of citizens' privacy, and the national law contains relevant provisions with a view to protecting citizens' personal information. It is also upheld by the national courts.

On the contrary, in France, the law of 2011 imposed transmitting information to the family members potentially concerned by the results of genetic testing in case of the detection of a severe genetic anomaly whose consequences might be prevented and organised an anonymous procedure through the doctor in case the patient did not want to know the results. Although an individual is free to decide whether or not to receive results of genetic testing, there is an obligation to transmit the results to the family members. Hence, contrary to the person who is undergoing genetic testing, the family member does not have a right not to know.

Right not to know

Some countries regulate expressly disclosure of genetic information (Germany, Netherlands). For example, in the Netherlands, a patient has the right to decide whether or not to be informed of the results of the genetic examination and the resulting consequences; this decision should be respected. This legislation is not specific for genetic testing but applies in general health situations so also in case of genetic tests.

Purpose of testing and secondary use of genetic data/information

Germany specifically regulates some types of testing, besides the use of genetic testing for medical purposes, the national law regulates the use in the field of insurance) and in working life. The law does not regulate genetic testing in basic research in any particular way. In the Netherlands, access to testing is related to a medical indication. Moreover, genetic information is also of interest to insurance companies. Therefore, Dutch legislation has limited the right to ask questions during the intake of an insurance agreement. For example, limitations are set forth for questioning about severe hereditable diseases and questions about the investigation to a predisposition of hereditable diseases or the outcomes of these tests. Questions related to the results of genetic tests are not legitimate to ask when the insured amount of money does not exceed to a certain extent, the "vraaggrens" (the limit for questions). For live insurance, this is an indexed amount of money; when it comes to disability insurances with periodic payments, the line is drawn at 70 percent of the income. In France, since 2002, a law is in place that forbids any discrimination based on genetics and to ban insurance from asking their clients to proceed with genetic testing. Regarding the secondary use of samples/data/information, France is among countries that note the permissibility of research in the absence of objection of the data subject.

Greece is among countries that have specifically noted using genetic testing for other purposes than health care. First, the national law sets forth an exception to the general prohibition of genetic data processing, when this processing relates to data to be used by judicial authorities to determine the execution of crimes, following the general principle of proportionality. This exception is founded upon public interest and justice, to identify a criminal. The national criminal law also allows the mandatory DNA testing by the state's criminal sanctions' authorities (police, customs, port authority, etc.) at their discretion, under the auspices of the competent district attorney. The test must be ordered in cases of a suspect when there are severe indications of a felony or a misdemeanor punished with at least one-year imprisonment; the DNA test is obligatory for the authorities. The suspect may appoint a technical counsellor who may be present during the experts' work and who may have access to all related documents and information these experts also have. The suspect may also ask for the repetition of a positive test. Secondly, the national law in Greece also dictates that in a civil paternity suit. If the defendant declines to undertake the appropriate scientifically test (in this case, the DNA paternity test), then the court is bound to recognize his paternity. The DNA test, in this case, is ordered as part of expert testimony, as a method of scientific proof. It follows that in this case, the defendant is practically obliged to take a paternity test or else, his paternity will be deemed proven in court.

Sanctions

In France, criminal sanctions apply for some violations relating to the regulations applicable to genetic testing.

6.4.4 Prenatal screening

Generally, prenatal screening/testing, including PGD, is in some way regulated in the surveyed national legal orders.

Generally, the surveyed national legal orders permit prenatal testing/screening, either through regulating it (for example, Sweden and the US), or addressing it as an exception from particular criminalized activities (for example, Greece, Germany). Considerable differences can be noticed regarding the extent to which it is permitted, and how detailed it is regulated. Some states restrict where PGD or screening can be carried out (for example, France), or set forth conditions for licencing (for example, the Netherlands, UK).

Different states address the question of conditions for which prenatal screening can be carried out differently. While some states list specific conditions, others provide criteria or envisage only certain bans

in using the technology. States generally have set forth counselling requirements. However, it differs whether the emphasis is on the pregnant woman only or the pregnant women and the other prospective parent (for example, China); it is common that counselling is provided.

Except for Brazil, in the surveyed states abortion is allowed. Nonetheless, it differs under which week and what circumstances. It is common that following the prenatal testing/screening abortion is possible, even though that is not *expressis verbis* indicated in the national law. In Greece, however, women are informed that if they do not intend to carry on with abortion, there is no purpose in carrying out a prenatal testing/screening.

In Greece, the question of the law on prenatal testing/screening is tightly connected to the regulation of termination of a pregnancy. The Greek Criminal Code, Art. 304, justifies (de-criminalizes) termination among other things until the 24th week of pregnancy, in case of a pathological foetus, whose "severe anomalies" are detected after current scientific screening methods. The law, however, does not specify when a foetus suffers from "severe anomalies" and is, therefore regarded as "pathological." Nonetheless, in 2007 the competent national body, the National Commission of Bioethics, recommended screening for trisomy (Down syndrome and others) and thalassemia as these are the diseases which could satisfy the legal requirement of "pathological" (child) under Art. 304 of the Criminal Code. The Commission has also recommended testing for the other monogenetic etc. diseases only in case of (proven) heredity. Case law in Greece highlights that cystic fibrosis is in the list of diseases allowing termination also (Supreme Court 13/2010 on liability for defective prenatal testing), anatomic anomalies such as the missing of limbs (due to phocomelia, etc.) do not fall within the exemption category.

Similarly, in France prenatal diagnosis is authorized to detect particularly serious illnesses *in utero* in the embryo or the foetus. The conditions in which this test is conducted has massively evolved over the past 20 years. While it initially only concerned couples who had already had a child with Down syndrome, the diagnosis was then extended to specific categories of women (in particular those who were over 38 years old) and was finally conducted as a screening. However, while new-born screening of cystic fibrosis exists in France since 2002 to ensure that care is provided as soon as possible, prenatal screening for this purpose is forbidden.

In Brazil, prenatal care is regulated by law. However, no genetic test is available in the list of prenatal tests offered as part of the public healthcare system. In Poland, a doctor is obliged to provide to a patient (or their statutory representative) information about their state of health, proposed and possible diagnostic and therapeutic methods, foreseeable consequences of their use or omission, the results of the treatment and the prognosis. Additionally, when any diagnostic or therapeutic doubts occur, the physician, if he/she considers it justified in the light of his/her medical expertise, should consult a competent specialist or arrange a medical consultation. Moreover, a doctor may refrain from providing health services incompatible with his/her conscience. However he/she is obligated to inform about the real possibility of obtaining the service from another physician or medical entity and note that fact in the medical records.

Regarding the use of PGD, Greece considers it ethically imperative to provide genetic counselling to both prospective parents, before the intervention can take place. In Greece, also other screenings are regulated, including amniocentesis, early amniocentesis, and second-trimester ultrasound, the administration of which requires informed consent and appropriate genetic counselling. Regarding amniocentesis, it can be provided in specific situations, and reception of risk information is mandatory. The pregnant woman must also be informed that if she has not decided that she will terminate her pregnancy if the foetus is shown as pathological, then, there is no meaning in prenatal testing.

In Germany, the use of PGD is sanctioned with up to one year's imprisonment or fine, except for when strict conditions outlined in the law relating to a high probability of a serious illness are met, or the pregnancy would highly likely lead to still-birth or miscarriage. A clarification of the terms high probability and serious genetic disease is missing, which is often criticized in the legal scholarship debate. Prenatal testing is only permitted if a written consent from the potential parents can be obtained. Furthermore, predictive testing is only allowed to be implemented when the parents become well informed about the medical, psychic and social consequences of the diagnostic, and when an ethics committee examined whether the mentioned requirements are met. For the implementation of prenatal testing, only qualified physicians in licensed centres are authorised to perform PGD. Prenatal testing in vivo, however, may be performed only for medical purposes and only if the examination is aimed at specific genetic characteristics of the embryo or foetus which, according to the generally accepted scientific and technical knowledge, adversely affect their health during pregnancy or after birth, or if a treatment of the embryo or foetus with a drug is provided, the effect of which is influenced by specific genetic properties and the pregnant woman has been duly informed.

In the Netherlands, the current prenatal screenings programme consists of two types of screening/testing. At first, the prenatal screening to risk factors of the pregnancy. This screening aims to provide a health benefit for the pregnant woman and the (future) child. The other part of the screening aims to detect foetal abnormalities (chromosomal abnormalities). Based on the results a well-considered decision could be made by the prospective parents about the continuation of pregnancy. In the Netherlands, access to abortion relates to a foetal viability criterion. Following that criterion, it is permissible to carry out abortion until the foetus is able to stay alive outside the body of the mother. Traditionally it has been a 24-week threshold; however, with the advances in science and technology, discussions over limiting this period have emerged. This approach can be compared with other states that expressis verbis set forth a specific gestational week in their national law. Spain is among those States, and abortion can be carried out within the first 14 weeks, but in case of a health condition, when there is a high risk for the life or health of the mother or there is a risk of severe anomalies of the foetus, it is possible to terminate the pregnancy in the first 22 weeks; if there are anomalies incompatible with life at any time.

In Sweden, a distinction is drawn between providing information about prenatal diagnosis and carrying out a prenatal diagnosis. While all pregnant women shall be offered general information about prenatal diagnosis, if a pregnant woman has a medically established increased risk of giving birth to an impaired child, she shall be offered further information on prenatal genetic diagnosis. PGD, however, may only be used if the man or woman has a predisposition towards a serious monogenetic or chromosomal hereditary disease, which entails a high risk of having a child with a genetic disease or impairment. In that case, the treatment may only be aimed at preventing the child from inheriting the predisposition towards the disease or impairment in question. In Sweden abortion is available at the woman's request if there is no serious risk to her health or life until the 18th week of pregnancy; later terminations are permitted only with the authorization of the competent authority.

In the UK, PGD is permissible, provided the license has been received, for the following purposes upon offering to the people receiving the care an appropriate genetic counselling: (a)establishing whether the embryo has a gene, chromosome or mitochondrion abnormality that may affect its capacity to result in a live birth; (b)in a case where there is a particular risk that the embryo may have any gene, chromosome or mitochondrion abnormality, establishing whether it has that abnormality or any other gene, chromosome or mitochondrion abnormality; and (c)in a case where there is a particular risk that any resulting child will have or develop— (i)a gender-related serious physical or mental disability, (ii)a gender-related serious illness, or (iii)any other gender-related serious medical condition, establishing the sex of

the embryo. Further to that, also sex selection can be carried out, where there is a risk that a woman will give birth to a child who will have or develop—(a) a gender-related serious physical or mental disability, (b) a gender-related serious illness, or (c) any other gender-related serious medical condition. Under the UK laws, also preimplantation tissue typing (PTT), known as "saviour siblings" can be carried out in restricted cases, provided conditions set forth in law are met.

All pregnant women in England, Scotland and Wales are offered antenatal genetic screening for Down syndrome and neural tube defects and have the opportunity to terminate a pregnancy if 'there is substantial risk that the child would suffer from physical or mental abnormalities as to be seriously handicapped' (Abortion Act 1967 s. 1(d)). Nonetheless, the Abortion Act 1967 does not extend to Northern Ireland. Sections 58 and 59 of the Offences against the Person Act 1861 and sections 25 and 26 of the Criminal Justice Act (Northern Ireland) 1945 provide for stringent requirements for abortion, where abortion is permitted only if the life of the woman, but not in case of fatal foetal abnormality. Based on the eighth amendment, a clause inserted into the Irish constitution restricting the abortion. This framework has been challenged by case law and a recent referendum in May 2018, requiring legislative changes.

Some states set forth conditions that are part of the screening. For example, in the Netherlands, at this moment two screenings programmes to detect foetal abnormalities exist: a screening to identify the down syndrome in the first trimester (week 11-13), with a focus on trisomy 21, 18 and 13 (the combination test; and a structural echoscopic examination (SEO), with an emphasis on structural abnormalities of the foetus, especially in regard to neural tube defects (spina bifida and anencephaly) in the second trimester (week 18-22).

NIPT is available in some states. In Greece, NIPT is not obligatory, but when it is carried out, it requires informed consent. In Spain, NIPT is not particularly addressed. In Brazil, prenatal care is generally regulated by law and NIPT is commonly provided by private healthcare providers for such purposes as Down syndrome, Edwards's syndrome, or Patau syndrome; the providers must ultimately comply with a thick layer of applicable national law. However, as has already been noted above, abortion is generally forbidden in Brazil. Unlike countries, for example, the UK and France, where prospective parents use NIPT to decide if they will terminate a pregnancy, in Brazil parents can only learn that their prospective child has, for instance, Down syndrome. They cannot legally request the termination of a pregnancy in this case.

In the Netherlands, NIPT is offered as part of screening for congenital abnormalities for Down's syndrome, Edwards' syndrome and Patau's syndrome. NIPT is offered to women over the age of 36 years old with an increased chance of having a child with congenital disabilities, and to women who previously have had several miscarriages, moreover, all pregnant woman can choose to have an NIPT test. In Sweden, NIPT is practiced, but it is not regulated in any specific way apart from the general requirements applicable to screening/testing. There are, however, guidelines for NIPT for trisomy 13, 18 and 21. In that regard, the Swedish Society of Obstetrics and Gynaecology (SFOG) has adopted guidelines for the use of NIPT as a means to decide whether an invasive procedure should be offered to patients. In the UK, NIPT is currently being offered through the NHS to determine foetal sex in pregnancies at risk of serious X-linked conditions, such as Duchenne muscular dystrophy, and those at risk of congenital adrenal hyperplasia. From 2018, women will be offered a safer screening test as an alternative to the invasive tests. A simple blood test will be offered which is then used to check for DNA fragments of these chromosomal syndromes, or Down's, Edwards' and Patau's syndromes. In France, prenatal screening of Down syndrome is authorised from the analysis of foetal DNA circulating in the maternal blood.

South Africa is among the states that have a somewhat liberal regulatory framework on the issue. Under the national law, only PGD and prenatal testing for sex selection except for serious sex-linked or sex-limited genetic conditions are prohibited. The law does not put other constraints on PGD and prenatal testing.

Generally, all states have reported that screening is voluntary and consent of the person/-s concerned is necessary. In the Netherlands, only in exceptional cases, presumed consent can be accepted.

6.4.5 Newborn screening

Commonly (with an exception for South Africa) newborn screening is regulated nationally, albeit different states have chosen different regulatory strategies.

In the UK, newborn screening involves the identification of a baby's risk of developing a disease that is preventable or treatable. The current new-born screening programme in the UK is based on the blood spot test (heel prick, dried onto a piece of filter paper), which screens for nine rare but serious conditions: sickle cell disease, cystic fibrosis, congenital hypothyroidism, phenylketonuria, medium-chain acyl-CoA dehydrogenase deficiency, maple syrup urine disease, isovaleric acidaemia, glutaric aciduria type 1 and homocystinuria (pyridoxine unresponsive). In Brazil, the following conditions are screened for in the heel prick test: Hypothyroidism, Phenylketonuria, Sickle cell disease, and other hemoglobinopathies, Cystic fibrosis, Biotin, Congenital adrenal hyperplasia. Other new-born screening tests offered within the public health care system in Brazil include Newborn Hearing Screening and the assessment of heart condition and eyesight. Further to that, also the private sector offers various new-born screening services. In France, newborn screening primarily concerns genetic illnesses: phenylketonuria, congenital adrenal hyperplasia, sickle cell disease, and cystic fibrosis. In Germany, screening includes inborn errors of amino acid metabolism; phenylketonuria, hyperphenylalaninemia, maple syrup urine disease; inborn errors of organic acid metabolism; glutaric acidemia type I, isovaleric acidemia; inborn errors of fatty acid metabolism; congenital hypothyroidism; biotinidase deficiency; classical galactosemia, as well as classical congenital adrenal hyperplasia. In Poland, it is recommended that screening includes screening for phenylketonuria, cystic fibrosis, congenital hypothyroidism and hearing, and heart defects. In the Netherlands, the following 21 conditions are included and the list is going to be expanded to additional 12 conditions in a near futrue: Adrenogenitaal syndrom (AGS), Alfa-thalassemie,Bèta-thalassemie, Biotinidase deficiency (BIO), Congenitale hypothyreoidy (CH), Cystic fibrosis (CF), Galactosemy (GAL), Glutaaracidurie type 1 (GA-1), HMG-CoA-lyase deficiency (HMG), Isovaleriaan-acidury (IVA), Long-chain hydroxyacyl-CoA dehydrogenase deficiency (LCHADD), Maple syrup urine disease (MSUD), Medium-chain acyl CoA dehydrogenase deficiency (MCADD), 3-Methylcrotonyl-CoA carboxylase deficiency (3-MCC), Multiple CoA carboxylase deficiency (MCD), Phenylketonury (PKU), Sickel Disease (SZ), Tyrosinemie type 1 (TYR-1), Very long-chain acylCoA dehydrogenase deficiency (VLCADD), Carnitine transporter (OCTN2) deficiency, carrier of the Sickel Disease (SZ). At present, China mainly screens CH and PKU for newborn blood samples, and increases glucose-6-phosphate dehydrogenase deficiency (G6PD) screening in Guangxi and Guangdong, with an incidence rate of 3.6%, and screening for congenital adrenal hyperplasia (CAH) has been added in the area of Jiangsu and Shanghai. Moreover, although the law in China does not explicitly prohibit the screening of incurable diseases, in practice, it is basically limited to the range of diseases that can be cured.

Poland has reported that a particular coordination mechanism is in place for newborn screening. The computer registry of labels and blood papers allows controlling all stages of screening, including blood collection from new-borns, tests, parents' notifications and diagnostic confirmation by the doctor. In

China, with the continuous advancement of screening for neonatal diseases, in 2008, 30 provinces (municipalities) and autonomous regions in China have conducted screenings. The number of new-borns screened has reached 5.6 million in Shanghai, Beijing, Zhejiang and other places. The screening rate of neonatal diseases has reached more than 95%. The Provincial New-born Disease Screening Center established by the Children's Hospital affiliated to Zhejiang University screened more than 500,000 new-borns in 2008 and established a screening network system with more than 1,200 pregnant women in the province.

Approaches differ whether or not the screening is compulsory. In the UK, newborn screening is not mandatory; “parents are asked for verbal consent for newborn screening. They can decline for sickle cell disease, cystic fibrosis, and congenital hypothyroidism individually.” They can decline phenylketonuria, medium-chain acyl-CoA dehydrogenase deficiency, maple syrup urine disease, isovaleric acidaemia, glutaric aciduria type 1 and homocystinuria as a group. Likewise, it is non-compulsory, for example, in France, Germany, the Netherlands, Spain, and Sweden. In Brazil, the heel prick test is mandatory and, depending on the municipal legislation; a birth certificate will not be issued before the test has been performed.

In Germany, in case the test is positive for a disorder parents will be informed immediately. In Poland, expedited access to health care is granted in case of risks identified in the screening. In the Netherlands, the law permits the paediatrician, who is involved (regarding the heel prick), permission to consult data of the child (up to the age of 6 months) without previously given consent from the parents to do so. This is only allowed if this is necessary for follow-up care for the child.

In the UK, samples are retained for quality assurance purposes in laboratories. Whereas, the results of the genetic screening form part of the child’s medical record and will be kept in line with records management guidance. Generally, for child screening records are retained until the 25th birthday or 10 years after the child has been screened whichever is the longer personal information is held. In Germany, the blood sample must be destroyed after the examination; therefore a biobank is not needed.

In the Netherlands, the blood of the heel prick card can be used for scientific research purposes. For this research, only anonymous heel prick card may be used. The parents need to object to anonymous use for scientific research purposes. If they do not object, the card is used for research. Sometimes it necessary to link the blood samples to personal information of the child. If this is the case then there needs to be an explicit consent from the parents in a written from. If the parents objected to the use of the card for scientific research, the blood will be destroyed by the laboratory one year after collection of the blood. If an implicit consent is given, the data/samples retained by the laboratory are allowed to keep the blood samples for one year to be able to check up on the test. After this period the blood may be kept for another 4 four years for scientific research (anonymous). After in total of five years, the blood will be destroyed. No specific legislation exists in the Netherlands in regard to biobanks, but according to effective control of the consent towards body material legal developments are going on with a concept legislative proposal.

In Brazil, data can be stored for five years. After this period the patient has the right to request to delete them. In case he/she does not use this right the data will be stored for the time needed to preserve the health of the patient or third persons related to her. In addition to these cases, the data can be retained for research if anonymized.

In Sweden, samples are retained in a special biobank, PKU biobank, unless the child's legal guardians have opted out of that. The following information may be included in the record: the mother's name, personal identity number, and town of domicile, as well as the length of pregnancy, the child's time of birth and gender, and the order in case of a multiple birth; the unit within the medical service that took specimen; diagnosis, information concerning the treatment of diagnosed diseases, and consent form the child's parent/guardian.

In the Netherlands, the data/samples that are retained, are used in the interest of the child but also for scientific research and identification of people after a disaster. The use of samples for identification is first used after the so-called "firework disaster" (which happened in the year 2000 in Enschede) to identify children. In Sweden, however, the tissue samples in the PKU biobank may only be used for analyses and other surveys to trace and diagnose metabolic diseases, retrospective diagnosis of other diseases of individual children, epidemiological studies, monitoring, evaluation and quality assurance of the business, as well as clinical research and development.

6.4.6 Direct-to-consumer advertising of genetic testing

In Brazil advertising of genetic testing or screening is not prohibited; however, for example, commercialization of biological material is. In connection with fertility treatments, genetic testing and screening are offered to the consumers. Although the practices need to comply with the law, it does not seem to preclude, for example, advertising of biospecimen for fertility purposes that relates to choosing donors pertaining to various features, including education, even though scientifically it is uncertain whether or not it increases the odds having a child with above average intelligence. However, the use of IVF and PGD for non-therapeutic purposes is prohibited.

In France, advertising of genetic testing directly-to-consumers is prohibited by law. Indeed, the results of genetic testing in France need to be delivered together with the appropriate medical counselling. In Spain, advertising of genetic testing, disregarding whether it is health related or non-health related is not permitted. Moreover, in Spain, a specific authority is tasked to control the advertisements relating to diagnosis, prevention or treatment of illness physiological development, weight loss, physical or psychological modification, restoration, correction or modification of the organic functions or other health aims. In Greece, while physicians are prohibited from advertising genetic testing, laboratories functioning as legal persons of private law offering services do not fall within the prohibition and enjoy the constitutional protection of the constitutionally protected freedom to advertise/economic freedom.

On the contrary, in other states, advertising of genetic testing is not regulated, and therefore possible. For example, Germany, South Africa, China, and the UK. Germany, nonetheless, following the national legal requirements, only physicians may carry out genetic testing for a medical purpose. In Poland, the Netherlands, and Sweden, even though there are statutes and regulations relating to advertisement, specific requirements for direct-to-consumer advertising of genetic testing do not exist. In China, similarly as in Sweden, misleading advertising, including false representation of the quality of commodities is not permitted. Additionally, in Sweden, a specific prohibition is made regarding the health impacts of advertised products, including genetic testing.

In the US, the advertising of genetic tests for health purposes directly to consumers is currently permitted at the federal level only if a company has received individual pre-market approval by the FDA.[352]

[352] Exempt from this pre-market approval requirement are genetic carrier screening tests: in 2017, the FDA published "an order to exempt autosomal recessive carrier screening gene mutation detection systems from the premarket

Specifically, a company that wishes to advertise and sell genetic tests directly to consumers for health purposes must first apply for approval through the FDA's "de novo premarket review pathway, a regulatory pathway for hte novel, low-to-moderate-risk devices that are not substantially equivalent to an already legally marketed device."[353]

Common to the EU Member States is the relevance of EU law regarding unfair commercial practices and *in vitro* diagnostic medical devices which shape the national legal environment. This law, however, does not place distinct requirements relating to the direct-to-consumer advertising of genetic testing. The general requirements that relate to CE marking are applicable (for example, Greece, UK). In the UK, as well as in Sweden, an emphasis has been placed on the *in vitro* diagnostic medical device meeting the requirements assigned by the manufacturer. Greece has placed emphasis on different types of illegal and immoral advertising which is also of relevance in advertising genetic testing directly to consumers.

7. Discussion and general analysis

7.1 Germline gene editing

7.1.1 International and regional human rights legal orders

Few legally binding human rights instruments *expressis verbis* address human germline editing. Among the regional human rights regimes, only CoE has taken a stand on human germline editing and creating embryos for research. On the one hand, it has limited effect, and it applies only to those states that have ratified CoE BMC, however, the signatories of the convention could be expected not to act contrary to its object and purpose. On the other hand, it should be recalled that the Parliamentary Assembly has indicated this approach also be relevant for the ECHR. The recommendation adopted by a Parliamentary Assembly remains a source of soft law, even though, it could have implications in certain situations, for example, cases brought before the ECtHR. The legal challenges arising in the context of germline gene editing have to be addressed by interpreting general human rights norms. However, interpretation has its limits (e.g., contra legam). More guidance is, to some degree, offered by soft law on genetics and genomics.

Legal orders of concern provide a general normative framework for protecting rights relevant in light of the legal issues raised by germline gene editing, such as the right to life, right to privacy including reproductive autonomy, prohibition of discrimination, right to health, right to science and access to justice. Their meaning and applicability in the context of germline gene editing is however debated and differs in different legal cultures. In the case of the right to health and the right to science their realization remains at the discretion of states. The international and regional human rights norms in this context do not provide clear guidance. For instance, none of the instruments that protect the right to health requires granting access to specific advances in the area of genomics. Similarly, a positive right to enjoy the benefits of scientific progress does not directly inform to what extent differentiation in access to benefits of germline editing would be acceptable; this differentiation could be seen as discretion that remains with the national legal orders which should be exercised with due regard to equality and the prohibition of

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notification requirements, subject to certain limitations." U.S. Food and Drug Administration, "Medical Devices; Exemption from Premarket Notification; Class II Devices; Autosomal Recessive Carrier Screening Gene Mutation Detection System", 82 FR 51567, 7 November 2017. https://www.federalregister.gov/documents/2017/11/07/2017-24162/medical-devices-exemption-from-premarket-notification-class-ii-devices-autosomal-recessive-carrier.

[353] U.S. Food and Drug Administration, "FDA allows marketing of first direct-to-consumer tests that provide genetic risk information for certain conditions", 6 April 2017. https://www.fda.gov/NewsEvents/Newsroom/PressAnnouncements/ucm551185.htm.

discrimination. In a similar vein, the legal status of the human embryo and the scope of its protection remain unclear and differentiated in studied systems.

The question of adequacy of the current regulatory framework raises a broader question on how scientific advances, including access to them, should be balanced against protections relating to the ethical, legal, societal concerns and technical challenges. While the principle of proportionality is a commonly used tool to balance competing rights and interests, the practical application could differ, depending on the doctrines relevant in the legal orders. Moreover, the outcome could differ, depending on, for example, values that are aspired to be protected and potential benefits that could be gained. In light of the diversity of concerns that have been raised, a fundamental question is whether the different legal orders, given different cultural and societal backgrounds, have similar views on the values that need to be protected.

The existing ambiguities, as well as potentially diverse regional regulatory approaches, could suggest a need for the introduction of a specific legal instrument that would comprehensively address different legal including human rights issues related to human germline gene editing. This would require addressing basic questions about global governance of human germline gene editing. Besides the substantive question of "how", a question about "who" would come into play. Since human germline editing gives rise to numerous human rights challenges, it would be justified to argue that all global and regional human rights orders may intervene. If an action would be taken at the regional level there is, however, a risk of substantial differences. This would speak in favour of a global regulatory response, which, going back to the question of "how", raises its own set of challenges. Global legal regulation of germline gene editing would necessitate reaching a consensus in ethically controversial issues related to the moral and legal status of the embryo or permissibility of intervening in the "natural order". This would require balancing competing rights and interests and, once again, might lead to different results in different legal cultures.

7.1.2 National legal orders

Among the countries included in this research, there is a variation of national legislation governing embryo research. In those countries that allow the use of embryos for research, it is occasionally unclear if that entails a possibility of germline modification. Germline modification is generally not permitted, however, in clinical application. Such a prohibition is also included in the EU Clinical Trials Directive and Regulation. Exceptionally, for example, the UK has enabled mitochondrial transfer for reproductive purposes. None of the countries has set any bans on animal use for germline gene editing in pre-clinical studies, which in the case of EU countries in light of the prohibition at the clinical research stage may raise questions about regulatory consistency.

On the other hand, it can also be seen as a means to obtain knowledge to revisit the approach taken in regard to regulating science eventually. It has been suggested elsewhere that a better understanding of categorical deontological objections to clinical germline gene editing is needed to inform future policy decisions. The gaps and ambiguities that are shown by the survey of the laws in countries included in this report speak in favour of this recommendation.

Differences emerge in regulating clinical trials and clinical care in regards to human germline editing. Some countries have highlighted that explicit bans do not exist. Some others have raised concern regarding the scope of application of the current legal frameworks, in particular, whether they apply to health-related or also non-health related germline-editing interventions.

The question of germline gene editing has received some attention in the academic legal debates at the national level. The main line of inquiry triggered by the development in the field and pursued by national

legal scholars seems to concern the effectiveness and appropriateness of existing legal frameworks. Scholars have suggested a range of possible regulatory responses ranging from a moratorium for germline experiments to more lenient approaches if the technology is deemed safe (enough). As far as actual regulatory responses are concerned they consist of legal and policy discussions on the existing legal frameworks and address challenges arising from the need to apply them in a technologically more advanced context.

7.2 Genetic screening

7.2.1 International and regional human rights legal orders

Genetic screening raises many specific legal and human rights issues. Among the surveyed legal orders only the CoE has a detailed framework on genetic screening and the protection of individual rights in that regard.

The challenge of availability, accessibility, acceptability and quality of the screening programmes can be seen as falling under the right to health which is guaranteed in the legal instruments of all surveyed human rights order. As already mentioned in the discussion to the previous section the actual scope of the right to health remains, however, within the broad discretion of the state that decides on how to allocate limited resources.

The risk of singling out and stigmatizing certain groups may be addressed under the existing prohibitions of discrimination common across the different legal orders. Protection against discrimination is also guaranteed in the instruments that specifically address genetic screening or questions relating to the human genome. As far as disability is concerned, existing legal instruments protecting the rights of people with disabilities could be relied on to overcome discrimination and stigmatization based on genetic features.

The nature of genetic screening may create pressure and challenge the voluntary character of the decision to participate in the procedure. Voluntariness of screening is protected by general norms protecting privacy and human integrity, and more specifically by the protection of informed consent. These instruments may not, however, be effective in protecting against pressure. Moreover only a few instruments, i.e., those that *expressis verbis* address human genetics and genomics, provide for the requirement of counselling. These add to doubts whether the current general protection of privacy is adequate in the context of genetic screening.

Questions related to reproductive freedom and the right to decide whether or not to proceed with conception/continuation of pregnancy following risk identification are not directly addressed by international or regional legal orders analysed in regard to this question. However, challenges related to the tension between guaranteeing reproductive freedom and respect for people with disabilities are discussed in greater detail in the section on prenatal screening.

Protection of genetic data and information is guaranteed across legal orders under general protection of privacy and/or specific provisions related to personal data; in AU, the Convention on Cyber Security and Personal Data Protection remains to enter into force. Among the legal orders that address the possibility of further use of data or samples (UNESCO, CoE) constraints emerge, permitting the use of samples on data generally for research purposes. While this is in line with the presumption of trust vested in research, it cannot be precluded that the increasing scientific advances can create pressure to revisit the current approaches.

7.2.2 National legal orders

Regulation of genetic screening as a matter of public health generally falls within the state's discretionary power. This explains why national regulatory responses to genetic screening differ significantly in terms of whether it is separately addressed or as a part of general screening regulation, the conditions under which screening is possible (also in terms of authorization and requirements), rights and obligations of participants, oversight and issues of liability. Relatively few recent regulatory developments in the area of genetic screening have been reported.

As far as the scholarly debate is concerned national legal scholars do not widely discuss the issues of screening. Specific issues that are addressed the concern, for instance, terminological confusion (i.e., the relation between "screening" and "testing"), the risk of discrimination and other undesirable consequences, as well as or the concept of "genetic sovereignty". As the science progresses and more preventive and early interventions can be taken to tackle diseases with genetic background, states might face a need to revisit their regulatory approaches in fulfilling the right to health obligations.

7.3 Genetic testing

7.3.1 International and regional human rights legal orders

Genetic testing, mainly due to the character of genetic data (e.g., its' complicated nature and serious implications for persons besides the one being tested) raises a specific set of legal issues. Human rights law provides a certain degree of guidance on how to address them. It is rather undisputed that it protects the right to receive results of the test. As far as the scope of information a person should be provided to make informed choices is concerned, it is not regulated in greater detail. Regarding the "right not to know" it is explicitly protected in acts adopted by UNESCO and CoE, though one could argue it is also protected under general provisions on the right to privacy, or self-determination as protected in the AU. Similarly, as far as rights of family members are concerned, this issue has been addressed in UNESCO and CoE, but more generally, while protection of genetic data is guaranteed, the right to data protection is not absolute, and thus under certain circumstances, genetic data could be disclosed to family members.

Human rights law provides limited guidance on how to avoid the negative effects of patenting genes. About this issue, exclusions from patentability on ethical and social grounds envisaged by the patent law need to be taken into consideration. However, the area of international and regional patent law has been beyond the focus of this report. Within the framework of human rights law question of patentability and access may be tackled under the right to health and the right to science and right to property; however, the normative content of the right to science is still debated, and the right remains underdeveloped. As far as the involvement of children's right is concerned, human rights instruments are consistent in guaranteeing children's privacy and their involvement in decision making adequate to their competency to make decisions. There is however little specific guidance on what constitutes "child's best interest" and how to balance conflicting interests in the context of genetic testing. The UNESCO and CoE in so far as having addressed the question, have tipped the balance over enabling testing for immediate benefits to the child concerned, in that way also attempting to safeguard genetic privacy and retain room for the child's self-determination once decision-making age has been achieved.

Genetic data is a valuable resource for research. Consequently, human rights norms generally enable further processing of these data for research purposes. This is usually anchored in the notion of trust in science. As science and technology advances, these data and sample banks are of greater interest for other purposes, including criminal investigations. Therefore, a potential area of concern is a question of maintaining trust in research, compatibility of research regulations with data protection frameworks that

enable further processing of these data, and enforcement of these approaches vis-à-vis the private and public data and sample banks.

7.3.2 National legal orders

Similarly to screening, states have taken different approaches in regulating genetic testing. In the case of some states, there is no regulation on genetic testing, and legal questions are tackled by reference to general provisions applicable to the area of health law. Other countries adopted specific laws. In both cases, domestic regulation is decisive in addressing legal issues raised by genetic testing. Adaptation of general rules, for example on informed consent, to specific nature of genetic testing may, however, not be enough to protect human rights at stake adequately. In all states included in the research genetic testing is offered both within the health care system as well outside the health care setting. Requirements tend to differ in those cases. This may lead to further gaps in the protection of individual rights.

National legal scholars focus on a variety of issues, though the human rights relevance of genetic testing is a common feature of national scholarship in different countries. The need to strike a balance between different interests at stake and deal with uncertainty has also drawn attention. More specifically scholars discuss issues of consent, counselling, and secondary findings. In those countries that have not adopted specific laws on generic testing, the need for such acts has been raised.

As far as legal developments are concerned, it is possible to distinguish certain trends. First, some countries are working on specific legislation or at least a need for such a law has been officially voiced (e.g., Poland) or reviewing existing legislation in light of new developments (e.g., France). Secondly, states have considered adoption of legislation for a specific context, e.g., use of DNA from crimes scenes (e.g., Germany) or for the area of reproductive health (e.g., Brazil). Lastly, EU Member States are waiting for the IVDMD Regulation to be applicable, which could, to some degree, tackle gaps and ambiguities relating to regulating the quality of genetic testing. However, as has been reviewed in Chapter 5, this concerns health-related testing and regulation of non-health genetic testing remains the responsibility of national legal orders.

7.4 Prenatal testing and screening

7.4.1 International and regional human rights legal orders

Among identified international standards, only CoE recommendations and UNESCO IBC Report - soft law instruments – explicitly refer to the prenatal genetic testing/screening. This shortcoming of direct references even in soft law standards may be perceived as a noticeable gap in the international human rights law. However, most human rights issues specific to the prenatal genetic testing/screening have been addressed in some form in the international and regional legal orders that have been reviewed. In literature, prenatal genetic testing/screening is often described in connection to increased reproductive autonomy, and the latter largely depends on the rules on termination of pregnancy. The UN and each of analysed regional systems refer to the right that is most often invoked in the abortion context (namely right to life, prohibition of cruel, inhuman or degrading treatment, right to privacy, right to health and prohibition of discrimination). Moreover, within each of the studied systems except the ASEAN, termination of pregnancy has been directly addressed within their human rights frameworks, either through hard or soft law measures or through the enforcement mechanisms. The identified UN and AU standards seem to be the most supportive for the pregnant women rights – the AU even regulates authorization of medical abortion (in certain cases) in a hard law instrument (AU Protocol to the African Charter on Human and Peoples' Rights on the Rights of Women in Africa). The CoE ECHR jurisprudence

tends to leave a wider margin of appreciation for the Member States in this context. It is unclear yet, how the UN ICCPR GC 36 will be accommodated in the regional legal orders.

A direct reference to conscientious objection in the medical care – which may raise problems with practical access to abortion services – has been identified in two cases: the CoE Parliamentary Assembly invited member states to guarantee a right to such an objection, but at the same time called for adequate guarantees for patients, while the CEDAW Committee more straightforwardly "expressed concern over the lack of access to abortion services due to laws permitting conscientious objection by hospital personnel".

The challenge of sex-selective abortions, which have been identified by some as one of the risks associated with the use of prenatal testing/screening, has been addressed in the UN and CoE soft law, i.e., by UN CEDAW and in the CoE Parliamentary Resolution – in both cases the bodies expressed concern and called the state to take measures to tackle these practices, a soft measure has also been taken in the AU. A hard law CoE instrument, the CoE BMC, generally prohibits sex selection, but only in relation to medically assisted procreation, not mentioning abortion. While reasons for that could be related to the conventions' silence on abortion, failure to take a stringent stand on sex-selection more generally can be seen as a shortcoming.

In the context of the possible tensions between reproductive rights and respect for persons with disabilities, the joint statement regarding abortions and disability, issued in 2018 by UN CEDW and UN CRPD, is especially worth noticing, as it strongly emprises that access to safe and legal abortions is a prerequisite for safeguarding women's human rights. This approach, arguably, marks also a limited effect for the CRPD in addressing abortion relating to discrimination and signifies tendencies to seek a harmonious interpretation of various co-existing regulatory tools within the same legal order.

The issues of access to prenatal testing/screening and the standard of care, besides of being grounded in more general right to the highest attainable standard of health (already discussed previously), may be related to some obligations of special care of pregnant women that are imposed in the each of the discussed system's hard law.

The prenatal testing/screening also raises the issues of genetic privacy and right not to know and informed consent, but these have been discussed in the context of international human rights law previously.

7.4.2 National legal orders

Generally, prenatal screening/testing is in some way regulated in all of the surveyed national legal orders. It is overall permitted in the surveyed national legal orders, but the regulatory approach varies – some states only guide conditions when it can be conducted (e.g., Sweden), whereas in some others it is addressed as an exception from criminalized activities (for example Greece). However, the greatest difference in practice is noticeable regarding the extent, to which it is permitted and under what conditions, and how that correlates with the accessibility of abortion. In general, it is usually permitted to detect serious illness or other genetic abnormalities of the child and health risks for the pregnant woman. However, what falls within these notions is often ambiguous, and this ambiguity has commonly been scrutinized by scholars. PGD is in some cases regulated separately and permitted under strict conditions and offered with counselling. In all surveyed states prenatal genetic testing/screening is voluntary, and the consent of the concerned person (or persons) is required. In all surveyed states, except for Brazil abortion is permitted under certain circumstances, but these conditions vary. However, neither of the

legal orders prohibit abortion relating to disability. Notably, while Brazil permits prenatal screening and PGD, it does not allow abortion.

The legal debates and developments differ among the surveyed states. Some of the discussed topics refer to the interpretation of unclear legal terms that determine the scope of application of prenatal testing/screening or PGD (Greece, Germany) or whether the current scope of tested/screened abnormalities or illness should be extended (France). Debates in a number of surveyed state oscillate around the access to prenatal screening/testing, either in the context of including it in the public healthcare system (Brazil, South Africa), in relation to unequal access to it in different regions of a country (Sweden) or in connection to the problems of conscience clauses and how in practice they negatively affect the access to the tests (Poland).

7.5 New-born screening

7.5.1 International and regional human rights legal orders

New-born screening poses a series of challenges related to the fact that new-borns are unable to consent. This leads to questions on how to safeguard their rights and interests in the context of making decisions about screening and what are the parents' and state actors' roles in this sphere. Beyond the decision about conducting screening itself, further questions arise regarding among others secondary uses of the samples stemming from new-borns, e.g., for research.

The analysed sources of international law do not address directly new-born screening. The closest relevant reference may be found in the CoE APGT and UNESCO Declaration on Human Genetic Data, which set forth conditions under which minor can undergo genetic testing. Both adopt a similar logic. According to the UNESCO Declaration on human genetic data, genetic screening or testing of a minor is acceptable only when it has important implications for the health of the person and have regard to his or her best interests. Similarly, while the CoE APGT *expressis verbis* addresses screening measures, in regards to individual rights it defers to the general provisions of the protocol.[354] Under the CoE APGT, as a rule, genetic testing should not be conducted on a minor, it should be deferred until she or he attains a capacity to consent – unless that delay would be detrimental to his or her health or well-being, which includes both negative effects on therapeutic and preventive measures, However, the differences in genetic screening and individual testing should also be kept in mind when regulating further access to new-born screening nationally.

The ASEAN and AU do not provide such specific conditions, but they include more general principles on children rights, from which such conditions could be possibly inferred, like principle of best interests of the child (AU) and of special assistance during this period of life (ASEAN), as well special obligation regarding protection of their health. The OAS system does not include any child-specific guarantees, and these issues would have to be tackled within a general framework.

The situation is similar about legal answers to the problem of research on samples stemming from new-born screening – the most directly informative norms were identified within the UN and CoE systems. They allow it only in exceptional cases, with a specific risk-health benefit relations: the concerned person may be exposed only to a minimal risk (or burden), and the research is intended to contribute to the health of another relatable person (in the same age category or with the same genetic conditions) – i.e., in a way bring some at least potential benefit.

[354] Council of Europe, Explanatory Report to the Additional Protocol to the Convention on Human Rights and Biomedicine concerning Genetic Testing for Health Purposes, ETS 203, para. 146.

7.5.2 National legal orders

In all surveyed countries but one (South Africa) new-born screening is regulated in national legislation – but the regulations are very differentiated between the states They differ largely as to the scope of screened conditions, as whether the screening is compulsory (e.g., UK, Brazil) or voluntary (for example France, Germany, Netherlands, Spain, Sweden). The survey also revealed a varied approach to data and samples retention – they are destroyed after examination in certain countries (for example Germany) or held for years in others (e.g., UK). Furthermore, in countries where data or samples are held, there are differences regarding the purposes they are used for (e.g., in the Netherlands they can not only for used for scientific research but also identification of people after disasters).

Similarly to the diversified regulatory landscape, the national debates also revolve around different topics, such as concerns about the use of newborn data and samples (Sweden) or privacy and ownership of stored materials (UK). There is, however, also a more common theme that stirs debates, that is the scope of conditions for which new-born screening in public healthcare should be offered (e.g., in UK, Poland, Netherlands).

7.6 Direct-to-consumer advertising of genetic testing

7.6.1 International

With concerns about its misleading effects, the legal discussion on the direct-to-consumer advertising revolves around the question whether it should be permissible at all and if yes, under what conditions or what are the adequate requirement for such advertising. The international law framework does not provide a straightforward answer to these challenges, but it sets forth some guiding principles. On the most general level, of particular relevance are the right to enjoy benefits of scientific advances and the right to the highest attainable standards of health (already discussed above). In the context of advertising of genetic testing or screening, they may speak at the same time in favour of a more permissible position (in order to respect individual autonomy in exercising these rights), as well for a more restrictive approach (in the context of state duty to actively protect individual from risks) – and therefore require a further balancing exercise from legislatures and courts.

Beyond these broadest principles, other relatively relevant norms have been identified in regional systems of CoE, OAS, and AU. The CoE APGT requires the provision of appropriate information when a genetic test is envisaged and reception of informed consent from the person concerned – and this could also cover cases of advertising of genetic testing. The AU does not address appropriate informing in the context of genetic testing as such, but it sets forth a Model Law for Medical Products Regulation, with special requirements on promotion and advertisement of medical products. The OAS, in turn, provides only some instruments envisaged for general consumer protection (by setting a platform for exchanging knowledge and experience in this field and a system for product safety warning).

7.6.2 National legal orders

There is no universal standard regarding the advertising of genetic testing or screening. In some states, it is not regulated and as such – it is possible (e.g., Germany, South Africa, China, UK), whereas in others it is explicitly prohibited (France). In many of the surveyed countries, more general consumer protection regulations prohibit misleading advertising (e.g., Poland, Sweden or China).

The legal debates are framed in similar terms: advertising of direct-to-consumer genetic advertising raises concerns, in particular in the internet context (France, Poland, and Germany), it is compared to

prescription drugs (Germany), and there have been calls for introducing a coherent regulation of this issue (Greece, UK).

7.7 EU legal order

For the field of genomics, in terms of questions considered in this report, EU law is specifically pertinent for human germline gene editing as well as genetic testing and screening. As far as specific legal issues are concerned, EU law significantly influences Members States' legislation when it comes to medical and genetic data protection and safety of *in vitro* diagnostic medical devices including devices used for genetic analysis. The relevance of CFREU, particularly provisions guaranteeing the protection of physical and mental integrity, privacy and data protection, is triggered whenever an issue falls within the competences of the EU.

As far as the EU and germline gene editing are concerned, EU legislation is particularly relevant for three stages of research. First, at the stage of allocating funding: research activities intended to modify the genetic make-up of human beings that could make such changes inheritable are not eligible for EU funding. Second, in the use of animals in pre-clinical trials. Third, at the stage of clinical trials: EU law precludes clinical trials that result in modifications to the subject's germ line genetic identity. This prohibition of germline gene modification is also endorsed in EU Advanced Therapy Medicinal Products Regulation. Although the EU law does not regulate clinical care, the fact that clinical trials resulting in changes to germline are prohibited implies that this kind of interventions should not take place at the stage of clinical care.

Although, it is not within the remit of EU competence to decide what genetic screening or testing services should be offered to the public and under what conditions, EU law needs to be considered in addressing many of the issues raised by testing and screening. EU law is of high relevance to the regulation of products used for testing and screening, e.g., *in vitro* diagnostic medical devices. EU law on *in vitro* diagnostic medical devices set forth safety and performance requirements. The EU law does not cover devices used for in-house and/or non-health regulated genetic analysis, and general safety provisions apply. In terms of ethics and protecting the rights of a person undergoing a procedure, IVDMD Directive refers to CoE Biomedical Convention. IVDMD Regulation refers to the CFREU. While EU Regulation is not intended to affect national law on health services and medical care, the new regulation addresses however the need to provide necessary information on the nature, significance and the implications of the genetic test to the individual being tested (or their legal representative). Nonetheless, this framework remains to have a limited effect on tackling devices only falling within its scope – generally, those relating to health.

In processing biological samples in genetic screening and testing, as well as other data relating to an individual so that genetic analysis can be provided, the applicability of GDPR is triggered. Medical and genetic data is considered sensitive data. Although the EU law has adopted extensive legislation that addresses legal issues raised by genomic (e.g., the data protection issues), specifics still can be determined (e.g., the use of health and genetic data for research). Different grounds for processing may be applicable. Furthermore, GDPR allows for further processing of data for scientific purposes. Member states can apply more specific conditions on the processing of genetic and health data, which leads to diverse genetic data protection frameworks at the national level. GDPR does not in any particular way expressly addresses protection of the embryo's or foetus's data. Finally, genetic testing is not exempted from the scope of the Cross-Border Healthcare Directive. Unfair Commercial Practices Directive prohibits misleading actions in advertising which should also apply to direct-to-consumer advertising of genetic tests, however, more

generally, EU law does not place any restrictions on advertising genetic testing directly to consumers as such.

8. Conclusions

The overall aim of this report was twofold. First, it was to examine how the law currently responds to challenges in the area of genetics and genomics, and identify what challenges, limitations and gaps emerge. Secondly, it was to identify key human rights norms and regulatory approaches that could be examined further for shaping legal responses to the new and emerging technology in the area with due regard to competences and authority of various actors. The two aims were interrelated.

As far as specific legal challenges (annex 1), legal and human rights questions are concerned (annexes 2), some of them are common across the field and have to do with the very nature of genetic data and information. These include, for example, questions about the rights of family members or the secondary use of samples and data. Other challenges, e.g., how to reconcile respect for people with disabilities and reproductive freedom are voiced in a more specific context, in this case, human germline gene editing and prenatal screening.

Regarding specific questions considered in this report, bearing in mind the current developments in the area of germline gene editing, one of the major challenges is related to the adequacy of the current restrictive approach towards germline gene editing.

The analysis of the national orders revealed a great variety of regulatory approaches. From the most fundamental decisions on whether to allow or to prohibit the use of some of the studied technologies, through the scope of regulations, the practical arrangements required for performing research, testing or screenings, up to the methods of regulations – the surveyed states presented a much-diversified landscape. To some extent, this also refers to the countries that are members of the CoE and member states of the EU.

As far as regional human rights regimes are concerned, CoE has been a “frontrunner” in addressing the ethical and human rights challenges raised by genomics. The scrupulous overview of the existing international and regional laws pointed to some possible challenges. First, when comparing different regional human rights orders and setting them against the international standards, a risk emerges that different regional cultures may, in the context of challenges posed by genomics, adopt a divergent conceptualization of the same rights (e.g., the right to privacy or self-determination (in AU), and the right to health). This could lead to diverse and fragmented national regulatory approaches, and difficulties for sustainable collaborations. Moreover, it remains to be seen whether general human rights provisions will provide enough guidance in addressing specific concerns.

More generally, with important, but limited exceptions of certain instruments of the EU, CoE, and UN, most of the analysed sources of international law do not explicitly address genetic editing, testing or screening, but rather sets forth general principles and human rights of a broader scope. To a certain extent, this might be justified – being technologically neutral makes them flexible and allows remaining applicable on a general level despite technological and societal developments. On the other hand, the identified general principles and human rights themselves usually do not give conclusive answers as to the adequate legal responses to genetics and genomics challenges. They often point to different or even

opposite directions and, as the literature review has shown, they may be used to advocate for different positions is the key controversies.

However, international human rights acquis offers at least three concepts that have been designed to help in such situations. First, there are methods of balancing competing rights and interests, with the principle of proportionality as probably the most common tool. Second, the idea of tripartite state obligations – to respect, protect and fulfil – applied to the key human rights norms from different legal orders, mapped in this report, may be used as a framework to include a wide variety of interests at stake in the face of challenges brought by developments in genetics and genomics. Thirdly, the indivisibility of human rights. For example, the duty to respect freedom of scientific research and the right to enjoy benefits of scientific advances would speak for a more permissive approach with fewer interventions from state and international actors. At the same time the duty to protect, among others, right to the highest attainable standard of health, right to privacy, freedom from discrimination, disability rights or principle of dignity, would allow to express many other ethical, legal and social concerns and set limitations for the duty to respect. Similarly, the duty to fulfil could enable to address concerns about equal access to the highest attainable standard of health, about equal enjoyment of benefits of scientific advances or connected to reproductive rights.

In this sense, the juxtaposition of the identified challenges related to genetics and genomics with the mapped relevant international human rights norms proves that human rights framework may provide for an important point of reference for shaping legal responses. At the same, the analysis suggests that in many aspects, the existing human right sources offer rather a starting point for further examinations and elaborations than a closing argument. In our view, the starting points should be grounded in the commonly shared civil and political rights and social, economic and cultural rights across the international human rights also enshrined in the CFREU. Simultaneously, due regard should be taken to the already found regional solutions; this, however, should not exclude the need to revisit them should that appear necessary. Although the EU could contribute to shaping the field of genomics within the EU and beyond, that has to be with due regard to the limits of competence, on the one hand, and aspirations for a social Europe on the other hand. The use of various regulatory tools, including soft measures, should be further scrutinized in SIENNA task 4.2 to maximize the effects of any incentives EU could possibly take in the field.

Specific considerations for task 4.2 regarding way forward

To work towards revisiting the existing frameworks, some additional considerations need to be made. Here, we present critical areas of concern that could affect future legal frameworks.

- A critical challenge that advances in genomics present relates to the understanding of human dignity, which is a cornerstone to the present-day human rights instruments. The challenges posed by human germline gene editing require reassessing what is the object these instruments seek to protect, and whether the human genome has any role to play in that regard. This understanding could have an impact on other rights relevant in the area.
- All persons are equal. Nonetheless, some individuals and their groups merit further protection and consideration, including persons with disability, women, children, minorities and marginalized groups. Deliberations over how any regulatory advances impact such groups as the listed is necessary.
- In the area of genomics, genetic information has a particular value. Reconceptualization and understanding of access to genetic information and the development of personalised medicine could give rise to considering whether access to one's genome should and could eventually be enshrined in the human rights instruments, either as a self-standing right or derivative from other

rights and acknowledged in the soft law tools. This understanding could affect legal frameworks on genetic screening and testing at various stages of human development.

- Genetic data and their future value and meaning in a globalised world is an area that should be further assessed. The reviewed legal orders have gone in the direction of setting forth protection to this information or revising the previously existing frameworks and setting stringent requirements. We have not identified any critical considerations regarding these approaches, namely, whether more protection/heightened protection it is the right or necessary way to go, or if other alternatives should be examined, for example, protections against misuse of genetic data or information.
- Research, use, and further use of biological material and genetic data is another area that needs further attention. In light of the diversity of public interest, it could be questioned whether is it valid to maintain the current approach many frameworks set, namely, research, on the one hand, and other uses, on the other hand? One can question whether the diversity of these "other uses" does not require further differentiation, which could serve as grounds for revisiting the existing regulatory frameworks.
- Genomics is a rapidly developing area. Whether everything that is scientifically possible, should be applied to humans? In that regard, the right to science is an area that should be further scrutinized. These findings could inform the right to health, as well as the protection of civil and political rights.
- Individual's perspective in both in light of various modalities, such as incidental findings, the return of individual research results, a copy of data undergoing processing. While currently policies and approaches vary, it should be duly considered that they can change over time and be empowerment tools that directly anchor in and further the right of health.
- Lastly, it is not only what to address but also how to address. National studies have shown challenges in light of responding to new technology in the area. These challenges emerge not because the lawmaker would have deliberately created then. They commonly appear as a deficiency in constructing a particular legal provision in a way that is not capable of accommodating technological developments and diverse applications, e.g., health versus non-health genetic testing, human germline editing per se or only that relating to health purposes. Finally, these modalities require also reconsidering periodic revisions in light of the duty to respect, protect, fulfil, as well as ensure oversight and enforcement of the legal requirements.

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Annex 1 Areas of inquiry

- Access to scientific advances and inequality in that regard
- Access, voluntariness and consent
- Advertising of genetic testing or screening
- Animal involvement in pre-clinical research in light of the overall legal framework
- Are patients making an informed choice and get appropriate counselling? How, if at all, return of results and informing family members, is balanced against the right not to know and confidentiality?
- Availability, accessibility, acceptability and quality of the screening programmes
- Decision-making about screening
- Elimination of certain features or diseases triggers disability, equality and diversity concerns
- Gene patents, impact on access to genetic tests
- Genetic testing in minors
- Governance of human germline gene editing
- Human embryo involvement in research
- Impact on future generations human germline editing could have
- Impact on people with disabilities
- Informed choice
- Protection afforded to human genome
- Reproductive autonomy and freedom to decide of to have an offspring with genetic impairments
- Reproductive choices, reproductive freedom, ability to decide whether or not to proceed with conception following risk identification
- Right not to know
- Risks of singling out and marginalizing some societal groups
- Safety considerations regarding of germline editing technologies
- Scope of application of human germline editing technologies
- Secondary use of genetic information, data and samples, genetic testing for non-medical purposes
- Standard of care and liability
- Storage and use of genetic data and information
- Voluntariness of screening and choice
- Whole-genome sequencing in new-born screening, conditions in new-born screening

Annex 2 List of human rights and legal concerns

- Access to genetic testing by minors
- Access to justice
- Beneficiary of human rights protection
- Child's best interests
- Competence and authority to regulate human germline interventions
- Conditions included in the screening programs
- Confidentiality
- Consistency of regulatory strategies
- Counselling
- Data protection
- Decision-making, about NIPT, about reproduction
- Dignity
- Disability as grounds for terminating pregnancy:
- Disability rights
- Discrimination
- Equality
- Family interests/ rights
- Genetic discrimination
- Genetic privacy
- Immediate and future health benefits as part of the right to health and right to science
- Incidental findings
- Information about the results of the test Counselling
- Informed consent
- Legal status of human embryo
- Legality of animal involvement and objective
- Liability
- Patenting
- Permissibility of direct-to-consumer advertising
- Private life/privacy
- Property rights
- Protection of integrity
- Protections afforded to embryo under such human rights, including principles, as dignity, right to life and right to private life.
- Public health
- Quality of genetic testing/screening
- Regulating the termination of pregnancy
- Requirements for direct-to-consumer advertising
- Respect for diversity
- Right not to know
- Right to information
- Right to private life/ privacy /liberty
- Right to science
- Right to sexual and reproductive health
- Right to the highest attainable standard of health, including for special groups of persons, for example, minors
- Scope and limits of self-determination
- Secondary use of genetic data, information, and biological samples
- Sex selection
- Stigmatization
- Voluntariness of screening programs
- Wrongful birth/life considerations

Annex 3 National reports

Brazil
China
France
Germany
Greece
Poland
South Africa
Spain
Sweden
The Netherlands
United Kingdom
United States (draft)