Cracking the code to individuality: DNA analysis in the service of self-knowledge and police investigations

Abridged version of the study «Neue Anwendungen der DNA-Analyse: Chancen und Risiken»

TA-SWISS, the Foundation for Technology Assessment and a centre for excellence of the Swiss Academies of Arts and Sciences, deals with the opportunities and risks of new technologies.

This abridged version is based on a scientific study carried out on behalf of TA-SWISS by an interdisciplinary project team under the leadership of Dr Erich Griessler and Alexander Lang from the Institute for Advanced Studies in Vienna. The association Open Science in Vienna and the University of Lucerne also participated in the study. The abridged version presents the most important results and conclusions of the study in condensed form and is aimed at a broad audience.

Neue Anwendungen der DNA-Analyse: Chancen und Risiken – Interdisziplinäre Technikfolgenabschätzung

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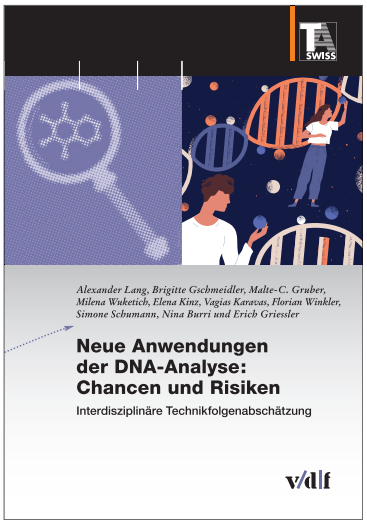
TA-SWISS, Stiftung für Technologiefolgen-Abschätzung (Ed.)
vdf Hochschulverlag an der ETH Zürich, 2020.

ISBN 978-3-7281-4037-1

Also available in open access: www.vdf.ch

This abridged version can be downloaded at no cost: www.ta-swiss.ch



New applications in DNA analysis in brief

Until a few years ago, genetic analysis was primarily reserved to the fields of research and medicine and was used to confirm medical diagnoses or the predisposition to a serious illness. Today, DNA testing can also be used to reveal physical characteristics that are not of particular medical relevance. As the prices for genetic tests decline, an ever broader public is gaining access to them.

DNA can tell us a lot about a person: which nutrients they metabolise especially well (and which ones their bodies will tolerate poorly), how resistant their body is to physiological stress and where their ancestors originated from. At least that is what the providers of genetic tests like to claim. These companies are usually located abroad and offer direct-to-consumer (DTC) genetic tests to Swiss consumers through the Internet. Police investigations, on the other hand, apply these new scientific findings to narrow down the pool of suspects based on genetic trace evidence collected at the scene of the crime.

Opportunities

DNA analysis tells us something about ourselves. What we find out can be helpful in many cases – for instance, when a test subject learns that their excess weight is associated with a gene that regulates how sugar is metabolised in fat tissue and is not simply a matter of lifestyle.

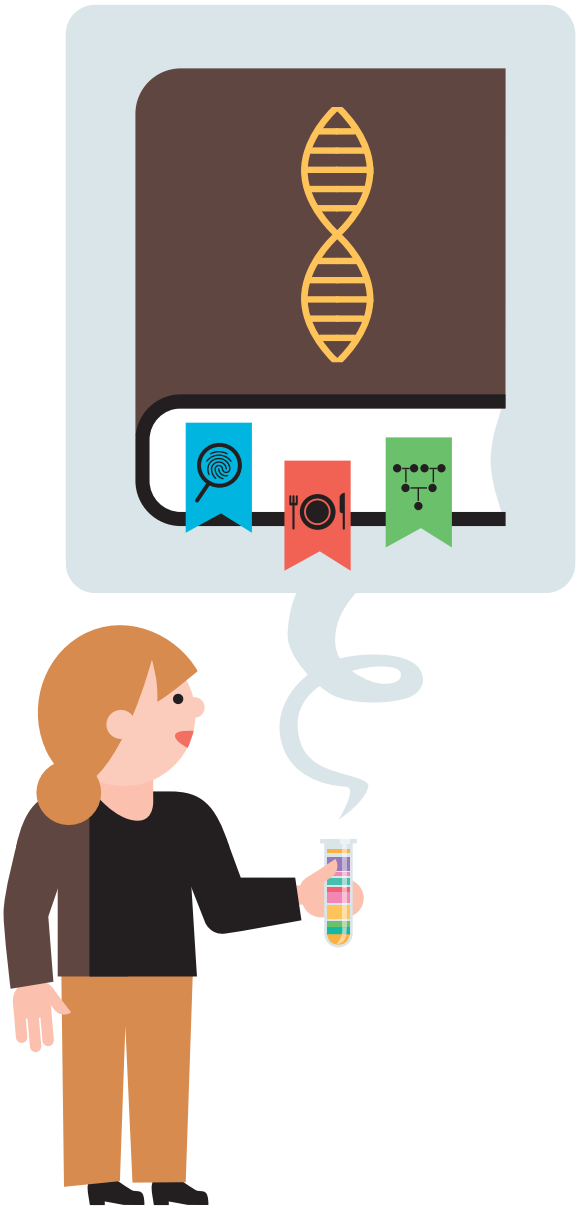
DNA evidence collected by police at the scene of a crime can reveal physical characteristics and the ancestry of the perpetrator and thus serve to exclude entire groups from suspicion or exonerate the wrongly accused.

Genome data are an important raw material in research. As part of their business models, many gene testing companies pass on the genetic data they analyse to research institutes and pharmaceutical companies, which may use them in the development of new therapies.

Risks

Providers of genetic tests typically work together with numerous partners. Customers are in many instances left in the dark about exactly who performs the analyses and who has access to which data, and they are often inadequately informed about the accuracy of testing – as well as about the measures in place to protect their data.

The law prohibits suppliers of direct-to-consumer genetic tests from revealing medically related information to customers. In practice, however, findings



classified as «not medically relevant» – such as those pertaining to metabolism or cell ageing – can nevertheless permit conclusions to be drawn about medical issues.

The law that currently regulates human genetic testing – the Federal Act on Human Genetic Testing, requires that persons who undergo genetic testing for medical issues receive medical counselling. Such counselling is often not available when a consumer purchases a genetic test online from a foreign supplier.

Recommendations

Transparent handling of data and informing customers in as much detail as possible have to be improved. Consumer protection organisations have an important role to play in this respect.

The personal and social impacts of genetic tests intended for lifestyle optimisation or ancestry research are unclear. They may, for instance, heighten bias against certain groups of people – or, in contrast, they may encourage people to take more responsibility for how they deal with their own strengths and weaknesses. Systematic research is therefore needed on the practical use of genetic tests and the related consequences.

Providers are not in the habit of ensuring that the sample submitted for testing is actually from the person who ordered the test. This is a particularly controversial issue when the genetic test is used as a cover for a paternity test. Providers of genetic tests should be forced to take precautionary measures to protect third persons, especially minors.

Genetic data not only reveal things about the individuals undergoing the testing, but about their relatives as well. For this reason, regulation models need to be devised that address the collective dimension of genetic information. For example, consideration should be given to endowing relatives with the rights of co-determination and consultation for the case that data are to be used for research purposes.

Understanding people through genetics

Until a few years ago, genetic testing was exclusively the purview of medicine. The predisposition for a number of diseases is detectable in the genome before the disease breaks out. DNA testing can now also be used to reveal physical characteristics that are independent of a person's medical condition. The cost of genetic tests has fallen dramatically, giving rise to testing offers that directly appeal to a broad consumer base.

Knowing oneself as well as possible and understanding one's strengths, weaknesses and predispositions are driven by the need to come to terms with one's own identity. For a long time, the illumination of personality was an endeavour mainly left to the fields of psychology and philosophy. In recent years, biology has also gained a voice.

Shrinking costs, growing customer base

The Human Genome Project was launched at the end of the 1990s, initiating the effort to map the human genome. The USA alone contributed over three billion US dollars to the ten-year international research campaign. In 2006, the cost of sequencing a single human genome was 14 million US dollars. By early 2015, advances in the analysis process had reduced the cost of genome sequencing to 4,000 US dollars, and by the end of 2015 the cost had dropped even further to less than 1,500 US dollars. At this price, the procedure was affordable for a broader public seeking to learn more about their physical make-up independently of any medical issues.

A variety of providers of genetic tests rushed to fill the niche. Operating mainly from abroad, they offer their services to Swiss clientele online. Having the test done is very simple. The customer orders the

test kit, which contains all the materials necessary for taking a cheek swab or saliva sample, and then submits the sample to the test provider. The results are available in anywhere between three and ten weeks. Depending on the customer's wishes, the results are geared either towards optimising lifestyle or learning more about one's ancestry.

Police investigations also make use of the new analysis capabilities, such as DNA phenotyping, to obtain clues about a potential suspect's physical characteristics based on DNA evidence from the crime scene. Phenotyping can provide information about hair, eye and skin colour, for instance. The study by TA-SWISS therefore addresses these three applications of genetic tests: ancestry research, lifestyle optimisation and forensic investigations.

Legal grey zone

The Federal Act on Human Genetic Testing has been in force since 2007 and governs medical applications as well as the potential use of genetic testing for employment-related purposes and by insurance companies. The law stipulates that the person being tested must be consenting. It also stipulates that genetic tests must be prescribed by a medical doctor and may only be performed by federally accredited laboratories.

Anyone taking a sample of their own saliva and independently submitting it to a company like ProGenom, GenePlanet, MyHeritage or 23andMe for a genetic test is thus, strictly speaking, acting in a legal grey zone. That is likely to change soon, however, because a revised version of the law is set to come into effect in 2021. The revised law will close the legal gaps surrounding commercial genetic tests that consumers can order directly.

The amended legal provisions will differentiate between genetic tests conducted for medical purposes and those done for the purposes of ancestry research and lifestyle optimisation, with different levels of regulation applying to each.

Expert study with self-experiment

The authors of «New applications in DNA analysis: Chances and risks - Interdisciplinary technology assessment», led by Alexander Lang and Erich Griessler (both from the Institute for Advanced Studies in Vienna, Austria), applied a variety of methods in this TA-SWISS study. They researched the literature and analysed the Internet platforms of various providers of genetic tests. They also conducted numerous interviews with professionals in the fields of genetics, nutrition, genealogy, ethics and forensics. Finally, they submitted themselves to different genetic tests in a self-experiment.

Human genetic material

The DNA (deoxyribonucleic acid) molecule is composed of two long, intertwined, spiral-shaped strands of chemical base pairs. The information it carries is encoded in the sequence of these base pairs. A gene is a section of DNA that contains the instructions for building one of the proteins found in the body. In specialist language it is said that the gene «codes for a protein». Variations in the «code» created by the base pairs can express as differences in human beings, such as in their metabolism. DNA tests read the information encoded in the DNA and try to interpret it.

Looking for meaning in the letters of the genetic code

Research is finding more and more genetic traits that are associated with specific characteristics of the human body. This facilitates the targeted and hence cost-efficient analysis of DNA. The legal regulation of genetic analyses differs from country to country.

The similarities among family members have always fascinated scientists. In 1875, Sir Francis Galton, a British physician, naturalist and cousin of Charles Darwin, published a scientific article entitled «The History of Twins» and thereby established the study of twins as a distinct area of research seeking to better understand the inheritance of personal traits – especially intelligence and talent. Owing to his focus on eugenics, however, Galton's work is not regarded positively today.

Scientific foundations are becoming ever broader

The possibilities offered by gene sequencing brought new momentum to the debate about the influence of genes on physical traits and behaviour. The sequence of all the building blocks – i.e. the base pairs – of the human genome is now known. Databases allow DNA traits to be linked to individual physical characteristics and some behaviours; scientists refer to this as genotype-phenotype correlations. The quantity of sequencing data is growing rapidly and is currently doubling in volume every seven months.

Human beings share around 99.5 percent of their genetic material. Given that DNA is comprised of more than three billion base pairs, even just 0.1 to 0.4 percent of the genome can still give rise to a huge number of individual differences (i.e. several million). One of the largest databases contains the data of over 2,500 sequenced genomes of people from different countries. Other databases store SNPs (pronounced «snips»), which are inherited together and are associated with certain physical or behavioural characteristics. This database input is the result of genome-wide association studies (GWAS), in which the genetic data of several thousand people are compared with one other. These studies result in numerical ratios that describe whether certain genetic traits systematically occur in combination with given physical characteristics. The providers of genetic tests also make use of the knowledge stored in these databases.

Landmarks in genetic material

Today, much is known about the variations in human genetic material and their effects on physical appearance and certain metabolic processes. For this reason, it is no longer necessary to sequence the whole genome of an individual in order to determine certain characteristics. Most commercial genetic tests concentrate on the analysis of «markers». In particular, SNPs and satellites serve as landmarks or markers in genetic material.

Procedures now exist for simultaneously analysing several thousand known markers. As only a relatively small portion of the entire genetic material has to be analysed under these circumstances, the test costs can be kept low – one of the reasons for the commercial success of genetic tests that are marketed directly to the customer (direct-to-consumer or DTC genetic tests). However, it is likely that sequencing of the entire genome will also become less expensive in the foreseeable future, thereby bringing it within the financial reach of a broad segment of the population. This would make more extensive analyses feasible.

The future is open despite one's genome

The evaluation of the enormous volumes of genome data relies on statistical methods. Hence, whether or not a given physical characteristic is linked with certain SNPs or other genetic traits – which is the question that genome-wide association studies address – is expressed as a probability. In other words, the predictions yielded by genetic analyses are not absolute, with the exception of predispositions to some diseases.

This fact is particularly significant in forensic investigations because, as of today, only a few physical characteristics – such as eye, hair and skin colour – can be predicted with a relatively high degree of certainty. Researchers are working at a high pace to zero in on other characteristics, such as facial structure and body size. Tests can yield results of varying accuracy for the expression of even a single characteristic, however. For example, brown hair can only be predicted with an accuracy of 74 percent, while the accuracy for predicting red hair is 93 percent.

Moreover, the probability for the occurrence of a given characteristic always refers to a group of individuals, not a particular person. So, if a genetic test indicates that someone is predisposed to obesity, it does not mean that that person will necessarily ever become overweight, because eating habits and exercise also play an important role.

Decrypting heritage and self

The first DTC genetic tests were sold through the Internet in the USA in the early 2000s. Ancestry analysis was offered as well as evaluations of disease risks. However, the direct sale of medical genetic tests was soon outlawed by the authorities, as the validity of the results was not reliable enough and the potential damage to worried customers was too great.

Ancestry or heritage analysis went on to become very popular, though, because in the ethnic «melting pot» of America many people are eager to learn more about their ancestors from far-away continents. In 2015, the legal situation changed. The Food and Drug Administration issued regulations that made it possible again for private people in the USA to order certain medical DTC tests that predict the risk for diseases such as Alzheimer's and Parkinson's.

In Europe, there is no uniform legal regulation of DTC genetic tests. Some countries have no laws regulating the use of such analyses, while other countries require the involvement of medical professionals or genetic counselling. In Switzerland, DTC tests are as a rule offered by companies that are domiciled abroad and operate via the Internet in cooperation with local partners. The regulations that apply in Switzerland probably contribute to making domestic companies cautious about offering testing to consumers directly, as Swiss suppliers are only allowed to market lifestyle tests directly if their use for medical purposes can be excluded.

From alleles to INDELs and SNPs

Genetic analyses identify changes in the genes as well as DNA polymorphisms. DNA polymorphisms refer to different alleles, i.e. different forms of a given gene. For instance, blossoms of carnations can be white, yellow or light to dark red, depending on a certain allele, the sequence of which determines blossom colour. There are also genetic changes referred to as «INDELs», which come about through the insertion or deletion of short gene sequences. This can suppress or heighten the expression of a gene – i.e. the new production of proteins. Point mutations, referred to as SNPs (pronounced «snips»), are also significant for genetic analyses. SNP is short for «single nucleotide polymorphism» and refers to a variation in a single base pair. SNPs are responsible for around 90 percent of the genetic variation in the human genetic make-up. Satellites are short gene segments that repeat but do not code for a protein. Genetic analyses primarily examine SNPs and satellites. If the location of a certain SNP or satellite is known, it is designated as a marker.

Exploring the boughs of the family tree

In royal households it was normal practice to legitimise claims to sovereignty through ancestry and to document them meticulously. Following the introduction of registry offices in the late 18th century, ordinary citizens were also able to trace their ancestors. Today they are able to do so with the aid of genetic analysis.

MyHeritage, a company established in Israel in 2003, has more than 100 million users throughout the world. It offers software that enables users to search billions of records to find relatives. In 2016, MyHeritage introduced genealogical research based on genetic testing. Other companies – for example FamilyTreeDNA, 23andMe and 24Genetics – are now also offering similar services. This trend is attracting a great deal of interest: the MIT Technology Review calculated that, from 2012 to the beginning of 2019, a total of 26 million people throughout the world had taken a commercial genetic test in order to trace their ancestry.

A search conducted as part of the TA-SWISS study showed that in June 2019 there were 14 companies offering genetic tests online for origin and genealogy purposes and which serve Switzerland. Today, the supplies needed for doing genetic tests to trace ancestry can even be purchased from Amazon – at least in Switzerland's neighbouring countries. These tests have thus become firmly established among Internet-savvy users for whom online shopping is a daily routine.

DNA depicted as a map

DNA test results, which users receive around two months after submitting their DNA sample, are somewhat reminiscent of a geography lesson: in addition to various diagrams, the documentation of the test results includes world and national maps on which the continents, countries and regions are depicted in a variety of colours which represent the proportions of the customer's familial origins. In other words, these maps show from where the customer's ancestors are likely to have originated and the relative number of ancestors from the geographical areas identified. Sometimes the results also refer to groups that previously lived in a given region. Thus, the genetic heritage of someone who is mainly Swiss may include some proportion of other ancestries, e.g. 20 percent German, five percent Belgian and three percent French. Depending on the provider, the time frame investigated may range from a few hundred years to as far back as the Bronze Age. In some cases, the tests even certify the presence of a small percentage of Neanderthal genes.

Providers advertise their ancestry tests as a means of facilitating a fascinating journey into family background and helping their customers discover their own roots. From a scientific point of view, these claims are questionable. This is because if, for example, a test traces the maternal or paternal lineage, only one individual per generation is identified. The further back the analysis goes, the greater the proportion of ancestors that remains hidden. In any case, the customer's ancestry becomes increasingly diverse the further back in the past it reaches. Instead of «origin» in the singular, the idea of «diverse origins» would be closer to reality.

Another approach involves considering all lines of descent based on certain markers on the autosomal DNA, which is not specific to biological gender. Here, the origin-specific markers are compared with the content of databases that have been fed with data pertaining to present-day populations. The drawback with this method is that it is based on the assumption that the people whose data are entered in the current reference database have been living in the same place for generations and thus represent the past population – a questionable hypothesis given the well-documented mobility of human beings.

Historians and genealogists have expressed concern that biologically derived ancestry is increasingly supplanting other concepts of heritage based on social, cultural or religious roots. It is improbable that even a historically well-circumscribed group such «the Vikings» would correspond to a uniform genetic sample set. Furthermore, it is not certain whether a population group that is retrospectively perceived by succeeding generations as a tribe or people (for example, the Celts) would have considered itself to have a shared identity.

On the other hand, what is certain is that ancestry analyses open up additional business opportunities for the providers. Through partner companies, for example, some providers urge customers to book accommodation in the places where their ancestors originated, so they can acquaint themselves with their material and cultural «heritage».

Search for previously unknown relatives

Thanks to the comprehensive databases to which the major providers of genetic tests have access, it is also possible to search for living but previously unknown relatives. By comparing analysed DNA segments with the data in the reference database it is possible to identify the degree of people's genetic relatedness. Biological parents, siblings and offspring, as well as grandparents, aunts, uncles and distant cousins can be identified this way – though in the latter case with a lower degree of accuracy. Only those persons are identified and contacted who have given their consent.

Providers advertise the search for relatives with the argument that customers may find family members who are otherwise untraceable. In the case of an adoption, they offer the prospect of establishing contact with the unknown biological parents or lost children. However, contrary to the message communicated in their advertising, the consequences of confrontations with unknown relatives are not always positive. For example, if someone discovers that the father they have known throughout their childhood and youth turns out not to be their biological parent, their trust in the dependability of family ties could be shattered.

In addition, under certain circumstances, a person's DNA may disclose something about relatives who were not tested and do not wish to know anything about their genetic make-up. As a general rule, it is possible to draw conclusions about untested third parties. A few years ago, for example, a forensic success story in the USA hit the headlines: decades after the crimes had been committed, a serial killer was finally caught after the DNA secured at a crime scene was compared with the data on an ancestry research platform onto which a third cousin had uploaded her genetic profile. After extensive inquiries, the authorities were able to locate and arrest the then elderly perpetrator. However, this coup by the police also gave rise to widespread outrage and concerns regarding data protection. At the same time it demonstrated that people who disclose their genetic data may also «out» their relatives.

Variety of search paths in the field of genealogical research

Autosomal (i.e. non-gender-related) DNA markers are inherited from both parents and can be used to draw conclusions about the geographic origin of ancestors on both the maternal and the paternal side. By contrast, markers on the Y chromosome are passed on by the father exclusively to his son(s) and provide information about the paternal male ancestors. Markers located in the mitochondria, on the other hand, are only passed on by the mother, though to offspring of both sexes equally, and thus enable conclusions to be drawn regarding the origin of female ancestors on the maternal side.

When genes determine our lifestyle

Genes co-determine how we utilise nutrients, for example, or cope with stress or perform in a marathon. The volume of genetic data is rapidly increasing, as are findings concerning certain genetic traits and thus related physical characteristics. DTC (short for «direct-to-consumer») genetic testing is based on this knowledge.

«Exploit your full potential»; «Get to know yourself better»; «Enhance your own well-being» – these are some of the slogans used by providers of genetic tests to advertise their products. According to one estimate, the global market for DTC genetic testing could be worth around a billion US dollars by 2021. Given the high level of purchasing power in Switzerland, the potential here is far from exhausted. Genetic testing is even more profitable for the providers because they are able to sell the data they receive from their customers to third parties, for example to pharmaceutical companies, which have a great deal of interest in this information resource.

Curiosity as main driving force

In a survey of scientists who had their genes tested, the majority stated that it was curiosity that prompted them to take this step. The second reason cited was that they wanted to use their data to support research – a motive that was undoubtedly associated with their vocation. A separate survey among students revealed that, in addition to supporting research, the search for predispositions to diseases was a major factor.

The urge to find out more about ourselves and identify potential susceptibility to sickness clearly reflects a lifestyle that is open to new technology – a lifestyle that embraces self-measurement using trackers and the constant striving for better performance and which values products tailored to our individuality (or products that at least claim to be).

Nutrition, wellness, sport

In the study carried out by TA-SWISS, eight providers of genetic testing for lifestyle optimisation were examined. All but one of them provide information in response to questions regarding nutrition and weight. On this basis they propose dietary plans, for example, that are tailored to the customer's individual genetic constitution. They analyse traits in the genes for metabolism, which provide information about the way in which the body utilises nutrients – for example, DNA variations that can give rise to difficulties in breaking down lactose («lactose intolerance»). Typical symptoms of lactose intolerance include flatulence and abdominal cramps. Another gene determines how sugar and fat are processed and in a certain form is associated with obesity, an increased risk of type two diabetes and cardiovascular diseases.

Well-being does not depend solely on nutrition. The wellness category includes tests that make it possible to determine the body's detoxification capability. How well a person is able to process alcohol, nicotine and environmental toxins, such as pesticides and insect sprays, depends on the gene variant their body carries. Some emotional states may also be influenced by DNA: a variant of a gene that steers the release of the «feel good» hormone dopamine is associated with anxiety and stress, as well as with the tendency towards drug addiction and alcoholism. Whether someone can hope to remain fit and relatively free of wrinkles into old age can be tested on the basis of genes that regulate how the body deals with oxidative stress and the capability of cells to regenerate themselves. DNA can even help determine whether someone is an early or late riser. Subjects then receive tips as to how they can adapt their biorhythm to their daily life and thus enhance their physical and mental well-being.

There are currently more than 150 genes that are believed to be capable of influencing athletic performance – above all, muscle strength and stamina. However, only few correlations have been clearly identified by the numerous scientific studies conducted on this topic to date. Genes that are associated with physical growth factors and thus with the more rapid healing of injuries are also of interest to athletes. It is mainly those participating in high-performance sport who stand to gain a genuine benefit from genetic testing, because in competitive sport, even the tiniest improvements can make the difference between winning and losing.

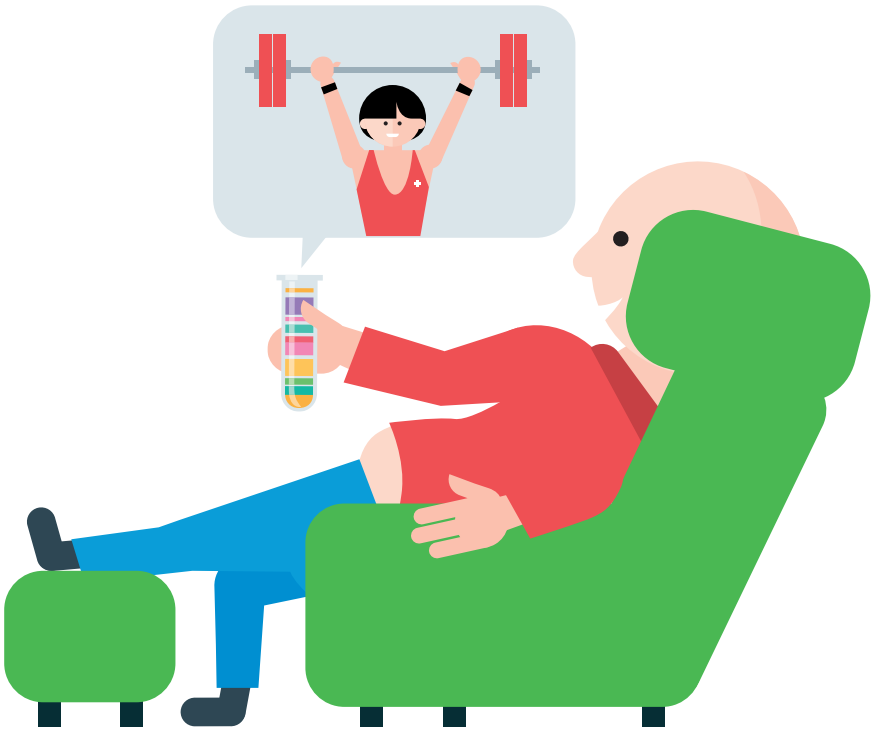
Science in the service of entertainment?

Healthcare professionals who were interviewed for the TA-SWISS study regard the majority of genetic tests carried out directly on behalf of clients as gimmicks and see them in a critical light. Some of the respondents acknowledge that while nutrition tips arising from lifestyle genetic tests can sometimes have a prophylactic value. However, experience has shown that information on its own does not suffice to change people's behaviour. Lovers of milk chocolate, for instance, are unlikely to stop eating it merely because a genetic test reveals they have a tendency to accumulate fat in their organs.

All the respondents were in agreement that no health risks should be communicated. This is precisely where DTC genetic tests become problematic, as the boundary between lifestyle analysis and medical test can be fuzzy. A severe case of lactose intolerance, for example, requires medical attention, and genes that influence the regeneration capability of cells and the ageing process are also associated with cancerous diseases.

Thus, the risk that the results of genetic testing could cause anxiety and distress should not be underestimated. A case in point is a customer who, based on a subsequent evaluation of the original test of her family's genetic raw data for non-medical purposes, discovered that her daughter had a predisposition for an uncommon connective tissue disorder. As a result of this discovery, she submitted her daughter to almost twenty medical examinations, none of which confirmed the suspicion of the uncommon syndrome. So, in addition to causing anxiety in entire families, DTC genetic tests can in some circumstances result in the unnecessary utilisation of medical resources.

Customers' exaggerated expectations can also turn into disappointment when it becomes apparent that the genetic test reveals little more than they had in any case assumed. Since other factors besides DNA, such as environmental conditions, influence a person's physical as well as psychological well-being, the usefulness of this type of genetic test is ultimately limited.



«Whodunnit?»: genetic testing as a forensic tool

For the past thirty years or so, genetic traces have also been analysed for the purpose of investigating crimes. Until recently, however, this trace evidence had to be compared with genetic profiles from specially created databases. With phenotyping it is possible to deduce the specific characteristics of individuals based solely on their genetic fingerprint.

In 1985, British geneticist Alec Jeffreys discovered that sequences in the human genome can be used for identifying people. He called his method «genetic fingerprinting», a designation he implicitly based on studies carried out by the already mentioned researcher Francis Galton, who had recognised the fact that the ridges on each person's fingertips are unique.

The UK began to compile a DNA profile database in 1995, and other countries soon followed suit. The method now familiar from TV programmes and crime films was thus born: genetic traces recovered at a crime scene – e.g. skin cells, hairs, sperm, saliva or blood – are compared with DNA profiles stored in a database. If a match is found, the culprit is identified.

Investigation without comparative data

Phenotyping takes the science of forensics – the reconstruction of criminal acts – a step further. Information about a wanted person is obtained from the trace DNA itself. Genetic traces for which no matching profile can be found in the reference database can thus also be used for investigative purposes. This method is used to localise the origin of an unidentifiable victim, for example, or approximate the physical features of an unknown suspect. This fundamentally changes the role of genetic analysis in police activity in that it no longer serves to identify or exclude suspects through DNA comparisons alone, but can also be used as an investigative tool for determining the physical features of unknown persons.

Variable accuracy

Contrary to the picture presented to us in the media, the results of DNA phenotyping are usually not clear and unequivocal. Only a person's biological gender can be determined with a high degree of certainty. Whether a female perpetrator has blonde or brown hair, or a male perpetrator has light or dark skin, can only be determined with a higher or lower degree of probability depending on the person's physical characteristics and their expression.

Of all a person's physical features it is the colour of the iris that has been scientifically studied the best. With the aid of IrisPlex, a statistical model for predicting eye colour, it is possible to determine with 95-percent probability whether a person has blue or brown eyes. Green or grey eyes are more difficult to identify and thus the probability of an accurate prediction is correspondingly lower.

Pigmentation of skin and hair has also been fairly well researched. The probability of precise identification ranges from 74 percent (brown hair), to 76 percent (light skin), 93 percent (red hair) and 99 percent (dark skin).

Research is currently being carried out on other genetic traits, such as hair type (curly, frizzy, straight), the presence of freckles and tendency toward baldness. Geneticists are also focusing attention on ear shape and body size. However, it is not yet possible to create a genuine «identikit» image based on genetic data.

DNA profiles only usable on a comparison basis

The Federal DNA Profiles Act entered into force in Switzerland in 2005. It addresses the comparison of genetic traces with samples stored in a database. In practice, investigators compare a trace collected at a crime scene with the content of the national DNA database, CODIS. Under the law, only the comparison of DNA sequences and the identification of biological gender are allowed; the law expressly prohib-

its searching for other personal traits. Typing based on a DNA sample alone is therefore not permissible.

This fact raised public awareness of a crime that was committed in 2015. On a summer evening in Emmen (canton of Lucerne) a young woman making her way home was thrown off her bicycle, raped, and left severely injured on the ground, with the result that she is now paralysed. The collected DNA sample did not yield a match in CODIS, and a mass DNA screening of more than 300 men with a potential link to the crime scene also failed to produce a result. This case set the debate on DNA phenotyping in motion. A member of the National Council submitted a motion to Parliament entitled «Kein Täterschutz für Mörder und Vergewaltiger» («No protection for murderers and rapists»), which called for DNA analysis to be extended to other physical features such as eye, hair and skin colour. The motion was adopted by Parliament and the Federal Council now has to revise the legislation accordingly. The public prosecution office of the canton of Lucerne has already announced that it will open the case again as soon as the revised legislation enters into force – probably in 2022.

Avoiding untenable suspicions

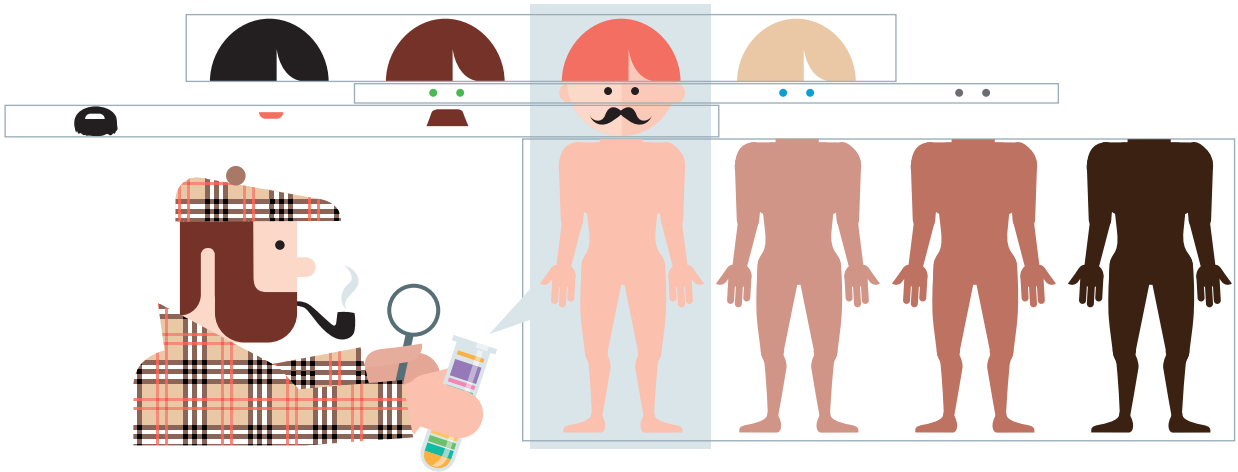
As the title of the above-mentioned motion makes clear, DNA phenotyping should only be used in the case of serious crime, i.e. for the investigation of criminal acts that are subject to terms of imprisonment of at least three years. In countries in which phenotyping is already established as an instrument for investigating crime, it is only used in connection with serious offences, and even then only if all the standard investigatory methods have failed to yield a result.

There are various reasons why phenotyping has to be used with caution, one of which is that the possibility exists that innocent people could come under general suspicion if they happen to possess certain traits (for example, red hair and blue eyes) that have been attributed to the perpetrator through phenotyping. Another reason concerns the possibility that identifying the bio-geographic origin of a perpetrator could foster racism. On the other hand, phenotyping can also help exonerate wrongfully suspected persons or exclude entire population groups from suspicion.

Even «conventional» DNA analysis may only be applied if sufficient suspicion exists and the identity of the perpetrator cannot be established through the application of milder measures. If DNA phenotyping is ordered as a mandatory measure in penal proceedings, it also has to meet these requirements.

DNA phenotyping: the dream of a genetic identikit

With DNA phenotyping, a person's physical features (for example, eye and skin colour) can be predicted on the basis of a DNA sample. However, the degree of accuracy varies according to the trait analysed, and predictions can only be expressed in terms of probability. Contrary to what is depicted in fictional films, however, it is not possible to produce a biological composite picture (identikit) with the aid of DNA phenotyping.



When the data trail is lost

Most providers of DTC genetic tests work together with numerous partners. The company that has contact with customers is not the same as the laboratories that analyse the genetic samples. Furthermore, the business model of many providers is based on getting further use from the samples. It is difficult to know who has access to which data.

Information obtained from a person's genetic material is sensitive personal data that should only be passed on to trusted specialists. Data protection is a matter of concern for many people who have undergone DTC genetic tests. In a survey regarding protection of the private sphere and confidentiality of data, more than half the people who had undergone this kind of analysis expressed major concerns, or at least some misgivings, in this respect. Around a quarter of the respondents also said they were worried that their data could get into the hands of third parties.

Who analyses what, and how?

Like numerous other providers, 23andMe uses its customers' genetic data to carry out its own research, but also places the data at the disposal of academic institutions and non-profit organisations for collaboration purposes. In addition, the comprehensive datasets compiled from DTC genetic tests attract a great deal of interest from profit-oriented pharmaceuticals companies.

Since, in the past, collaboration between providers of DTC tests and pharmaceuticals companies resulted in criticism because customers were of the opinion that they had not been given sufficient information regarding the use of their data, the providers of these tests now indicate which data they use, and how. The conditions of use and the documentation concerning the consent for research to be carried out on personal data cite potential risks, including those that could arise from future technological developments. Nonetheless, specialists doubt that customers can truly give their full «informed consent» for research to be conducted on their personal data. Furthermore, it is often difficult for customers to find out how they can refuse to agree to the use of their data by third parties by means of an opting out clause.

It is also often not clear that, as a rule, it is an external laboratory that carries out the analysis, not the provider of the DTC test. The authors of the TA-SWISS study criticise the complex structure involving partner companies, which usually handle different parts of the analysis, and the fact that the companies' websites do not clearly indicate which analyses are carried out, and where. It is also sometimes the case that comprehensible and comprehensive information is lacking with respect to measurement accuracy and technical standards.

Genetic data are not private property

Data relating to genetic disposition inevitably reveal information about biological relatives. This means that, even if people allow their genetic data to be passed on to third parties, their consent does not apply to their family members, who share many of their hereditary traits. In this context, geneticists speak of «common biosocial destinies».

Also, providers of genetic tests cannot be certain that a submitted sample truly originates from the customer concerned. It is conceivable that parents could be tempted to have their children's genes tested. This is a clear violation of the law (and essentially also infringes the conditions of use specified by most companies), which stipulates that test subjects must be autonomous and be fully informed before they consent to a genetic analysis. In the case of minors, this violation is even more serious in that the minors concerned are denied the «right to not know» and thus deprived of a wholly open future of their own.

Raw material for further interpretation

Once their sequenced genetic data are available, customers are not obliged to simply accept the non-medical results permissible by law. If they wish, they may have their genetic data reinterpreted by companies or through online platforms – without

having to submit a sample for a second test – to obtain information of relevance to their health. The boundaries between lifestyle or ancestry research and medical analysis are thus indistinct.

It is even the case that those who do not want to have their test medically evaluated may nevertheless be confronted with medically relevant information as a result of their lifestyle or ancestry analysis. The media now report on SNPs and genes with a pathogenic effect, and this means that even people with limited medical knowledge can recognise the potential of a health problem if a mutation in one of these genes shows up in their genetic profile. The fact that this is not synonymous with a genuine health risk, because risk is codetermined by other genetic traits and personal circumstances, should be explained to anxious customers by a healthcare professional.

Lack of support in the face of uncertainties and anxieties

When genetic tests are ordered by a medical practitioner – and the genetic analyses are thus carried out in accordance with the applicable legislation – a medical consultation must take place both before and after the test. Providers of DTC genetic tests cite various contact options, and some of them operate forums in which customers can exchange information and advice.

However, none of the companies specialising in ancestry analysis covered in the study offer a specific Swiss phone number where customers can get answers to their personal questions – this includes companies that supply genetic tests in Switzerland. The situation is a little better with respect to providers of lifestyle genetic tests: most offer advice online or by phone. However, the more detailed information on many websites to which customers are referred by their provider is only available in English. Furthermore, the advice provided is restricted to non-medical issues. Thus, customers who have their data reinterpreted to obtain health-relevant information and then receive worrying results are left without the support they need.

Specific legislation – vague reality

DTC tests are governed by a variety of laws, the most important of which is the Federal Act on Human Genetic Testing. The processing of collected data is regulated by the Federal Data Protection Act, while the handling of genetic traces at crime scenes is subject to the provisions of the Federal DNA Profiles Act. In addition, overlying legal texts such as the provisions of the Swiss Federal Constitution concerning human dignity also have to be complied with.

The Federal Act on Human Genetic Testing, which entered into force in 2007, governs medically prescribed tests. It therefore cannot regulate commercial DTC tests and is currently undergoing revision. The revised version is expected to enter into force in 2021.

Original legislation adapted to new reality

The main provisions of the original Federal Act on Human Genetic Testing will be retained in the revised version. For example, tested persons have to consent to the analysis and must receive pertinent information in advance («informed consent»). The existing version upholds the «right to not know», while the revised text also insists on the «right to information».

The revised Act will contain a new chapter regulating non-medical genetic analysis. This chapter will specify the traits which DTC tests will be allowed to identify. With a view to ensuring the necessary clarification for customers, the revised version will stipulate that laboratories and companies that carry out the tests must be named, and contact details must be provided for personnel who can answer customers' questions. This means that in order for currently active providers of DTC genetic tests to comply with the new legislation they will have to bring about improvements in terms of transparency and counselling services. The revised legislation also stipulates that customers may only be provided with information that is consistent with the intended aim of the test at the outset. In other words, if it should become apparent from a non-medical DTC test that a customer is at high risk of a treatable disease, even if the condition is preventable with the aid of suitable precautionary measures or medical attention, it is not permitted to communicate this information to the customer. Unlike medically indicated DNA analyses, DTC tests may be actively advertised.

When medical tests are carried out, many other legal provisions have to be complied with which, generally speaking, correspond to those contained in the original version of the Act. Medically indicated analyses must be prescribed by a doctor, the patient must be provided with information and advice, and neither employers nor insurers are permitted access to the results.

Vague distinctions

A fundamental problem still exists, however, which even the revised Act will not be able to eliminate: genetic data that are collected for the purpose of optimising lifestyle can also be of relevance to health, because a clear distinction cannot be made between medical and non-medical information. The revised Act aims to circumvent this problem by introducing the purpose of the analysis as a supplementary construct. If a genetic test is to be carried out for the purpose of optimising a person's diet, this is regarded as a non-medical analysis, but if a severely obese person is to be tested in order to determine the most suitable treatment, the same test is regarded as a medical analysis.

The definition problem is underscored by the fact that communicating additional information of relevance to the customer's health is prohibited. This means that the legislator implicitly concedes that DTC tests do in fact encroach on medical issues. The lack of clear distinctions between medical and non-medical uses of DTC genetic tests is likely to be the main difficulty with respect to the implementation of the revised Act.

Challenges of data protection

With respect to the processing of genetic data, the existing Federal Act on Human Genetic Testing refers to the observance of professional secrecy and compliance with the Federal Data Protection Act.

The revised version will regulate this issue in greater detail. It will call for the protection of not only genetic data, but also genetic samples. It will also specify the permissible retention period for genetic data and samples, which as a rule will have to be destroyed not later than two years after they have fulfilled their purpose. Another new article of the law addresses the reuse of genetic data and emphasises the importance of obtaining the prior informed consent of the «donor» of the data. In other words, if a company uses its customers' genetic data tacitly, without informing them or obtaining their consent, this will violate Swiss law.

Given that DNA samples collected by the police cannot be regarded as being «voluntarily» provided, the legal provisions governing the destruction of samples and deletion of data are of particular significance. Here the Federal DNA Profiles Act specifies the criteria that have to be met. It also regulates the right to information to which everyone is entitled if their genetic profile is fed into the information system.

With respect to DTC genetic tests, two challenges arise in the context of data protection. Firstly, it is likely to prove difficult to enforce Swiss law if the genetic data are placed in the hands of a company outside the country. And secondly, the «collective identities» resulting from genetic tests give rise to problems: genetic data are not a tangible object that is at a person's disposal like other property. Rather, these data also belong to the person's relatives, who in their turn have a genetically legitimised right to have a say in the way the data are used.

Circumspect handling of genetic data is essential

Information obtained from a person's genetic material also pertains to other people (i.e. relatives) and as such has to be treated with special care. This needs to be acknowledged by both customers and providers of genetic tests, as well as consumer protection organisations and decision-makers in the fields of politics and science.

Surveys among customers who have undergone genetic testing have revealed that curiosity is a strong motive for having their DNA analysed. Specialists point out that genetic data should not be used merely for «entertainment» purposes. Even if a DTC genetic test initially yields information that is of no consequence with respect to the person's health, the possibility can never be ruled out that a subsequent reinterpretation could reveal a predisposition to a serious disease, or identify a previously unknown family relationship.

Need for the introduction of accompanying research

As non-medical genetic analyses can also be problematic, there is a need to find out which genetic tests are used by which circles, and what impacts they have. It would be particularly important to learn how often use is made of the option of having raw DNA data analysed in greater detail (often retrospectively). Here, the necessary quantitative and qualitative research needs to be promoted.

Improving transparency

The information practices of providers of genetic tests are often insufficient. Here, consumer protection organisations should step into the breach in order to ensure that the clientele of genetic test providers receive the necessary information. At the same time, the companies concerned should be urged to provide more transparent information about the services they offer. It is particularly important that they explain in greater detail how they handle their customers' data.

Researchers should not be given a blank cheque

Researchers have a great deal of interest in data resulting from genetic tests. Various providers of DNA analyses pass on the collected data to scientific organisations or pharmaceutical companies. Customers must consent to the use of their data for this purpose. However, the content of the «informed consent» form is too broadly formulated and needs to be more specific. Genetic data are not private property in the conventional sense and, in view of this, the possibility of granting involved family members the right to co-determination and consultation should be taken into consideration.

Exceptions to the restrictions on communicating results

The revised Federal Act on Human Genetic Testing prohibits providers of genetic tests from communicating information to their customers that goes beyond the extent of the originally specified analysis. This means that if a DTC genetic test reveals a predisposition for a given disease, the test provider may not pass on this information to the customer concerned. In this absolute form, the regulation is problematic, however. Precise criteria for permitting exceptions should be defined, so that customers can receive their test results and go on to initiate urgently required medical clarification and treatment.

Protection of minors and third parties

Generally speaking, providers of DTC genetic tests do not take steps to ensure that the sample they are asked to test genuinely originates from the person who submitted it. It is possible, for instance, that adults could conceal the fact that they want to carry out a parentage test. This is prohibited and the result would not be legally recognised, but it could nonetheless have a negative impact on the family. In view of this, providers of DTC genetic tests should be required to verify that the samples they receive do not originate from unaware third parties. Here, special attention has to be paid to the protection of minors.

Keeping pace with scientific developments

The field of genetics is developing at a rapid pace. Much of what we regard as today's state-of-the-art knowledge will already be outdated tomorrow. Correspondingly high expectations are placed on genetic testing, which the tests are often unable to meet. Providers of genetic tests should clearly distinguish between scientifically established findings and those that are of a more speculative nature, and the ongoing developments in the field of genetics must be continuously taken into account.

Appropriate structuring of police investigations

Phenotyping can be used to predict certain physical features based on a DNA sample, and it yields indicators for identifying suspects or other wanted persons. This also means that entire population groups can become the focus of an investigation, which can be problematic considering the preconceptions and prejudices that may exist towards specific groups. When phenotyping is to be used, the associated benefits and risks need to be discussed at the social level and negotiated with a view to ensuring its appropriate use. Here, it is necessary to precisely define the applicable circumstances and parameters for the use of phenotyping.

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Impressum

Cracking the code to individuality: DNA analysis in the service of self-knowledge and police investigations
Abridged version of the study «Neue Anwendungen der DNA-Analyse: Chancen und Risiken»
TA-SWISS, Bern 2020
TA 74A/2020

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Layout and graphics: Hannes Saxer, Bern
Printed by: Jordi AG – Das Medienhaus, Belp

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