The use of genetic data by private parties: how does the new Swiss legislation address the human rights threats?

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KEYWORDS genetic data – new technologies – human rights – private parties.

ABSTRACT In the last years, researchers have made considerable progress in the analysis of the human genome. These advances open new perspectives in the development of precision medicine, but they also raise legal and ethical questions. In light of these new issues and the European human rights standards applicable to this field, this article demonstrates the main weaknesses of the revised Federal Act on Human Genetic Analysis.

ZUSAMMENFASSUNG Forschende haben in den letzten Jahren erhebliche Fortschritte bei der Analyse des menschlichen Genoms gemacht. Diese Fortschritte eröffnen neue Perspektiven für die Entwicklung der Präzisionsmedizin, aber werfen auch rechtliche und ethische Fragen auf. Im Lichte dieser neuen Fragen und der europäischen Menschenrechtsstandards in diesem Bereich zeigt die Autorin die wesentlichen Schwachstellen das revidierte Bundesgesetze über genetische Untersuchungen beim Menschen auf.

RÉSUMÉ Des progrès considérables ont été réalisés dans la recherche sur le génome humain ces dernières années, ouvrant de nouvelles perspectives dans le développement de la médecine de précision, mais soulevant également des problématiques juridiques et éthiques. À la lumière de ces nouvelles questions et des standards européens des droits de l'homme, cette contribution démontre les principales faiblesses de la version révisée de la Loi fédérale sur l'analyse génétique humaine.

I. Introduction

In recent years, there has been an increasing interest in the topic of genetic analysis. The latest developments in genetics and genomics[1] now allow researchers to overcome the two biggest challenges faced in the past, allowing for a more widespread application of genetic analyses – namely, high cost and technological limitations caused by the vast amount of data.[2] In parallel, the direct-to-consumer («DTC») genetic testing industry is growing, and private actors can now obtain genetic tests results without medical supervision.

These advances constitute interesting prospects for the future of medical research and need to be pursued. However, they also raise significant ethical and legal challenges and can pose a serious threat to human rights – notably protected by the Convention for the protection of Human Rights and Dignity of the Human Being with regard to the Application of Biology and Medicine («Oviedo Convention»)[3].

These new developments had an impact in Switzerland as well. In 2018, the Federal Assembly adopted the revised

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1 Genetics is the study of genes and their roles in inheritance. Genomics is a more recent term that defines the study of the entire genome, including interactions of those genes with each other and with the person's environment (Genetics vs. Genomics Fact Sheet, https://www.genome.gov/about-genomics/fact-sheets/Genetics-vs-Genomics, accessed 16 September 2021).

2 M. Naveed/E. Ayday/E. W. Clayton/J. Fellay/C. A. Gunter/J. P. Hubaux/B. A. Malin/X. Wang, Privacy in the Genomic Era, ACM Computing Surveys 2015, 1 et seqq., 2; A. Lang/ B. Gschmeider/M. Gruber/M. Wuketich/ E. Kinz/V. Karavas/F. Winkler/S. Schumann/N. Burri/ E. Griessler, Neue Anwendungen der DNA-Analyse: Chancen und Risiken. Interdisziplinäre Technikfolgenabschätzung, 2020, https://vdf.ch/neue-anwendungen-der-dna-analyse-chancen-und-risiken-e-book.html, accessed 16 September 2021, 55.

3 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 of 4 March 1997 (Oviedo Convention, CC 0.810.2).

version of the Federal Act on Human Genetic Testing («HGTA»)[4], which is expected to come into force in 2022.[5]

The purpose of this paper is to analyse how the revision addresses threats to human rights emerging from the latest developments in genetic analyses. This paper is structured in five parts. First, I will outline the latest progress in the field of genetic analysis and the ensuing implications for human rights. Secondly, I will examine the relevant European human rights standards in this field. I will then provide a critical analysis of the HGTA and the 2018 HGTA revision. This analysis will reveal a series of rather theoretical solutions to the human rights issues outlined previously. Finally, the fifth part of this paper concludes by discussing the approach adopted by the Swiss HGTA in the fragmented European legal framework.

II. Developments in genetic analysis and human rights implications

A. Latest developments in the field of genetic analysis

Just over twenty years ago, the announcement that the Human Genome Project had completed its goals, led to the beginning of the genomic era.[6] On that occasion, the Prime Minister of the United Kingdom and the President of the United States announced the completion of the first draft of the human genome[7].[8] Since then, a lot has changed and the latest developments are now allowing faster and cheaper genetic testing and whole genome sequencing («WGS»).[9] To provide a context, the first attempt to WGS, which was sponsored by the United States National Institute of Health, was launched in 1990 and took thirteen years and 3 billion USD to be completed.[10] Now, using new generation sequencing methods, it is possible to sequence the genome of an organism in a single day.[11] Furthermore, the costs have significantly dropped. Today, the sequencing of a human genome can be carried out for just over 1 000 USD.[12]

Traditionally, genetic analyses were services provided by large research laboratories or in clinical settings. However, in the last decade the decreasing price of this technique has led to an increasing offer of DTC genetic testing by private companies.[13] While the providers of DTC tests are often based abroad, they offer their services to Swiss customers on the Internet – without the involvement of medical professionals.[14] In this scenario, the customer orders the genetic test online and receives the material to collect a biological sample (usually, a mouth swab). The individual then sends the genetic material to the company and his or her DNA is analysed. A few weeks later, the customer is provided with the results via e-mail or through a secure website.[15] Three of the most renowned DTC companies (23andMe[16], AncestryDNA[17] and MyHeritage[18]) are experiencing remarkable international success. For instance, in 2019 AncestryDNA reported having a database of tests from more than 10 million people.[19] Ugalmugle and Swain argue that the DTC genetic testing market ex-

4 Federal Act on Human Genetic Testing of 8 October 2004 (HGTA, CC 810.12)

5 Révision de la loi fédérale sur l'analyse génétique humaine, https://www.bag.admin.ch/bag/fr/home/medizin-und-forschung/genetische-untersuchungen/aktuelle-rechtsetzungsprojekte1.html, accessed 16 September 2021.

6 Naveed/Ayday/Clayton/Fellay/Gunter/Hubaux/Malin/Wang (fn. 2), 2.

7 The complete set of genes of a human being i.e., the entirety of the genetic information of an individual (A. De Paor, Genetics, Disability and the Law: Towards an EU Legal Framework, Cambridge 2017, 17).

8 A. M. Lesk, Introduction to Genomics, 3rd edition, Oxford 2017, 63.

9 The process of mapping the full DNA sequence of an individual's genome. In: Naveed/Ayday/Clayton/Fellay/Gunter/Hubaux/Malin/Wang (fn. 2), 2.

10 Naveed/Ayday/Clayton/Fellay/Gunter/Hubaux/Malin/Wang (fn. 2), 2.

11 S. Behjati and P. S. Tarpey, What Is Next Generation Sequencing?, Archives of Disease in Childhood – Education and Practice 2013, 236 et seqq., 236.

12 The cost of sequencing a human genome, https://www.genome.gov/about-genomics/fact-sheets/Sequencing-Human-Genome-cost, accessed 16 September 2021.

13 Naveed/Ayday/Clayton/Fellay/Gunter/Hubaux/Malin/Wang (fn. 2), 7.

14 Lang/Gschmeider/Gruber/Wuketich/Kinz/Karavas/Winkler/Schumann/Burri/Griessler (fn. 2), 9.

15 L. Kalokairinou/H. C. Howard/S. Slokenberga/E. Fisher/M. Flatscher-Thoni/M. Hartlev/R. van Hellemondt/J. Juskevicius/J. Kapelenska-Pregowska/P. Kovac/L. Lovrecic/H. Nys/A. de Paor/A. Phillips/L. Prudil/E. Rial-Sebbag/C. M. Romeo Casabona/J. Sandor/A. Schuster/S. Soini/K. H. Sovig/D. Stoffel/T. Titma/T. Trokanas/P. Borry, Legislation of Direct-to-Consumer Genetic Testing in Europe: A Fragmented Regulatory Landscape, Journal of Community Genetics 2018, 117 et seqq., 118.

16 23andMe, https://www.23andMe.com/?ada=on&prs=car, accessed 16 September 2021.

17 AncestryDNA, https://www.ancestry.com/dna/, accessed 16 September 2021.

18 MyHeritage, https://www.myheritage.com, accessed 16 September 2021.

19 S. A. Mahmoud-Davis, Direct-to-Consumer Genetic Testing – Empowering EU Consumers and Giving Meaning to the Informed Consent Process Within the IVDR and GPDR

ceeded 1 billion USD in 2019 and predict that it will exceed 3.4 billion USD by the end of 2028.[20] In Switzerland, the interest in DTC genetic testing is growing as well. In 2017 the number of tests ordered doubled compared to the previous year.[21]

A wide range of both health-related and non-health-related information can be drawn from an individual's DNA. These can be grouped into two broad categories. The first group (health-related information) includes genetic variations that *will cause* an adverse effect on the individual's well-being, such as the mutation of the HBB gene for Sickle cell anaemia. The first group also encompasses genetic variations associated with an individual's *increased susceptibility* to a particular disease (the large majority), like the breast cancer predisposition variant BRCA 1/2.[22] Moreover, this group also includes pharmacogenomic tests (which determine the influence of an individual's genome on his or her reactions to drugs)[23] and nutrigenetic tests (which analyse how genetic variants can influence the body's metabolism and its responses to nutrients)[24]. The second group (non-health-related information) concerns ancestry, relatedness (paternity, maternity, or relative's identification)[25] or other «fun traits».[26] With regards to the latter, the website of 23andMe offers its clients services to discover «more insights into what makes them unique», like «cilantro taste aversion», predisposition to liking «sweet vs. salty» but also «odds of hating the sounds of chewing».[27]

B. What is the interest in genetic data collection and analysis for DTC companies?

The collection and analysis of genetic data is extremely important for the development of new fields like precision medicine[28] and pharmagenomics – and DTC companies are very well aware of it. If, at first glance, these companies provide a simple service (genetic analysis in return to a fee), the reality is slightly more complicated.

These companies' business strategy is primarily based on the collection of their clients' genetic data for other purposes, namely profit-oriented secondary transfer of information to research institutes and pharmaceutical companies[29].[30] Case in point: the contract concluded between Genentech[31] and 23andMe, which allows the Roche group's subsidiary to analyse the data from 3'000 Parkinson patients in order to develop new treatments against this disease. According to some accounts, Genentech is said to have paid around 60 million USD to have access to these data. From Genentech's point of view, one of the advantages is that the information has already been collected, genotyped and clinically annotated.[32] Moreover, this process breaks the traditional recruitment process for subjects to participate in clinical studies and allows the research to advance more rapidly.[33]

Thus, the long-term goal of these companies is the mass gathering of genetic information. Sounds familiar? As underlined by Jordan, this business strategy is very

Frameworks Washington University Global Studies Law Review 2020, 1 et seqq., 8.

20 S. Ugalmugle/R. Swain, DTC Genetic Testing Market to exceed US $3.4 Bn by 2028, Global Market Insights 2020, https://www.gminsights.com/pressrelease/direct-to-consumer-dtc-genetic-testing-market, accessed 16 September 2021.

21 L. Jorio, Quand quelques gouttes de salive peuvent changer votre vie, Swissinfo.ch 2018, https://www.swissinfo.ch/fre/tests-adn_quand-quelques-gouttes-de-salive-peuvent-changer-votre-vie/44117364, accessed 16 September 2021.

22 Naveed/Ayday/Clayton/Fellay/Gunter/Hubaux/Malin/Wang (fn. 2), 6.

23 E. Hafen, The key lies in the genes, Zukunftsblog 2020, https://ethz.ch/en/news-and-events/eth-news/news/2020/06/zukunftsblog-ernst-hafen-the-key-lies-in-the-genes.html, accessed 16 September 2021.

24 M. Floris/A. Cano/L. Porru/R. Addis/A. Cambedda/M. L. Idda/M. Steri/C. Ventura/M. Maioli, Direct-to-Consumer Nutrigenetics Testing: An Overview, Nutrients 2020, 1 et seqq., 3.

25 A. M. Phillips, Only a Click Away – DTC Genetics for Ancestry, Health, Love ... and More: A View of the Business and Regulatory Landscape, Applied & Translational Genomics 2016, 16 et seqq., 18.

26 Kalokairinou/Howard/Slokenberga/Fisher/Flatscher-Thoni/Hartlev/van Hellemondt/Juskevicius/Kapelenska-Pregowska/Kovac/Lovrecic/Nys/de Paor/Phillips/Prudil/Rial-Sebbag/Romeo Casabona/Sandor/Schuster/Soini/Sovig/Stoffel/Titma/Trokanas/Borry (fn. 15), 118.

27 23andMe, Ancestry + Traits Service, https://www.23andMe.com/dna-ancestry?ada=on, accessed 16 September 2021.

28 Precision medicine uses relevant information (notably genetic data) about a person to further personalize his or her healthcare – as opposed to a «one fits all» approach (G. O. Schaefer/E. S. Tai/S. Sun, Precision Medicine and Big Data: The Application of an Ethics Framework for Big Data in Health and Research, Asian Bioeth Rev 2019, 275 et seqq., 275).

29 Pharmaceutical companies have notably a significant interest in obtaining the kind of information. As mentioned above, the rise of pharmagenomics has the potential to identify «the right drug, in the right dose, at the right time for each patient» (Hafen [fn. 23]).

30 Lang/Gschmeider/Gruber/Wuketich/Kinz/Karavas/Winkler/Schumann/Burri/Griessler (fn. 2), 121.

31 Genentech, https://www.gene.com, accessed 16 September 2021.

32 B. Jordan, Chroniques genomiques: 23andMe ou comment (très bien) valoriser ses clients Médecine/Sciences (Paris) 2015, 447 et seqq., 447.

33 Mahmoud-Davis (fn. 19), 10.

similar to the one used by Google or Facebook.[34] In this regard, a member of the 23andMe board admitted in 2013 that «the long game here is not to make money selling kits (…) once you have the data (23andMe) does actually become the Google of personalized health care».[35] Stoeklé and others argue that it is a classic example of two-sided market, where two different user groups (in this case, the customers on the one hand and the research laboratories or pharmaceutic companies on the other) interact via an intermediary (the DTC company).[36]

C. Threats to human rights

The growing popularity of DTC genetic testing raises several ethical and legal issues, of which the following three are particularly salient.

1. Absence of medical supervision and genetic counselling and the right to health

Firstly, the *absence of medical supervision and genetic counselling for health-related genetic tests* raises concerns about the possible misinterpretation of the results by the customers, which may cause inappropriate healthcare decisions.[37] This situation raises concerns under the right to health, since the individual may not have access to all the necessary health-related information to take an informed decision. In a limited number of cases, receiving genetic analysis results without the necessary guidance may cause anxiety and depression, interfering in turn with the right to mental health.[38]

In Europe, health-related direct-to-consumer genetic tests are often not allowed.[39] However, once the customers have access to their raw genetic data through a DTC company, they can find other ways to obtain health-related genetic test results. In fact, the advent of regulation over this sector led to the emergence of an increasing number of third interpretation websites,[40] allowing users to re-interpret their own genetic data online and obtain the (health) information desired.[41]

2. Threats to privacy

Secondly, further concerns focus on *the privacy of the customers*. The complex structure of DTC companies and the two-sided business model make it difficult to understand who has really access to the client's data. Often, the company in contact with the clients is not the same as the one that owns the laboratories where the samples are analysed.[42] According to Hazel and Slobogin, out of the DTC companies based in the United States allowing some kind of third-party sharing, 71 % use data for other purposes than provide results to costumers, 62 % of them use data for research, and 78 % provide genetic information to third parties in de-identified or aggregate forms without additional consumer consent.[43]

The special features of genetic data[44] are well-known. In light of the sensitive nature of this information, third party interest in accessing this data may result in privacy violations. In the European countries where only non-health-related tests can be shipped, the threat is still relevant. In fact, the customers are encouraged to allow

34 Jordan (fn. 32), 447.

35 C. Seife, 23andMe is terrifying, but not for the reasons the FDA thinks, Scientific America 2013, https://www.scientificamerican.com/article/23andMe-is-terrifying-but-not-for-the-reasons-the-fda-thinks/, accessed 16 September 2021.

36 H. C. Stoekle/M. F. Mamzer-Bruneel/G. Vogt/C. Herve, 23andme: A New Two-Sided Data-Banking Market Model, BMC Medical Ethics 2016, 1 et seqq., 3–4.

37 Kalokairinou/Howard/Slokenberga/Fisher/Flatscher-Thoni/Hartlev/van Hellemondt/Juskevicius/Kapelenska-Pregowska/Kovac/Lovrecic/Nys/de Paor/Phillips/Prudil/Rial-Sebbag/Romeo Casabona/Sandor/Schuster/Soini/Sovig/Stoffel/Titma/Trokanas/Borry (fn. 15), 118.

38 In light of recent studies, it is important to underline that this claim has been proven only for a limited category of findings, notably to Huntington disease (S. Oliveri/F. Ferrari/A. Manfrinati/G. Pravettoni, A Systematic Review of the Psychological Implications of Genetic Testing: A Comparative Analysis Among Cardiovascular, Neurodegenerative and Cancer Diseases, Frontiers in Genetics 2018, 1 et seqq., 16).

39 The website of 23andMe only ships health tests to five EU countries (Denmark, Finland, Ireland, Sweden and the Netherlands) (Mahmoud-Davis [fn. 19], 9–10).

40 See for example: Promethease, https://www.promethease.com/ or Codegen.eu, https://codegen.eu/, accessed 16 September 2021.

41 L. Badalato/L. Kalokairinou/P. Borry, Third party interpretation of raw genetic data: an ethical exploration, Eur J Hum Genet 2017, 1189 et seqq., 1189.

42 Lang/Gschmeider/Gruber/Wuketich/Kinz/Karavas/Winkler/Schumann/Burri/Griessler (fn. 2), 179.

43 J. Hazel/C. Slobogin, Who Knows What, and When? A Survey of the Privacy Policies Proffered by U.S. Direct-to-Consumer Genetic Testing Companies, Cornell Journal of Law and Policies 2018, 1 et seqq., 22.

44 Notably, the «DNA contains information about an individual's health and behaviour, (…) it does not change much over time in an individual. (…) The DNA of any two individuals can be easily distinguished from one another. (…) (There is a) public perception of mystery about DNA. (…) (it) contains information about an individual's blood relatives.» (Naveed/Ayday/Clayton/Fellay/Gunter/Hubaux/Malin/Wang [fn. 2], 41).

the company to use their data for research in order to «become part of something bigger than themselves».[45]

Moreover, the unique nature of genetic data makes it difficult to protect the individual's privacy. For traditional health records, the privacy of research's subjects is generally protected by the removal of explicit attributes – for example, the name or the date of birth.[46] The same solution does not apply to genetic data: it has been demonstrated on several occasions that it is possible to re-identify individuals in large genetic datasets.[47]

Considering the threat posed to the individuals' privacy, this also raises concerns about their *informed consent*. As underlined by Mahmoud-Davis, the biggest threat to an individual's autonomy when it comes to informed consent is his or her «lack of clear understanding of the choices presented, including the potential risks, benefits and consequences».[48] The wrap contracts used by DTC companies for the purchase of these test are often extremely complex.[49] This issue, combined with the technical nature of the subject matter, contribute to this dangerous situation where customers may get an inaccurate view about the secondary use of their data.

3. Discrimination and stigmatisation concerns

Thirdly, disclosure of genetic data entails *discrimination and stigmatisation concerns*, notably in the sectors of insurance and employment. Genetic data can be used by insurance companies as a tool to distinguish low and high-risk customers, which might result in some individuals not being able to afford their premiums or even being deprived of some kind of private healthcare access.[50] Moreover, there is an obvious financial incentive for the employer to gain insights into the future perspective health of his or her employees through their genetic data, allowing him or her to select who appears to be the healthiest workers in order to reduce the risks of compensation claims or reduced productivity.[51]

III. Regional human rights standards: the Oviedo Convention and Additional Protocols

The idea that advances in genetic technologies might cause a violation to an individual's human rights is not new and the Council of Europe has been active in this field since the 1980's.[52] The only legally binding international instrument explicitly dealing with the protection of human rights in biomedicine, the *Oviedo Convention*, was ratified by Switzerland in 2008. It elaborates some of the rights enshrined in the ECHR[53] and specifically addresses the topic of genetic testing. In this regard, it protects the right to privacy (Art. 10) and prohibits discrimination based on genetic heritage (Art. 11). Furthermore, the Oviedo Convention restricts the use of «predictive genetic tests»[54] to health purposes or scientific research and mandates appropriate genetic counselling (Art. 12).

The *Additional Protocol to the Convention on Human Rights and Biomedicine concerning Genetic Testing for Health Purposes*,[55] which only applies to tests carried out for health reasons (Art. 2), goes one step further. According to Art. 7(1), genetic tests for health reasons may only be performed under medical supervision. Exceptions to this principle are possible – as long as that all the other provisions of the Protocol are met[56] – only in absence of «important implications for the health of the persons concerned or members of their family» (Art. 7([2]). Mahmoud-Davis observes that this condition undermines the utility of the exception. In fact, it seems difficult to determine when such a subjective condition is met.[57] This Protocol has only been ratified by five member states (Czech Republic, Montenegro, Norway, Portugal, Republic of Moldova and Slovakia) and is therefore not binding

45 Mahmoud-Davis (fn. 19), 9–10.
46 Naveed/Ayday/Clayton/Fellay/Gunter/Hubaux/Malin/Wang (fn. 2), 13.
47 Phillips (fn. 25), 22.
48 Mahmoud-Davis (fn. 19), 37.
49 Phillips (fn. 25), 16–17.
50 De Paor (fn. 7), 64; S. Fatos, Genetic Data Misuse: Risk to Fundamental Human Rights in Developed Economies, Legal Issues Journal 2019, et seqq., 58.
51 Fatos (fn. 50), 58; De Paor (fn. 7), 57–58. See for example the U.S. military interest in genetic testing allowing identify profiles «of special relevance to military performance and medical cost containment» and, notably, the decision of an officer's promotion based on genetic testing (M.J. Mehlman/T.Y. Li, Ethical, Legal, Social and Policy Issues in the Use of Genomic Technology by the U.S. Military, J Law Biosci 2014, 244 et seqq., 247–248).
52 De Paor (fn. 7), 214.
53 Convention for the Protection of Human Rights and Fundamental Freedoms of 4 November 1950 (ECHR, CC 0.101).
54 I.e., «tests 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» (Art. 12 Oviedo Convention).
55 Additional Protocol to the Convention on Human Rights and Biomedicine concerning Genetic Testing for Health Purposes of 27 November 2008 (Additional Protocol on Genetic Testing, CETS No. 203).
56 Notably, the quality of the tests (Art. 5), standards of genetic counselling and informed consent (Art. 8–9).
57 Mahmoud-Davis (fn. 19), 32.

upon Switzerland.[58] However, it is interesting to note that the Committee of Ministers of all the 47 Member States of the Council of Europe adopted the Protocol.[59] Moreover, the Protocol received the endorsement of several highly relevant stakeholders, such as the European Society of Human genetics, which welcomed its entry into force in 2018.[60]

Finally, since the business model of DTC companies is often based on the transfer of their client data for research purposes, it is important to address the principles underlined in the *Additional Protocol to the Convention on Human Rights and Biomedicine, concerning Biomedical Research*.[61] The Protocol has neither been signed nor ratified by Switzerland.[62] Particularly relevant for DTC companies is the rule prescribed at Art. 13(2)(vii), according to which persons participating in research projects should be informed of «any foreseen potential further uses, including commercial uses, of the research results, data or biological materials». Moreover, Art. 13(1) prescribes that research subjects should be given «adequate information in a comprehensible form». The same concept is reiterated by the *UNESCO Declaration on Bioethics and Human Rights*, which stipulates that information should be provided to participants in a «comprehensible» manner (Art. 6[2]).[63] In fact, from a human rights perspective, the consent is free and informed if not only is given based on objective information, but also if the information is «sufficiently clear and suitably worded» for the subject.[64]

Building on the legal framework set out by the Council of Europe, I will discuss the latest developments in the Swiss legislation.[65]

IV. The 2007 HGTA and why the revision was necessary

Based on the mandate enshrined at Art. 119(2)(f) of the Federal Constitution of the Swiss Confederation,[66] HGTA came into force on 1 April 2007.[67] This federal act aims to protect human dignity and personality; to prevent improper genetic testing and the improper use of genetic data; as well as to ensure the quality of genetic tests and the way their results are interpreted (Art. 2 HGTA). In light of these goals, it regulates the conditions under which genetics tests can be carried out in the medical field. The tests may only be prescribed by medical doctors (Art. 13 HGTA) and they may not be performed in absence of informed consent (Art. 5 HGTA). The laboratories that perform these tests are subject to authorisation from the competent federal authority (Art. 8 HGTA).

In the last few years, the HGTA was subject to criticism. Firstly, recent scientific developments have radically changed the quality and rapidity of genetic testing, leading to new commercial offers in the field. Consequently, the HGTA was no longer adequate.[68] Secondly, the HGTA only regulated the analysis of hereditary or embryon-

58 Chart of signatures and ratifications of Treaty 203, https://www.coe.int/en/web/conventions/full-list/-/conventions/treaty/203/signatures?p_auth=WnN2s8OP, accessed 16 September 2021.

59 L. Lwoff, Council of Europe adopts protocol on genetic testing for health purposes, Eur J Hum Genet 2009, et seqq., 1374.

60 ESHG welcomes the Council of Europe's new protocol on genetic testing, https://www.eshg.org/index.php?id=910&tx_news_pi1%5Bnews%5D=11&tx_news_pi1%5Bcontroller%5D=News&tx_news_pi1%5Baction%5D=detail&cHash=6fb03b4669aef46b5b46ffe0c41faa8f, accessed 16 September 2021.

61 Additional Protocol to the Convention on Human Rights and Biomedicine concerning Biomedical Research, Council of Europe of 25 January 2005 (Additional Protocol on Biomedical Research, CETS No.195).

62 Chart of signatures and ratifications of Treaty 195, https://www.coe.int/en/web/conventions/full-list/-/conventions/treaty/195/signatures?p_auth=mBoZxP6r, accessed 16 September 2021.

63 Universal Declaration on Bioethics and Human Rights of 19 October 2005 (2005 UNESCO Declaration). Contrary to the Oviedo Convention, the 2005 UNESCO Declaration is an instrument of soft law i.e., has a non-binding nature) (R. Andorno, Global bioethics at UNESCO: in defence of the Universal Declaration on Bioethics and Human Rights, Journal of Medical Ethics 2007, 150 et seqq., 151).

64 A. Constantin/R. Andorno, Human Subjects in Globalized Health Research, in: Gostin/Mason Meier (eds.), Foundations of Global Health and Human Rights Oxford 2020, 395 et seqq., 405.

65 It should be noted that the Oviedo Convention was conceived as a framework convention, containing broad and general principles supposed to be developed through the years. (Oxford International Organizations headnote on the 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, https://opil.ouplaw.com/view/10.1093/law-oxio/e635.013.1/law-oxio-e635?prd=OPIL, accessed 16 September 2021). These principles are addressed to Member States having signed the Convention. Thus, they do not directly apply to private parties. It is each Member State's obligation to provide adequate protections. (Constantin/Andorno [fn. 64], 407).

66 Federal Constitution of the Swiss Confederation of 18 April 1999 (FC, CC 101).

67 L. Gallegos Lopéz, Information génétique, une nouvelle source de discrimination PhD thesis University of Lausanne 2011, 263–264.

68 Message concernant la loi fédérale sur l'analyse génétique humaine, 5 July 2017, FF 2017 048, 5261–5263.

ic characteristics in the medical field. It was not clear whether analysis outside this field (for example, the ones concerning ancestry) were permitted or not.[69] On 15 June 2018, the Federal Assembly adopted the revised version of the HGTA («nHGTA»).[70] The nHGTA and the two ordinances which were revised accordingly[71] are expected to come into force in 2022.[72]

V. The new HGTA project as a response to new developments in genetic analysis

A. The new scope

The revision has considerably expanded the scope of the HGTA. The Act now covers genetic analyses carried out *outside the medical field* (Art. 1[2][b] nHGTA) as well as those *relating to non-hereditary characteristics* (Art. 2[1] nHGTA).[73] The latter are subject to more lenient regulation, because they may only directly affect the individual concerned.[74] The extension of the scope was the necessary answer to the insecurity regarding the genetic analyses not covered in the previous version of the law.

B. Exploring the key criterion of « medical field »

The HGTA's new scope has to be seen in conjunction with the following idea: different types of genetic analysis need to be regulated differently depending on the possible consequences of their misuse.[75] In light of this principle, I will explore the distinction formulated by the law between genetic analysis in and outside the «medical field» *(medizinischer Bereich / domaine médical / ambito medico)*.

Concerning genetic analysis *in the medical field*, the requirements remain essential unchanged from the previous version of the law. However, two innovations merit to be highlighted. Firstly, under certain conditions, the Federal Council has now the option to authorise other healthcare specialists – like pharmacists or dentists – to carry out these tests (Art. 20[3] nHGTA).[76] Secondly, the nHGTA regulates the fate of «surplus information» *(Überschussinformationen / informations excédentaires / informazioni in eccesso)* obtained from a genetic analysis: the individual concerned has the option to decide which bits of information he or her would like to know (Art. 27 nHGTA). These requirements are partially in line with the Additional Protocol on Genetic Testing, which also prescribes medical supervision for health-related genetic tests. However – conversely to Art. 7(2) of this Protocol – the nHGTA does not provide any exceptions in the case of absence of «important implications for the health of the persons concerned or members of their family».

When it comes to genetic tests *outside the medical field*, the nHGTA draws a distinction between the tests aiming to find particularly sensitive characteristics and the others. The *first group* concerns physiological characteristics that may influence an individual's lifestyle; personal characteristics such as character, behaviour, intelligence, preferences or abilities; and characteristics relating to the individual's origins (Art. 31[1] nHGTA). These characteristics may not contain any medically relevant information, but they are still considered sensitive, and they need a special protection.[77] Consequently, as required for tests in the medical field, they may only be prescribed by a healthcare professional (Art. 34 nHGTA) and the laboratories that perform these tests are subject to authorisation (Art. 35 nHGTA). However, it is not allowed to communicate any surplus information to the individual in question. The *second group* concerns characteristics that have nothing to do with health and that the authorities considered to entail a minimal risk of abuse (Art. 31[2] nHGTA). In this case, the genetic analysis can be delivered directly to the

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69 FF 2017 048 (fn. 68), 5263.

70 Projet de la Loi fédérale sur l'analyse génétique humaine, https://www.bag.admin.ch/bag/fr/home/medizin-und-forschung/genetische-untersuchungen/aktuelle-rechtsetzungsprojekte1.html → Documents concernant la révision totale de la LAGH → Projet concernant la LAGH, accessed 16 September 2021.

71 Ordonnance sur l'analyse génétique humaine of 14 February 2007 (OAGH, CC 810.122.1); Ordonnance sur l'établissement de profils d'ADN en matière civile et administrative of 14 February 2007 (OACA, CC 810.122.2). For more details on the OAGH, see: M. Boller, Revision of the Regulation on Genetic Testing Life Science Recht 2020, 177 et seqq., 177–182.

72 Révision de la loi fédérale sur l'analyse génétique humaine (fn. 5).

73 FF 2017 048 (fn. 68), 5271.

74 FF 2017 048 (fn. 68), 5276.

75 As well as the different needs of the persons concerned, notably when it comes to children and persons incapable of judgement (Révision totale de la loi fédérale sur l'analyse génétique humaine [LAGH] – Questions et réponses, https://www.bag.admin.ch/bag/fr/home/medizin-und-forschung/genetische-untersuchungen/aktuelle-rechtsetzungsprojekte1.html → Documents concernant la révision totale de la LAGH → Questions et réponses sur la révision totale de la LAGH, accessed 23 16 September 2021).

76 Rapport explicatif concernant la révision totale de l'ordonnance sur l'analyse génétique humaine (OAGH), https://www.bag.admin.ch/bag/fr/home/medizin-und-forschung/genetische-untersuchungen/aktuelle-rechtsetzungsprojekte1.html → Documents concernant la révision totale de la LAGH → OAGH – Rapport explicatif, accessed 16 September 2021.

77 FF 2017 048 (fn. 68), 5273.

customer without medical supervision and the laboratories are not subject to any authorisation.[78]

Applying this new regulation to the DTC genetic testing market analysed in the previous sections, the Swiss response to the problems of *medical supervision and genetic counselling* seems clear. The only genetic tests that Swiss customers will be able to purchase on the Internet will be those concerning non-medical and non-sensitive information.[79]

While the different categories may seem clear in the law, it will be challenging to apply them in practice. The distinguishing criterion between the medical and the non-medical field is the purpose of examination. The same test aiming to optimise the nutrition of a person can be qualified as medical or non-medical depending on the situation in which it is ordered.[80] Moreover, most companies are not limited to offer one type of genetic test and the line between the different tests is often blurred.[81]

Furthermore, this distinction fails to address the issue of third-interpretation websites.[82] Will these websites become completely unavailable in Switzerland in 2022? Will it be possible to have a partial access to the services, which do not concern health and sensitive information? Overall, the solution proposed by the nHGTA runs the risk of becoming more theoretical than practical. If the aim of the drafters was to prohibit direct access to sensitive genetic information, it will be extremely difficult to implement it in practice.

78 FF 2017 048 (fn. 68), 5274.

79 This means that you will, for instance, be able to buy a genetic tests promising insight about your predisposition to like sweet food better than salty. However, you will not be able to discover which percentage of ancestors were Scandinavian. (Similar conclusion: J. Vasella, Totalrevision des GUMG, Swissblawg 2017, https://swissblawg.ch/2017/08/totalrevision-des-gumg.html, accessed 16 September 2021).

80 For example, a nutrigenetic test imposed to an overweight individual in order to define an appropriate therapy opposed to a test bought by an individual in perfect health who is curious about its metabolism (Lang/Gschmeider/Gruber/Wuketich/Kinz/Karavas/Winkler/Schumann/Burri/Griessler ([fn. 2], 258–259).

81 Phillips (fn. 25), 18.

82 In their 2019 study, Nelson and others found that 89% of the DTC customers participating in the survey downloaded their raw data. Among them, 94% used at least one third interpretation website (S. C. Nelson/D. J. Bowen/S. M. Fullerton, Third-Party Genetic Interpretation Tools: A Mixed-Methods Study of Consumer Motivation and Behavior, Am J Hum Genet 2019, 122 et seqq., 122).

C. The processing of genetic data

I will now analyse how the standards outlined in the nHGTA address the informed consent criterion.

Concerning *the right to privacy* and the processing of genetic data, the current legislation refers to the obligation of professional secrecy as stipulated in Art. 321–321a of the Swiss Criminal Code[83] as well as to the federal and cantonal data protection regulations (Art. 7 HGTA). The new law goes one step further and introduces an article regulating specifically the re-use of genetic data. Art. 12(1) nHGTA (as the Additional Protocol on Genetic Research) stipulates that genetic data and genetic samples may only be re-used if the person concerned has given his or her *free*, *express and informed consent*. The law also states that this data may be used for other purposes in an anonymised form if the subject has been informed in advance and has not objected (Art. 12[2] nHGTA). Consequently, a DTC company that transfers its client's genetic data without their consent would breach Art. 12 nHGTA.[84]

According to FF 2017 048, in order to fulfil the condition of «free, express and informed consent», the intended purpose must be sufficiently defined and explained, which also entails providing the location and duration of this planned re-use.[85] This solution pursues the principle of the purpose of data processing, according to which personal data may only be processed for the purpose indicated at the time of collection, that is evident from the circumstances, or that is provided for by law (Art. 4[3]of the Federal Act on Data Protection).[86]

However, the inclusion of these elements may not be sufficient to consider the criterion of informed consent completely fulfilled from a human rights perspective. While Switzerland has not ratified the Additional Protocol on Biomedical Research and the 2005 UNESCO Declaration does not impose binding obligations, some important lessons can nonetheless be drawn from these instruments.

As we have seen above, the information required to consider consent «free and informed» should be sufficiently

83 Swiss Criminal Code of 21 December 1937 (SCC, CC 311.0).

84 For the use of genetic data and genetic samples in research concerning human diseases and the human body, the individual's right to self-determination is already protected by Art. 16–17 of the Federal Act on Research involving Human Beings of 30 September 2011 (HRA, CC 810.30). It was therefore justified to introduce a similar standard for purposes other than research within the meaning of the nHGTA (FF 2017 048 [fn. 68], 5326).

85 FF 2017 048 (fn. 68), 5325.

86 Federal Act on Data Protection of 19 June 1992 (FADP, CC 235.1).

clear and comprehensible to the specific person undergoing the genetic analysis. It has already been acknowledged that this may be difficult in several situations (e.g., subjects not familiar with science or with limited education)[87] and the use of online contracts (as the ones proposed by DTC companies) came with new challenges. The volatility of «tick this box for approval» formulas makes it easier to consent without having achieved full comprehension of terms and conditions.[88] The required details should not be hidden under a mound of other information – as is often the case in contracts for smartphone applications.[89] The contracts need to be shorter and formulated using simple concepts that every customer can understand.[90] In addition, online consent should always be opt-in (express consent) rather than opt-out (presumed consent).[91]

D. Threat of genetic discrimination: the sectors of employment and insurance

Art. 4 HGTA states that «no one may be discriminated against on grounds of his or her genetic material». Discrimination is prohibited only when it is not objectively justified.[92] This provision clarifies Art. 8(2) FC which does not explicitly mention genetic heritage as a discriminatory criterion.[93] However, the nHGTA does not provide a special civil or criminal sanction to protect those who consider themselves victims of genetic discrimination.[94] For this reason, FF 2017 048 underlines that Art. 4 HGTA acquires a practical significance only in connection with other provisions. For instance, one could imagine a contractual clause that provides for unjustified unequal treatment based on genetic characteristics. In this case, the consequence would be the invalidity of the clause based on Art. 20 of the Swiss Code of Obligations[95].[96]

This issue regulated in more detail in the field of employment (Art. 38–41 nHGTA) and insurance (Art. 42–44 nHGTA).

In the field of insurance, insurance companies are in principle forbidden to request a genetic test from their clients (Art. 42 nHGTA). However, they may request the disclosure of prior tests results for private insurance contracts that do not fall under the scope of Art. 43 nHGTA – for example private health insurance, as well as for life and voluntary invalidity insurance above a certain amount.[97] In these cases, discrimination based on genetic heritage is therefore accepted.

In the field of employment, the employer can request the employee to undergo genetic analysis if the purpose is to determine characteristics relevant for the position occupied (Art. 38 nHGTA). As underlined by Junod, the genetic analysis should «target the suspicion of a given existing disease».[98] The employer cannot require the employee to undergo a predictive genetic test (Art. 39 nHGTA), except when the strict requirements set out by Art. 40 nHGTA are met.

If this issue generated a significant amount of discussion in 2007, this was not the case in the latest revision of the law and the media devoted little attention to this topic. There is no available statistics regarding the extent to which genetic analyses are used in these sectors.[99] Beside a few minor changes, the nHGTA remains in line with the existing law,[100] and continues to avoid tackling specific sanctions to protect individuals against genetic discrimination.

87 Constantin/Andorno (fn. 64), 405.

88 M. van Hoof/G. Widdershoven, Informed Consent in Online Research: The Need for New Ways of Addressing Research Subjects, AME Medical Journal 2017, 109 et seqq., 1.

89 According to a recent study, it takes nearly one hour to read the terms and conditions of a smartphone application (Mahmoud-Davis [fn. 19], 38).

90 Kalokairinou/Howard/Slokenberga/Fisher/Flatscher-Thoni/Hartlev/van Hellemondt/Juskevicius/Kapelenska-Pregowska/Kovac/Lovrecic/Nys/de Paor/Phillips/Prudil/Rial-Sebbag/Romeo Casabona/Sandor/Schuster/Soini/Sovig/Stoffel/Titma/Trokanas/Borry (fn. 15), 127.

91 H. C. Stoekle/J. F. Deleuze/G. Vogt/C. Herve, Vers un consentement éclairé dynamique, Med Sci (Paris) 2017, 188 et seqq., 189.

92 FF 2017 048 (fn. 68), 5311.

93 Gallegos Lopéz (fn. 67), 265.

94 FF 2017 048 (fn. 68), 5311–5312.

95 Federal Act on the Amendment of the Swiss Civil Code (Part Five: The Code of Obligations) of 30 March 1911 (SCO, CC 220).

96 FF 2017 048 (fn. 68), 5311–5312.

97 C. Bram/T. Szucs, Is It Desirable That I Must Disclose My Genetic Data to Swiss Private Medical Insurances?, Public Health Genomics 2016, 251 et seqq., 252.

98 V. Junod, Genetic Analyses in the Insurance and Employment Contexts in Switzerland, in: Khoury/ Blackett/Vanhonnaeker (eds.), Genetic Testing and the Governance of Risk in the Contemporary Economy – Comparative Reflections in the Insurance and Employment Law Contexts Cham 2020, 317 et seqq., 294.

99 Juond (fn. 98), 307.

100 FF 2017 048 (fn. 68), 5289–5290.

E. The international market and the applicability of the nHGTA

The offer of DTC genetic testing is predominately international, and in the majority of cases the purchase of these tests takes place on the Internet. In the previous sections, we concluded that only tests revealing non-health-related and non-sensitive characteristics could be proposed to clients directly on the Internet.

As already mentioned by Schott and Mayoraz, the enforcement of the nHGTA provisions concerning DTC genetic testing could be difficult.[101] The nHTGA does not explicitly address the question of how the authorities will guarantee that the provisions will be respected in case of a transfer of genetic data abroad.[102] The only possibility would be to proceed against the individuals themselves who order non-authorised genetic tests from foreign companies.[103] Moreover, as already stressed by Junod, the problem of the international applicability of the nHGTA also concerns genetic analysis in the sector of employment and insurances.[104]

VI. Where does Switzerland stand in light of the fragmented European legal framework?

Since 2015, the Food and Drug Administration has granted authorisation to order health-related and non-health-related DTC genetic tests in the United States.[105] In Europe, the situation is more complicated: according to the extensive study conducted by Kalokairinou and others in 2018, there is a wide spectrum of laws regarding genetic testing.[106] On one end, we find France, where only certain types of health-related genetic tests can be carried out, and the supervision of a healthcare professional is always mandatory. Hence, DTC tests are completely banned.[107] On the other end, we find the United Kingdom, where there is very little regulation on the topic,[108] and different types of DTC tests can be purchased online or through pharmaceutical chains.[109]

Switzerland seems to find itself somewhere in the middle. DTC genetic tests are not completely banned, but only the ones giving results on non-health-related and non-sensitive information can be delivered directly to the customer without medical supervision. The Swiss solution therefore seems comparable to the one adopted in Germany or Austria, where only genetic DTC testing in the medical field is explicitly banned.[110]

However, as we have seen before, this solution runs the risk of becoming rather theoretical and the customers may find other ways to obtain the desired information online. Other authors have also underlined a series of questions that remained open after the revision. Some of these questions are directly relevant for DTC testing and have been mentioned in this contribution (like international applicability),[111] while others concern other aspects of genetic testing (like DNA profiling to determine filiation or identity[112] or other issues concerning genetic testing in employment and insurance contexts[113]).

VII. Conclusion

The DTC genetic testing market is in rapid expansion, and it is characterised by a two-sided business model. Furthermore, the collection of customer's genetic data for secondary purposes triggers several human rights issues.

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101 M. Schott/J. Mayoraz, Totalrevision des Bundesgesetzes über genetische Untersuchungen beim Menschen: Neue Regelungen für Untersuchungen ausserhalb des medizinischen Bereichs und für Direct-to-Consumer Gentests, Life Science Recht 2018, 267 et seqq., 273.

102 Lang/Gschmeider/Gruber/Wuketich/Kinz/Karavas/Winkler/Schumann/Burri/Griessler (fn. 2), 288.

103 Révision totale de la loi fédérale sur l'analyse génétique humaine (LAGH) – Questions et réponses (fn. 75).

104 Junod (fn. 98), 310.

105 Stoekle/Mamzer-Bruneel/Vogt/Herve (fn. 36), 9.

106 Kalokairinou/Howard/Slokenberga/Fisher/Flatscher-Thoni/Hartlev/van Hellemondt/Juskevicius/Kapelenska-Pregowska/Kovac/Lovrecic/Nys/de Paor/Phillips/Prudil/Rial-Sebbag/Romeo Casabona/Sandor/Schuster/Soini/Sovig/Stoffel/Titma/Trokanas/Borry (fn. 15), 126.

107 Kalokairinou/Howard/Slokenberga/Fisher/Flatscher-Thoni/Hartlev/van Hellemondt/Juskevicius/Kapelenska-Pregowska/Kovac/Lovrecic/Nys/de Paor/Phillips/Prudil/Rial-Sebbag/Romeo Casabona/Sandor/Schuster/Soini/Sovig/Stoffel/Titma/Trokanas/Borry (fn. 15), 119.

108 A. Hall/T. Finnegan/S. Chowdhury/T. Dent/M. Kroese/H. Burton, Risk Stratification, Genomic Data and the Law, Journal of Community Genetics 2018, 195 et seqq., 197.

109 Kalokairinou/Howard/Slokenberga/Fisher/Flatscher-Thoni/Hartlev/van Hellemondt/Juskevicius/Kapelenska-Pregowska/Kovac/Lovrecic/Nys/de Paor/Phillips/Prudil/Rial-Sebbag/Romeo Casabona/Sandor/Schuster/Soini/Sovig/Stoffel/Titma/Trokanas/Borry (fn. 15), 127.

110 FF 2017 048 (fn. 68), 5293.

111 Schott and Mayoraz (fn. 101), 273–274; Junod (fn. 98), 310.

112 M. Zieger, Abstammungsuntersuchungen nach dem revidierten GUMG, Jusletter, 2 September 2019, 1 et seqq., 25–26.

113 Junod (fn. 98), 310–313.

The nHGTA made strides in the protection of the individual's rights and is in accordance with the relevant provisions of the Oviedo Convention. The revised law now also regulates genetic analysis outside the medical field, going one step further than the Additional Protocol on Genetic Testing. Finally, both the Additional Protocol on Biomedical Research and the nHGTA prescribe free and informed consent in case of further use of the subject's data.

Nevertheless, some questions remain open and the assessment of the legal framework suggests that the nHGTA is still insufficient to ensure the protection of human rights against the misuse of genetic data by private parties. In particular, the law provides a rather theoretical distinction of regimes when it comes to health-related data, sensitive data and data that falls under other characteristics. The Act also provides limited guidance on the question of informed consent and no specific criminal or civil sanction has been imposed to protect individuals against genetic discrimination. Moreover, it is still unclear to what extent it will be possible to enforce the regulation – considering the cross-border nature of the DTC genetic testing sector.

The collection of genetic data is essential to progress in precision medicine. Moreover, DTC platforms expedite the process compared to the traditional recruitment of patients.[114] These services should therefore not be banned. However, to ensure the continuous development of new treatments that may benefit everyone's health, it is crucial that individuals feel encouraged to participate in research (*via* DTC companies or more traditional platforms). To reach this goal, all challenges to human rights must be extensively addressed at a national and international level.

My proposed recommendations for the next round of reform focus on three aspects.

The limitations pertaining to the criteria of «medical field» should be seriously reconsidered. The presentation of some categories of genetic tests as more dangerous than others can become rather problematic. The boundaries between different types of genetic analyses are often blurred depending on the final use of the results. Moreover, the genetic data drawn from the DTC tests can be re-interpreted to obtain the information desired. The focus should shift to the protection of individuals' genetic data in all scenarios – rather than prohibiting direct access to tests in certain situations. To achieve this result, the next two point should be addressed.

Further reflections on the notion of informed consent in this field are crucial and specific guidelines should be issued to call upon DTC companies operating in Switzerland. The online DTC industry has amplified the challenges of informed consent in healthcare-related situations and customers should always understand the consequences of their purchase. It should not be considered sufficient that the use of genetic data is defined and explained, but it should be ensured that this information is realistically comprehensible and accessible to the individual client. E.g., previous research suggests employing clear and objective third parties' information in the form of diagrams or audio-video technology during the ordering process.[115]

Finally, the difficulties relating to the regulation of industries operating internationally should not only be taken into consideration, but also directly addressed in the legislation. The question of the decisive criterion for applying the nHGTA to DTC companies operating internationally – notably in the case of transfer to genetic data aboard – should be separately mentioned in the law.

114 Mahmoud-Davis (fn. 19), 35.

115 Mahmoud-Davis (fn. 19), 44–45.