Legal Analysis on the Genetic Privacy Protection: A Comparative Perspective from the United States and the European Union

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Abstract

With the rapid development and easy accessibility of gene sequencing, people can have various types of genetic testing by both public institutions and private companies. The concerns about genetic privacy are following the proliferation of genetic tests. On account of the sensitivity of genetic information and its vulnerable status, the United States (the US) and the European Union (EU) have established different legal framework for genetic privacy protection.

The US adopts the genetic-specific legislation, which has been criticized by a lot of scholars. The US genetic privacy law only focuses on certain fields, such as insurance and employment. It protects genetic privacy from nondiscrimination perspective. On the contrary, the EU has provided protection for genetic privacy through a general regulation, namely the *General Data Protection Regulation* (GDPR), which covers all identified or identifiable personal data. To highlight the sensitivity of genetic data, the GDPR classifies genetic data into special categories and offers more stringent protection. The GDPR regards genetic privacy as a fundamental right. Therefore, the genetic privacy is endowed with more universal meaning.

Despite of the differences between the EU and the US regulations with regard to the genetic privacy, they have the same goal, that is to respect the personal interests in genetic privacy. Personal autonomy is one of the most vital interests contained in genetic privacy, which represents the personal control of their own genetic information. Both the US and the EU design various mechanisms to guarantee personal autonomy in genetic information, but the mechanisms are subject-centered and ignore an important party in the relationship — family members, who possess significant position in the context of genetic privacy. Therefore, the US and the EU legislations for genetic privacy protection not only can be complementary, but also have the same problem to solve.

Key words: Genetic privacy, legal protection, fundamental right, nondiscrimination, personal interest, autonomy

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Chapter 1 Introduction

With the rapid development and easy accessibility of gene sequencing, people can have various types of genetic testing by both public institutions and private companies. The concerns about genetic privacy are following the proliferation of genetic tests. Taking the United States (the US) as an example, there are over 60,000 tests under almost 12,000 conditions at over 500 registered laboratories in the US.[1] A great amount of genetic information and uncertain controllers thereof make individual genetic information in an unsafe status. Besides, genetic information has some special features. Firstly, genetic information is often metaphorized as "blueprint for life"[2] or "compact disc."[3] It not only reveals the information needed to create proteins,[4] but also implies a person's health condition and relevant behavior. Therefore, it is the unique biological identifier of individuals. Secondly, genetic information is likened to "future diary."[5] It means genetic information describes an important part of a person's unique future and, as such, can affect or undermine an individual's view of his/her life's possibilities.[6] Furthermore, genetic information, as inherited information, does not matter one person only, it also contains the information of the carrier's blood relatives.

On account of the importance of genetic information and its vulnerable status, it has caused increasing concerns about how to regulate the collection, use and transmission of genetic information. In the US, 50 states and multiple federal departments make a patchwork of laws. Though there are certain scenarios in which nondiscrimination related to genetic information is taken into consideration, like employment and insurance, it is in lack of a broad and systematic legal framework of genetic privacy protection. With more and more private genetic testing companies appearing, genetic privacy needs a comprehensive protection, not only in certain situations. The choice of legislative approach, namely the genetic-specific legislation or general (medical) privacy legislation, is also under debate. The European Union (EU) has adopted a general legislative approach and it specially addresses some sensitive data, including genetic data. The 1995 *Data Protection Directive* has been updated by the *General Data Protection Regulation* (GDPR), which entitles data subjects with more control over their personal data. With respect to genetic data, the GDPR has provided stringent protection which generally prohibits the processing thereof.[7] However, processing sensitive data is allowed in particular circumstances. In addition to the consent and

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[1] National Center for Biotechnology Information, Genetic Testing Registry, available at: https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=all%5Bsb%5D (lase assessed: 09 September 2020).

[2] E. Richard Moxon and Christopher F. Higgins, "A Blueprint for Life", *Nature*, 389 (September 1997).

[3] Jacquelyn Ann K. Kegley, "Using Genetic Information: The Individual and the Community", *Medicine and Law*, 15 (1996).

[4] Dror G. Feitelson and Millet Treinin, "The Blueprint for Life?", *Computer*, 35, 7 (2002).

[5] George J. Annas, "Privacy Rules for DNA Databanks Protecting Coded 'Future Diaries'", *Jama*, 270, 19 (1993).

[6] George J. Annas, Leonard H. Glantz and Patricia A. Roche, "Drafting the Genetic Privacy Act: Science, Policy, and Practical Considerations", *Journal of Law, Medicine & Ethics*, 23, 4 (1995).

[7] See Article 9 GDPR.

anonymization approach for processing, the GDPR has established research exemption for the utilization of genetic data, which creates great freedom for genetic research.[8]

There are amounts of literature mainly introducing and analyzing the US and the EU genetic privacy legislation respectively, but they rarely make a specific and systematic comparison. This thesis not only compares the legal framework of the US and the EU with regard to genetic privacy from different aspects, but also puts the genetic privacy issue in the context of personal interests, trying to sort out the relationship between individual autonomy and family members' right. The author tries to answer how the differences are reflected in the two legal systems and the theoretical basis behind them. Moreover, the law needs to draw a balance between individual right and others' interests. To solve these problems, the thesis, based on comparative approach, has adopted the research methodology of theoretical analysis and case study. The theoretical analysis helps to understand the theory foundation and logic of genetic privacy legislation, and the case study is necessary for explaining the complex definitions through judicial interpretation.

This thesis consists of five chapters. The first is the introduction part. The second chapter respectively introduces the main genetic privacy law of the US and the EU and analyzes their advantages and disadvantages. Chapter 3 makes the comparison between the two legal approaches from attribution of genetic privacy and the notions used in legislation and reflects their legislative conceptions. Then Chapter 4 extends the theme beyond genetic privacy, analyzing the interests and values behind it and makes suggestions for legislative improvement. The last chapter draws a conclusion that both of the US and the EU legal approaches for genetic privacy have pros and cons, and they provide valuable experience for each other. Furthermore, the genetic privacy law shall not be limited within personal context. Rather, it would be better to take the familial and social relationship into consideration.

[8] See Article 9(2)(j) GDPR.

Chapter 2 The Genetic Privacy Legislation in the United States and the European Union

2.1 The Genetic Privacy Protection in the US

The US policy consideration about genetic information issues began with the Human Genome Project (HGP), which has revealed that there are probably about 20,500 human genes and given the world a resource of detailed information about the structure, organization and function of the complete set of human genes,[9] causing widespread concern with genetic information among the scientists and the public. Since 1990 when the HGP started, the federal government has passed a series of legislation to struggle with genetic discrimination, especially stressing the genetic information of employees and the insureds. Up to 2008, almost every state has established legal protection against genetic discrimination in health insurance; 34 states and Washington, D.C. have enacted legislation to prohibit genetic discrimination in employment.[10]

2.1.1 The Americans with Disabilities Act

The *Americans with Disabilities Act* (ADA) "is one of the most comprehensive pieces of civil rights legislation in the US that prohibits discrimination and guarantees that people with disabilities have the same opportunities as everyone else to participate in the mainstream of life, including enjoying employment opportunities, purchasing goods and services, and participating in State and local government programs and services".[11] The ultimate purpose of the ADA is to ensure that people with disabilities enjoy equal rights and preserve them from discrimination from employers (Title I), state and local governments (Title II) and public accommodations and commercial facilities (Title III).

Individuals protected by the ADA are those having disability. The ADA defines the term "disability" as: (1) a physical or mental impairment that substantially limits one or more major life activities of such individual; (2) a record of such an impairment; or (3) being regarded as having such an impairment.[12] In other words, the objectives of the law include the people who is suffering from disabilities affecting his or her major life activities adversely, who used to be disabled and those who are regarded as having a physical or mental impairment. This definition covers the present and past situations and the subjective judgment by others. However, there is an obvious incompleteness in the dimension of time, that is, it does not cover the possible impairment in the future. As mentioned above, the predictive function of genetic information makes it possible to foresee the potential genetic disorders. For example, if individuals carry a mutated form of BRCA1 or BRCA2, they are at a great risk of developing breast or ovarian cancer.[13] Genetic testing is now available to determine whether an individual has such

[9] National Human Genome Research Institute, What is the Human Genome Project?, available at: https://www.genome.gov/human-genome-project/What (last accessed: 09 September 2020).

[10] See National Conference of Legislatures, Genetics and Health Insurance State Anti-discrimination Laws, available at: https://www.ncsl.org/research/health/genetic-nondiscrimination-in-health-insurance-laws.aspx (last accessed: 09 September 2020) and National Conference of Legislatures, Genetic Employment Laws, available at: https://www.ncsl.org/research/health/genetic-employment-laws.aspx (last accessed: 09 September 2020).

[11] Introduction to the ADA, available at: https://www.ada.gov/ada_intro.htm (last accessed: 09 September 2020).

[12] The ADA §12102. (1).

[13] What do we know about heredity and breast cancer?, available at: https://www.genome.gov/Genetic-

genetic mutation.[14] Considering that the ADA does not prohibit employers from requiring employees to make a medical examination and making a conditional offer on the results of such examination in some circumstances, it cannot prevent the employers from withdrawing the offer because of the possibility of impairment in the future showed by the examination results.[15]

According to its definition of disability, the ADA does not cover genetic predisposition which might develop into actual impairment. It also does not directly address manifested genetic disability. In spite that the Equal Employment Opportunity Commission (EEOC) suggested a broader interpretation of the term "disability" and interpreted genetic disorders as the third prong of the definition, *i.e.* "being regard as having such an impairment",[16] the Supreme Court seemed not to agree with this interpretation. In *Bragdon v. Abbott*, the respondent Abbott filed suit against the petitioner Bragdon under the ADA. Abbott went to Bragdon's office for a dental examination and imparted her human immunodeficiency virus (HIV) infection, even though not manifested. The dentist declined to provide treatment in his office. Abbott claimed that Bragdon violated the non-discrimination requirement stipulated in 42 U.S.C. § 12182(a) that "[n]o individual shall be discriminated against on the basis of disability in the full and equal enjoyment of the … service of any place of public accommodation by any person who … operates a place of public accommodation". After reviewing the ruling that respondent's HIV infection constituted a disability under the ADA, which defines disability in three subsections, the Supreme Court attributed the respondent's HIV infection into subsection (A) (a physical or mental impairment limiting one's major life activities), without considering the applicability of subsections (B) (a record of such impairment) and (C) (being regarded as having such an impairment).[17]

The Court's attribution makes this case fall into the first prong of the definition of "disability," but it cannot be interpreted that the Court has made a broad explanation of the term including the future situations. The Court concluded that the respondent's HIV infection constituted a disability because it imposed substantial limitations on the infected person's major life activities, which mainly referred to reproduction in this case. To explain HIV infection constitutes physical impairment, the court held "it is an impairment from the moment of infection…in light of the immediacy with which the virus begins to damage the infected person's white blood cells and the severity of the disease".[18] Therefore, it is improper to draw an analogy between an asymptomatic genetic preposition and non-manifested HIV infection because the latter causes immediate damage to the body, while the former does not.[19] The court took a conservative view with regard to making discrimination of asymptomatic genetic

Disorders/Breast-Cancer (last accessed: 09 September 2020).

[14] Is there a test for hereditary breast cancer?, available at: https://www.genome.gov/Genetic-Disorders/Breast-Cancer (last accessed: 09 September 2020).

[15] The ADA §12112. (d)(3).

[16] See Mark A. Rothstein, "Currents in Contemporary Ethics: GINA, the ADA and Genetic Discrimination in Employment", *Journal of Law, Medicine & Ethics*, 36, 4 (2008); EEOC Compliance Manual, vol. 2, EEOC Order 915.002, Definition of the Term "Disability", 902-45 (1995).

[17] *Bragdon v. Abbott*, 524 U.S. 624, 118 S.Ct. 2196 (1998).

[18] *Ibid.*

[19] Robert B. Lanman, "An Analysis of the Adequacy of Current Law in Protecting Against Genetic Discrimination in Health Insurance and Employment", *A Report Commissioned by the Secretary's Advisory Committee on Genetics, Health, and Society*, May 2005.

preposition (*i.e.* future genetic condition) illegal. In Chief Justice Rehnquist's opinion, the asymptomatic condition does not presently limit one's major life activities.[20] If the court supports the argument that asymptomatic condition has potential damage to physical or mental health and thus causes disability, then "every individual with a genetic marker for some debilitating disease" is disabled "because of some possible future effects".[21]

2.1.2 The Health Insurance Portability and Accountability Act Privacy Rule

The *Health Insurance Portability and Accountability Act* (HIPAA) was enacted in 1996. Title II of HIPAA, as known as the Administrative Simplification provisions, requires the Secretary of the U.S. Department of Health and Human Services (HHS) to promulgate standards for the electronic exchange of health care transactions, as well as privacy and security standards for safeguarding and protecting the privacy of an individual's personal health information.[22] Accordingly, the *Standards for Privacy of Individually Identifiable Health Information* (*Privacy Rule*) was issued in 2000, which addresses the use and disclosure of protected health information (PHI).[23] It is an evident progress in the *Privacy Rule* that PHI explicitly includes the future conditions. The HHS has definitely answered that genetic information is health information protected by the *Privacy Rule*[24] and the *Privacy Rule* has provided the definition of "genetic information."[25]The *Privacy Rule* establishes, for the first time, a set of national standards for the protection of certain health information[26] and HIPAA is the first federal legislation to address directly the problem of genetic discrimination, but is narrowly limited both in its protections and covered entities.[27] As the *Privacy Rule* states, it only applies to three types of organizations: health plans, health care providers and health care clearinghouses. This implies that other entities except from those mentioned above, such as direct-to-consumer (DTC) genetic testing companies, are excluded from the application of the *Privacy Rule* with regard to the use and disclose of PHI. Many covered entities do not perform the health care functions themselves, rather entrust the business to third parties which is called "business associate" (BA).[28] BA is a person or organization "that performs certain functions or activities on behalf of, or provides certain services to, a covered entity that involve the use or disclosure of individually identifiable health information".[29] HIPAA requires covered entities to enter into contractual agreements with BA, namely BA contract, assuring that the BA will safeguard and not misuse the information.[30] On account that the HIPAA *Privacy*

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[20] *Bragdon v. Abbott, supra* note 17.

[21] *Ibid.*

[22] Amy E. Kempfert and Benjamin D. Reed, "Health Care Reform in the United States: HITECH Act and HIPAA Privacy, Security, and Enforcement Issues", 61, 3, *FDCC Quarterly* (2011).

[23] See Summary of the HIPAA Privacy Rule, Office for Civil Rights, HHS.

[24] Does the HIPAA *Privacy Rule* protect genetic information?, Frequently Asked Questions for Professionals, HHS, available at: https://www.hhs.gov/hipaa/for-professionals/faq/354/does-hipaa-protect-genetic-information/index.html (last accessed: 09 September 2020).

[25] See 45 CFR § 160.103.

[26] Summary of the HIPAA *Privacy Rule*, *supra* note 23.

[27] Elizabeth Hutton and Devin Barry, "Privacy Year in Review: Developments in HIPAA", *A Journal of Law and Policy for the Information Society*, 1 (2005).

[28] Kempfert and Reed, *supra* note 22.

[29] Summary of the HIPAA *Privacy Rule*, *supra* note 23.

[30] Kempfert and Reed, *supra* note 22.

Rule does not directly apply to the BA, the available remedy for PHI misuse by BA would be that covered entities sue BA for breach of contract.[31]

Concerned with genetic information, the HIPAA prohibits a health plan, excluding an issuer of a long-term care policy, from using or disclosing genetic information for underwriting purposes.[32] This has great implication on those who need long-term care, especially people with Alzheimer's disease. Researchers have found that a genetic variant of the apolipoprotein E (APOE) gene on chromosome 19 can increase the risk of late-onset Alzheimer's disease.[33] APOE testing can identify "participants who may have an increased risk of developing Alzheimer's"[34]. People who find that they have the variant of such genetic marker are more likely to purchase long-term care insurance, then the insurance price would float based on who possesses the APOE information.[35] In the four scenarios illustrated by Donald Taylor *et al*., once insurers realize the increased risk of Alzheimer's, fair premium based on APOE risk is likely to be assigned.[36] Unlike the higher premiums charged from smokers for life insurance, the increasing price on the basis of APOE genotype might be discriminatory and unfair because people cannot choose their genotype.[37]

2.1.3 The Genetic Information Nondiscrimination Act

One of the original intentions to protect personal information is to eliminate discrimination. Only when people possess certain information of others, can the foundation of discrimination be set up. Privacy protections can cut off access to certain information which might result in discrimination, thus eliminate the basis of it.[38] There was a period when genetic discrimination developed extremely in the history of the US. In 1924, Virginia adopted eugenic sterilization law to authorize compulsory sterilization of the intellectually disabled. In *Buck v. Bell* case, Carrie Buck is the daughter of a feeble-minded mother and the mother of a feeble-minded child[39], and she was forced to receive a salpingectomy according to Virginia sterilization law because she was deemed to have genetic threat to the society. The Supreme Court denied the Virginia law to be unconstitutional. An Act of Virginia "recites that the health of the

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[31] *Ibid.*

[32] See 45 CFR § 164.502(a)(5)(i).

[33] Alzheimer's Disease Genetics Fact Sheet, National Institute on Aging, available at: https://www.nia.nih.gov/health/alzheimers-disease-genetics-fact-sheet (last accessed: 09 September 2020).

[34] *Ibid.*

[35] Donald H. Taylor Jr., Robert M. Cook-Deegan, Susan Hiraki, J. Scott Roberts, Dan G. Blazer, and Robert C. Green, "Genetic Testing for Alzheimer's and Long-term Care Insurance", *Health Affairs*, 29, 1 (2010).

[36] *Ibid.* In this article, the authors exhibited four scenarios depending on who possesses the information: (1) neither insurers nor individuals know APOE genotype; (2) only consumers know their genotype; (3) only the insurer knows individuals' genotypes; (4) insurers and individuals know APOE status. The long-term care insurance markets have different responses in different scenarios.

[37] *Ibid.*

[38] Jessica L. Roberts, "Protecting Privacy to Prevent Discrimination", *William and Mary Law Review*, 56 (2014).

[39] *Buck v. Bell*, 274 U.S. 200, 47 S. Ct. 584 (1927).

patient and the welfare of society may be promoted in certain cases by the sterilization of mental defectives".[40] In his report, Justice Holmes concluded that:

> "it is better for all the world, if instead of waiting to execute degenerate offspring for crime, or to let them starve for their imbecility, society can prevent those who are manifestly unfit from continuing their kind. The principle that sustains compulsory vaccination is broad enough to cover cutting the Fallopian tubes" and "three generations of imbeciles are enough."[41]

The case legitimizes those eugenic sterilization laws in the US which deprived the intellectually disabled of their rights of reproduction for the purpose of eugenics. It is actually unequal treatment only aiming at those who carry defective genetic makeup, because "heredity plays an important part in the transmission of insanity, imbecility, etc."[42]. There is the possibility that people carrying certain genetic material can give birth to intellectually defective babies, so they are operated upon against their will, while other people without such genetic material would not. Such eugenic sterilization laws have in fact constituted genetic discrimination.

With the advanced discoveries of genetic secrets, the new eugenics and genetic discrimination might be more sophisticated and have more detrimental impact.[43] Unlike the basis of eugenic sterilization laws, the genetic-related disease can be predicted through genetic testing without outside manifestation. In other words, even if an individual performs the same as other normal people extrinsically, he or she can still be treated differently on account of his or her intrinsic and "abnormal" gene. That is why genetic information needs to be protected as privacy. Given the history of eugenics, the increasing potential for discrimination as genomic research moves forward, actual instances of genetic discrimination, and the continuing concerns of the populace, the Congress took a vital step in this important realm by passing the *Genetic Information Nondiscrimination Act* (GINA) of 2008.[44]

GINA was signed into law in 2008 to protect individuals against discrimination based on their genetic information in health insurance and in employment.[45] The main part of GINA consists of two Titles: Title 1 is concerned with genetic nondiscrimination in health insurance and Title 2 is about prohibiting employment discrimination on the basis of genetic information. The passage of GINA has experienced a long and controversial period.[46] After "proposed-and-sent back" over and over again, the GINA

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[40] *Ibid.*

[41] *Ibid.*

[42] *Ibid.*

[43] Morse Hyun-Myung Tan, "Advancing Civil Rights, the Next Generation: The Genetic Information Nondiscrimination Act of 2008 and Beyond", *Health Matrix: Journal of Law Medicine*, 19, 1 (2009).

[44] *Ibid.*

[45] See Genetic information, HHS, available at: https://www.hhs.gov/hipaa/for-professionals/special-topics/genetic-information/index.html (last accessed: 09 September 2020).

[46] "To safeguard individual privacy of genetic information from the misuse of records maintained by agencies or their contractors or grantees for the purpose of research, diagnosis, treatment, or identification of genetic disorders, and to provide to individuals access to records concerning their genome which are maintained by agencies for any purpose", the Human Genome Privacy Act was introduced in the 101st Congress, but was not enacted. See "H. R. 5612 – 101st Congress: Human Genome Privacy Act.",

was finally signed into law by President George W. Bush in 2008. In the Privacy and Confidentiality Section of Title I, GINA expressly restates that genetic information shall be treated as health information. It provides a broad definition of genetic information, including information about genetic tests of individuals and their family members, the manifestation of a disease or disorder in their family members, and request for, or receipt of, genetic services and participation in genetic research.[47]

Compared to HIPAA, the covered entities of GINA expand to group health plan, health insurance issuer and issuer of a medicare supplemental policy of protected health information that is genetic information, which are prohibited from using or disclosing genetic information for underwriting purposes.[48] GINA establishes a uniform standard about how insurers and employers should use and treat individual's genetic information. The provisions make genetic discrimination illegal in the practice of health insurance and employment,[49] protecting individuals from unfavorable treatment based on their genetic conditions: the group health plan and the health insurance issuer are prohibited from making decisions about insurance, such as premium or contribution amounts, on the basis of genetic information;[50] and it would be unlawful for employers to make employment decisions, like hiring, discharging, placing or promotion, just because of genetic information of the employees.[51] By guaranteeing the confidentiality of genetic information and banning genetic discrimination, GINA would encourage individuals to participate in genetic research, thus promoting the studies.[52]

There also exist some limitations on GINA's privacy protection. The coverage of GINA is only confined to genetic information, not involving other health information. Disadvantaged decisions by employers or insurers on the basis of genetic information is unlawful under GINA, but there is no such assurance for other health-related information, like the condition of mental disorder or a check of lipid levels.[53] Besides, only the pre-symptomatic genetic information is protected.[54] In other words, the

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GovTrack.us., available at: https://www.govtrack.us/congress/bills/101/hr5612 (last accessed: 09 September 2020).

Genetic nondiscrimination legislation has been debated since the 103rd Congress. The introduced bills caused lots of debates in terms of genetic nondiscrimination legislation. "Supporters of nondiscrimination legislation feel that current laws are not sufficient to protect individuals from discrimination in health insurance or employment. Further, without protection, individuals are hesitant to seek potentially beneficial genetic services or participate in much needed clinical research. At this stage of debate, opponents believe that current laws provide sufficient protection. They are primarily concerned that new legislation will provide further incentives and additional opportunities for litigation against employers." See Michele Schoonmaker and Erin D. Williams, "Genetic Testing: Scientific Background and Nondiscrimination Legislation", CRS Report for Congress, Order Code RL32478, 21 March 2005.

[47] GINA § 201(4)(A) and (B).

[48] GINA § 105(a).

[49] Daniel Schlein, "New Frontiers for Genetic Privacy Law: The Genetic Information Nondiscrimination Act of 2008", *George Mason University Civil Rights Law Journal*, 19, 2 (2009).

[50] GINA § 101(a)(3)(A).

[51] GINA § 202(a).

[52] Jennifer L. Lee, "The First Civil Rights Act of the 21st Century: Genetic Information Nondiscrimination Act of 2008", *I/S: Journal of Law and Policy for the Information Society*, 4, 3 (2008).

[53] *Ibid.*

[54] See Mark A. Rothstein, "HIPAA *Privacy Rule* 2.0: Currents in Contemporary Bioethics", *The Journal of Law, Medicine & Ethics*, 41, 2 (2013); GINA § 101(a)(3)(B); and GINA § 102(a)(3)(B).

manifested health conditions are not covered by GINA.[55] It takes a process for genotype to develop into disease.[56] "It is quite likely … that these biomarkers, protein expression profiles, epigenetic marks, endophenotypes, and preliminary symptoms would be considered 'manifestations' of disease under GINA because these discrete markers extend beyond health risk factors based on genetic information."[57] Then they cannot be protected by GINA if these biological markers are regarded as manifestation.

2.2 The Genetic Privacy Protection in the EU

Article 8 of the *European Convention on Human Rights* (ECHR) recognized the right to privacy,[58] which is the basis of the privacy protection legislation in the EU. In 1995, the *European Data Protection Directive* was passed, "establishing minimum data privacy and security standards, upon which each member state based its own implementing law."[59] The GDPR is passed to update the *Data Protection Directive* in order to adapt to the Internet era and harmonize the data protection law throughout the EU.

2.2.1 From the Directive/95/46/EC to the General Data Protection Regulation

The GDPR entered into force on 24 May 2016 and has been applied since 25 May 2018,[60] which has evolved from and eventually replaced the Directive/95/46/EC (the *Directive*). The GDPR aims at enhancing effectiveness and harmonization of personal data protection law in the EU.[61]

The *Data Protection Directive* is built on the seven principles of the Organization for Economic Cooperation and Development (OECD)'s *Recommendations of the Council Concerning Guidelines Governing the Protection of Privacy and Trans-Border Flows of Personal Data*.[62] Created in 1980, those seven principles include: notice; purpose; consent; security; disclosure; access; and accountability.[63] These guidelines were non-binding and the application of law was quite restricted by location.[64] Impeded by inconsistent and disparate data privacy laws throughout the EU states, data flows were faced with obstacles.[65] Therefore, the European Commission "adopted the OECD guidelines into the *Data Protection Directive*, a binding set of data protection requirements for EU member states".[66]

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[55] *Ibid.*
[56] *Ibid.*
[57] *Ibid.*
[58] Article 8(1) European Convention on Human Rights: "Everyone has the right to respect for his privacy and family life, his home and his correspondence."
[59] What is GDPR, the EU's new data protection law?, available at: https://gdpr.eu/what-is-gdpr/ (last accessed: 09 September 2020).
[60] Data protection in the EU, European Commission, available at: https://ec.europa.eu/info/law/law-topic/data-protection/data-protection-eu_en#legislation (last accessed: 09 September 2020).
[61] Mahsa Shabani and Pascal Borry, "Rules for Processing Genetic Data for Research Purposes in View of the New EU General Data Protection Regulation", *European Journal of Human Genetics*, 26 (2018).
[62] Nate Lord, What is the Data Protection Directive? The Predecessor to the GDPR, *Digital Guardian's Blog*, 12 September 2018.
[63] *Ibid.*
[64] *Ibid.*
[65] *Ibid.*
[66] *Ibid.*

Article 2(a) of the *Directive* defined "personal data" as "any information relating to an identified or identifiable natural person ('data subject')".[67] An identifiable person can be identified, directly or indirectly, through identifiers, which are listed in the *Directive* such as identification number or one or more factors specific to his physical, physiological, mental, economic, cultural or social identity.[68] Contrast with the *Directive*, the GDPR defines personal data through non-exhaustive list of identifiers, explicitly including genetic factor.[69] According to Article 29 of the *Directive*, Data Protection Working Party (the Working Party) was set up.[70] In its 2004 Working Document on Genetic Data, the Working Party took into account the orientation of international instruments, referred to the legislation of the US, and highlighted the importance of protection of genetic data.[71] The Working Document provided the definition of genetic data by reference to three different instruments and analyzed its characteristics, then it interpreted the applicability of the *Directive* to genetic data.[72] On top of all, genetic data certainly falls within the scope of the *Directive* according to Article 2 (a) on account of its identifiability.[73] After that it considered genetic data providing a person's physical disposition and health condition as "data concerning health", therefore classified it into special categories of data according to Article 8 (1).[74] In contrast, Article 4 of GDPR provides definition of genetic data and data concerning health respectively, paralleling the two sorts of data.[75] It also clearly puts genetic data into special categories of personal data and prohibits the processing of it.[76]

2.2.2 Genetic Data under the GDPR

2.2.2.1 Genetic Data as Personal Data

The key element of the definition of personal data in the GDPR is "identified or identifiable". In Recital 26, anonymous data is defined as information which does not relate to an identified or identifiable natural person or to personal data rendered anonymous in such a manner that the data subject is not or no longer identifiable, which means there is no possibility to identify a natural person. Therefore, GDPR does not apply to the processing of such anonymous data, including for statistical or research purposes.[77] Another way to hinder identification is pseudonymization. However,

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[67] Article 2(a) Directive/95/46/EC.

[68] *Ibid.*

[69] See Article 4(1) GDPR.

[70] Article 29(1) of the Directive states: "A Working Party on the Protection of Individuals with regard to the Processing of Personal Data … is hereby set up."

[71] See Working Document on Genetic Data, Article 29 Data protection Working Party, 12178/03/EN WP 91, adopted on 17 March 2004.

[72] *Ibid.*

[73] Article 2(a) of the Directive states: "'personal data' shall mean any information relating to an identified or identifiable natural person('data subject'); an identifiable person is one who can be identified, directly or indirectly, in particular by reference to an identification number or to one or more factors specific to his physical, physiological, mental, economic, cultural or social identity." The Working Document on Genetic Data give an example that samples of DNA may constitute a source of personal data in so far as it may be possible to associate samples of DNA with a given person.

[74] See Working Document on Genetic Data, *supra* note 71; Article 2(a) Directive/95/46/EC.

[75] See Articles 4(13) and 4(15) GDPR.

[76] See Article 9 GDPR.

[77] Recital 26 of GDPR: "The principles of data protection should apply to any information concerning an identified or identifiable natural person. Personal data which have undergone pseudonymisation, which could be attributed to a natural person by the use of additional information should be considered to be information on an identifiable natural person. To determine whether a natural person is identifiable,

pseudonymization can be done both in retraceable way and a way that no re-identification is possible.[78] According to the Working Party, using a pseudonymized means that it is possible to back track to the individual, so that the individual's identity can be discovered. Therefore, "retraceably pseudonymized data may be considered as information on individuals which are indirectly identifiable".[79] Recital 26 resonates that pseudonymized data should be considered to be information on an identifiable natural person and therefore should be regulated by the GDPR. Both anonymous data and pseudonymized data are relevant in the context of research and statistics. Nevertheless, recognizing pseudonymized data in the GDPR as personal data will affect the practices of those research studies that are currently considering pseudonymized data as non-personal data.[80] When pseudonymized data was not in the scope of personal data, the GDPR was not applicable to it. However, in the circumstance that pseudonymized data is regarded as identifiable personal data, the research using it have to rely on exemption rules of GDPR.

Personal data under the GDPR can be linked, directly or indirectly, to a specific person. To determine whether certain personal data is subject to the GDPR, the determinant is whether the data is identifiable. The identifying attribute might change with the technological development and legal context. In current situation, genetic data itself can be regarded as identifying without additional identifiers.

From the scientific perspective, an individual's identification can be determined depending on his or her DNA profile.[81] In addition, the worldwide researchers can access and share genetic data they have obtained and publish their discoveries.[82] "Computing power not only has continued to grow at an astonished pace itself, but also has accelerated the development of ever more powerful algorithms that are capable of more thorough data analysis and, in a number or contexts, allowing identification of individuals where it was previously not thought possible."[83]

From the legal perspective, some scholars argue that genetic data is identifying just like fingerprints which are recognized as personal data by case law.[84] The Court of Justice

account should be taken of all the means reasonably likely to be used, such as singling out, either by the controller or by another person to identify the natural person directly or indirectly. To ascertain whether means are reasonably likely to be used to identify the natural person, account should be taken of all objective factors, such as the costs of and the amount of time required for identification, taking into consideration the available technology at the time of the processing and technological developments. The principles of data protection should therefore not apply to anonymous information, namely information which does not relate to an identified or identifiable natural person or to personal data rendered anonymous in such a manner that the data subject is not or no longer identifiable. This Regulation does not therefore concern the processing of such anonymous information, including for statistical or research purposes."

[78] Opinion 4/2007 on the concept of personal data, Article 29 Data Protection Working Party, 01248/07/EN WP 136, adopted on 20 June 2007.

[79] *Ibid.*

[80] Shabani and Borry, *supra* note 61.

[81] Kärt Pormeister, "The GDPR and Big Data: Leading the Way for Big Genetic Data?", *in* Schweighofer E., Leitold H., Mitrakas A., Rannenberg K. (eds.), *Privacy Technologies and Policy: 5th Annual Privacy Forum, APF 2017 Vienna,Austria June 7-8, 2017* (Cham: Springer, 2017).

[82] Paul Quinn and Liam Quinn, "Big Genetic Data and Its Big Data Protection Challenges", *Computer Law & Security Review*, 21, 46 (2018).

[83] *Ibid.*

[84] Pormeister, *supra* note 81.

of the European Union (CJEU) held the opinion that "fingerprints constitute personal data, as they objectively contain unique information about individuals which allows those individuals to be identified with precision"[85]. Even though fingerprints are unintelligible to the untutored eye and without a comparator fingerprint, the European Court of Human Rights (ECtHR) confirmed that this consideration cannot deny fingerprints' ability to identify specific individuals with precision.[86] The ECHR added that reservation of genetic data has more severe impact on private life than fingerprints.[87] Therefore, genetic data is identifying *per se* without additional links to individuals. This means that genetic data, compared to other kinds of data, has more outstanding identifying characteristics which contributes more to accessing privacy.[88]

2.2.2.2 GDPR Protection for Genetic Data

An obvious modification of the GDPR is that it provides a clear definition for genetic data and expressly lists it in the special categories of personal data.[89] Therefore, the processing of genetic data is subject to the general prohibition of processing of sensitive data as stipulated by Article 9(1) GDPR. Except that, there are also a series of exceptional circumstances for processing sensitive data in the rest sub-provisions of Article 9. In this regard, the GDPR's solution is fairly similar to the *Directive*.[90] The GDPR maintains the original exceptions with slight modification, including explicit consent; employment and social security; vital interests of the data subject or others; data which are manifestly made public by the data subjects or exercise of judicial capacity; occupational medicine, or health or social care, or the management of health or social care systems and services with safeguards; substantial public interest.[91] In addition, the GDPR introduces further grounds for legitimate processing of sensitive data[92]: public interest in the area of public health; and archiving purposes in the public interest, scientific or historical research purposes or statistical purposes. Through comparison, it can be found that the GDPR, in fact, does not make a substantial reform compared to the *Directive*. Both of them set a special category of data beyond common personal data, and genetic data falls into such category.

Generally, the processing of special categories of data is forbidden, but they leave legal room for various possible exceptional circumstances in which the processing is necessary. The GDPR, with respect to genetic data protection, deviates from its purpose of creating a uniform standard for data protection to some extent, because it leaves large room for Member States to establish their own rules. The GDPR just covers genetic data in form without safeguarding it more rigorously, which seems to have little effect in terms of genetic data protection.[93] Even though it introduces genetic data specially, it does not provide a more stringent protection for genetic data compared to other sensitive data. "The GDPR generally strengthens data subjects' rights over their

[85] C-291/12 *Schwarz v. Bochum* (2013) ECLI:EU:C:2013:670, para 27.
[86] *S. and Marper v. United Kingdom* (2008) ECLI:CE:ECHR:2008:1204JUD003056204, para 84.
[87] *Ibid.*, para 86.
[88] Pormeister, *supra* note 81.
[89] Article 9 GDPR.
[90] Pormeister, *supra* note 81.
[91] Article 9 GDPR.
[92] *Ibid.*
[93] Pormeister, *supra* note 81.

personal data and demands greater transparency and accountability from processors."[94] Whilst these features are not specific to genetic data.

2.2.3 Processing of Genetic Data under Research Exemption

Beyond the general prohibition of processing sensitive data, the GDPR has introduced "research exemption" for processing special categories of data, including genetic data, without the strict requirements. The research exemption supplements the *Directive*'s binary approach -consent and anonymization- in the research context.

2.2.3.1 The Binary Approach in the Directive: Consent and Anonymization

Article 8(2)(a) of the *Directive* recognized "explicit consent" as one of the exceptions of the general prohibition of processing special categories of data. However, the consent requirement seems to be not ideal when performed. First, it is too burdensome and inefficient to obtain consent every time the genetic data is processed,[95] which does not adapt to data-intensive research. Second, even if the data subjects give explicit consent to the physicians to collect their genetic data for treatment or some other reasons, they cannot predict the potential further processing by the physicians. For example, a patient agree to take a genetic testing as a part of physical examination and permits the doctor to store the data as healthy record, but he cannot prevent the doctor from utilizing his data for research under research exemption, even though it goes against his original purpose for which he gives consent. The validity and legality of the one-off consent are skeptical.[96]

Anonymization is an alternative to the requirement of consent. The objective of the data protection law is identified or identifiable personal data. Recital 26 of the *Directive* stated that the principles of protection do not apply to anonymous data which is no longer identifiable. According to the Working Party, anonymization results from processing personal data in order to irreversibly prevent identification, having regard to all the means "likely reasonably" to be used for identification.[97] The conventional research model has been evolving. More and more scientific researches are utilizing pre-existing personal health data.[98] Using the health data stored in datasets or electronic health records (EHRs) makes the step of pre-collection omitted. Researchers might be inclined to take advantage of these sources, for example, combining genetic data with EHRs, which can allow information concerning phenotype and genotype to be linked.[99] However, this form might invoke privacy issues as the gathering of such

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[94] Dara Hallinan, Michael Friedewald, Paul De Hert, "Genetic Data and Protection Regulation: Anonymity, Multiple Subjects, Sensitivity and a Prohibitionary Logic Regarding Genetic Data", *Computer Law & Security Review*, 29, 4 (2013).

[95] See Menno Mostert, Annelien L. Bredenoord, Monique CIH Biesaart and Johannes JM van Delden, "Big Data in Medical Research and EU Data Protection Law: Challenges to the Consent or Anonymise Approach", *European Journal of Human Genetics*, 24 (2016).

[96] *Ibid.*

[97] See Opinion 05/2014 on Anonymisation Techniques, Article 29 Data Protection Working Party, 0829/14/EN WP216, adopted on 10 April 2014.

[98] See Paul Quinn, "The Anonymisation of Research Data —— A Pyric Victory for Privacy that Should Not Be Pushed Too Hard by the EU Data Protection Framework?", *European Journal of Health Law*, 24 (2017).

[99] *Ibid.*

data and its further processing may often not be based on the explicit and informed consent of data subjects,[100] so the researchers turn to the another approach other than explicit consent, *i.e.* anonymization.

Nevertheless, anonymization does not provide a legitimate basis for processing genetic data in research. It is just a way to escape the application of personal data protection law.[101] From the research perspective, anonymization is far from ideal because truly anonymous data is of little value for research in practice.[102] "Very often data is only of use where it contains personal (or *quasi* personal) identifiers that allow the data in question to be analyzed within specific contexts."[103] To say the least, it is hard to meet the true anonymization standards in the context of genetic research. Genetic data denotes a link to specific individual and the family members so that it is not practical to truly anonymized it.[104] Moreover, given that anonymization helps avoid the application of GDPR, it relieves the data collectors or processors from the legal obligation to respect individuals' interests.[105] These are why anonymization, despite attractive, is not of great practicability.

Due to the deficiencies of the consent and anonymization approaches to processing genetic data for research purpose, new approach which more benefits genetic research is in need. The *Directive* did not explicitly introduce research exemption, but it permitted the Member States to lay down exemptions for substantial public interest with suitable safeguard,[106] and in Recital 34, it recognized that scientific research counted as public interest, which paved the way for the research exemption in the GDPR.

2.2.3.2 Research Exemption in the GDPR

The GDPR has adopted a research-friendly approach[107], which directly establishes research exemption with regard to processing sensitive data, including genetic data. Article 9(2)(j) stipulates that the general prohibition of processing sensitive data shall not apply when processing is necessary for scientific research purposes with suitable safeguards. This provision is separate from other exceptions of processing sensitive data, including explicit consent. Through systematic interpretation, it can be inferred that sensitive data can be processed for research purposes without explicit consent from data subjects.[108] In terms of genetic data, the GDPR allows the Member States to "maintain or introduce further conditions, including limitations"[109]. Therefore, the

[100] *Ibid.*

[101] *Ibid.*

[102] See R. Fears, H. Brand, R. Frackwiak, P. Pastoret, R. Souhami and B. Thompson, "Data Protection Regulation and the Promotion of Health Research: Getting the Balance Right", *Quarterly Journal of Medicine*, 107 (2014).

[103] Quinn, *supra* note 98.

[104] See H. Schmidt and S. Callier, "How Anonymous is 'Anonymous'? Some Suggestions towards a Coherent Universal Coding System for Genetic Samples", *Journal of Medical Ethics*, 38, 5 (2012).

[105] Pormeister, *supra* note 81.

[106] Article 8(4) the Directive.

[107] Shabani and Borry, *supra* note 61.

[108] Pormeister, *supra* note 81.

[109] Article 9(4) GDPR.

Member States can establish stricter protections for processing genetic data for research. To avoid breaking the harmonization of regulations and encourage cross-border cooperation in scientific research, Recital 53 warns that the "further conditions" "should not hamper the free flow of personal data with the Union". In addition, Recital 159 requires interpreting scientific research purposes in a broad manner, including privately funded research.[110] "Thus, in a simplified manner one could conclude that the research exemption in the GDPR covers all sorts of research."[111]

Regarding genetic research, the broad definition might include the DTC genetic testing companies as long as they have research groups inside.[112] After collecting genetic data from individuals and summitting the testing report, the companies might process the genetic data again without consent.[113]

Article 5 sets principles of processing personal data, including purpose limitation for further processing and storage limitation for duration of keeping data. However, research can exempt from these limitations. Article 5(b) requires that personal data shall not be further processed in a manner that is incompatible with the original specified, explicit and legitimate purposes, but processing for research purposes shall "not be considered to be incompatible with the initial purposes"[114]. It can be interpreted into that no matter for what purposes the data was originally collected, secondary processing for research purpose is deemed as compatible with the initial purposes. "In terms of genetic data, unless further processing serves a purpose of discrimination, it will most likely serve a research purpose of some kind since the (commercial) value of genetic data lies in its potential to advance science in genetics, pharmacogenetics, clinical medicine, etc."[115] Thus, further processing of genetic data is not restricted by purpose limitation, which means retrospective and back-and-forth use genetic databases is permitted.[116] The inherent investigative feature of genetic data collection makes it immensely convenient to future research, because the collection of genetic data for any purposes would, intentionally or not, serve for the future unknown research.

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[110] Recital 159 of GDPR: "Where personal data are processed for scientific research purposes, this Regulation should also apply to that processing. For the purposes of this Regulation, the processing of personal data for scientific research purposes should be interpreted in a broad manner including for example technological development and demonstration, fundamental research, applied research and privately funded research. In addition, it should take into account the Union's objective under Article 179(1) TFEU of achieving a European Research Area. Scientific research purposes should also include studies conducted in the public interest in the area of public health. To meet the specificities of processing personal data for scientific research purposes, specific conditions should apply in particular as regards the publication or otherwise disclosure of personal data in the context of scientific research purposes. If the result of scientific research in particular in the health context gives reason for further measures in the interest of the data subject, the general rules of this Regulation should apply in view of those measures."

[111] Kärt Pormeister, "Genetic Data and the Research Exemption: Is the GDPR Going too Far?", *International Data Privacy Law*, 7, 2 (2017).

[112] Heidi C. Howard, Pascal Borry and Bartha Maria Knoppers, "Blurring Lines: The Research Activities of Direct-to-Consumer Genetic Testing Companies Raise Questions about Consumers as Research Subjects", *EMBO Reports*, 16 July 2010.

[113] *Ibid.*

[114] *Article* 5(b) GDPR.

[115] Pormeister, *supra* note 111.

[116] Shabani and Borry, *supra* note 61.

Article 5(1)(e) requires the personal data to be kept in a form which permits identification of data subjects for no longer than is necessary for the purposes for which the personal data are processed, but personal data may be stored for longer periods for research purpose with proper safeguard. Therefore, researchers can retain genetic data with identification for an uncertain time. As a result, the data subjects and their relatives "can be identified potentially throughout their lifetime along with an undetermined amount of additional personal data"[117].

2.3 A Summary and Analysis of the Legal Framework of the US and the EU

The legal protection for genetic information in the US has experienced a process from generalization to specification. In ADA, it relies on legal interpretation to extend the protection from disabled conditions to genetic conditions, with no explicit provisions to genetic information. Such protection is unstable and changes case by case. The HIPAA includes genetic information into the PHI, regulating the use and disclose of it by the covered entities, even though the coverage is limited. GINA is a genetic-specific law specially addressing genetic discrimination in health insurance and employment. It stipulates how insurers and employers shall treat personal genetic information in detail, prohibiting them from making any disadvantaged decisions solely based on genetic conditions in order to avoid discrimination.

The 1995 *Directive* of the EU did not explicitly address genetic data, while the GDPR definitely provides the definition of genetic data. The GDPR is a general and comprehensive legislation covering all kinds of personal data so long as it is identifiable. In addition to the protection for common personal data, it sets special categories of data and provides more stringent protection, which includes genetic data. For the convenience of scientific research, the GDPR introduces research exemption in terms of general prohibition of sensitive data. Processing sensitive data for research purposes is allowed, and research can exempt from purpose and storage limitation. Processing genetic data and research purposes have a strong connection because genetic data processing contains scientific value for the advance of genetics. Therefore, research exemption has greater impact on genetic research.

2.3.1 The Analysis of the US Genetic Privacy Legislation: Shifting Focus from Genetic Exceptionalism to System Improvement

2.3.1.1 Justification and Skepticism to Genetic Exceptionalism

As reviewed above, the policy concerned with genetic information has come into being from nothing and become gradually specific. At the beginning, genetic information was mentioned just as a subtitle or a few provisions. Recently, a whole statute named as genetic information, *i.e.* GINA, has started to be introduced. Genetic privacy is thought to be different from other kinds of privacy and therefore deserves special treatment. This idea is called "genetic exceptionalism." [118] According to the genetic exceptionalism, genetic information has some unique characteristics. Firstly, gene has been covered with a mysterious veil for a long time. Knowledge about gene is too professional and complicated to be understood broadly and correctly by the public. In the public perception,

[117] Pormeister, *supra* note 111.

[118] Thomas H. Murray, "Genetic Exceptionalism and 'Future Diaries': Is Genetic Information Different from Other Medical Information?", *in* Mark A. Rothstein (ed.), *Genetic Secrets: Protecting Privacy and Confidentiality in the Genetic Era* (New Haven, CT: Yale University Press, 1997).

> "genes appear to explain obesity, criminality, shyness, directional ability, intelligence, political leanings, and preferred styles of dressing. There are selfish genes, pleasure-seeking genes, violence genes, celebrity genes, gay genes, couch-potato genes, depression genes, genes for genius, genes for saving, and even genes for sinning. These popular images convey a striking picture of the gene as powerful, deterministic, and central to an understanding of both everyday behavior and the secret of life."[119]

The genetic determinism puts gene in a sensible and essential position. It seems like once an individual's genetic information is disclosed, all his or her secret of body like character, behaviors, habits and health status would be revealed to the public. Secondly, genetic information not only records an individual's past life activities such as a diary, but also predicts the future potential diseases such as Alzheimer's disease. This means one's future health conditions can be known about by medical professionals through his or her genetic information, even the information subject might not know it himself or herself. The prediction is just like a time bomb: no matter in what situation the subject is currently, there always exists the possibility that he or she would catch a disease in some time. Besides, genetic information is not just the subject's own business. Genetic information is a kind of inherited information. According to EEOC,

> "[g]enetic information includes information about an individual's genetic tests and the genetic tests of an individual's family members, as well as information about the manifestation of a disease or disorder in an individual's family members (i.e. family medical history). Family medical history is included in the definition of genetic information because it is often used to determine whether someone has an increased risk of getting a disease, disorder, or condition in the future."[120]

An individual's genetic information contains his or her family health history. Correspondingly, people's physical characteristics can also be inferred from their family members' genetic information. In view of these features, genetic information is so important and special that it is different from other medical information. Therefore, some legal scholars in favor of genetic exceptionalism call for special and stringent protection of genetic information. The legislative process in the US exactly reflects this approach. It is based on the argument of genetic exceptionalism that the genetic-specific legislation such as GINA is proposed and promulgated.

Other experts, however, hold opposite opinions. They not only deny the unique features of genetic information, but also suspect the feasibility of genetic exceptionalism. With regard to the uniqueness of genetic information, they firstly put forward that gene is not

[119] Dorothy Nelkin and M. Susan Lindee, *The DNA Mystique: The Gene as a Cultural Icon* (Michigan: University of Michigan Press, 2004).
[120] Definition of "Genetic Information", U.S. Equal Employment Opportunity Commission, available at: https://www.eeoc.gov/laws/types/genetic.cfm (09 September 2020).

the only factor which influences and reflects one's health status. Disease rates and personal attributes are influenced substantially by social, behavioral, and environmental factors.[121] Interaction between gene and environment is a factor which cannot be ignored when scientists analyze the causes of disease. "Although a person's genetic makeup cannot be altered, some lifestyle and environmental modifications (such as having more frequent disease screenings and maintaining a healthy weight) may be able to reduce disease risk in people with a genetic predisposition."[122] Moreover, genes can indeed tell the genetic predispositions in the future, but often this can be modified by many factors, including diet, environment, and exercise. "Non-genetic factors, such as lifestyle habits, may be a better predictor of one's future health."[123] Observable diagnostic conditions can also play the role of predictor. Even the link to other family members is not unique to genes. Family members not only share same genes, but also have similar lifestyle and habits. It is not rare that infectious disease spreads among family members. If an individual catches a disease, his or her family members might also be discriminated or stigmatized because family is a strong link *per se*.

With regard to the feasibility of genetic exceptionalism, the opponents point out that it is hard to separate genetic information and other medical information.[124] "Genetic information and medical information are so intimately intertwined that they cannot be segregated legislatively or by regulation in any way that would prove operationally feasible."[125] To treat genetic information specially, the first step is to define "genetic information". Legislators will find it difficult to give a clear definition of genetic information so as to draw a line between it and other medical information. As mentioned above, the scope of genetic information given by EEOC only includes the result of genetic tests. However, this scope is too narrow to cover all genetic information, especially those which does not come from genetic tests, such as family history. Excluding family history, some diseases, such as Huntington Disease (HD) which indicates fifty percent of the suffering condition from the parents, would not be protected.[126] However, it would be over-inclusive to adopt a broader definition including family history.[127] Some unnecessary and not so sensitive information such as eye color, height and gender are physiological traits related to gene, but they are not

[121] Lawrence O. Gostin and James G. Hodge, "Genetic Privacy and the Law: An End to Genetics Exceptionalism", *Jurimetrics*, 40 (1999).

[122] What Does It Mean to Have a Genetic Predisposition to a Disease?, Your Guide to Understanding Genetic Conditions, *U.S. National Library of Medicine-National Institute of Health*, available at: https://ghr.nlm.nih.gov/primer/mutationsanddisorders/predisposition (09 September 2020).

[123] Lainie Friedman Ross, "Genetic Exceptionalism vs. Paradigm Shift: Lessons from HIV", *Journal of Law, Medicine & Ethic*, 29, 141 (2001).

[124] Amy L. McGuire and Mary Anderlik Majumder, "Two Cheers for GINA?", *Genome Medicine*, 1, 1 (2009).

[125] Clarisa Long, *Genetic Testing and the Use of Information* (Washington, D.C.: American Enterprise Institute, 1999).

[126] Mark A. Rothstein, "Genetic Exceptionalism and Legislative Pragmatism", *The Journal of Law, Medicine & Ethics*, 35, 2 (2007).

[127] Sonia M. Suter, "The Allure and Peril of Genetics Exceptionalism: Do We Need Special Genetics Legislation?", *Washington University Law Quarterly*, 79, 669 (2001).

the information which the privacy law intends to protect.[128] Therefore, it is not practical to isolate genetic information from other health information, and there is no convincing reason to treat genetic information specially because of its "uniqueness".

Apart from the feasibility of genetic-specific legislation, some scholars also express the concern about the adverse effect of genetic exceptionalism. Legislation to protect genetic privacy means blocking access to genetic information, which, at the meantime, blocks the opportunity of treatment. However, genetic information about some diseases sometimes contains the value of timely and effective treatment of the diseases. For example, the diagnosis of phenylketonuria contributes to treat it as early as possible for the healthy future of the infant.[129] For the newborn screening, the rapider the results are spread, the more appropriate treatment can be provided.[130]

Furthermore, stressing the differences too much might bring about *de facto* discrimination. Treating genetics as distinct from the rest of medicine may enhance the stigma of genetic testing, even as legislators attempt to remove its stigmatizing effects.[131] Since there is no explicit distinction between genetic information and other medical information, legally specific treatment would make genetic information more unusual and increase misunderstanding in public perception. Simultaneously, only singling out genetic information implies disregarding other medical information. Now that many functions of genetic information are not so unique and can be operated through other kinds of information, there is no reason to provide special treatment for genetic information. Genetic-specific law makes information of patients with genetically caused disease receiving more rigorous protection, but it is unfair to patients with nongenetic disease. For example, medical information about a woman who has developed breast cancer of genetic origin (e.g. BRCA 1 or 2) can obtain greater protection based on specific law, whereas a woman who has developed breast cancer because of behavioral factors (e.g. smoking) cannot, because there is no such law.[132]

2.3.1.2 Shifting Focus from Genetic Exceptionalism to System Improvement

The justification for genetic exceptionalism is untenable under the attack from the opponents, but genetic information is indeed particularly important and sensitive, at least at the level of clinical medicine. Genetic exceptionalism can be an obstruction for prevention or cure when it is beneficial to reveal genetic information, just as the example of phenylketonuria laid out above. On the other hand, when there is possibility to lead to discrimination or breach of privacy, special treatment for genetic information might be necessary. For instance, lacking of curative therapy, the diagnosis of HD might bring about discrimination.[133] If access to genetic information is conducive to treatment, then it might be reasonable to provide strict protection for the genetic information about some special diseases which cannot be cured for the time being.[134] However, such special protection is not immutable. "In the hoped-for future, when effective therapies are developed to prevent or treat HD, barriers to the dissemination

[128] *Ibid.*

[129] James P. Evans and Wylie Burke, "Genetic Exceptionalism. Too Much of a Good Thing?", *Genetics in Medicine*, 10, 7 (2008).

[130] *Ibid.*

[131] Gostin and Hodge, *supra* note 121.

[132] *Ibid.*

[133] Evans and Burke, *supra* note 129.

[134] *Ibid.*

of that information would be inappropriate."[135]

The core of privacy law is to protect privacy of individuals, which shows respect to human dignity. The aim is not only to eliminate discrimination in order to reach equality, but also contains esteem for individual autonomy. It is beyond doubt that genetic information should be endowed with legal protection as a kind of personal information. The debate is concerned with how genetic information is different from other medical information. Even though the opponents of genetic exceptionalism win the argument, it seems to be difficult for us to equate genetic information with other medical information entirely. After all, genetics indicates the most intimate relationship, and it is the continuity of the kinship. Besides, some people hold the cultural belief "that genetics largely determines who we are (despite many observations to the contrary)"[136]. Setting aside the controversial question, the key problem is not which legislative paradigm is better, but how to prohibit misuse of genetic information and utilize it effectively.

The US legal approach, first of all, is too narrow in terms of privacy law. The purpose of its legislation is solely from the perspective of nondiscrimination. However, nondiscrimination is not the only reason to protect privacy. Genetic information is a kind of identifier just as other personal identifiable information, such as ID number or phone number, through which a natural person can be identified. Discrimination would definitely not occur because of certain number, rather because of the information it contains. Likewise, normal genetic information may not incur discrimination, but they can also be the subject-matter of privacy law. Legislation prohibiting genetic discrimination seems to only protect problematic genetic information which indicates physical or mental disorder, excluding the genetic information signifying good health. Apart from nondiscrimination, privacy law maintains other value. For example, bank account includes economic value; phone number is a social tie; and browsing history records one's all actions on the internet. People just do not want to make some of their information known by others, whether or not it causes discrimination. In other words, individuals have the right to decide what information is made public or not.

Furthermore, the current law only limits health insurers' and employers' access to individual's genetic information. The scope of the limitation is narrow. "With the burgeoning use of genetic testing and advances in understanding hereditary links for disease, laws need to address how the broader society - from government to educational institutions to researchers to nosy neighbors - can use an individual's genetic information in contexts outside of health insurance and employment."[137] It is hard to list all industries in which genetic information needs to be protected. Rather, it shall be considered how to create a common environment to respect and protect genetic information. Comprehensive regulation is needed to protect genetic privacy.

As with the existing traits of genetic information, especially the inherent belief of people (regardless of whether they are true), it is not easy to draw a firm conclusion about whether genetic information is different from other health information. In that case, let us suspend the dispute, shifting the focus to establishing an effective privacy

[135] *Ibid.*

[136] *Ibid.*

[137] Anya E. R. Prince, "Comprehensive Protection of Genetic Information: One Size Privacy or Property Models May Not Fit All", *Brooklyn Law Review*, 79, 175 (2013).

system, in which medical health including genetic information is protected effectually and "legitimate medical providers have quick and reliable access to it."[138] The HIPAA and GINA are respective example of general protection for genetic information and genetic-specific law, while both of them are not an integrated system for genetic privacy. No matter whether regarding genetic information as separate category of information or not, it needs the comprehensive protection under which genetic information can flow freely and safely.

2.3.2 The Analysis of the EU Genetic Privacy Legislation: Research Exemption Should Not Be the Universal Umbrella

2.3.2.1 The Derogation of General Principles and Rights

The EU is not entangled in genetic exceptionalism. The GDPR puts all identifiable personal data into its basket. As for the sensitive and vulnerable data, it requires stricter protection. However, the stricter protection seems not so solid because it can be avoided through research exemption. In the current legal framework of GDPR, it seems like research exemption serves as a universal umbrella, under which any data processing can be justified and legitimized, and it enjoys large priority and convenience. As analyzed above, scientific research can change the purpose of processing personal data without consent of data subjects and considering the new purpose is compatible with the original one for which the data was collected, and the variety of research is broadly covered. However, according to a survey carried out by Ipsos MORI, a social research institute in the United Kingdom (UK), only a minority of people (4%-7%) highly trust private entities, such as insurance, telecommunications and internet companies, to use personal data appropriately.[139] Main reasons of the low trust are misuse and loss of data.[140] On the contrary, only a few respondents (17%) are opposed the government to share anonymized data with researchers in universities for government-funded research.[141] Around half of the respondents agree with data-sharing within government so long as the safeguards are in place.[142] The survey indicts that the majority of respondents are more willing to contribute their data for scientific research in anonymized form and trust the public entities more than the privately funded research organizations. Therefore, further processing without permission might run counter to the data subjects' wishes. In terms of genetic data, as explained above, it can be further processed for any research and stored for undefined time. To keep the data subjects' sense of security about their personal data, robust safeguards shall be established in the context of scientific research.

As for the exemption from purpose and storage limitation, it is hard to prejudge whether the further processing is going to serve a purpose of discrimination in practice. Privately funded research is included in research exemption, and the private entities can serve for

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[138] Evans and Burke, *supra* note 129.

[139] Ho, Chih-Hsing, "Challenges of the EU General Data Protection Regulation for Biobanking and Scientific Research", *Journal of Law, Information and Science*, 25, 1 (2017). Royal Statistical Society, Royal Statistical Society research on trust in data and attitudes toward data use/data sharing, 2 October 2014.

[140] *Ibid.*

[141] *Ibid.*

[142] *Ibid.*

any person for any purpose. If the results derived from the genetic test conducted by private entities are used by employers to select employees, it might cause the issue of discrimination. As to the private entities, it is difficult to separate research from profit purpose. In addition, storing genetic data with identification information is always permitted as long as it is out of research purpose, and the researchers can hold such data for an unspecified time. It is quite unsecure for the data subjects and their relatives because there is no guarantee that the researchers would not repurpose, *i.e.*, the genetic data stored for research originally is not necessarily used for research. The follow-up supervision is lacking.

Research is not only unconstrained by purposes and storage limitation, but also can derogate some rights of data subjects. Article 14 entitles data subjects with rights to be informed of relevant details of their data where the data have not been obtained from the data subject. In terms of genetic data, those which not been obtained from the data subject must be further processed, because the original source of genetic data derives from biological samples, namely it must come from the data subject. At the same time, Article 5(b) releases scientific researchers from providing information for data subject. With these conditions, it can be inferred that genetic research, even where the genetic data is used for further processing, is not constrained by the requirement of providing information. Combining the regulation that further processing for research purpose does not abide by purpose limitation, the data subject might have no right to determine whether their genetic data is going to be used for research once it has been collected, they even have no opportunity to know how their genetic data would be further processed. Likewise, the data subjects' right to be forgotten is also derogated with regard to scientific research. Article 17(3)(d) weighs "the achievement of objectives" of processing for research purpose over the right to be forgotten.[143] Naturally, it is quite difficult if not impossible to retroactively remove a person's data from research conclusions. "However, in terms of prospective research this should not be the case (*i.e.* it should be possible for the data subject to ask for their data to be removed from any databases to avoid future processing for research purposes)."[144] Therefore, the right to be forgotten can be achieved at least in prospective research. However, since scientific research does not observe purpose and storage limitation, the data subjects are not able to stop their data from being transferred to third party or being further processed.

2.3.2.2 Safeguards for Research Exemption

The *Directive* generally requested Member States to furnish suitable safeguards for further processing of personal data for scientific purposes.[145] The Working Party criticized its shortcoming of not containing any specific rules.[146] Then it recommended additional organizational and technical safeguards like the introduction of Information Security Managements Systems (e.g. ISO/IEC standards), and additional legal safeguards could reinforce information rights of data subjects, accentuate strict

[143] Article 17(1) stipulates that the data subject shall have the right to obtain from the controller the erasure of personal data concerning him or her without undue delay and the controller shall have the obligation to erase personal data without undue delay. However, Article 17(3)(d) allows the exemption of the paragraph 1 for archiving purposes in the public interests, scientific or historical research purposes or statistical purposes.

[144] Pormeister, *supra* note 111.

[145] See Recital 29 and Article 17 of the *Directive*.

[146] Advice paper on special categories of data ("sensitive data"), Article 29 Data Protection Working Party, Ref. Ares(2011)444105 – 20/04/2011.

relevance of processing or introduce other specific safeguards.[147] However, the new GDPR does not take the advice. Article 89 outlines the required safeguards and permitted derogation in terms of processing personal data for research purpose. With regard to the safeguards, Article 89(1) just raises the principle of data minimization,[148] and provides a suggestion of pseudonymization, but pseudonymization, as mentioned above, has the risk of re-identification. The GDPR addresses no further rules about the standards of pseudonymization, much less distinguishes the retraceable and non-retraceable means. Pseudonymization is not equal to anonymization, because it merely makes identification of the data subject more difficult, not impossible.[149] "Those in control of pseudonymized datasets may be able to take certain measures to allow 're-identification' of the data subjects in question. This could for example include referencing pseudonymized datasets to master datasets where cross referencing will allow data subjects to be identified."[150]

The derogations, instead, in Article 89 are quite clear. Article 89(2) allows the Member States to discount the rights referred to Articles 15 (right of access), 16 (right to rectification), 18 (right to restriction) and 21 (right to object) for research purpose when these rights impede the achievement of such purpose. Genetic researchers are likely to invoke these articles as defense of not informing data subjects as the genetic research is usually involved with volumes of genetic data. The rules seem to be concerned more with derogations and less with safeguards.[151] Both the safeguards and derogations are at the discretion of the Member States, which leaves room for Member States to carry out the national law. Considering the primary goal of the GDPR is to harmonize the data protection law among the Member States and to promote the efficiency of data transfer, such discretion might be little beneficial to achieve it. The discretion "might create a forum-shopping syndrome where data processors are attracted to conduct their activities in those Member States that provide for the most derogation."[152] Furthermore, stricter approaches in individual Member States might prove to be of little practical efficacy with the easy digital data transfer.[153]

Both the legal framework of the US and the EU are not impeccable in terms of genetic privacy protection. One of the most frequently discussed issues with regard to genetic legal protection in the US is whether genetic information shall be provided with special protection. The GINA and HIPAA can respectively represent the legal paradigm of genetic-specific law and genetic-nonspecific law to some extent. However, it seems that both of the two models do not deal with the issue perfectly. The protection provided by the two regulations is too limited to totally protect genetic privacy. The legislators could probably not dwell on the problem of genetic exceptionalism. On the contrary, they could shift the focus to how to establish a comprehensive legal protection for genetic

[147] *Ibid.*

[148] Data minimization is explained by Article 5(1)(c): "Personal data shall be adequate, relevant and limited to what is necessary in relation to the purposes for which they are processed."

[149] Quinn, *supra* note 98.

[150] *Ibid.*

[151] Pormeister, *supra* note 111.

[152] *Ibid.*

[153] *Ibid.*

information, no matter in the form of a general personal information act which contains genetic information, or a comprehensive genetic information act covering all fields. Different from the US, the EU is faced with another problem. Apart from the general protection for common personal data, the GDPR sets prohibition for processing sensitive data, including genetic data. Along with the strict protection, it also leaves large room for research exemption, which acts as a universal umbrella. A lot of rights and principle can be derogated in the name of research exemption. The processing of genetic data is closely connected with scientific research. In that case, the research exemption could be taken great advantage. Therefore, the corresponding safeguards shall be established so that the research exemption can be utilized in a reasonable extent.

Chapter 3 The Comparison Between the Legal Approaches of the United States and the European Union

The US and the EU have adopted different legal models regarding genetic privacy protection, which reveals their respective rationales of the legislation. Genetic privacy is regarded as disparate attributes of right. In the EU, genetic privacy originates from fundamental human right to respect for private life, while the US protect genetic privacy as a mean of antidiscrimination. Furthermore, they employ different notions in terms of genetic privacy, containing distinct meaning. With the GDPR coming into effect, the genetic data has a uniform and broad definition throughout the EU. However, the connotations of genetic privacy and means of definition in the US change with the statutes, leading to the problem of coordination between different statutes. Similar to the definitions, the legal frameworks of the US and the EU also reflect the distinction of uniformity and fragmentation. The EU has built a unified legal framework for all kinds of personal data through the GDPR, while the US has no consistent legislation with regard to genetic information. The federal government and states acts do not coordinate with each other, and each industry also observes respective rules. Through the comparison, the characteristics in protecting genetic privacy of the two legal systems can be more apparently revealed, so do the shortcomings. Thus, the US and the EU could improve their respective legislation by comparing with each other.

3.1 Attribution of Genetic Privacy

Genetic privacy is endowed with different attributions in the EU and the US. The attribution of the right to genetic privacy implies the right hierarchy, then further determines the origin of rights, the extent of protection, and the remedy of rights.

3.1.1 The EU: Genetic Privacy as Fundamental Right

The right to protection of genetic data originates from international instruments, which guarantee the fundamental rights and freedom in the Europe. Article 8 of the ECHR enshrines the right to respect for private and family life, mainly focusing on the private and family life, home and correspondence.[154] The *Charter of Fundamental Rights of the European Union* (CFREU) draws a distinction between the conventional "right to respect for his or her private and family life"[155], which is modelled after the ECHR and "the right to the protection of personal data"[156], which becomes thereby a new and autonomous fundamental right.[157] The 1995 *Directive* restates its standpoint of respecting individuals' fundamental rights, "notably the right to privacy".[158] One of its objectives is to preserve and strengthen peace and liberty and promoting democracy on

[154] Article 8 European Convention on Human Rights.

[155] Article 7 Charter of Fundamental Rights of the European Union.

[156] Article 8 Charter of Fundamental Rights of the European Union.

[157] Stefano Rodotà, "Data Protection as a Fundamental Right" *in* Gutwirth S., Poullet Y., De Hert P., de Terwangne C., Nouwt S. (eds.), *Reinventing Data Protection* (Dordrecht: Springer, 2009).

[158] Recital 2 of the 1995 Directive.

the basis of the fundamental rights recognized in the ECHR.[159] "The GDPR by and large continues the regulatory approach that has been in place since the *Directive*."[160] Article 1(2) of the GDPR specially addresses the fundamental right to the protection of personal data. More importantly, apart from the right to data protection, EU data protection law also contributes to the protection of other individual rights, "notably personal autonomy, non-discrimination, freedom of expression and thought, and therefore, ultimately, human dignity".[161] As a fundamental right, the right to genetic data protection is universal to all data subjects in the EU. The force of protection is also fairly vigorous. Data subjects who suffer damages resulting from infringement of the GDPR can ask for compensation from data controller or processor.[162] The compensation not only covers material damage, but also includes non-material damage.[163] Apart from active safeguards by data subjects, independent supervisory authority is also responsible for oversight of the right protection. Supervisory authority could impose administrative fines in respect of violation of the GDPR.[164] Furthermore, infringements of the principle of processing sensitive data (*i.e.* Article 9), including genetic data, are subject to higher administrative fines ("up to 20 million EUR" or "up to 4% of the total worldwide annual turnover of the preceding financial year") than invasion of other kinds of personal data.[165] As for the cause of action, data subjects do not have to prove that they have suffered discrimination or harms; they only need to prove that the processing of their sensitive data violates the relevant regulations.[166]

Fundamental human rights are always closely interrelated with the core concepts of human dignity and integrity.[167] These core concepts are inevitable considerations "in addressing advancing science and technology and the issue that arise, encouraging the appropriate application of these new technologies and avoiding the creation of a genetic underclass"[168]. More specifically, by identifying the right to privacy and data protection, the EU essentially opts for a privacy approach to protect genetic data, which means that access to genetic data is hindered in a general way at an early stage. Anyone who intends to obtain genetic data must provide legal basis for processing, which breaks the limitations targeted to industries. All processors are in the same position. Data subjects enjoy high autonomy about whether to undergo a genetic testing and whether to make the results public. They can also determine who is permitted to have access to their

[159] Recital 1 of the 1995 Directive.

[160] Manon Oostveen and Kristina Irion, "The Golden Age of Personal Data: How to Regulate an Enabling Fundamental Right?", *in* Bakhoum M., Conde Gallego B., Mackenrodt MO., Surblyė-Namavičienė G. (eds.), *Personal Data in Competition, Consumer Protection and Intellectual Property Law* (Berlin, Heidelberg: Springer, 2018).

[161] *Ibid.*

[162] Article 82(1) GDPR.

[163] *Ibid.*

[164] Article 83(1) GDPR.

[165] Article 83(5) GDPR.

[166] Kristi Harbord, "Genetic Data Privacy Solutions in the GDPR", *Texas A&M Law Review*, 7, 1 (2019).

[167] Aisling de Paor, *Genetics, Disability and the Law* (Cambridge: Cambridge University Press, 2017).

[168] *Ibid.*

genetic data.

3.1.2 The US: Genetic Information Protection for Antidiscrimination

On the contrary, Professor Frederick Schauer argued that the right to privacy is the social construction in the US, without direct constitutional provisions.[169] Therefore, "it is frequently asserted that there is no fundamental right to privacy in the United States".[170] It is believed that protecting privacy can prevent discrimination by restricting access to information discriminators use to discriminate, since a person cannot consider information that she does not have. "Unlawful discrimination, therefore, frequently requires discriminators to have knowledge about protected status."[171] There is such an opinion that privacy protection and antidiscrimination are complementary[172]: protection personal information can help achieve the purpose of nondiscrimination, and correspondingly, antidiscrimination policies also contributes to prevent unwanted intrusion of personal information. On one hand, privacy regulations restrict the access to some information, thus undermine the basis of discrimination. On the other hand, discrimination means treat individuals differently in the same situation on the basis the knowledge the discriminators have. "Depending on whether the protected information is readily ascertainable, antidiscrimination laws can be understood as prohibitions on certain extrinsic privacy harms because those laws prevent decision makers from using certain kinds of information to an individual's detriment."[173] It means even if one has possessed certain knowledge about others, he or she cannot use the information to differentiate them. As a result, the possession of information becomes meaningless because it cannot be used.

Different from other categories of information, such as race and sex, which is quite apparent and easy to recognize, genetic information usually contains less visible traits where privacy law can come into play to prohibit discrimination.[174] The US chooses to protect personal genetic information in the name of anti-discrimination law or subordinate genetic privacy to other social interests. For example, the HIPPA *Privacy Rule*'s goal is to balance the interest of individuals in maintaining the privacy of their health information with the interests of society in obtaining, using, and disclosing health information to carry out a variety of public and private activities.[175] "Unlike privacy, antidiscrimination protections do not require positive conduct on the part of health insurers and employers, such as disclosure agreements."[176] Instead, it utilizes negative

[169] Frederick Schauer, "Free Speech and the Social Construction of Privacy", Social Research, 68,1 (2001); Khadija Robin Pierce, "Comparative Architecture of Genetic Privacy", *Indiana International Comparative Law Review*, 19, 89 (2009).
[170] *Ibid.*
[171] See Roberts, *supra* note 38.
[172] *Ibid.*
[173] *Ibid.*
[174] *Ibid.*
[175] Stacey A. Tovino, "The HIPPA Privacy Rule and the EU GDPR: Illustrative Comparisons", *Seton Hall Law Review*, 47, 973 (2017).
[176] Jessica L. Roberts, "The Genetic Information Nondiscrimination Act as an Antidiscrimination Law", *Notre Dame Law Review*, 86, 2 (2011).

prohibition to require the covered entities not to access to the protected genetic information. Although it forbids insurers or employers to require or request individuals to take genetic tests, the starting point and ultimate purpose of the genetic privacy law is to eliminate genetic discrimination, just as the title of GINA. After all, the restrictions on access to genetic information diminish the basis of discrimination to some extent.[177] At the same time, the opportunity for insurers and employers to misuse genetic information is also reduced. Therefore, it is not so much genetic *discrimination* that is prohibited, as well as the unjustifiable *use* of genetic information.[178]

3.1.3 Problems for Protecting Genetic Privacy under Antidiscrimination Law

"Discrimination is generally understood to mean a form of detrimental or less favourable treatment, in comparison to other forms of treatment, which is based on certain actual or perceived human features."[179] There are two principles behind the intrinsic meaning of antidiscrimination: antisubordination and anticlassification.[180] Antisubordination principle contends that pervasive social stratification impedes the realization of equal citizenship and "law should reform institutions and practices that enforce the secondary social status of historically oppressed groups".[181] While anticlassification principle holds that "the government may not classify people either overtly or surreptitiously on the basis of a forbidden category: for example, their race".[182]

GINA, as "the first civil rights act of the 21st century"[183], can be understood in line with anticlassification.[184] It is opposed to classify genetic information and forbids to make any decisions according to the classification, regardless of favorable or disadvantaged decisions.[185] GINA explicitly forbids employers to "limit, segregate, or classify the employees" "that would ……adversely affect the status of the employee as an employee, because of genetic information with respect to the employee".[186] It also does not allow insurers to establish rules for eligibility or premium conditioned on genetic information.[187] In other words, GINA does not permit to differentiate individuals based on genetic information and requires equal treatment. In the statutes, disparities between genetic profiles are erased. Thus, "by outlawing positive, as well as negative, differential treatment, GINA takes a formal equal treatment approach to protecting

[177] Janneke H. Gerards and Heleen L. Janssen, "Regulation of Genetic and Other Health Information in a Comparative Perspective", *European Journal of Health Law*, 13 (2006).
[178] *Ibid.*
[179] Aart Hendriks, "The UN Disability Convention and (Multiple) Discrimination: Should EU Non-Discrimination Law Be Modelled Accordingly", *European Yearbook of Disability Law*, 2 (2010).
[180] See Jack M. Balkin and Reva B. Siegel, "The American Civil Rights Tradition: Anticlassification or Antisubordination?", *University of Miami Law Review*, 9 (2003).
[181] *Ibid.*
[182] *Ibid.*
[183] Lee, *supra* note 52..
[184] Roberts, *supra* note 176.
[185] *Ibid.*
[186] See Public Law 110-233 § 202. (a)(2).
[187] See Public Law 110-233 § 2753. (a) and (b).

genetic information"[188]. From another perspective, antisubordination principle seems to be able to facilitate antidiscrimination law better at the source. Antisubordination does not deny the differences among the traits; rather, it encourages diversity. At present, there seems no lower social status divided by genetic information. However, once social perception or practice favors particular genetic profiles, everyone can be risk of genetic underclass. Therefore, antidiscrimination law could "preempt the formation of a genetically disadvantaged social group"[189].

As stated above, the premise of discrimination is to possess certain information. Thus, only after someone have known the personal information, can the antidiscrimination law exert its influence. In other words, antidiscrimination law only inhibits misuse of personal information, but cannot prevent the access to it. However, once personal information is disclosed, the trespass has occurred, no matter how the information is used. The antidiscrimination laws like HIPAA and GINA operate at an earlier stage. They refrain employers and insurers from acquiring genetic information in advance, thus cut off the access to such information. But they only create negative restraints on the covered entities, but confer no positive rights on information subjects.[190] Besides, as antidiscrimination law, the extent is only limited within health insurance and employment, which decide the enforcement is relatively weak. Moreover, the antidiscrimination statute prohibits decisionmakers from considering based on genetic information in order to combat discrimination. However, the basis of discrimination is the genetic information which indicates disability or the potential of disability. That is, only the problematic genetic information is under the protection of antidisctimination law, those normal information, instead, is not covered.

3.1.4 Superiorities and Shortcomings of the EU Protection Model

The GDPR requires positive respect for personal data as fundamental right. It gives data subjects more control over their personal data. Data subjects have high autonomy in deciding whether to disclose the personal data or to whom the data are disclosed. Even though the disclosure is based on explicit consent, the data subjects could "revoke their consent if the changes make it something they no longer want to share"[191]. At the same time, the GDPR also imposes positive obligation on governments to take active measures to protect personal data.[192] The role of governments shall transform from passive defender into active guarder. The dual protection strengthens individual's right to privacy.

With regard to genetic data, the GDPR provides stronger protection not because of its uniqueness, but considering its nature "could create significant risks to the fundamental

[188] Roberts, *supra* note 176.
[189] *Ibid.*
[190] *Ibid.*
[191] Harbord, *supra* note 166.
[192] Menno Mostert, Annelien L. Bredenoord, Bart van der Slootb and Johannes J.M. van Delden, "From Privacy to Data Protection in the EU: Implications for Big Data Health Research", *European Journal of Health Law*, 25 (2018).

rights and freedoms"[193]. Genetic privacy risk is universal to everyone. With the increasing data sharing and easier access to genetic testing, everyone is affected by potential genetic data disclosure.[194] The privacy model employed by the GDPR to protect genetic data enhances the applicability of the regulation, which is not limited within certain sectors or directed at specific groups, but apply to every data subject, protecting them from breach of privacy.

After right to privacy, the right to data protection has been recognized and acquired a prominent position in the GDPR.[195] Traditionally, the right to privacy is violated by interfering one's private life.[196] This scope is very limited, not including all information in personal life. According to the GDPR, any operation performed on any information relating to an identified or identifiable natural person can constitute "processing of personal data".[197] "Consequently, almost all forms of personal data processing fall under the scope of the right to data protection, regardless of whether the right to privacy is interfered with."[198] Therefore, the right to data protection under the GDPR can be invoked more widely. The GDPR obviously expand the application of right to data protection, affording genetic data explicit data protection safeguards.[199] Thus, genetic data not only enjoys protection from general principles set for personal data, but also applies to the stringent protection as sensitive data.

The GDPR closes the gate of obtaining genetic data at an early stage, inhibiting the opportunity of access to personal data, but "it does not provide for any continuing control over personal matters once they enter the public sphere".[200] This cannot avoid misuse of personal data. In other words, privacy approach alone does not address discriminatory use of genetic data.[201] The privacy regime must be combined with other antidiscrimination regulations in order to protect genetic data completely. The privacy regulation intervenes beforehand, controlling the access to genetic data, then the antidiscrimination statute works subsequently, regulating the utilization of it. If one acquired others' genetic data through legitimate approach, there is no regulation about how he or she will use or treat such data. For instance, an employer is usually prohibited from requesting employees' genetic data, but if he receives it through legitimate means such as individuals' consent, he would make decisions based on their genetic data, even if it might result in discrimination. Moreover, not all disclosure count as harm to personal data. It is recognized that personal genetic data is of vital value to scientific

[193] Recital 51 of the GDPR.

[194] Fatos Selita, "Genetic Data Misuse: Risk to Fundamental Human Rights in Developed Economies", *Legal Issues Journal*, 7, 1 (2019).

[195] *Ibid.*

[196] See Article 7 Charter of Fundamental Rights of the European Union.

[197] Article 4 (1) and (2) GDPR.

[198] Mostert *et al.*, *supra* note 192.

[199] See De Paor, *supra* note 167.

[200] Graeme Laurie, *Genetic Privacy: A Challenge to Medico – Legal Norms* (Cambridge: Cambridge University Press, 2002).

[201] Aisling de Paor, "Regulating Genetic Information—Exploring the Options in Legal Theory", *European Journal of Health Law*, 21, 5 (2014).

research and public health.[202] The advances in genetic science have relied upon not only access to genetic information, but also a willingness of individuals to participate in clinical trials.[203] The data protection as fundamental right reduces possibility of getting to personal genetic data and might hinder beneficial disclosure. Furthermore, genetic data indicating disability is not only the basis of discrimination, it can also account for preferential treatment. Under the overprotected status, even if an employer wishes to obtain genetic information to provide accommodations or promote diversity, it cannot do so by law.[204] Imagine a factory worker with a genetic proclivity for carpal tunnel syndrome. This person could perhaps benefit from a longer workday with more frequent breaks to avoid repetitive stress on her wrists, effectively a genetic-information accommodation.[205]

3.2 Notions Relevant to Genetic Privacy in the EU and the US

From the text of the EU and the US regulations, it is obvious that they adopt different notions when it comes to the privacy issues. The different notions imply different definitions and the definition determines the scope of protection by law. "Too narrow a definition may result in reducing the protection of the individual. Too broad a definition may either impede scientific research and the distribution of benefits across society or have an opposite effect of confusion and ambiguity."[206] Personal data, in the context of GDPR, covers a much wider range of information than personally identifiable information (PII), commonly used in North America. In other words, while all PII is considered personal data, not all personal data is PII.[207] For example, if a hacker acquired your credit card number, the hacker might trace this number directly to your name, address and bank account.[208] Hence, the credit card number is a kind of PII. But if a hacker accessed your searched location history from Google Maps, "this information cannot be linked directly to your identity without considerable effort and is therefore not likely to be considered PII"[209], but it is under the protection of the GDPR as personal data.[210]

3.2.1 Processing Genetic Data under the GDPR

3.2.1.1 The Definition of Genetic Data

The GDPR provides a quite broad definition for personal data. Any identified or identifiable personal information can fall into its concept of personal data. The GDPR seeks to expand the material scope of protection in order to apply to the changing

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[202] *Ibid.*

[203] *Ibid.*

[204] Roberts, *supra* note 176.

[205] *Ibid.*

[206] Atina Krajewska, "Conceptual Quandaries about Genetic Data – A Comparative Perspective", *European Journal of Health Law*, 16 (2009).

[207] Malia Thuret-Benoist, "What is the difference between personally identifiable information (PII) and personal data?" *Tech GDPR*, 27 June 2019.

[208] See Sara Kassabian, What's the difference between PII and personal data?, *Truevault Blog*, 30 October 2018.

[209] *Ibid.*

[210] *Ibid.*

situations. “This is due to the in-built possibilities for the evolving interpretation of the concept itself, exploding generation and aggregation of data, as well as advances in data analytics.”[211] The broad concept of personal data equips the GDPR with flexibility, adaptability, as well as uncertainty.[212]

With regard to the definition of genetic data, however, the GDPR appears to take a more cautious and restrained position. In fact, the proposal for GDPR defined genetic data broadly: “‘genetic data’ means all data, of whatever type, concerning the characteristics of an individual which are inherited or acquired during early prenatal development.”[213] This pattern is similar to the definition of personal data, namely combining all data with specific characteristics. According to the proposal *Regulation*, genetic data incorporate all inherited or acquired information, including “family medical history and other health-related information”[214]. However, the final version of the GDPR slightly narrow the definition: “ ‘Genetic data’ means personal data relating to the inherited or acquired genetic characteristics of a natural person which give unique information about the physiology or the health of that natural person and which result, in particular, from an analysis of a biological sample from the natural person in question.”[215] This definition only includes the data obtained from the results of analysis biological sample, namely genetic testing,[216] and focuses more on the identification function of genetic data, less considering the biological connections with other individual (as it only stresses the unique information revealing the physiological status of data subjects, but not addresses the information embodied in their family members). Compared to the broad concept of personal data, this narrow definition seems to be less inclusive, thus restricting the application of the GDPR in terms of genetic data protection. “This was a missed opportunity to formulate a comprehensive definition of genetic data to include not only the results of genetic testing but also family medical history.”[217]

3.2.1.2 Performance on Genetic Data: Controlling and Processing

The performance on genetic data reveals the relationship between different parties in the processing of data. Under the GDPR, there are two roles involving with data processing: controller and processor. Controller is the one who “determines the purposes and means of the processing of personal data”[218]. The controller shall consider the “nature, scope, context and purposes of processing as well as the risks of varying

[211] Nadezhda Purtova, “The Law of Everything. Broad Concept of Personal Data and Future of EU Data Protection Law”, *Law, Innovation and Technology*, 10, 1 (2018).
[212] *Ibid.*
[213] Proposal for a REGULATION OF THE EUROPEAN PARLIAMENT AND OF THE COUNCIL on the protection of individuals with regard to the processing of personal data and on the free movement of such data (General Data Protection Regulation), European Commission, COM(2012) 11 final, 2012/0011 (COD).
[214] De Paor, *supra* note 167.
[215] Article 4(13) GDPR.
[216] De Paor, *supra* note 167.
[217] *Ibid.*
[218] Article 4(7) GDPR.

likelihood and severity for the rights and freedoms of natural persons"[219] and ensure "that processing is performed in accordance with" the GDPR[220]. Processor is the one who "processes personal data on behalf of the controller"[221]. The processor shall carry out the processing with the authority from the controller based on the contract relationship.[222] To put it simply, controller is in charge of the whole processing, while processor is the actual operator.

In the context of genetic data processing, there are two models of relationship. The first is patient-clinician-laboratory relationship, in which the clinician gains genetic data from patient, and outsources the analysis of data to the laboratory. In this model, the clinician acts as the controller and the laboratory as the processor.[223] The relationship between patient and clinician is based on a contract, so does the relationship between clinician and laboratory. "The processor assists the controller, by appropriate technical and organizational measures, insofar as this is possible, to satisfy his obligation to respond to requests from data subjects for exercising their rights."[224] Another model is consumer-genetic testing company, which usually exists in the DTC genetic testing. The genetic companies directly obtain genetic data from the consumers and feedback with the analysis report of their genetic profile. In this situation, the roles of controller and processor roll up into one, because the testing companies not only determine the purposes and means of the genetic data processing, but also undertake the analysis thereof.

Setting the roles of controller and processor, the GDPR makes the division of responsibilities relatively clear. The controller steers the processing within the framework of regulations and assesses the potential risks, while the processor performs actions on the data obtained. Moreover, this setting avoids the distinction between publicly medical care purpose and private commercial purpose, as it can adapt to both circumstances.

3.2.1.3 The Exclusion of Anonymous Data: Can Genetic Data Be Anonymized?

The data that are excluded from the protection of the GDPR can partly reflect the actual scope of its protection. The GDPR is directed at identifying data, which determines that the anonymous data cannot be protected, namely "information which does not relate to an identified or identifiable natural person or to personal data rendered anonymous in such a manner that the data subject if not or no longer identifiable"[225]. In order to determine whether the data is nonidentifiable, "account should be taken of all the means reasonably likely to be used……either by the controller or by another person to identify

[219] Article 24(1) GDPR.
[220] *Ibid.*
[221] Article 4(8) GDPR.
[222] Article 28 GDPR.
[223] Jasper A. Bovenberg and Mara Almeida, "Patients v. Myriad or the GDPR Access Right v. the EU Database Right", *European Journal of Human Genetics*, 27, 2 (2019).
[224] *Ibid.*
[225] Recital 26 GDPR.

the natural person directly or indirectly"[226]. To ascertain all the possible means to identify the natural person, "account should be taken of all objective factors……taking into consideration the available technology at the time of the processing and technological developments"[227]. In short, the anonymous data shall be stripped of any identifying information, and the process must be irreversible.[228]

The crucial question therefore becomes whether genetic data can be truly anonymized, or whether the de-identified genetic data can always be re-identified. Previous studies have attempted to demonstrate that "individual genomic data, even if included in datasets of aggregated information, cannot be irreversibly de-identified"[229]. Aside from the experimental evidence, multiple factors shall be taken into consideration to assess the likelihood of re-identification of genetic data. The re-identifiability shall be assessed in contexts in which the genetic data are being processed.[230] The processing of genetic data for kinds of purposes, in various institutional settings and at different stages are subject to "distinct policy, ethical and regulatory frameworks that may prescribe different duties for data controllers, and different safeguards for data subjects"[231]. "The adoption of technical safeguards and the implementation of adequate governance frameworks"[232], which are in the charge of data controllers, could affect the assessment of re-identifiability of genetic data. Under the high standards of anonymization, data controllers might "take a conservative approach and consider genomic data as, in principle, always identifiable"[233]. The GDPR therefore establishes a quite high threshold for anonymization of genetic data, which, as a result, can hardly escape from being regulated.

3.2.2 Using Genetic Information in the US

3.2.2.1 Genetic Information in the US

In the US, "information relating to an individual is typically referred to as 'personal information' (rather than personal data)"[234]. Compared to the EU, the US offers various definitions of personal information, without a coherent and consistent notion.[235] The definition of personal data varies across states and regulations.[236] "Certain data may be considered personal information for one purpose but not for another."[237]

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[226] *Ibid.*

[227] *Ibid.*

[228] Mahsa Shabani and Luca Marelli, "Re-identifiability of Genomic Data and the GDPR", *EMBO reports*, 20, 6 (2019).

[229] *Ibid.*

[230] *Ibid.*

[231] *Ibid.*

[232] *Ibid.*

[233] *Ibid.*

[234] USA: Data Protection 2019, ICLG.com, 03 July 2019, available at: https://iclg.com/practice-areas/data-protection-laws-and-regulations/usa (last accessed: 22 September 2020).

[235] Paul M. Schwartz and Daniel J. Solove, "Reconciling Personal Information in the United States and European Union", *California Law Review*, 102, 4 (2014).

[236] *Supra* note 234.

[237] *Ibid.*

As for genetic information, there are specific laws to provide detailed definition. HIPAA gives stratified definition of genetic information. First of all, it points out that genetic information means: (i) the individual's genetic tests; (ii) the genetic tests of family members of the individual; (iii) the manifestation of a disease or disorder in family members of such individual; or (iv) any request for, or receipt of, genetic services, or participation in clinical research which includes genetic services, by the individual or any family member of the individual.[238] The situations listed above focuses on specific actions or manifestation concerned with genetics. Then it stipulates that any reference to genetic information "concerning an individual or family member of an individual shall include the genetic information of: (i) A fetus carried by the individual or family member who is a pregnant woman; and (ii) Any embryo legally held by an individual or family member utilizing an assisted reproductive technology"[239]. At last, information about the sex or age of any individual are excluded.[240]

Likewise, genetic information obtains similar definition in GINA. Both Titles define genetic information with respect to any individual in general as: "(i) such individual's genetic tests, (ii) the genetic tests of family members of such individual, and (iii) the manifestation of a disease or disorder in family members of such individual"[241]. They also incorporate "any request for, or receipt of, genetic services, or participation in clinical research which includes genetic services, by such individual or any family member of such individual"[242]. Besides, genetic information of a fetus or embryo are also recognized under GINA.[243] Same as the HIPAA, "the term 'genetic information' shall not include information about the sex or age of any individual"[244]. With regard to genetic tests, they do not only refer to "an analysis of human DNA, RNA and chromosomes"[245], but also cover the analysis of "proteins, or metabolites that detects genotypes, mutations, or chromosomal changes"[246],which broadens the applicable forms of genetic tests, recognizing the indirect detection of genetic risks.[247] Considering the difficulties to distinguish genetic information from other medical information, GINA defines family history in a general way. It does not emphasize "the manifestation of an *inheritable* disease or disorder in the family members"[248], but offers the concept of family history in an inclusive manner. Thus, the term "family history" does not necessarily have predictive nature.[249]

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[238] 45 CFR § 160.103.
[239] *Ibid.*
[240] *Ibid.*
[241] Public Law 110-233 § 101. (d)(6)(A) and § 201. (4)(A).
[242] Public Law 110-233 § 101. (d)(6)(B) and § 201. (4)(B).
[243] Public Law 110-233 § 101. (c) and § 209. (b).
[244] Public Law 110-233 § 101. (d)(6)(C) and § 201. (4)(C).
[245] Public Law 110-233 § 101. (d)(7)(A) and § 201. (7)(A).
[246] *Ibid.*
[247] Sonia, M. Suter, "GINA at 10 Years: The Battle over 'Genetic Information' Continues in Court", *Journal of Law and the Biosciences*, 5, 3 (2018).
[248] *Ibid.*
[249] *Ibid.*

Therefore, with regard to genetic information, the US regulations adopt a broad definition, which not only recognized the various forms of genetic information (including the results of genetic tests of individuals and their family members, and the family medical history), but also expand the subjects of application (including genetic information of a fetus or embryo). However, the definition of genetic information under GINA can be narrow in the sense that it does not apply to "the manifestation of a disease or disorder of an individual"[250]. It means that health insurers and employers are only discouraged from accessing pre-symptomatic genetic information, which is a potential risk and has not developed into the manifested condition, while the manifestation of genetic disease is not protected by GINA. Such distinction between manifested and pre-symptomatic conditions might, to some extent, destroy the goal to provide full protection,[251] even if the manifested information can be covered by other acts, such as the ADA. In addition, the differentiation raised difficulties for enforcement because it requires the covered entities to make a medical judgment of whether the genetic information in question is predictive of future disease while the judgement is not immutable with the evolvement of medical discoveries.[252]

3.2.2.2 Access to, Uses and Disclosure of Genetic Information by Covered Entities

The US regulations distinguish the performances and performers (*i.e.* covered entities) on genetic information. There are three kinds of actions by different covered entities in the US regulations: accessing, using and disclosing, and the regulations differentiate involved parties by industries.

GINA firstly prohibits insurers and employers from collecting genetic information from individuals or their family members. The methods of collection include requesting or requiring individuals or their family members to undergo a genetic test and purchasing genetic information.[253] However, incidental collection is not considered in violation of the regulation.[254] It seems that the incidental collection provision leaves room for accessing to individual genetic information, but this exception is restricted by the purposes of using the information: only if the information which is incidentally obtained is not used for underwriting purposes, can the incidental collection be permitted.[255]

Therefore, use is the core action that is emphatically regulated, which reflects the purposes of collecting genetic information and the attitude of the covered entities toward the information. GINA does not forbid any use of genetic information, but misuse thereof, namely making unfavorable decisions to individuals on the basis of their genetic information. The insurance issuers are prevented from establishing rules for the eligibility and adjusting premium or contribution amounts based on genetic

[250] Public Law 110-233 § 101. (a)(2), § 102. (a)(1)(B), § 102. (b)(1)(B), § 103. (a)(2) and § 104. (a).
[251] Suter, *supra* note 247.
[252] *Ibid.*
[253] Public Law 110-233 § 101(b).
[254] *Ibid.*
[255] *Ibid.*

information[256]; employers shall not "fail or refuse to hire, or to discharge employee" or "limit, segregate, or classify the employees……in any way that would deprive or tend to deprive any employee of employment opportunities or otherwise adversely affect the status of the employee"[257]. Except for the general prohibition of acquiring genetic information, health care professionals or counselors could obtain individual genetic information when the employees are receiving genetic services, but they are prohibited from disclosing the information to employers except in the way that does not "disclose the identity of specific employees"[258].

As an antidiscrimination act, GINA not only regulates the discriminatory treatment of genetic information, but exerts itself ahead of time – from the stage of access, even if the limitation of acquiring genetic information serves for the purpose of prohibiting the misuse thereof. However, GINA expressly lists the covered entities, thus the non-covered entities are not regulated by it. As a result, the scope of application is limited to large extent.

3.2.2.3 The Lenient Standards for De-identification of Genetic Information

The US privacy laws also exclude the information that are of less identifiable value. The HIPAA, has established the standard for de-identification of PHI: "Health information that does not identify an individual and with respect to which there is no reasonable basis to believe that the information can be used to identify an individual is not individually identifiable health information." [259] Such nonidentifiable health information is thus not regulated by the HIPAA. The wording used is quite tolerant, just requiring the information cannot identify an individual, without further requirements. To implement the requirements for de-identification, the HIPAA suggests two methodologies: "the 'statistical' (or expert) method" [260] and "the 'safe harbor' method"[261]. The statistical method needs "a person with appropriate knowledge of and experience with generally accepted statistical and scientific principles and methods"[262] to render information "not individually identifiable"[263]. The ideal result shall be that the risk of identifying information subject is "very small"[264] with the nonidentifiable information. The safe harbor method requires removing the following 18 identifiers so that "the covered entity does not have actual knowledge that the information could be used alone or in combination with other information to identify an individual who is a subject of the information"[265]: (A) Names; (B) All geographic subdivisions smaller than a State; (C) All elements of dates (except year) for date directly related to an individual;

[256] Public Law 110-233 § 102.
[257] Public Law 110-233 § 202(a).
[258] Public Law 110-233 § 202(b)(2).
[259] 45 CFR § 164.514 (a).
[260] Deven McGraw, "Building Public Trust in Uses of Health Insurance Portability and Accountability Act De-Identified Data", *Journal of the American Medical Informatics Association,* 20 1 (2013).
[261] *Ibid.*
[262] 45 CFR § 164.514 (b)(1).
[263] *Ibid.*
[264] 45 CFR § 164.514 (b)(1)(i).
[265] 45 CFR § 164.514 (b)(2)(ii).

(D) Telephone numbers; (E) Fax numbers; (F) Electronic mail addresses; (G) Social security numbers; (H) Medical records numbers; (I) Health plan beneficiary numbers; (J) Account numbers; (K) Certificate/license number; (L) Vehicle identifiers and serial numbers; (M) Device identifiers and serial numbers; (N) Web Universal Resource Locators; (O) Internet Protocol address numbers; (P) Biometric identifiers, (Q) Full face photographic images and any comparable images; and (R) Any other unique identifying number, characteristic, or code.[266] The former method adopts an objective standard, which relies on the operations by experts; the latter depends on the subjective judgment by the covered entity itself.

There were comments that true de-identification is impossible "because more and more information about individuals is being made available to the public"[267]. In response to the comments, the HHS acknowledged that it is impossible to de-identify to absolutely zero risk and this is neither the purpose of the provisions. It expressly explained that the statutory standard "envisions a reasonable balance between risk of identification and usefulness of the information"[268]. The legislators actually make a compromise between the statutory requirements of de-identification and the practical limitations. The de-identifying process is not entirely for the purposes of protecting privacy of information subjects, but providing excuses for utilizing the information without the regulation by HIPAA.

3.2.3 Analysis of the Scope of Notions Adopted by the EU and the US

The notions accepted by the EU and the US reveal the differences in scope. I will analyze the scope of definitions from three aspects: genetic privacy, relevant performance on genetic privacy and the information that is excluded from the regulation.

The GDPR defines genetic information in a prudent manner. It closely relates to the data subjects themselves, and does not incorporate information obtained from family history. Proceeding from the perspective of data protection law, the GDPR principally considers whether the genetic data can be used to identify specific person. However, the US law adopts a broad definition of genetic information. It not only addresses the genetic predisposition of the subjects themselves, but also takes the genetic status of their family members into consideration, because the family history might influence the opinions toward individuals and result in discrimination. In another sense, the definition in GDPR can be broader because it does not limit the status of genetic data. The GDPR covers the genetic data in all periods, regardless of presymptomatic or manifested. Conversely, the application scope of GINA is restricted within the presymptomatic genetic information. Once the genetic propensity develops into actual genetic disease or disorder, it is not protected by GINA anymore. Therefore, neither of the two definitions is perfect. It would be better if the connotation of the two definitions can be combined, then the genetic privacy would be overall protected.

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[266] 45 CFR § 164.514 (b)(2)(i).

[267] Department of Health and Human Services, *Standards for Privacy of Individually Identifiable Health Information; Final Rule*, 45 CFR Parts 160 and 164, 65 FR 82462-82829, 2000.

[268] *Ibid.*

With regard to the performance on genetic information, the EU and the US diverge fairly from each other. The GDPR integrates the different parties and roles in the processing of genetic data, which simplifies the relationship between the involving parties and improves the applicability of the regulation. Through setting the role of "controller" and "processor", the GDPR does not necessarily need to classify the numerous fields or industries. In the context of genetic data, it is not necessary to determine in which the genetic privacy breach might occur, such as insurance, employment or education, because every party taking part in the processing of genetic data can correspond to their respective roles. Therefore, this model is quite flexible and applicable. By contrast, the US adopts the model to make rules by industries. It does not create a uniform regulation to apply to all fields, but makes rules according to the need of each industry. Such model is too burdensome and impractical, because it is hard to list all industries which call for genetic privacy protection with the deep penetration into more and more fields by genetic technology. On the other hand, legislation by industries might result in waste of efforts. It is easy to find that there are a lot of repeated wording in GINA, because the provisions in the insurance section need to be formulated once more in the employment section with no substantial distinctions. As a result, the legislative language appears to be complicated.

As for the exclusion of de-identified genetic information, the EU and the US set different standards. To render data anonymous, the GDPR establishes means-exhausted and zero-risk standard. It fully considers the future development of technology to identify specific person with genetic data in order to maximally reduce the risk of re-identification. As data protection law, the GDPR focuses more on whether the data can be used to identify data subjects and tries to avoid unlawful identification. The high threshold of anonymization makes data hardly excluded from the GDPR, and thus the scope of covered data is expanded. Furthermore, requiring exhausting all methods and considering all factors, the GDPR evaluates the effect of anonymization based on the result – the anonymization of genetic data shall be irreversible. However, the US acknowledges that it is impossible to truly de-identify genetic data, and it permits the existence of risk of re-identification. Compared to the result-oriented standards in the GDPR, the US law employs formal standards for de-identification. It only requires the covered entities to take actions of de-identification, but not pursues a high level of de-identifying effect. In that case, the de-identification of genetic data actually becomes a means of evading regulation.

The definition of genetic privacy, the enforcement of regulations and the exclusion of application reveal respective attitudes of the EU and the US laws. The GDPR emphasizes the rights of data subjects, trying to avoid the disturbance to individuals resulted from unlawful access to their genetic data. While the US genetic privacy laws pay more attention to coordinating the relationship between genetic privacy and other values, such as antidiscrimination and reasonable utilization. The discrepancy reveals the related values and respective perspectives from which the EU and the US start to

protect genetic privacy.

Chapter 4 Balancing Interests of Parties Relate to Genetic Privacy

Despite of the differences between the EU and the US regulations with regard to the genetic privacy, they have the same goal, which is to respect the personal interests in genetic privacy. One of the most important interests is the individual autonomy, which represents the personal control of their own genetic information. Both the EU and the US laws seek to enhance personal autonomy interest through various designs in regulations, especially through the informed consent mechanism. However, considering the peculiarities of genetic information, it is not only the personal issue which completely pertains to private sphere, but would have implication on others, for example, the relatives of information subjects. Therefore, the genetic privacy law shall consider balancing the interests of relevant parties while protecting personal privacy.

4.1 Personal Interest behind Genetic Privacy: Autonomy

The definitions of autonomy are not uniform among scholarship. As Graeme Laurie puts it, there are four common elements contained in this concept. The first is "choice"[269]. To respect one's autonomy generally means respect his choices. The second one is "non-interference"[270]. An autonomous individual shall make choices without intervention or constraint by others. The third element is "capacity"[271]. An individual can freely make choices on condition that she possesses such capacity, for example, to be a rational person. The last is "informed"[272]. The will of an autonomous individual is reflected on the understood information. In general, autonomy is concerned about one's capability to informedly make choice for herself without interference. "In the health care setting respect for individual autonomy is required by the common ethical principles that constitute medical ethics and which dictate the appropriateness of the conduct of health care professionals toward their patients."[273] This principle is even regarded as a fundamental principle "from which other ethical principles derive their authority"[274]. In the context of genetic privacy, autonomy might mean that individual have the right to control over their genetic information, deciding whether to know about their own genetic information, whether to disclose the information, to whom the information could be disclosed, and how their genetic information shall be used.

The principle of privacy, as a moral and legal principle, requires people not to probe others' genetic information.[275] Therefore, the right to privacy creates a space inhibiting

[269] Laurie, *supra* note 200.
[270] *Ibid.*
[271] *Ibid.*
[272] *Ibid.*
[273] *Ibid.*
[274] *Ibid*; R. S. Downie, K. C. Calman, R. A. Schröck and M. Macnaughton, *Healthy Respect: Ethics in Health Care* (Oxford: Oxford University Press, 2009).
[275] Matti Häyry and Tuija Takala, "Genetic Information, Rights, and Autonomy", *Theoretical Medicine and Bioethics*, 22, 5 (2001).

trespass by others, in which individuals can autonomously make choices with regard to their genetic information. However, given the fourth element of autonomy, *i.e.* informed, the private space is not absolutely closed, because in health care relationships, professionals would inevitably step into the private sphere of patients, and it is also essential for individuals to know about their own genetic information with the assistance of professionals. "If a central ethical value behind privacy is respect for personal autonomy, then individuals from whom data are collected must be afforded the right to know about and to approve the uses of those data."[276] Thus, the conducts of professionals need to be justified. "Consent can entitle medical professionals to seek knowledge regarding the genetic makeup of their patients."[277] Consent can be a form of exercising autonomy and create a "privilege"[278] through contracts or commitments. In turn, valid consent also represents the spirit of autonomy because it must be given autonomously without coercion or deception.[279]

4.2 Protection of Personal Interest in the EU and the US Laws

The protection of genetic privacy in the EU and the US reflects their respective value orientation and nature of regulation. The GDPR attaches paramount importance to data subjects' rights, strengthening personal control over their personal data. It enables data subjects a variety of rights so that their autonomy can be adequately guaranteed. By contrast, the US tends to protect people's genetic privacy through stressing the duty of confidentiality of relevant parties. By establishing the duty of confidentiality between certain relationships, an external safeguard could be formed.

4.2.1 Autonomous Data Subjects under the GDPR

4.2.1.1 Data Subjects' Right

The entitlement of a series of rights by the GDPR highlights the subject status of individuals. With these rights, data subjects could better control over the content, the processing and the storage of their genetic data.

Article 15 GDPR grants data subjects the right to know "whether or not personal data concerning him or her are being processed" and essential information related to the processing.[280] This right, as the Recital 63 explains, includes:

> "the right for data subjects to have access to data concerning their health, for example the data in their medical records containing information such as diagnoses, examination results, assessments by treating physicians and any treatment or interventions provided."[281]

With the right of access, the data subjects are not only clearly aware what happens to

[276] Lawrence O. Gostin, "Genetic Privacy", *in* Sheila A.M. McLean (ed.), *Genetics and Gene Therapy* (Abingdon: Routledge, 2017).
[277] *Ibid.*
[278] *Ibid.*
[279] The Collecting, Linking, and Use of Data in Biomedical Research and Health Care: Ethical Issues, Nuffield Council on Bioethics, 2013.
[280] Article 15 GDPR.
[281] Recital 63 of the GDPR.

their personal data, but also further understand the complicated personal data such as medical data.

The individual genetic data generated in genetic research include not only the genomic raw data[282], but also the individual research findings[283]. Individuals' right of access to their genetic data has been previously recognized. Article 8(2) the CFREU entitles everyone with "the right of access to data which has been collected concerning him or her"[284]. The *Additional Protocol to the Convention on Human Rights and Biomedicine, concerning Genetic Testing for Health Purposes* addresses respect for right to information, stating "everyone undergoing a genetic test is entitled to know any information collected about his or her health derived from this test"[285]. The *Additional Protocol to the Convention on Human Rights and Biomedicine, concerning Biomedical Research* further stipulates that the person participating in a research project shall be informed of "arrangements for access to information relevant to the participant arising from the research and to its overall results"[286]. The GDPR more explicitly clarifies the content of the right of access. The individual genetic data are definitely incorporated in the ambit of Article 15(3) which requires the controller to provide "a copy of the personal data undergoing processing"[287].

The right of access can be regarded as the premise of individual autonomy, as it affects people's ability to freely decide how their personal data are processed or transferred.[288] In the genetic research context, the initial forms of what collected from data subjects are just biological samples, while they do not thoroughly understand the self-knowledge contained in the samples and the inherent information about physical and psychological disposition is possessed by the research institutions. To enable the data subjects assess to the genetic data might to the extent avoid the informational asymmetry and therefore promote the informed exercise of autonomy.

The right of access, however, is limited to some degree, (especially in genetic research). The GDPR allows the EU or the Member States to place restrictions on this right in so far as "such a restriction respects the essence of the fundamental rights and freedoms and is a necessary and proportionate measure in a democratic society to safeguard" other important interests.[289] Accordingly, the power of restriction can be highly elastic and largely at the discretion of the Member States. "This is most likely intended to refer

[282] Christoph Schickhardt, Henrike Fleischer and Eva C. Winkler, "Do Patients and Research Subjects Have a Right to Receive Their Genomic Raw Data? An Ethical and Legal Analysis", *BMC Medical Ethics*, 21, 1 (2020).

[283] Kristien Hens, Herman Nys, Jean-Jacques Cassiman and Kris Dierickx, "The Return of Individual Research Findings in Paediatric Genetic Research", *Journal of Medical Ethics*, 37, 3 (2011).

[284] Article 8(2) CFREU.

[285] Article 16(2) Additional Protocol to the Convention on Human Rights and Biomedicine, concerning Genetic Testing for Health Purposes.

[286] Article 13 (2)(v) *Additional Protocol to the Convention on Human Rights and Biomedicine, concerning Biomedical Research*.

[287] Article 15(3) GDPR.

[288] Schickhardt, Fleischer and Winkler, *supra* note 282.

[289] Article 23(1) GDPR.

to cases where there is a therapeutic necessity to do so. For example, Member States would have the power to provide that raw data could only be released by way of genetic counselling."[290] Moreover, the right of access to genetic data can be further restricted by the research exemption provided by Article 89(2) of the GDPR. Under research exemption, research institutions are able to refuse return the genetic data to the participants in defense of the realization of research purpose.

Genetic data are sensitive and vulnerable because of some particularities. They "can be easily obtained without the knowledge of the individual"[291]. The right of access facilitates the data subjects to exactly know the flow of their genetic data and decide the subsequent processing actions, laying the foundation for them to consent to the processing of genetic data or not. Therefore, the right of access could to a certain extent guarantee the personal interest of autonomy.

4.2.1.2 Consent by Data Subjects

"Consent has been the cornerstone of the personal data privacy regime."[292] It is the source of valid authorization of lawful processing on personal data. In terms of genetic data, consent is the prerequisite for the collection of biological specimens and the analysis thereof. "The doctrine of consent is premised on the liberal tenets of individual autonomy, dignity, and integrity, rooted in the fundamental respect to a person, and intertwined with the right to respect for privacy."[293]

According to Article 4(11), consent means "any freely given, specific, informed and unambiguous indication of the data subject's wishes by which he or she, by a statement or by a clear affirmative action, signifies agreement to the processing of personal data relating to him or her"[294]. Article 7 lays out strict conditions for consent. For processing genetic data which pertain to special categories of data under Article 9, explicit consent for specified purposes is first of all necessary condition under the GDPR.[295] Data subjects can also withdraw the consent at any time.[296] Thus, the respect for people's autonomous choice runs through the processing of genetic data, because the data subjects could give or refuse to give consent at any moment, implying that the start and end of processing are based on the data subjects' wishes.

It seems easy to determine the rights and obligations between the simple patient-clinician or participant-research relationships. However, when it comes to the secondary use of genetic data, things would be more complex. For example, the genetic data collected from data subjects might be stored in public databases, through which

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[290] Schickhardt, Fleischer and Winkler, *supra* note 282.

[291] Marilyn Cech, "Genetic Privacy in the 'Big Biology' Era: The 'Autonomous' Human Subject", *Hastings Law Journal*, 70, 3 (2019).

[292] Anne S.Y. Cheung, "Moving Beyond Consent for Citizen Science in Big Data Health and Medical Research", *Northwestern Journal of Technology and Intellectual Property*, 16, 1 (2018).

[293] *Ibid.*

[294] Article 4(11) GDPR.

[295] Article 9(2)(a) GDPR.

[296] Article 7(3) GDPR.

clinical laboratories or research institutions could share the data and conduct secondary processing.[297] However, the secondary use is not authorized by consent. The consent-based processing needs to "ensure that consent is valid and appropriate at every stage of the process and with respect to future possibilities"[298].

Considering that the possible future need for genetic data in research context, Recital 33 of the GDPR suggests:

> "data subjects should be allowed to give their consent to certain areas of scientific research when in keeping with recognized ethical standards for scientific research. Data subjects should have the opportunity to give their consent only to certain areas of research or parts of research projects to the extent allowed by the intended purpose"[299].

It appears to provide a flexible solution of broad consent to secondary use of genetic data for genetic research, relaxing the requirement of specification. Broad consent authorizes genetic researchers to collect genetic data "for use in unspecified future research projects"[300]. However, as the Working Party explains, Recital 33 shall not be interpreted in favor of broad consent:

> "Recital 33 does not disapply the obligations with regard to the requirement of specific consent. This means that, in principle, scientific research projects can only include personal data on the basis of consent if they have a well-described purpose. For the cases where purposes for data processing within a scientific research project cannot be specified at the outset, Recital allows as an exception that the purpose may be described at a more general level."[301]

The sensitive data, such as genetic data, the processing of which is based on explicit consent would comply with "a stricter interpretation" and "a high degree of scrutiny" in terms of adopting the flexible position of Recital 33.[302] When it is indeed difficult to obtain explicit consent in advance due to the uncertainties of research purposes, the Working Party still insists the "subsequent rolling granular consents over one, *ex ante*, broad consent"[303]. Therefore, according to the guidance of the Working Party, the

[297] Mahsa Shabani, Stephanie O.M. Dyke, Luca Marelli and Pascal Borry, "Variant Data Sharing by Clinical Laboratories through Public Databases: Consent, Privacy and Further Contact for Research Policies", *Genetics in Medicine*, 21, 5 (2019).

[298] Susan E. Wallace, Elli G. Gourna, Graeme Laurie, Osama Shoush and Jessica Wright, "Respecting Autonomy over Time: Policy and Empirical Evidence on Re-Consent in Longitudinal Biomedical Research", *Bioethics*, 30, 3 (2016).

[299] Recital 33 of the GDPR.

[300] Dara Hallinan, "Broad Consent under the GDPR: An Optimistic Perspective on a Bright Future", *Life Sciences, Society and Policy*, 16, 1 (2020).

[301] Article 29 Working Party Guidelines on consent under Regulation 2016/678, 17/EN WP259 rev.01, at last revised and adopted on 10 April 2018.

[302] *Ibid.*

[303] Hallinan, *supra* note 300; Article 29 Working Party Guidelines, *supra* note 301.

processing of genetic data is still pursuant to the standard of specific informed consent for secondary use. As a result, it strengthens the power and importance of the consent of data subjects.

Patient autonomy is primarily "manifested through the doctrine of informed consent"[304]. Through the stringent requirement of informed consent, the GDPR maximally sticks to its original aim to increase data subjects' control over their personal data. For sensitive data, including genetic data, the GDPR endorses explicit consent as the foremost condition for legitimizing the processing, holding a conservative position for broad consent, thus avoiding the infringement of individual autonomy as much as possible.

4.2.2 Protecting Individual Autonomy under the US Laws

4.2.2.1 Ethical Principles of Autonomy in the US

Before the legal responses to the risks to individual rights in biomedical research, especially the potential infringement of individual autonomy, the professional ethics takes the lead in dealing with the vulnerable status of participant rights. The first U.S. Federal Policy for the protection of human subjects was put into place in 1953 for research conducted at the Clinical Center, National Institutes of Health.[305] With the lessons from Syphilis Study at Tuskegee[306], the National Commission for the Protection of Human Subjects of Biomedical and Behavioral Research was formed which issued *Ethical Principles and Guidelines for the Protection of Human Subjects of Research*, namely the Belmont Report.[307] The Belmont Report established three principles relevant to research involving human subjects: (1) Respect for persons: this principle acknowledges individual autonomy and protects people with diminished autonomy;[308] (2) Beneficence: it requires that individuals shall be treated "in an ethical manner not only respecting their decisions and protecting them from harm, but also by making efforts to secure their well-being"[309]; (3) Justice: it discussed the balance between the benefits and burdens of research.[310] These ethical principles have guided the federal regulations to consider the legal protection for human subject rights in researches. Even though these ethical principles have provided good basis for the doctrine of informed consent, the legal system in the US seems still far from encompassing.

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[304] Laurie, *supra* note 200.

[305] NIH Office of Extramural Research, Protecting Human Research Participants.

[306] The Syphilis Study at Tuskegee refers to the long-term study of black males conducted by the United States Public Health Service in Tuskegee, Alabama, which was initiated in the 1930s and continued until 1972. The Study involved approximately 600 African-American men: about 400 with syphilis (cases) and about 200 without syphilis (controls). These men were recruited without informed consent, in fact, were led to believe that some of the procedures done in the interest of research were actually "special free treatment". See Protecting Human Research Participants.

[307] *Ibid.*

[308] The National Commission for the Protection of Human Subjects of Biomedical and Behavioral Research, The Belmont Report: Ethical Principles and Guidelines for the Protection of Human Subjects of Research, Department of Health, Education, and Welfare, 18 April 1979.

[309] *Ibid.*

[310] *Ibid.*

4.2.2.2 Legal Protection for Individual Autonomy

The individual autonomy is more stressed in regulation which focuses on the rights of human subjects in scientific research – the *Federal Policy for the Protection of Human Subjects* (the *Common Rule*). With the Belmont Report as foundational background, the *Common Rule* was published in 1991 and outlined the basic provisions for Institutional Review Boards, informed consent, and Assurances of Compliance, applying to human subject research conducted or supported by federal department or agency.[311]

(1) *Pre-2018 Requirements*

Under the *pre-2018 Requirements*, the legally effective informed consent is the essential precondition for conducting research involving human subjects.[312] In accordance with the definition of human subject, it means "a living individual about whom an investigator (whether professional or student) conducting research obtains (1) Data through intervention or interaction with the individual, or (2) Identifiable private information"[313]. As a result, there are some kinds of researches not pursuant to the *Common Rule*, such as those using publicly available information or nonidentified biospecimens.[314] In that case, using leftover samples might fall outside the *Requirements*, thus such researches do not need to obtain informed consent. However, the possibility could cause concern "given the ability to match an anonymous sample with the original donor through DNA analysis and further derive meaning from the DNA"[315]. Furthermore, the identifiable private information means "the identity of the subject is or may readily be ascertained by the investigator or associated with the information"[316]. Therefore, informed consent is not necessary if researchers use personal information from subjects whose identity cannot be readily ascertained, such as de-identified information.[317]

(2) *The Notice of Proposed Rulemaking*

The HHS and 15 other Federal Departments and Agencies have announced proposed revisions to the regulations for protection of human subjects in research.[318] A *Notice of Proposed Rulemaking* (NPRM) was published in 2015, which seeks comment on proposals to update the *Common Rule*.[319]

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[311] Federal Policy for the Protection of Human Subjects ('Common Rule'), Office for Human Research Protections, available at: https://www.hhs.gov/ohrp/regulations-and-policy/regulations/common-rule/index.html (last accessed: 22 September 2020).

[312] 45 C.F.R. §46.116.

[313] 45 C.F.R. §46.102(f).

[314] Mark E. Sobel and Jennifer C. Dreyfus, "Disruptive Influence on Research in Academic Pathology Departments: Proposed Changes to the Common Rule Governing Informed Consent for Research Use of Biospecimens and to Rules Governing Return of Research Results", *The American Journal of Pathology*, 187, 1 (2017).

[315] Marilyn Cech, *supra* note 291; John Bohannon, Genealogy Databases Enable Naming of Anonymous DNA Donors, *SCIENCE*, 339, 262 (2013).

[316] 45 C.F.R. § 46.102(f).

[317] Marilyn Cech, *supra* note 291.

[318] NPRM for Revisions to the Common Rule, Office for Human Research Protections, available at: https://www.hhs.gov/ohrp/regulations-and-policy/regulations/nprm-home/index.html (last accessed: 22 September 2020).

[319] *Ibid.*

One of the fundamental changes proposed in the NPRM is the expansion of the definition of human subject to cover research with non-identified biospecimens.[320] It proposed that all clinical trials, regardless of identifiability shall be subject to the *Common Rule*. If that is the case, researches involving non-identified biospecimens are covered by the *Common Rule*. This change is of great significance for genetic research, which could make up the flaw of HIPAA's narrow focus on informational risk to research subjects.[321] The *Havasupai Tribe v. Arizona Bd. Of Regents* case demonstrated that "participants have serious concerns about the use of their samples that are not limited to the release of identifiable information"[322]. The Havasupai tribe accused an Arizona State University of using their members' stored genetic samples to conduct further researches without consent.[323] "The Havasupai were concerned not just that their tribe may have been identifiable based on supposedly anonymized samples and data but that their samples had been used, without their consent, for research that ran counter to important cultural and religious tribal values."[324] The proposal of the NPRM might provide an overall protection from genetic sample to the information it contains, expanding and strengthening individuals' autonomy during the genetic research.

More importantly, the NPRM sought to change the informed consent form and process in order to "facilitate prospective subjects' decision about whether or not to participate in a research study, thereby enhancing autonomy"[325]. It suggested that essential information with sufficient detail relating to the specific research shall be provided to enhance the subjects' understanding so that "a reasonable person"[326] could make an informed decision with regard to whether to participate in the research. The proposal sufficiently considers the autonomy interests of subjects to decide whether or not to take part in the researches on the basis of adequate comprehension. On the other hand, to encourage the growth of genetic analyses and other similar researches involving biospecimens, the NPRM also included exemptions "to facilitate storage, maintenance, and secondary research use of biospecimens and identifiable private information"[327] by means of broad consent. Instead of research-specific requirement for informed consent, the NPRM permitted broad consent for "the storage or maintenance for secondary research of information and biospecimens that were originally collected for either research studies other than the proposed research or non-research purposes"[328]. The mechanism of broad consent is meaningful for genetic research considering the widespread use of biobanks and databases, and the common practice of sharing data

[320] Notice of Proposed Rulemaking, *Federal Register*, 80, 173, 8 September 2015, II.A.1.

[321] Megan Allyse, Katrina Karkazis, Henry T. Greely, Mildred K. Cho and David Magnus, "Is HIPAA Enough? Informational Risk, Institutional Review, and Autonomy in the Proposed Changes to the Common Rule", *IRB*, 34, 3 (2012).

[322] *Ibid.*

[323] Amy Harmon, Indian Tribe Wins Fight to Limit Research of Its DNA, *The New York Times*, 21 April 2010.

[324] Allyse *et al.*, *supra* note 321.

[325] Notice of Proposed Rulemaking, II.B.1.a.

[326] *Ibid.* II.B.1.d.

[327] *Ibid.* II.A.3.d.iv.

[328] *Ibid.* II.B.2.d.

among researchers.[329] Such flexible changes tried to reduce the procedural and administrative burden for researches, while recognizing the individual autonomy, for institutions and investigators would "give individuals the choice to either allow or disallow the use of their biospecimens and identifiable privacy information for secondary research"[330].

The NPRM has put forward proposals which would enhance the protection of genetic data in research. These proposals are selectively accepted by the final Revised *Common Rule*.

(3) The Revised *Common Rule*

The final revised *Common Rule* has adopted the proposal of broad consent, but made several changes. The final rule does not accept the NPRM's proposal to cover all biospecimens regardless of identifiability. Therefore, the suggestion of obtaining broad consent for secondary research using non-identified biospecimens will not be implemented.[331] Besides, broad consent is allowed "only for secondary research and no other types of research"[332]. Given that the NPRM covered all biospecimens used in researches, whether identifiable or not, the researchers could not avoid the application of the *Common Rule* through de-identification methods, but could only choose to obtain broad consent. However, under the final rule, researchers have opportunity to do secondary research with non-identifiable samples, as was the case under the *pre-2018 Requirements*.[333] In addition, the option of IRB waiver of informed consent is still applicable. Therefore, even though the route of broad consent gives subjects to say no to the secondary research, the final revised *Common Rule* provides other options for institutions to avoid the requirement of obtaining consent from subjects. Compared to the NPRM, the final rule eases the burden of institutions, but relatively undermines the autonomy of subjects.

The consent requirements scatter among several US rules. The mechanism of consent is more dynamic and flexible to adapt to the future need of researches, especially those involving large amounts of data and biospecimens, such as genetic researches which take advantage of databases or tissue repositories. Despite of public comments and proposed changes, the final *Common Rule* does not make substantial change with regard to the consent requirements. Just like the criticism in the NPRM, the consent provisions are more likely to mitigate the liability of institutions, rather than help subjects to better understand the research and enhance their autonomy.

Protection genetic privacy contributes to respect for individual autonomy, for it facilitates people to make choices without interference. On the other hand, individual autonomy is also the basis for privacy values. Nonetheless, the genetic privacy is not

[329] Marilyn Cech, *supra* note 291.
[330] *Ibid.*
[331] Feral Policy for the Protection of Human Subjects, Final Rule, *Federal Register*, 82, 12, 19 January 2017, XIV.D.4.
[332] *Ibid.*
[333] *Ibid.*

only involved with personal interest of autonomy. There are other interested parties who might be influenced by one's genetic information. However, there seems to be a common deficiency in the EU and the US regulations that they only emphasize the personal interests in genetic privacy, ignoring to consider the link between personal rights and others' rights. The family members also share certain interests in genetic privacy based on the inherited relationship.

4.3 Family Members Interests in Genetic Privacy

Genetic testing tells people information about their DNA, which is shared with other family member.[334] Sometimes a genetic testing result may have implications for blood relatives of the person who had testing.[335] This fact makes genetic privacy go beyond personal sphere. Imagine that the genetic testing result indicates one's genetic disorder risk, whether her family members have the right to be informed of the risk so that they can take preventive measures in advance? The professionals are encountered with the dilemma of the conflicts of the patients' genetic privacy and their family members' right to know, which correspond to the duty to confidentiality and the duty to warn.

4.3.1 Safeguarding Genetic Privacy: The Duty of Confidentiality

If we regard privacy as a "right", then there are corresponding obligations that should be undertaken by the counterparty. In terms of genetic privacy, professionals shall keep the duty of confidentiality and not disclose the genetic information without consent.

4.3.1.1 The Foundation of Confidentiality

Hippocratic Oath is the origin of medical ethics to keep confidentiality, which has become the rules that regulate medical practices: "What I may see or hear in the course of the treatment in regard to the life of men, which on no account one must spread abroad, I will keep to myself holding such things shameful to be spoken about."[336] Confidentiality is firstly rooted in the recognition and protection of personal autonomy, by which the individuals could control access to the genetic information about themselves.[337] The duty of confidentiality exists in specific relationships. In professional and patient relationship, the duty of confidentiality is kept by professionals in order to assure that the information they have obtained from patients would not be disclosed to third parties without consent. Now that individuals entitle professionals certain privilege to assess to their genetic information, expectations for professionals to keep confidential in order to protect personal privacy are well established.[338] Confidentiality is seen as the only one tool used to achieve and maintain privacy.[339]

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[334] Genetic Testing FAQ, What can I learn?, National Human Genome Research Institute, available at: https://www.genome.gov/FAQ/Genetic-Testing (last accessed: 22 September 2020).

[335] *Ibid.*

[336] Sani Ibrahim Salihu, Yuhanif Yusof and Rohizan Halim, "Disclosing HIV Status: Confidentiality, Right to Privacy and Public Interest", *Social Sciences & Humanities*, 26, 2 (2018).

[337] Charles Ngwena and Ruth Chadwick, "Genetic Diagnostic Information and the Duty of Confidentiality: Ethics and Law", *Medical Law International*, 1, 1 (1993).

[338] Nuffield Council on Bioethics, *supra* note 279.

[339] *Ibid.*

4.3.1.2 Confidentiality and Respect to Autonomy

Medical confidentiality is another implication of respecting people's autonomy.[340] The duty of confidentiality contributes to the protection of privacy of individuals, consolidating the space in which patients can exercise their autonomy. The absence of promise of confidentiality would discourage people to take part in genetic testing which is necessary for treatment and might reveal some sensitive information, thus might decrease the likelihood of understanding the genetic information by themselves. The fear of information leakage in fact restricts the patients' choices with regard to undergoing genetic consult or testing. With the assurance of confidentiality, individuals could make choices more autonomously in an informed condition.

The principle of autonomy also provides justification for protecting personal health information through the concept of confidentiality. The autonomous control over information by individuals requires that others shall not step into their private sphere. At the same time, the health care professionals, despite of possessing patients' information, is still in lack of authority of disclosure without consent. Otherwise, the breach of confidentiality equals with furtherance of the action of invasion and undermines personal autonomy.

4.3.2 Family Members' Right to Know

"If confidentiality is supported on the basis of autonomy then what has to be taken into account is the fact that respecting the autonomy of one person may have implications for the autonomy of others."[341] With the genetic information of the individuals who have gone through the genetic testing, their relatives could make decisions about receiving treatment or adjusting life styles, even the non-blood relatives, such as their spouses, are able to make reproductive decisions based on the testing result. However, the absence of such knowledge would deprive them of the opportunities to make autonomous choices.

Even though the family members' right to know has not been recognized by law, it cannot be denied that moral justifications could be provided for the relatives assess to genetic information. "The importance of autonomy, combined with the prospective health benefits, give relatives some ethical claim to genetic information."[342]

4.3.2.1 Harm Prevention

To increase health or prevent harm is the most direct justification for disclosing patients' genetic information to the at-risk family members. With the previous testing result of patients, the genetic risk of other relatives could be identified earlier and they would consciously seek individual testing.[343] "The result of this increased testing could range from cure, through prevention and treatment, to awareness and management of

[340] Raanan Gillon, "Medical Ethics: Four Principles Plus Attention to Scope", *British Medical Journal*, 309, 6948 (1994).
[341] Ngwena and Chadwick, *supra* note 337.
[342] Ana Mayne, "Genetics and the Family: A Right to Know", *Edinburgh Student Law Review*, 3, 4 (2019).
[343] *Ibid.*

disease."[344] Therefore, entitling relatives with the right to know enhances the timeliness of coping with genetic risk and promote health in family context.

However, the reason of harm prevention is conditional. Only when the benefits of disclosing genetic information to at-risk relatives outweigh the patients' genetic privacy interests, can the disclosure be justified.[345] As Roy Gilbar concluded, the limited situation occurs "where the doctor (a) can identify the relatives; (b) is aware that they are at serious risk; and (c) knows that by disclosing the information he/she could prevent serious physical harm to them".[346]

4.3.2.2 Relational Autonomy

Relational autonomy, a conception deriving from feminism, places individuals in the social context, upholding that others' interests shall be taken into consideration in the decision-making process.[347] Interdependence, instead of independence, is the core notion in the theory of relational autonomy.[348] The proponents of relational autonomy make their argument based on the point that "social surroundings and relationships enable us to flourish and develop a robust capacity for self-determination and identity formation."[349] Social relationship is bound with personal capacity to implement autonomy.[350] Therefore, individuals shall consider "the interests of those who will be affected by his or her choice"[351].

The notion of relational autonomy is also pertinent to clinical genetics because in genetic context individuals are intertwined with others, especially with the family members and their interdependent relationship is particularly true. Sometimes patients might wish to keep their genetic information confidential or does not explicitly allow clinicians to disclose the information. Then clinicians would fall into conflict of duties. On one hand, the clinicians shall keep the duty to confidentiality to show respect for patients' genetic privacy; on the other hand, they undertake the duty to warn the family member of the risk in order to prevent potential harm. In that case, the conception of relational autonomy provides a justifiable solution for balancing the genetic privacy and health benefits. It adopts a two-tiered approach to deal with the privacy problems: Genetic information about certain mutation that causes disease is conceptualized as

[344] *Ibid.*

[345] Inside Information: Balancing Interests in the Use of Personal Genetic Data, *Human Genetics Commission*, 2002.

[346] Roy Gilbar, "Medical Confidentiality within the Family: The Doctor's Duty Reconsidered", *International Journal of Law, Policy and the Family*, 18, 2 (2004).

[347] Jennifer Nedelsky, "Reconceiving Autonomy: Sources, Thoughts and Possibilities", *Yale Journal of Law & Feminism*, 7, 11 (1989).

[348] Edward S. Dove, Susan E. Kelly, Federica Lucivero, Mavis Machirori, Sandi Dheensa and Barbara Prainsack, "Beyond Individualism: Is There a Place for Relational Autonomy in Clinical Practice and Research?", *Clinical Ethics*, 12, 3 (2017).

[349] *Ibid.*

[350] Roy Gilbar, "Patient Autonomy and Relatives' Right to Know Genetic Information", *Medicine and Law*, 26, 4 (2007).

[351] *Ibid.*

"owned" by both the patient and family members.[352] "It is thus considered as confidential at the familial level."[353] Specific genetic diagnosis and disease status are individual-level information thus regarded as confidential to individual.[354] This approach actually subdivides the genetic information and sets confidentiality level, extracting those only "belonging" to patients themselves and providing high level privacy protection.

4.4 The Legal Respond to Family Members' Interests in the EU and the US

It seems that the EU and the US emphasize highly on the autonomous control of the genetic privacy by individuals. Yet, neither of them provides an explicit solution in terms of the conflicts of the interests on genetic privacy between individuals and their family members. They are cautious about the problem whether family members could claim a right on genetic privacy.

4.4.1 The EU Position to the Right to Know and Multiple Data Subjects

Some scholars have criticized the restrictive definition of personal data in the GDPR, in the sense that it only admits the data protection right in individual perspective, neglecting the circumstances where several subjects claim rights to certain data.[355] The definition of personal data implies the information is related to an independent identified or identifiable person, namely the data subject, without the consideration for the relationship with others.

The Working Party recognized the characteristic of genetic data:

> "While genetic information is unique and distinguishes an individual from other individuals, it may also at the same time reveal information about and have implications for that individual's blood relatives (biological family) including those in succeeding and preceding generations, Furthermore, genetic data can characterize a group of persons (e.g. ethnic communities);"[356]

Considering this characteristic, the Working Party notably raised the concern of right to know the genetic information by family members:

> "One of the fundamental features of genetic data consist both in its marking out an individual from others and the fact that this data – and more precisely: the characteristics to which it refers – is structurally shared by all the members of the same biological group – whereas

[352] Dove *et al*., *supra* note 348.
[353] *Ibid.*
[354] *Ibid.*
[355] J. Lyn Entrikin, "Family Secrets and Relational Privacy: Protecting Not-So-Personal, Sensitive Information from Public Disclosure", *University of Miami Law Review*, 74, 3 (2020); Ugo Pagallo, "The Group, the Private and the Individual: A New Level of Data Protection?", *in* Linnet Taylor, Luciano Floridi, Bart van der Sloot (eds.) *Group Privacy: New Challenges of Data Technologies* (Cham: Springer, 2017).
[356] Article 29 Working Party, *supra* note 71.

> other mechanisms by which personal data are shared depend on the data subjects' will, on social custom, or on legal rules."[357]

Therefore, it supported that "to the extent that genetic data has a family dimension, it can be argued that it is 'shared' information, with family members having a right to information that may have implications for their own health and future life"[358].

Yet, the Working Party did not expressly answer whether family members have the right to know as *data subject*[359]. It only approved the interest of family members ethnically but "seemed decidedly unsure about how any 'right' to access 'shared' genetic data ought to be reflected within law"[360]. If it is too radical to directly regard family members as equivalent data subjects of genetic data, the Working Party provided a conservative alternative that "that other family members would have a right of information of a different character, based on the fact that their personal interests may be directly affected"[361].

Unfortunately, with regard to the requirement to find a balance between a data subject's right to genetic privacy and the family members' interests on genetic data, the GDPR does not provide explicit solutions. A series of questions raised by this concern, for example, "is it possible for data to 'relate' to more than one identifiable person in a particular context?"[362], could neither find answers in the GDPR.

4.4.2 The US Position to the Duty to Warn

The case *Tarasoff v. Regents of University of California* held that when the patient presents a serious danger of violence to another, a doctor incurs an obligation to warn the intended victim.[363] In that case, Prosenjit Poddar killed Tatiana Tarasoff.[364] The plaintiffs, Tatiana's parents, alleged that before the murder, Poddar once told his intention to kill Tatiana to Dr. Lawrence Moore, a psychologist employed by the University of California. The campus police detained Poddar on Moore's request for a short time and released him when he appeared rational.[365] Dr. Harvey Powelson, Moore's superior, then directed that no further action be taken to detain Poddar. "No one warned plaintiffs of Tatiana's peril"[366]. The justice explained that the fact Tatianna is not the patient of the therapists cannot release them from the liability for failing to warn the endangered Tatiana.[367] Following this case, the clinicians' duty to warn has

[357] *Ibid.*
[358] *Ibid.*
[359] Mark Taylor, *Genetic Data and the Law: A Critical Perspective on Privacy Protection* (Cambridge: Cambridge University Press, 2012.)
[360] *Ibid.*
[361] Article 29 Data Protection Working Party, *supra* note 88.
[362] Taylor, *supra* note 359.
[363] *Tarasoff v. Regents of University of California*, 551 P.2d 334, 17 Cal.3d 425, 1 July 1976.
[364] *Ibid.*
[365] *Ibid.*
[366] *Ibid.*
[367] *Ibid.*

been established in the US.[368] Similarly, in the medical practice, the clinicians would know exactly the existence of the at-risk family members who share the genetic information of their patients.[369] But the essential difference is that the clinicians does not cause the occurrence of the genetic risk. Whether clinicians inform the relevant family members only impacts when they *find out* the genetic disorder.[370] Despite of the distinction of situations, the case has made the duty of clinicians go beyond the doctor-patient relationship and extend to the relevant parties who are affected by the diagnosis.

Then in the case *Molloy v. Meier*, Kimberly Molloy and her husband, Glenn Molloy, brought a medical malpractice against three doctors, claiming they were negligent in failing to diagnose a genetic disorder in Molloy's daughter and their negligence caused Molloy to conceive another child with the same genetic disorder.[371] Molloy alleged that the doctors and their employers were negligent in the care and treatment rendered to her daughter by failing to order Fragile X testing on her daughter, failing to properly read those lab tests that were performed, mistakenly reporting that her daughter had been tested for Fragile X, and failing to provide counseling to Molloy regarding the risk of passing an inheritable genetic abnormality to future children.[372] The court held recognized that physicians owe a duty to a third party who is not a patient of the physicians where patient threatens foreseeable harm to the third party and physicians have the ability to control the risk of harm.

In the US, some states have legislated to address the disclosure of information to family members. For example, in *Safer v. Estate of Pack* case, the New Jersey appellate court declared that "physician has duty to warn those known to be at risk of avoidable harm from genetically transmissible condition"[373] and "physician's duty extends to members of immediate family of patient who may be adversely affected by breach of duty". However, "in 2001, the New Jersey Legislature effectively overturned the decision in *Safer* by enacting a broad genetic privacy statute".[374] It forbids disclosure of genetic information which permits identification of an individual.[375] Pursuant to the New Jersey law, it is illegal to disclose genetic information to family members without the consent of patients.

On the federal level, the HIPAA states clearly various situations in which disclosure of PHI is permitted or prohibited. Under HIPAA, a covered entity shall not disclose the PHI beyond treatment, payment or health care operations without the individual's

[368] Melodie Nothling Slabbert, The Genetic Ties that Bind Us and the Duty to Disclose Genetic Risks to Blood Relatives, *De Jure*, 40, 1 (2007).
[369] *Ibid.*
[370] *Ibid.*
[371] *Molloy v. Meier*, 679 N.W.2d 711, Supreme Court of Minnesota, 20 May 2004.
[372] *Ibid.*
[373] *Safer v. Estate of Pack*, 667 A.2d 1188, N.J. Super. Ct. App. Div., 11 July 1996.
[374] Mark A. Rothstein, "Reconsidering the Duty to Warn Genetically at-risk Relatives", *Genetics in Medicine*, 20, 3 (2018); New Jersey Code § 10:5-47 (2001).
[375] *Ibid.*

written authorization or the permission or requirement of the Privacy Rule.[376] The *Privacy Rule* also contains 12 priority purposes exceptions for public interest and benefit which do not require authorization.[377] Among these exceptions, only one can be related to the warning to endangered family members, that is "use and disclosures to avert a serious threat to health or safety"[378]. The use and disclosure shall be judged by the covered entity to be necessary and reasonably able to lessen the threaten to the health or safety.[379] In other words, whether to warn is at the discretion of physicians. The Office for Civil Rights of the Department of HHS interprets that "a health care provider may share genetic information about an individual with providers treating family members of the individual who are seeking to identify their own genetic health risks, provided the individual has not requested and the health care provider has not agreed to a restriction on such disclosure"[380]. The HIPAA *Privacy Rule* allows physicians to disclose PHI to another physician of the family members and the latter can inform the relatives of possible risk,[381] *i.e.*, it is permitted that the PHI is shared among physicians and the duty to warn can be undertaken indirectly with assistance of another physician. Therefore, the HIPAA actually recognizes relatives' access to genetic information, but it does not straightly address the relationship between physicians and family members rather adopts a roundabout approach to deal with the conflict.

The EU and the US regulations takes a prudent attitude toward the family members' interests. They appear conservative when faced with conflicts of interests between individuals and their family members, not expressly entitling family members with the right to know. Generally speaking, the regulations still presents a subject-centered form in terms of genetic privacy protection, addressing less the relationship between subjects and others. For the individualistic notion of both the EU and the US law, it would be a good start point for perfection to put subjects in a relational context. It is too idealistic to treat genetic privacy in an isolated environment. The relational notion of privacy might be more practical to balance interests of relevant parties with regard to genetic

[376] 45 C.F.R. § 164.508.
[377] 45 C.F.R. § 164.512.
[378] 45 C.F.R. § 164.512(j).
[379] 45 C.F.R. § 164.512(j)(1)(i)(A) and (B).
[380] Modifications to the HIPAA Privacy, Security, Enforcement, and Breach Notification Rules under the Health Information Technology for Economic and Clinical Health Act and the Genetic Information Nondiscrimination Act; Other Modifications to the HIPAA Rules; Final Rule, US Department of Health and Human Services, Federal Register 5566-5702, 5668 (2013).
[381] 45 C.F.R. § 164.502(a)(1)(ii); FAQ: Under the HIPAA Privacy Rule, may a health care provider disclose protected health information about an individual to another provider, when such information is requested for the treatment of a family member of the individual?, Office of Civil Rights, US Department of Health and Human Services, available at: https://www.hhs.gov/hipaa/for-professionals/faq/512/under-hipaa-may-a-health-care-provider-disclose-information-requested-for-treatment/index.html (last accessed: 22 September 2020).

privacy.

Chapter 5 Conclusion

The legal frameworks of genetic privacy law in the US and the EU are quite different. The US has gradually separated genetic information from the other medical information and regulated them separately. The main fields covered by genetic privacy law are insurance and employment. By contrast, the EU puts all kinds of data into the general system, and the general regulations contain special rules for sensitive data. The debate around genetic exceptionalism has lasted for years. The genetic-specific law indeed has various weakness, but if genetic information is just simply included in medical information, its sensitivities might be ignored. In other words, there is no perfect answer for the controversy about genetic exceptionalism up till now. Therefore, the author advises to suspend the dispute on genetic exceptionalism and focus on the discussion of how to establish a comprehensive legal system for genetic privacy. No matter through genetic-specific regulation or through omnibus privacy law, the genetic privacy shall be protected comprehensively. The EU GDPR applies to all kinds of personal data. It is not special for genetic privacy, but it does have great implication on genetic research. The research exemption could break the restriction of processing sensitive data. At the same time, it also derogates personal rights of data subjects, but the corresponding safeguards are not clear. Considering the possible derogations of rights and principles, the research exemption shall be limited to reasonable extent and the safeguards shall be in place.

The specific differences contained in the legal framework are the attribution of genetic privacy and the notions related to genetic privacy adopted by the US and the EU legislation. The GDPR keeps a foothold on respecting individual fundamental rights, which regards genetic privacy as a fundamental right. As a result, genetic privacy, as a branch of personal privacy, is endowed with high hierarchy among various rights. The privacy approach is the first step of genetic privacy protection, which must be assisted by other approaches such as nondiscrimination to completely protect genetic privacy. On the contrary, the US protects genetic information from the nondiscrimination prospective. The limitations on access to genetic information is a means to prevent genetic discrimination. Instead of entitling individuals with positive rights, the nondiscrimination law intends to impose obligations on the insurers and employers. Besides, the protection for genetic information provided by nondiscrimination law is not fully covered. It only works in circumstances where discrimination is most likely to happen.

By comparing the legislative language, it is found that the EU and the US have accepted different notions, which indicate different scope of privacy protection. The GDPR, even though provides a broad definition for personal data, narrows the definition of genetic data, mainly focusing on the data originating from genetic testing. In the processing of genetic data, controller and processor play different roles. As thus, the division of responsibility is clear and the adaptability of the regulation is promoted. Corresponding to the restrictive scope of genetic data, the exclusion of anonymous data shall meet the rigorous standard. All reasonable accounts are required to be taken into consideration

in order to evaluate the anonymization. The EU expresses cautious attitude toward inclusion of genetic data. On the other hand, the US regulations usually determine the covered range through enumeration. Even though the scope includes the family history which is excluded by the GDPR, it is limited in the sense that manifested conditions are not covered. It is worthy to note that GINA works from the information collection stage, thus the access to, use and disclosure of genetic information is strictly regulated. However, due to the lenient standard for de-identification of genetic information, it is not difficult to escape from the genetic privacy law in the US. The EU and the US show different standards with regard to the scope of genetic privacy regulations, both of which do not provide the complete protection. However, they could be complementary to each other.

Despite of the differences in various aspects, both of the EU and the US highly stress the respect for individual autonomy. The GDPR sets different rights for data subjects so that they can better control over their genetic data. The US laws also protect the autonomy of information subjects through consent mechanism during scientific research. However, considering the special link of genetic privacy with family members, the interests of family members cannot be ignored. When the duty of confidentiality, which guarantees the privacy rights, conflicts with family members' right to know, the law must draw a balance between them. Nevertheless, the EU and the US focus more on the genetic privacy from personal perspective. There are few solutions for conflicts between different rights. It seems that the subject-centered regulations consider genetic privacy in an isolated condition, in lack of the adjustment of different relationships. Therefore, the balance between personal genetic privacy and the family members' interests could be a reform target of the legislation.

The EU and the US show many differences in terms of genetic privacy legislation. However, they also provide experiences worthy of reference for each other. The comprehensive legislative framework of the GDPR offers a valuable example for the US to perfect its legal system. The US regulations are more practical in specific circumstances such as insurances and employment. The two legal frameworks reflect different logics and ideas, but both of them attach great importance to respect for personal autonomy. They stress the main role of the subjects of genetic privacy, but one common deficiency is that they ignore the position of family members in this context. Therefore, when continuously improving respective legal framework for genetic privacy, the EU and the US shall take the balance of interests into consideration.

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