Legal Issues Regarding Genetic Information in the US Genetic Information Nondiscrimination Act (GINA) and Need for a New Legislation for Prohibiting Genetic Discrimination in Japan

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ABSTRACT

In 2023, Japan passed The Genome Medicine Promotion Act to achieve its goal of contributing to the preservation of the health of the people by enabling the provision of optimal medical care through the development of genomic medicine in Japan (Houritsu, No. 57, 2023). However, there is no explicit prohibition of genetic discrimination in Japan yet. Even though the Act on the Protection of Personal Information in Japan includes DNA for individuals but not their families. For this reason, genetic information is not fully kept private. In addition, there are no clear guidelines for genetic information in non-medical fields such as insurance and employment, nor are there laws prohibiting discrimination based on genetic information in these fields. On the other hand, the United States Congress passed the Genetic Information Nondiscrimination Act (GINA) in 2008. This act prohibits genetic discrimination in health insurance and employment. However, since the enactment, there have been legal issues in the judiciary regarding the interpretation of genetic information. In addition, a survey conducted in 2020 found that even 12 years after GINA was implemented, it was still not well known among US citizens. The results point out the interrelation between concerns about genetic discrimination and insufficient awareness of GINA. Furthermore, with the development of research on genetics in the post-genome era, there is a call for a revision of existing laws emphasizing genes. In this paper, we address the limitations of the legal protections of GINA and issues with the interpretation of genetic information in the US judicial system, the issue of the recognition of GINA among US citizens, and necessitating new legal developments of epigenetic information in line with the development of human genome research to consider the Japanese model act to prevent genetic discrimination.

I. Introduction

We have been engaging in a research project to create a model act for a law prohibiting genetic discrimination in Japan. In order to provide the task, we are researching laws against genetic discrimination in the United States, as well as new legal developments relating to genetic information. As far as we understand the content of the US Genetic Nondiscrimination Act (GINA), and the US state insurance, employment, and privacy laws, it appears that an act of genetic discrimination is strictly regulated in each law. However, the dispute over the limitation of GINA protection, and the battle of interpretation of genetic information in the US judiciary has pointed out issues on the practicality of genetics laws in America. Even with legal protection by GINA, it is difficult to prove genetic discrimination in employment in a court of law. Also, a survey conducted 12 years after the establishment of GINA shows the insufficient awareness by US citizens of GINA and their concerns about genetic discrimination interference. This issue also highlights the problem of not being aware of existing laws regarding genetics among Japanese citizens.

In June 2023, Japan passed the Genome Medical Care Promotion Act (注. https://laws.e-gov.go.jp/law/505AC1000000057). There were not many active public debates on the law, and public awareness among Japanese citizens was not high. However, if this law will contribute to developing genetic medicine for Japan to maintain its citizens' health by utilizing the genomic information of its citizens, it proactively needs to be discussed by not only professionals but the entire nation. One of the factors behind this problem is that, unlike in Europe and the United States, there is not much discussion about bioethics among the general public in Japan. For this reason, the development of laws related to genetic discrimination in Japan is more than 30 years behind that of America that have developed such laws (Maruyama, et al., p.155,2024).

Unlike the development of laws that prohibit discrimination based on gender, age, race, etc., there is no historical record of activism against genetic discrimination. However, the background to the enactment of GINA was the Human Genome Project ('HGP'). Due to the project and the discovery of genetic risk in the human genome through the project, it created a public fear of genetic discrimination. As a result, it was essential to enact this law without strong evidence of existing genetic discrimination (Suter, p. 497, 2019). In this respect, Japan also needs to consider enacting genetic laws to protect against genetic discrimination and enforce privacy in order to its development of genetic medicine for the future. Nevertheless, we are entering a post-genome era, where research and technologies are being developed after the human genome has been decoded. Recently, there have been concerns about health insurance services and asylum policies that utilize information about the ability to infer personal information based on epigenetic information. (Davidson, p. 164, 2023). Because epigenetic information has an impact due to environmental factors without genetic mutation. For this reason, the information reveals components of individual lifestyle, it may violate an individual's privacy and leave them open to discrimination.

II. The limited scope of GINA's legal protection

GINA is a two-part law that prohibits genetic discrimination. The first part of the law prohibits genetic discrimination in health insurance, and the second part prohibits genetic discrimination in employment. The scope of GINA's protection is limited to these two areas. For example, the first part prohibits insurance companies from accessing genetic information. Therefore, it cannot request, or require genetic information or genetic testing. The second part prohibits the use of genetic information in employment, such as hiring, discharging, determining terms, conditions, etc. However, even though the law prohibits genetic discrimination in employment, it is difficult to prove genetic discrimination in employment in a court of law. Since the scope of protection differs from state to state, such as some states covering life insurance and disability, it cannot be said that there is consistent protection of genetic privacy and discrimination. Yet, these protections do not apply to military personnel, nor is it applicable to companies with 15 or more employees. Moreover, GINA does not prohibit the use, acquisition, or disclosure of medical information that is not genetic information related to a disease or a disorder that has displayed external symptoms. Once a genetic risk develops into a manifested condition, GINA no longer applies (Suter, pp. 503-504, 2019). For these reasons, it points out GINA's goal of full protecting against genetic discrimination has still not been achieved.

III. Legal issues on interpretation of Genetic Information

When GINA was being drafted, there was debate about the definition of genetic information. Suter (2019) mentions that "...attempting to distinguish genetic information from other medical information is conceptually fraught because there are no bright lines between what is genetic or non-genetic medical information; the real distinction is the degree to which genetics or environment play a role." (p. 499). Therefore, the following broad definition of genetic information was ultimately decided upon:

> "(6) GENETIC INFORMATION. — "(A) IN GENERAL. —The term 'genetic information' means, with respect to any individual, information about— "(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. "(B) INCLUSION OF GENETIC SERVICES AND PARTICIPATION IN GENETIC RESEARCH. —Such term includes, with respect to any individual, 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 (United States Congress, 2008)."

More than 10 years after the enactment of GINA, employment-related genetic discrimination cases had grown in number. However, these cases have also brought to light the problem of interpreting genetic information in the judicial system. One approach to genetic information is a narrow interpretation that mainly follows legal terminology. For example, in the case of Poore v. Peterbilt Bristol, L.L.C, Poore claimed that the information Peterbilt Bristol, L.L.C obtained regarding his wife's multiple sclerosis diagnosis was a violation of GINA, which is a violation of the acquisition of genetic information. However, the court did not accept the definition of genetic information claimed by the plaintiff. The court stated that the main purpose of GINA is to prohibit an employer from making predictive assessments of an individual's or family member's predisposition to a genetic disease or disorder based on the occurrence of a genetic disease or disorder. For the reasons stated above, the court dismissed the claim, stating that the genetic information claimed by Poore did not predict the possibility of manifesting from the same disease as his wife, and did not fall under the category of information that takes into account the plaintiff's health condition. Suter (2019) points out that "the Poore court set the groundwork for a two-tiered interpretative approach that other courts soon followed: ie a determination of (1) whether a manifested disease or disorder exists in a family member and (2) whether information about a family member's disease or disorder is 'taken into' account in determining whether the employee has a propensity for disease" (p. 508). GINA includes family members who are related by marriage or adoption. However, in this case, the court did not discuss this point and issued its ruling.

The other approach is a broad interpretation including understanding the fundamental purpose of GINA. For example, in the case of Jackson v Regal Beloit America, the doctor who conducted the medical examination for employment purposes was asked to provide medical information that included genetic information under the guise of family history. The court found this to be a violation of GINA. However, unlike the Poore case, it did not indicate that an investigation was required to assess whether the family history was taken into account regarding the employee's propensity for illness (Sutter, p. 513, 2019). In another case, Punt v. Kelly Service, the court did not use a two-tiered interpretative approach and recognized a family history of breast cancer as genetic information. However, the plaintiff was unable to prove that was the reason for dismissal, and the claim was dismissed. In any case, the court decisions that either accept or reject genetic information that the plaintiff claims, even if genetic information is accepted by the court, proving employment discrimination had been another difficult problem. Yet defining and interpreting genetic information may become legal issues when Japan enacts a similar law.

IV. Insufficient awareness of GINA among US citizens

Twelve years after the enactment of GINA, in April 2020, a survey was conducted on the awareness of GINA and concerns about genetic discrimination among the general public in the United States. Respondents were recruited through Qualtrics Research Services, and the survey was open to US residents aged 18 and over. To ensure that the characteristics of the respondents were consistent with the overall population, restrictions were placed on gender, age, race, ethnicity, education level, and total household income. The questions were developed based on a review of GINA-related literature and research studies, and new questions were also added by the research team. To calculate demographic data on knowledge of GINA, concerns about genetic discrimination, and its impact, questions on political ideology, religiosity, and health status were added to the questionnaire.

Out of the 586 respondents to the survey, 96 did not complete the survey, and 69 were invalidated due to low-quality responses. The final number of respondents was 421, giving a response rate of 71.8%. Secondly, as a result of the survey on GINA awareness, of those who said they had a high level of knowledge about GINA, only 7.4% answered correctly about the scope of GINA protection. In addition, 54.4% of respondents incorrectly answered that GINA also covers car insurance and property insurance. Also, many respondents reported that they were more familiar with other healthcare-related laws such as ACA (Affordable Care Act) and HIPAA (Health Insurance Portability and Accountability Act) than with GINA. Based on this survey results, the research team pointed out that the impact of media coverage related to these laws compared to GINA may be influenced by a lack of awareness of GINA (Lenartz, et al., p. 2331, 2021). The survey results showed that there was a low level of awareness of GINA and that there was a widespread misconception about the scope of its protection. Surprisingly, among those who did not correctly answer the question about the scope of GINA's protection, there were people who claimed to have Huntington's disease.

As a result of their analysis of concerns about genetic discrimination, the research team concluded that the respondents' reported concerns about genetic discrimination may be based on a lack of knowledge or misunderstanding of GINA protections, given that GINA was less well known than expected (Lenartz, et al., p. 2332, 2021). In addition, 60% of respondents said they were likely to refuse genetic testing due to concerns about how the results would be used to make decisions about employment and insurance. In response to these survey results, the research team concluded that this high level of concern would increase even further if people were aware that GINA protections are limited to health insurance and employment, and that this would increase the likelihood of refusing genetic testing. In the end, based on the above survey and analysis results, the research team concluded that to resolve the lack of awareness of GINA, it is necessary to conduct effective dissemination activities, research and surveys on policies, and expand the scope of protection of GINA. In the future, when enacting laws similar to GINA in Japan, we must consider the importance of measures to promote public awareness of the law and a correct understanding of it, as well as public debate influenced by the media.

V. Need for New Legislation on Genetic Information

Epigenetics is a mechanism by which cells control the activity of genes without changing the base sequence of DNA, and it is affected by both genetic and environmental factors. This information is used not only to predict an individual's disease but also to estimate their age, life expectancy, living environment, and diet. Because this information also reflects factors in an individual's lifestyle, there are serious concerns about the ability to estimate personal information, and some scientists have called it "life-intrusive information" (Davidson, p. 164, 2024).

In addition, there are social and ethical issues regarding the use of epigenetic information for non-medical purposes. In the US, for example, FOXO Technologies is using a test that measures the level of DNA methylation for which it has obtained a patent to predict biological age in life insurance underwriting examinations. This test is being used to predict mortality and aging results such as cancer, physical function, and Alzheimer' s disease from the saliva samples of insurance policyholders. In addition, the German immigration office has commissioned Zymo Research in the US to conduct tests to estimate the age of young asylum seekers based on DNA methylation. However, due to issues with the credibility of the data related to DNA methylation mentioned above, and the lack of legal development related to epigenetics, there are concerns about new discrimination and privacy violations. For this reason, it is considered urgent to revise existing genetic discrimination and privacy laws.

Nevertheless, in the midst of inadequate legislation regarding epigenetics, there are interdisciplinary experts who point out the legal, ethical, and social issues of epigenetic research with an eye to military use: "For example, research is being conducted to identify epigenetic signatures associated with post-traumatic stress disorder (PTSD) in war-zone exposed veterans and active-duty soldiers. In one pilot study, a DNA methylation signature of PTSD (a high-severity biotype termed G2) was identified to characterize the biological and clinical heterogeneity of PTSD, along with the development of an improved panel of PTSD diagnostic markers in risk assessment for soldiers, accompanied by the inclusion of a psychotherapy follow-up for a subset of individuals.” (Dalpé, et al., p. 4, 2024). As we mentioned earlier, military personnel do not fall under the protection of GINA. In other pilot tests, the researchers have used data on DNA methylation to predict cognitive decline with age and to predict the brain health of soldiers.

Since GINA, US state laws, and international declarations only protect limited information such as genetic sequences, the results of genetic testing, chromosomes, and single-gene disorders, an approach that focuses on genetics excludes epigenetic information, so legal amendments are needed to protect individuals’ epigenetic information (Dalpé, et al., p. 175, 2024). The law states that genetic data, as defined by the California Genetic Information Privacy Act (GIPA), is not limited to the following:

> any data . . . that results from the analysis of a biological sample from a consumer, or from another element enabling equivalent information to be obtained, and concerns genetic material. Genetic material includes, but is not limited to, deoxyribonucleic acids (DNA), ribonucleic acids (RNA), genes, chromosomes, alleles, genomes, alterations or modifications to DNA or RNA, single nucleotide polymorphisms (SNPs), uninterpreted data that results from the analysis of the biological sample, and any information extrapolated, derived, or inferred therefrom. (Cal. Civ. Code § 56.18(b)(7)(A).

However, as there is no explicit definition of epigenetics, it is difficult to consider it as a supplementary law. In addition, the availability of direct-to-consumer epigenetic testing is growing, there are concerns about the privacy protection of the data. Overall, “this post-genomic era disrupts our existing conceptions of biological causality, responsibility, identity, and justice, necessitating a reassessment of the existing laws and principles that guide how such information is governed (Dalpé et al., p. 189, 2024). In terms of the development of laws related to genomic information in Japan, the development of laws related to epigenetics will be a future issue.

VI. Conclusion

As mentioned above, there is a need to consider a model for a legal system in Japan that addresses issues of discrimination and privacy related to genetic information, considering such factors as legal issues related to genetic information, public awareness of GINA, and the need to develop a legal system in a post-genome era. The first issue to consider is the definition of genetic information. How will the legislative and judicial branches understand the problem of distinguishing between genetic information and non-genetic information, and how will they define the scope of legal protection? In addition, it is necessary to discuss whether the definition of family history contained in genetic information, which is also an issue in the GINA case, should be limited to blood relations or whether it should include dependents. In any case, these discussions should be held not only by experts but by the general public as well.

The definition of genetic discrimination in insurance and employment also needs to be fully considered. In this case, since Japan's employment system and social security system differ from those of the United States, it is important to discuss the definition of genetic discrimination in light of the Japanese market and existing laws. For example, because insurance companies cannot request genetic information due to the protection of genetic information in GINA, it is possible for insurance subscribers to buy the insurance plan based on the genetic information they obtained. This is called the problem of adverse selection (Maruyama, et al., p. 164, 2024). In response to this problem, for example, in Europe, such as in the UK and Germany, life insurance policies have a set amount and regulations for specific genetic diseases. In any case, research into the human genome is constantly progressing. Given the legal and social issues in the United States after the enactment of GINA, if a similar law is enacted in Japan in the future, there will be a need for the whole nation to consider fully the purpose and functionality of the law even after it is enacted. In the end, as Japanese genomic medicine develops, we do hope that the public will increasingly discuss the legal protection of genetic information as the ultimate privacy issue.

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The authors declare no conflicts of interest in this article.

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