Robert F. Rich

Robert F. Rich is a Professor of Law, Political Science, Medical Humanities and Social Sciences, Community Health, and Health Policy and Administration at the University of Illinois at Urbana-Champaign. He is currently a "Faculty Fellow" in the Office of Vice Chancellor for Public Engagement and Institutional Relations and Director of the Office of Public Management within the Institute of Government and Public Affairs (IGPA). He was the Director of IGPA from 1986 to 1997. In the 1993-94 academic year, he was on sabbatical leave in Washington, D.C. at the Johns Hopkins University Center for the Study of American Government; he was also a "Special Guest" at the Brookings Institution. In the 2002-03 academic year Rich was the Mercator Professor at the Humboldt University in Berlin, Germany where he has also been appointed as a Permanent Fellow in the European Center for Comparative Government and Public Policy. In 2004, he was a Visiting Scholar at the Max Planck Institute for Foreign and International Social Law in Munich.

Before coming to the University of Illinois, Rich was on the faculty of the Heinz School of Urban and Public Affairs at Carnegie-Mellon University (1982-86), the Woodrow Wilson School of Public and International Affairs at Princeton University (1976-82), the University of Michigan Institute for Social Research (1975-76), and the University of Chicago (1974-75).

He is the author of six books and over 50 articles in the areas of health law and policy, federalism, information policy, and science and technology policy.

Julian Ziegler

Julian Ziegler was born in 1978 in Cambridge (UK), but spent most of his youth in Germany.

After graduation from high school in Berlin in 1997 and obligatory military service, he joined the Free University of Berlin to study law. During his time as a student he worked part-time on a scientific project for ethical and legal issues of disease in the context of social health insurance, at the Institute for Ethics in Medicine at the Eberhard-Karls-University of Tübingen. After graduation in spring of 2003 (1. State-Examination), he was accepted as a visiting scholar by Prof. Rich at the University of Illinois at Urbana-Champaign, College of Law as part of the BMEP/IALS exchange program, Biomedical Sciences Exchange Program—Academic Year Program. For this research program he received a fellowship from the German Academic Exchange Service (2003–2004). During that time (since January 2004), his dissertation project predictive genetic testing in health insurance, was accepted by Professor Kirchner (LL.M. Harvard) at the Humboldt-University of Berlin, Law Faculty. In summer 2004, he was awarded a Ph.D. fellowship by the Senate of the State Berlin. Currently, he is working on this dissertation project.

GENETIC DISCRIMINATION IN HEALTH INSURANCE – COMPREHENSIVE LEGAL SOLUTIONS FOR A (NOT SO) SPECIAL PROBLEM?

Robert F. Rich*
Julian Ziegler**

INTRODUCTION

In the course of the last decade, the technologies encompassing the field of human genetics have been constantly developed and improved. The most notable achievement was probably the completion of the Human Genome Project (HGP) in 2003.[1] Along with the completion of the mapping of the human genome came the discovery that many diseases are gene-related, being either the effect of chromosomal aberrations, or monogenic or multifactorial diseases.[2] Currently, over 5000 gene-associated diseases have already been discovered, with the number increasing constantly.[3] While this scientific progress offers great possibilities for the improvement of medical treatment, it also provides the possibility to analyze the DNA of single individuals for forensic purposes or the diagnosis or treatment of medical conditions. In fact, predictive genetic testing is already possible for several gene-associated diseases, such as cystic fibrosis.[4] Even though these tests are expensive, it is expected, the development of so called "Gene-Chips" or "DNA-Microarrays," which are capable of analyzing a large number of genes in one process,[5] will eventually lead to a significant decrease in cost. Thus, in the near future these

---

* Professor of Law and Political Science, University of Illinois, and Professor in the Institute of Government and Public Affairs, University of Illinois. B.A. 1971, Oberlin College; M.A. 1973, Ph.D. 1975, University of Chicago.

** Dr.jur.-candidate, Humboldt-University of Berlin, 2004-2005; 1. State-examination, Free University Berlin, 2003; Visiting scholar at the University of Illinois, College of Law, 2003-2004. (I would like to thank Prof. Dr. med. Hilmar Stolte from the International Academy of Life Sciences (IALS) and the Biomedical Sciences Exchange Program (BMEP) for supporting this research project).

1. Press-release, National Human Genome Research Institute, International Consortium Completes Human Genome Project: All Goals Achieved; New Vision for Genome Research Unveiled (Sept. 30, 2004), *at* http://www.genome.gov/11006929 (last visited Apr. 16, 2005) (on file with the Indiana Health Law Review).

2. *See* Scientific Appendix *infra* Part VI.

3. Jennifer S. Geetter, Coding for Change: *The Power of the Human Genome to Transform the American Health Insurance System*, 28 AM. J. L. & MED. 1, 4 (2002).

4. *See e.g.*, Wylie Burke, *Genetic Testing*, 347 N. ENG. J. MED. 1867, 1869 (2002).

5. Memorandum by the Senate Commission on Genetic Research, Predictive Genetic Diagnosis, Scientific Background, Practical and Social Implementation, 23 (Mar. 2003), *available at* http://www.dfg.de/aktuelles_presse/reden_stellungnahmen/2003/download/predictive_genetic_diagnosis.pdf (last visited Apr. 16, 2005) (on file with the Indiana Health Law Review) [hereinafter DFG].

tests could be affordable and available to the public at large. This development is quite important for medical purposes, such as "paternity testing" or criminal investigations, and also for employers and the insurance industry. Employers and insurance companies could use genetic tests in order to screen out job or insurance applicants who have a genetic predisposition to develop specific fatal diseases. If costs could be decreased, then large scale testing of employees or individuals seeking health insurance coverage could become a supplement to common medical examinations and tests. Consequently, the insurance industry might be interested in the use of genetic testing and genetic information in the process of underwriting.[6] This possibility is quite controversial in the legal and public policy community.

The use of genetic information by employers and insurance companies is far from being a mere utopian thought. This can be viewed both as an opportunity and a risk. In 1989, a study conducted by the U.S. Congress Office of Technology Assessment (OTA) revealed that twelve Fortune 500 companies were already using genetic monitoring or screening techniques to screen their employees.[7] In this context it is not surprising that law and public policy are concerned with "genetic discrimination;" indeed, the literature describes many examples of "genetic discrimination."[8] An example of this type of "discrimination" is well illustrated by Theresa E. Morelli's experience.[9] Her health insurance coverage was terminated because she had disclosed to her insurance company that her father suffered from Huntington's Disease.

It is feared that employers might single out employees with a high risk of developing certain diseases in order to lower their health care expenditures.[10] An additional concern is that health insurance companies offering coverage in the individual market might use predictive genetic testing as a means of underwriting, which could—in the case of an unfavorable test result —lead to an increased number of people who are virtually uninsurable because of their genetic risk profile.[11] One could describe the primary fear of many

---

6. Eric Mills Holmes, *Solving the Insurance/Genetic Fair/Unfair Discrimination Dilemma in Light of the Human Genome Project*, 85 KY. L. J. 503, 539 (1997).

7. Office of Tech. Assessment, U.S. Cong., Genetic Monitoring and Screening in the Workplace, 171, 176 (1990); *see also* Larry Gostin, *Genetic Discrimination: The Use of Genetically Based Diagnostic and Prognostic Tests by Employers and Insurers*, 17 AM. J. L. & MED. 109, 117 (1991); Jennifer Krumm, *Why Congress Must Ban Genetic Testing in the Workplace*, 23 J. LEGAL MED. 491, 495 (2002); Kimberly A. Steinforth, Book Note, *Bringing Your DNA to Work: Employers' Use of Genetic Testing Under the Americans with Disabilities Act*, 43 ARIZ. L. REV. 965, 970 (2001).

8. Krumm, *supra* note 7, at 496; Gostin, *supra* note 7, at 117; Robyn B. Nicoll, *Long-term Care Insurance and Genetic Discrimination – Get it While You're Young and Ignorant: An Examination of Current Discriminatory Problems in Long-term Care Insurance Through the Use of Genetic Information*, 13 ALB. L.J. SCI. & TECH. 751, 752-53 (2003).

9. Theresa E. Morelli, *Genetic Discrimination by Insurers: Legal Protections Needed From Abuse of Biotechnology*, Health Span, Sept. 1992, at 8.

10. Jeremy A. Colby, *An Analysis of Genetic Discrimination Legislation Proposed by the 105th Congress*, 24 AM. J. L. & MED. 443, 453-54 (1998).

11. *Id.* at 454-55.

authors as a fear of genetic discrimination.[12] While most legal scholars have concentrated on the applicability of Title I of the Americans with Disabilities Act of 1990 (ADA)[13] and Title VII of the Civil Rights Act of 1964[14] to genetic discrimination in health insurance, neither Act directly addresses this issue. The Health Insurance Portability and Accountability Act of 1996 (HIPAA),[15] represents the first health insurance nondiscrimination law, which partially deals with genetic information. Although HIPAA offers certain protections against discrimination based on genetic information,[16] these protections were not considered to be sufficiently protective, as the response of interest groups to more recent legislative proposals illustrates.[17] Therefore, the discussion about genetic discrimination in health insurance continued after the enactment of HIPAA, and interest groups and associations called for new, more comprehensive legislation. Consequently, several pieces of federal legislation concerning genetic discrimination have recently been introduced.[18] On October 14, 2003, one of those bills, the Genetic Information Nondiscrimination Act of 2003 (GINA),[19] was passed unanimously by the Senate and is being actively considered by the House of Representatives. It is worth noting, the President urged Congress to enact GINA,[20] and its passage by the Senate has been widely applauded by lobbying organizations such as the American Medical Association (AMA)[21] and the American Civil Liberties Union (ACLU).[22] Thus, it is safe to assume that GINA will not face further roadblocks on its way to become the first true federal genetic nondiscrimination law in the United States

In this Article, we argue the common legal conception of "genetic discrimination" is fundamentally flawed. Consequently, the current legal and public policy approach, based on this concept and reflected in the enactment of HIPAA and GINA, is neither sound nor fully defensible.

---

12. Kristie A. Deyerle, *Genetic Testing in the Workplace: Employer Dream, Employee Nightmare – Legislative Regulation in the United States and the Federal Republic of Germany*, 18 COMP. LAB. L. J. 547, 555 (1997); Krumm, *supra* note 7, at 491.

13. 42 U.S.C. §§ 12,111–12,117 (2004).

14. 42 U.S.C. § 2000(e) (2004).

15. 42 U.S.C. § 210 (2004).

16. *See infra* Parts IV.B.1, IV.C.1.

17. *See, e.g., infra* notes 20-23.

18. *See* Human Genome Project Information, General Privacy and Legislation, *at* http://www.ornl.gov/TechResources/Human_Genome/elsi/legislat.html (last visited Apr. 16, 2005) (on file with the Indiana Health Law Review).

19. S. 1053, 108th Cong. (2003).

20. Press Release, White House, President Pleased with Health Insurance Bill, (Mar. 6, 2002) *at* http://www.whitehouse.gov/news/releases/2002/03/20020306-10.html (last visited Apr. 16, 2005) (on file with the Indiana Health Law Review).

21. Press Release, Yank D. Coble Jr., AMA Applauds Senate Passage of Genetic Information Nondiscrimination Bill, (Oct. 14, 2003) (on file with author).

22. Press Release, ACLU, ACLU Urges House to Move Senate-Passed Bill Prohibiting Genetic Discrimination, (Apr. 1, 2004) *at* http://www.aclu.org/Privacy/Privacy.cfm?ID=15407&c=129 (last visited Apr. 16, 2005) (on file with the Indiana Health Law Review).

HIPAA and GINA respond to fears of a "genetic determinism," which rely on incomplete information about the nature and invasiveness of genetic information. Furthermore, genetic information as expressed and defined in the provisions of both statutes, is a concept that evades a precise definition; it is difficult, if not impossible, to distinguish it from regular medical information. Nevertheless, both statutes provide differing definitions of genetic information and try to distinguish between medical and genetic information. Consequently, we conclude that HIPAA and GINA do not provide a clear statutory basis for legal decisions concerning discrimination based solely on the use or application of genetic information. The language of these statutes stems from a biased public policy conception of genetic information, which seems to be based on a misperception or misunderstanding of genetic testing methods and the empirical results derived from such testing. The current statutory approach also neglects the legitimate information needs of the private health insurance market. The common fear of health insurers seems to be that if genetic test kits become widely available, adverse selection resulting from an asymmetry of information could occur. In turn, this could eventually lead to an increase in the pricing of health insurance policies and consequently lead to a higher number of people without health insurance coverage.

In order to support our arguments, we will analyze the potential influence of HIPAA and GINA on genetic discrimination in private health insurance. In the first Part of this Article we analyze what is meant by the term "discrimination." We will examine the scientific basis underlying the concept of "genetic discrimination" in order to pinpoint what exactly makes "genetic discrimination" different from "normal" discrimination in health insurance. The second Part of the Article focuses on whether genetic discrimination, as described in the first Part, is likely to occur and if so, in which market segment. The third Part examines protections against genetic discrimination in health insurance offered by the ADA and Executive Order 13,145. In the fourth Part, the impact of HIPAA and GINA on "genetic discrimination" in health insurance will be analyzed. Since the idea of "genetic discrimination" is rooted in the definition of genetic information, we will examine the various meanings of the term "genetic information" provided by HIPAA, the HIPAA final interim regulations, and GINA. We will show that HIPAA and GINA do not even offer comprehensive protection against "genetic discrimination", as defined by the laws themselves. The fifth and final Parts of the Article present our overall conclusions.

I. WHAT IS UNFAIR "GENETIC DISCRIMINATION" IN HEALTH INSURANCE?

In order to analyze the influence of HIPAA and GINA on unfair "genetic discrimination" in the market for health insurance, it is imperative to understand what the term "genetic discrimination" actually means. As "discrimination" is widely used to describe a behavior that is perceived in a negative fashion, it is important to emphasize the difference between this kind of

"negatively connotative" discrimination on the one hand,[23] and insurance discrimination on the other hand.

Despite the semantic difficulties in the use of the term "discrimination," the legal view of discrimination is founded in the Fourteenth Amendment to the U.S. Constitution. In Section 1, the Fourteenth Amendment provides for a general legal framework of appropriate and inappropriate differentiation.[24] Under the Fourteenth Amendment, "[c]lass legislation, discriminating against some and favoring others, is prohibited, but legislation which, in carrying out a public purpose, is limited in its application, if within the sphere of its operation it affects alike all persons similarly situated, is not within the amendment."[25] Thus "it is only invidious 'discrimination' which offends the Constitution."[26] Given this foundation, the issue is, how one differentiates between permissible and invidious discrimination. The rules which are used to determine that question are summarized in Lindsley v. Natural Carbonic Gas Co.,[27] whereby a differentiation is unconstitutional if it is without any reasonable basis and therefore purely arbitrary. The court also developed another standard which stipulates that "the classification must be reasonable, not arbitrary, and must rest upon some ground of difference having a fair and substantial relation to the object of the legislation, so that all persons similarly circumstanced shall be treated alike."[28] Therefore, the core principle of the legal definition of unlawful (e.g. unconstitutional) discrimination is that similarly circumstanced individuals have to be treated alike in the absence of a reasonable basis for a differentiated treatment.

Since insurance discrimination is legally permissible, only as long as it is based on reason and not purely arbitrary, we will at first explain the use of

---

23. For an example of the derogatory connotation of the term "discrimination" see OXFORD ADVANCED LEARNER'S DICTIONARY, at 343: "treating a person or group differently (usually worse) than others . . . ." (Anthony P. Cowie et al. eds., Oxford University Press, 1989).

24. The Constitutional language of the Fourteenth Amendment only focuses on whether a state statute conforms to the Constitution; *see* CBS v. Democratic Nat'l Comm., 412 U.S. 94 (1973). Although it is the Fifth Amendment which offers the due process clause in regard of federal action (such as the enactment of HIPAA and GINA), unlike the Fourteenth Amendment, the Fifth contains no equal protection clause and it provides no guaranty against discriminatory legislation by Congress; *see* Detroit Bank v. United States, 317 U.S. 329, 337 (1943). Thus, the "equal protection of the laws" is a more explicit safeguard of prohibited unfairness than "due process" of law; *see* Bolling v. Sharpe 347 U.S. 497, 499 -500 (1954).

Thus, it is the Fourteenth Amendment that has to be employed in order to illustrate the legal idea of wrongful discrimination.

25. Barbier v. Connolly, 113 U.S. 27, 32 (1884).

26. Ferguson v. Skrupa, 372 U.S. 726, 732 (1963); *see* Williamson v. Lee Optical Co., 348 U.S. 483, 488-89 (1955).

27. Lindsley v. Natural Carbonic Gas Co., 220 U.S. 61, 78 -79 (1911); Morey v. Doud, 354 U.S. 457, 463-64 (1957) (stating that classifications which are purposefully discriminatory fall before the equal protection clause without more); *e.g.*, *Barbier*, 113 U.S. at 30; Yick Wo v. Hopkins, 118 U.S. 356, 373-74 (1886); *cf.* New York City Transit Auth. v. Beazer, 440 U.S. 568, 593 (1979).

28. F.S. Royster Guano Co. v. Virginia, 253 U.S. 412, 415 (1920); *see also* Brown-Forman Co. v. Kentucky, 217 U.S. 563, 573 (1910).

discrimination in health insurance. Next, we will analyze if the insurers' use of genetic testing procedures justifies a different legal treatment of genetic discrimination in insurance.

The health insurance market in the United States is divided into several market segments. According to the U.S. Census Bureau, in 2002, 84.8% of the American population had health insurance coverage.[29] That left 15.2% of the population (43,574,000 people) without health insurance coverage. Approximately, 25.7% of the insured population was covered through government issued health insurance, such as Medicare, Medicaid and Military health care. The remaining 69.6% of the population received health care through private health insurance, either employment based or individually purchased.[30]

Since eligibility for governmentally issued health insurance coverage is not based on the applicants' health status, this market segment is not likely to be affected by genetic discrimination. Private health insurance, in contrast to governmentally subsidized plans, is financed solely by individuals seeking coverage, such as employers, employees and private individuals. In this market segment, health insurers commonly use medical information for underwriting purposes, which is fully permissible. On the one hand, insurance companies ask for the family history of applicants in order to determine if any hereditary disease is likely to manifest itself in the person of the applicant.[31] On the other hand, medical examinations are conducted in order to screen out already manifest diseases.[32]

Usually health insurance is obtained as an employment benefit. In 2002, roughly 85% of the privately insured American population (175,296,000 of 198,973,000 people) received health insurance coverage through employment in policies written for large groups.[33] This means that most medical insurance coverage is provided on a group basis and financed jointly (in some "shared arrangement") by employers and employees. The insurance industry classifies the risk of such groups based on a group's size, on its past claims experience or on data of the claims experience of similar groups in the same industry or

---

29. U.S. CENSUS BUREAU, U.S. DEP'T OF COMMERCE, HEALTH INSURANCE COVERAGE IN THE UNITED STATES: 2002, 20 (2003), *available at* http://www.census.gov/prod/2003pubs/p60-223.pdf.

30. *Id.* at 20 (In the author's opinion, the discrepancy between the total of insured people and the sum of governmentally and privately insured people is apparently due to overlapping coverage, such as Medigap).

31. David J. Christianson, *Genetic Testing: Risk Classification and Adverse Selection*, 15 J. INS. REG. 75, 76 (1996); Colby, *supra* note 10, at 462; Deborah A. Stone, *The Rhetoric of Insurance Law: The Debate over AIDS Testing*, 15 LAW & SOC. INQUIRY 385, 389 (1990) [hereinafter *Rhetoric*].

32. Christianson, *supra* note 31, at 76; Colby, *supra* note 10, at 462.

33. U.S. CENSUS BUREAU, U.S. DEP'T OF COMMERCE, HEALTH INSURANCE COVERAGE IN THE UNITED STATES: 2002, 20 (2003), *available at* http://www.census.gov/prod/2003pubs/p60-223.pdf.

region.[34] The group's premium rates are also set according to these factors.[35] Since the risk incurred in employment-based health insurance plans is spread among the whole group, premium rates can be maintained at a relatively low price level. Furthermore, employer sponsored group health plans are usually subsidized by the employer.[36] Because of these subsidizations, the pricing of the group's policy does not increase due to high claims experiences, which may occur if many high-risk individuals are enrolled in the plan. As this subsidization practice helps to maintain stable premium rates, low-risk individuals are also attracted to enroll in the policy. Consequently, the same average number of low-risk and high-risk individuals are attracted by the stable premium rates.[37]

As employment-based group health insurance plans are experience rated, individual insurance underwriting practices are not used in calculating the group's premium rates. The risk of individuals can be spread among the whole group. This is only different in small group health plans with less than twenty-five employees.[38] When the group size is too small to qualify for experience rating, the individual underwriting practices are used.[39] Given the rate-setting practices just described, health-status related information is of no actuarial value in the larger group market where underwriting practices are not used. Consequently, any attempt to obtain and use such information for underwriting in this market segment should be considered both unnecessary and unfairly discriminatory.

In contrast to employment-based large group health plans, small group health plans and individual insurance plans are underwritten in order to calculate appropriate, i.e. actuarially fair,[40] premium rates. Risk-classification is one method utilized in the underwriting process. Risk-classification enables insurers to divide individuals into groups with similar projected claims and obtain an estimate of the projected costs associated with an individual.[41] Thereby, insurers are able to calculate the lowest premium rates compatible

---

34. Holmes, *supra* note 6, at 534.

35. *Id.*; Geetter, *supra* note 3, at 45.

36. NATIONAL CONFERENCE OF STATE LEGISLATURES, GENETICS POLICY REPORT: INSURANCE ISSUES 12 (2001).

37. *Id.*

38. Deborah A. Stone, *The Struggle for the Soul of Health Insurance*, 18 J. HEALTH POL., POL'Y & L. 287, 302 (1993); *Rhetoric*, *supra* note 31, at 390.

39. Geetter, *supra* note 3, at 45; Stone, *supra* note 38, at 302.

40. Geetter, *supra* note 3, at 49.

41. Christopher M. Keefer, *Bridging the Gap between Life Insurer and Consumer in the Genetic Testing Era: The RF Proposal*, 74 IND. L. J. 1375, 1383 (1999); Marcelita C. Anderson, *Genetic Testing in Insurance Underwriting: A Blessing or a Curse? An Examination of the Tension Between Economics and Equity in Using Genetic Testing in Risk Classification*, 25 CREIGHTON L. REV. 1499, 1510 (1992); Christianson, *supra* note 31, at 75; Roberta B. Meyer, *General Position of the American Council of Life Insurance on Genetic Information and Genetic Tests and Life and Disability Income Insurance*, 15 J. INS. REG. 66, 67 (1996).

with the projected costs (reasonable discrimination). Furthermore, risk-classification allows insurers to offset the impact of adverse selection.[42]

Consumers, when faced with different insurance plans, must weigh the premium rates against the value they expect to receive. If the insurance applicant has critical information about his or her health status which is unknown to the insurer, this asymmetry of information might lead to insurance becoming less affordable. Because high-risk individuals would be more likely to buy insurance, the insurer would be forced to eventually raise the premium rates to cover the losses, which occur due to the higher claims costs. With increasing premiums, low-risk individuals would discontinue the plan, which would lead to a further increase in premium rates to keep the plan balanced.[43] This process, which can result in cases of asymmetric information, is called adverse selection.[44] By contrast, a situation of asymmetric information is not likely to occur if applicants disclose all relevant information to insurers. Thus, the possibility of adverse selection could be offset and premium rates could be stabilized through fair risk classification.

As mentioned above, insurers differentiate among individuals seeking health insurance coverage. The purpose of this practice is maintaining a system in which each insured person's premium is rated according to the amount of his or her risk. Implementation of this notion of actuarial fairness requires that insurers are allowed to create classifications that represent real differences between individuals. In order to stay in business, insurers must be allowed to risk-classify, which means, that they must be allowed to discriminate or distinguish among individual health insurance applicants. Because the private health insurance system covers nearly seventy percent of the American public without discrimination through risk-classification, the cornerstone of health insurance in the United States would be endangered.[45] Although discrimination is important in the health insurance market, it is necessary to distinguish between reasonable, legally permissible discrimination, which is vital to the health insurance market, and discrimination that is not. The legal permissibility of insurance discrimination depends upon its foundation of "reason," which in this case refers to medical, clinical, or statistical data about the insurance applicant. Therefore, in the absence of this reasonable basis, insurance discrimination would be arbitrary and, thus, unlawful.

---

42. Christianson, *supra* note 31, at 75.

43. George A. Akerlof, *The Market for "Lemons": Quality Uncertainty and the Market Mechanism*, 84 Q. J. ECON. 488, 492-93 (1970).

44. *Id.* at 493; Keefer, *supra* note 41, at 1384; Holmes, *supra* note 6, at 543; Anderson, *supra* note 41, at 1511; Kimberley Nobles, *Birthright or Life Sentence: Controlling the Threat of Genetic Testing*, 65 S. CAL. L. REV. 2081, 2089 (1992).

45. Holmes, *supra* note 6, at 535 (stating that without risk classification, the insurance system will cease to be a private insurance system).

Reasonable legally permissible discrimination and unreasonable discrimination can also be described as fair and unfair discrimination.[46] In determining unfair discrimination, it is important to analyze whether insurers' use of genetic information for underwriting purposes justifies a substantially different legal treatment than common methods of risk-classification in the process of insurance underwriting. As genetic discrimination arises from the "discriminatory" use of genetic information, we will examine the sources of this kind of information; genetic defects, gene-associated diseases, and the scientific and clinical methods used to detect them. In an effort to determine what is "reasonable," we will briefly explain genetic defects and the testing methods used to identify such defects.

A. The Nature of Diseases

From a legal perspective, in order to define "genetic information" it is essential to understand the role of genes in the development of diseases. If a person has a genetic defect, this defect can cause the development of a certain disease. While certain genetic defects translate into various diseases, others do not. In some cases, the presence of a particular genetic defect is connected to the inevitable development of a disease associated with that defect.[47] In most cases though, this relationship is more complex.

There are three large and heterogeneous groups of gene-associated diseases: (1) Monogenic diseases, which are caused by defects affecting a single gene;[48] (2) Polygenic-multifactorial diseases, which stem from the interaction of one or more genes and environmental factors;[49] and (3) Chromosomal aberrations, which are due to a change in the structure or number of the chromosomes.[50] These diseases, and the genetic predisposition to develop such diseases, can be discovered or diagnosed by several differing methods. The most widely recognized method in this context is the so called gene-analysis.

B. Gene-Analysis

Depending on the underlying purpose of gene-analysis, several differing definitions of "gene-analysis" are offered.[51] In the insurance industry, obtaining information about the "genetic" health-status of applicants or enrollees is the basis of underwriting, and thus, should be the basis for the definition of the term "gene-analysis" in this paper. Thus, "gene-analysis" should be interpreted to include all methods that reveal information about the genotype of an

---

46. Keefer, *supra* note 41, at 1384; Colby, *supra* note 10, at 459.
47. DFG, *supra* note 5, at 25.
48. *See* Scientific Appendix *infra* Part VI.
49. *See* Scientific Appendix *infra* Part VI.
50. *See* Scientific Appendix *infra* Part VI.
51. *See, e.g.*, Burke, *supra* note 4, at 1867; Nicoll, *supra* note 8, at 756-57.

individual. As such, the term "gene-analysis" encompasses four levels of analysis with several analytical techniques at each level. Methodologically, two types of genetic tests can be distinguished: a) the direct genetic test, which tries to identify one or more disease-associated mutation(s); and[52] b) the indirect genetic test, which tries to identify the risk-haplotype.[53]

Both direct and indirect testing methods have methodological problems associated with them. The direct test requires the identification and at least partial characterization of the relevant gene.[54] The other method, the indirect gene-analysis, uses the principle of genetic linkage. Relying on the use of cloned deoxyribonucleic acid (DNA) segments exhibiting sequence variability (polymorphism), which reside near a disease-associated gene, the method is used to track a chromosome carrying the disease-associated gene through a family.[55] The goal is to predict whether an individual in the family is likely to inherit the disease-associated gene.[56] The accuracy of this testing method depends on the informativeness[57] of the individual's family for the considered gene loci, as well as the proximity of the marker genes to the disease gene locus on the chromosome.[58]

As the list of known disease-associated genes grows rapidly, the direct testing method is of greater relevance for insurance companies. This is because the indirect testing method requires additional medical information concerning the family of the tested individual which may or may not be forthcoming.

Information about an individual's genotype can be derived using four different analytical approaches: (1) phenotype-analysis; (2) cytogenetic analysis; (3) protein-chemical analysis; and (4) DNA-analysis.[59] In phenotype-analysis, the outward appearance of an individual, the phenotype, is examined in order to reach conclusions concerning certain genetic traits of an individual. In cytogenetic analysis, abnormalities in the number or structure of the chromosomes are examined by the use of microscopic techniques. The protein-chemical analysis is an examination method by which one tries to discover gene-associated diseases linked to the presence, absence, or alteration of proteins, which can lead to metabolic defects. In contrast to the three preceding methods, the DNA-analysis tries to discover genetic defects by examining the DNA itself. If a genetic defect is known to be associated with the

---

52. H. WILLIAM TAEUSCH ET AL., SCHAFFER AND AVERY'S DISEASES OF THE NEWBORN 102 (Lisette Bralow ed., 6th ed. 1991).

53. *Id.*

54. *Id.* at 101-02.

55. *Id.* at 102.

56. *Id.*

57. MARGARET W. THOMPSON ET AL., THOMPSON & THOMPSON: GENETICS IN MEDICINE 180 (Martin J. Lawrence ed., 5th ed. 1991).

58. *Id.*

59. For more information on the described methods of gene-analysis, see Scientific Appendix *infra* Part VI.

development of a certain disease, it is possible to estimate the likelihood of the development of this disease, if the genetic defect is present. DNA-analysis, therefore, does not predict the development of a disease. It only serves as a means to discover whether a genetic defect is present or not. The "penetrance," the likelihood of the development of a certain disease due to a genetic defect, differs according to whether the disease is monogenic or polygenic-multifactorial.[60] Except for the DNA-analysis, all these methods are commonly used in regular medical examinations and only use different strategies in uncovering genotype-related information.

C. Is Genetic Information Different?

As summarized above, the methods used to unveil genetic information are not, from a scientific perspective, fundamentally different from the methods used in gathering other medical information. One could say that all medical information has a genetic basis.[61] As already indicated, genetic information can be derived by various methods, which are commonly used during regular medical examinations. Phenotype-analysis, for example, can provide information about an individual's genetic makeup and is part of every routine medical examination. Cytogenetic analysis and protein-chemical analysis are commonly used in the diagnosis of hereditary diseases. Thus, the only "new" form of gene-analysis is the DNA-analysis. Although the basic techniques used in DNA-analysis were developed before 1990, DNA-analysis is not widely used in medical examinations. This is primarily because of the laborious, and thus, expensive character of this technique, rather than because of its "exceptional" or "unusual" character. Therefore, even DNA-analysis cannot be labeled a truly "new" or "unusual" form of examination that has to be regarded as strictly set apart from the other methods of gene-analysis, which are already used during regular medical examinations.

Consequently, we conclude the methods used in gathering genetic information are not fundamentally different from, but rather inseparably linked, to the methods used in gathering regular medical information. Therefore, from a legal and public policy perspective, "genetic" discrimination cannot be fundamentally different from "normal" discrimination based on regular medical information. If this is true, then like commonly used underwriting criteria, genetic testing procedures have to be considered reasonable sources of data and, as such, are a reasonable basis for legally permissible insurance discrimination.

Nevertheless, critics of gene-analysis in health insurance maintain their opposing view by arguing there is a difference between regular medical infor-

---

60. *See* Scientific Appendix *infra* Part VI.

61. See, e.g., George J. Annas, *The Limits of State Laws to Protect Genetic Information*, 345 N. ENG. J. MED. 385, 387 (2001).

mation and genetic information, which consists of several factors resulting in a "genetic exceptionalism."[62] One argument is that a person's genotype is beyond that person's control.[63] Other arguments stipulate that only a small number of people would actually be affected by a genetic predisposition,[64] and that genetically "unlucky" people would be stigmatized.[65] The critics maintain the position that genetic information is different in its depth, scope and influence on the affected individual. It is argued, therefore, that the use of genetic information leads to genetic discrimination.

Although these arguments seem convincing at first, it is questionable that they can be employed to justify a different legal treatment of genetic tests as compared to regular medical.

1. Only a Small Number of Individuals Will be Negatively Affected by Insurers Use of Genetic Information

One of the first arguments used to support prohibitive legislation in the field of genetic testing is that the outcome of such tests would only adversely affect a small number of people. Greely, a proponent of the argument, who reasons that while genetic predispositions will most likely be concentrated in a small group of "extremely unlucky" high-risk individuals, it would constitute a cruel policy to allow genetic discrimination in health insurance.[66] To support his view, Greely offers three arguments: first, health insurance is an important part of life in contemporary America;[67] second, providing protection from genetic discrimination would highly benefit those groups of high-risk individuals;[68] and third, "we can afford" this protection.[69]

Greely's argument is morally appealing. Nevertheless, the mere affordability, which itself is very questionable, does not constitute a duty for private health insurance to support those "unfortunates" with unfavorable genetic makeup. Furthermore, it is not understandable why individuals with a predisposition for a certain disease should be subsidized through a prohibition of the use of genetic information in health insurance, and individuals who actually have developed a disease should be exempt from that subsidy. Why people with a predisposition to develop a disease should be treated more favorably than people who developed such diseases remains unexplained. Moreover, if only a small group of people would be affected by negative test results, it

---

62. Geetter, *supra* note 3, at 56.

63. Deborah Hellman, *What Makes Genetic Discrimination Exceptional?*, 29 AM. J.L. & MED. 77, 87 (2003).

64. Henry T. Greely, *Genotype Discrimination: The Complex Case for Some Legislative Protection*, 149 U. PA. L. REV. 1483, 1500 (2001).

65. Hellman, *supra* note 63, at 89.

66. Greely, *supra* note 64, at 1500.

67. *Id.*

68. *Id.*

69. *Id.*

would constitute discrimination against the unaffected majority to deprive them of the possibility to lower their health insurance premiums through the use of their favorable test results.

2. Genetic Determinism

Another argument employed by the critics of the insurer's use of genetic information is that an individual has no control over her genotype.[70] The principle on which this argument is founded is in itself very convincing, namely that a person should be judged on the basis of what he or she does and not on what he or she is.[71] Despite the important moral appeal of this argument, it fails for one reason. As shown above, the importance of genes in the development of certain diseases differs according to whether the disease is monogenic or multifactorial. While the penetrance, the certainty, that a genetic defect will translate into a disease, of a genetic defect in monogenic diseases may in some instances be close to absolute certainty, as in the case of Chorea Huntington,[72] the penetrance of genetic defects in multifactorial diseases can only reach the concordance-rates of identical-twin-studies[73] (40-60%).[74] The remaining percentage consists of non-genetic or environmental factors. Therefore, a clear predictability of the onset of a multifactorial disease is not possible. Consequently, as the idea of a genetic determinism lacks an evidentiary basis; genetic determinism cannot serve as an argument against the use of genetic testing and genetic information.

3. Stigmatization

The idea of genetic determinism is closely related to another argument employed by the supporters of prohibitive legislation; that genetic predispositions to disease are intrinsically stigmatizing.[75] As genetic predispositions to disease stigmatize an affected individual, the use of information concerning that predisposition is stigmatizing, and thus, discriminatory. The underlying claim is that genetic discrimination should be prohibited, either, because of the impact it has on the individual, or, because of the idea it expresses.[76] The effect of genetic testing on the tested individual can be one of psychological

---

70. Gostin, *supra* note 7, at 110-11; Hellman, *supra* note 63, at 87.

71. Gostin, *supra* note 7, at 110-11; Hellman, *supra* note 63, at 87.

72. DFG, supra note 5, at 25; Richard A. Epstein, *The Legal Regulation of Genetic Discrimination: Old Responses to New Technology*, 74 B.U. L. REV. 1, 2 (1994); Michael J. Smith, *Population-Based Genetic Studies: Informed Consent and Confidentiality*, 18 SANTA CLARA COMPUTER & HIGH TECH. L. J. 57, 64-65 (2001).

73. DFG, *supra* note 5, at 19 (the concordance-rates in identical-twin-studies can reach forty to sixty percent).

74. *See* Scientific Appendix *infra* Part VI.

75. Hellman, *supra* note 63, at 89.

76. *Id.* at 90.

trauma.[77] Confronted with the elevated chance to develop a possibly life-threatening disease, individuals might experience an adverse influence of their "death sentence" on major life decisions.[78] The risk of suicide, substance abuse, job loss or marriage problems are mentioned as possible effects of predictive genetic testing.[79] Even though Holmes argues these risks distinguish predictive genetic tests from diagnostic tests, which reveal the presence of a fatal disease,[80] it is hard to understand why a person with a fatal disease should feel less stigmatized than a person with a test that expresses a mere probability of disease development.

The "expressivist" theory also argues that the use of genetic information will create a genetic underclass. This theory correlates with the idea of a genetic determinism,[81] which is mostly based on fear rather than on scientifically based rationality.[82]

Nevertheless, both theories of genetic stigmatization fail to address the core of the problem. It is not the genetic predisposition itself that exerts a stigmatizing effect. As long as the information about a disposition is not known to a third party, stigmatization is unlikely to occur. Therefore, only the disclosure of the information to third parties, that an individual has a genetic predisposition, might cause stigmatization. Insurance companies could represent such third parties.

4. Right to Privacy

It is also argued, that the protection of the right to privacy requires the prohibition of genetic testing and the subsequent use of genetic information in health insurance. As every human's genotype is that person's unique property, a use of genetic information which could culminate in centralized genetic information databases would violate that person's civil rights.[83] Though describing a justifiable fear, this argument does not provide a substantial reason for a different treatment of genetic information, because the same argument could be employed to protect regular medical information. From a legal or clinical perspective, there is no warrant to treat medical and genetic information differently. Thus, the right to privacy should not be claimed for one type of information as compared to the other.

---

77. Krumm, *supra* note 7, at 506.

78. Holmes, *supra* note 6, at 573.

79. Ronald M. Green & A. Mathew Thomas, *DNA: Five Distinguishing Features for Policy Analysis*, 11 HARV. J. L. & TECH. 571, 572 (1998); Krumm, *supra* note 7, at 506.

80. Holmes, *supra* note 6, at 573.

81. Hellman, *supra* note 63, at 90.

82. Greely, *supra* note 64, at 1483.

83. Holmes, *supra* note 6, at 569; Tara L. Rachinsky, *Genetic Testing: Toward a Comprehensive Policy to Prevent Genetic Discrimination in the Workplace*, 2 U. PA. J. LAB. & EMP. L. 575, 585 (2000); Smith, *supra* note 72, at 84.

Another form of the right to privacy is the "right not to know."[84] A person has the right to know about his or her genetic predisposition(s) to certain diseases.[85] This corresponds to the right not to know about those predispositions. As shown above, the knowledge of a genetic predisposition triggers anxieties and might have adverse effects on life decisions of the affected person. Thus it is arguable that as part of the right to privacy, a person should be able to refuse this "burden" of knowledge. Still, this does not explain why genetic information should be treated differently from regular medical examinations and the medical information gained thereby. When faced with this concern, critics of genetic testing argue that genetic testing is more invasive than regular medical examinations.[86] Moreover, the predictive value of genetic information makes it more vulnerable to abuse than regular medical information.[87] As shown above, genetic testing techniques do not differ fundamentally from regular medical examinations. On the contrary, in many cases they are already part of normal medical examinations. In sum, the information derived from genetic tests constitutes "predictive data," while the information obtained from medical examinations represents diagnostic information which has a much higher level of certainty associated with it. Nevertheless, the distinction between these two types of information is not meaningful from a legal or public policy perspective.

D. Discussion

In summary, insurers offering health insurance in the individual market and consumers of such health insurance usually have diametrically opposing arguments regarding unfair discrimination in health insurance based on genetic predispositions. Insurers argue genetic information, like any other health status related information, should be shared with them in order to prevent the effects of asymmetric information.[88] In order to maintain equity among policyholders, the premiums must reflect the individuals' loss probabilities.[89] As a major goal of insurance underwriting is equity, the use of genetic

---

84. George J. Annas, *Genetic Privacy: There Ought to be a Law*, 4 TEX. REV. L. & POL. 9, 11 (2000); Colby, *supra* note 10, at 458; Patrik S. Florencio & Erik D. Ramanathan, *Secret Code: The Need for Enhanced Privacy Protections in the United States and Canada to Prevent Employment Discrimination Based on Genetic and Health Information*, 39 OSGOODE HALL L. J. 77, 88 (2001) (who refer to this as "choice"); Holmes, *supra* note 6, at 574; Krumm, *supra* note 7, at 505; Rachinsky, *supra* note 83, at 583.

85. Colby, *supra* note 10, at 458; Krumm, *supra* note 7, at 505 Rachinsky, *supra* note 83, at 584.

86. Colby, *supra* note 10, at 459.

87. Ellen Wright Clayton, *Ethical, Legal, and Social Implications of Genomic Medicine*, 349 N. ENGL. J. MED. 562, 563 (2003).

88. Colby, *supra* note 10, at 461.

89. Deborah A. Stone, *The Implications of the Human Genome Project for Access to Health Insurance*, *in* THE HUMAN GENOME PROJECT AND THE FUTURE OF HEALTH CARE 133, 138 (Thomas H. Murray et al. eds., 1996).

information as a basis for a sound actuarial calculation of premium rates would lead to a more accurate reflection of the individuals' risk, thus to a more equitable treatment of applicants and policyholders.[90] The opposite, an equal treatment of individuals in premium rates, would result in an unfair "forced" subsidization of high-risk individuals by low-risk individuals.[91] This practice would in itself constitute unfair discrimination, which can be avoided by sound actuarially fair risk-classification.[92]

On the other hand, supporters of prohibitive legislation argue that the use of genetic information by insurers is subject to abuse resulting in unfair discrimination.[93] Furthermore, consumers question the reliability and the significance of currently available genetic tests for the purpose of insurance underwriting.[94] Also, considering the special nature of genetic information, the use of such information could lead to a stigmatization of individuals solely because of their genetic nature.[95] As the genetic makeup of an individual is beyond that individual's choice, and the purpose of insurance being risk distribution among all policyholders, insurance should distribute such involuntary risks among all policyholders.[96] Therefore, a system of equal access to health insurance through fair risk distribution should be adopted rather than a system of equitable access through risk-classification.[97]

From our point of view, despite the current lack of simple, inexpensive genetic tests which possibly will be mitigated by technological improvements such as DNA-Chips or DNA-Microarrays, the use of genetic testing improves the underwriting process in health insurance by allowing insurers to calculate premiums more accurately. As noted earlier, insurers use probabilistic data in order to calculate fair premium rates; if there was certainty about a health condition, insurance would be pointless. Thus, the probabilistic character of genetic information, based on the varying reliability of genetic tests, should not be used as an argument against the inclusion of genetic testing in health insurance underwriting.

Moreover, as Geetter shows, the argumentation in itself is contradictory.[98] The argument that genetic information, based on its predictive value, has the potential to cause stigmatization, is counteracted by the assessment that genetic information is too unreliable to be of much predictive value in the underwriting process.[99]

---

90. Colby, *supra* note 10, at 460; Holmes, *supra* note 6, at 539.
91. Holmes, *supra* note 6, at 544.
92. *Id.*
93. *Id.* at 557; Colby, *supra* note 10, at 454.
94. Geetter, *supra* note 3, at 59.
95. Hellman, *supra* note 63, at 89.
96. Colby, *supra* note 10, at 457.
97. Holmes, *supra* note 6, at 563.
98. Geetter, *supra* note 3, at 59-60.
99. *Id.* at 60.

Furthermore, in contrast to the anecdotally[100] fueled fears regarding the availability of health insurance coverage, more refined risk-classification would not necessarily lead to less insurability. Insurers, when faced with an individual who has an unfavorable genetic makeup, can react with premium increases for full coverage, with coverage exclusions for that particular trait, or by denying coverage at all.[101] Overall it is clear that the supporters of prohibitive legislation have a biased view of genetic testing which focuses mainly on individuals with positive, e.g. unfavorable test results. But discrimination is a two-edged sword. The protection of individuals with poor genetic makeup could come at the expense of individuals with favorable genetic traits, resulting in forced insurance premium subsidies. This would be equally unjust, because it would result in actuarially unfair discrimination of the insurance applicants and enrollees with favorable genetic makeup. Unfairness occurs when unequal risks are treated equally. In this context, Stone argues that, given perfectly predictive medical information, following this definition of fairness would eventually lead to a system in which "each person *would* pay exactly the cost of his or her own medical care."[102] Thus insurance would be transformed into an individual medical savings account.[103] Stone's argumentation is convincing except for one major point: as shown above, medical information is not perfectly predictive. Although the insurance industry relies on risk-pooling, as Stone rightfully states, this does not mean, that insurers should not be able to differentiate between groups of different risk-categories. Inside these different risk-categories, the risk (that is the risk which remains due to the imperfect medical or genetic predictability) represented by the individuals in the risk-category is sufficient to justify a differentiated treatment in terms of premium calculation in relation to members of the other risk-categories.[104] As genetic information is not perfectly predictive, there will always remain individually different risks which can be classified into different categories, which prevent insurance to be transformed into individual medical savings accounts.

All the above arguments employed by the critics of genetic testing are centered on the idea of a genetic exceptionalism. The idea is that genetic information is new, not only technically, but because it is more revealing than regular medical information. Moreover, it is argued the unique predictive

---

100. Greely, *supra* note 64, at 1489 (stating that the shortage of well-documented examples of genetic discrimination in health insurance is [indeed] noteworthy); Paul Steven Miller, *Genetic Discrimination in the Workplace*, 26 J. L. Med. & Ethics 189,190 (1998) (referring to cases of workplace discrimination).

101. The insurance industry classifies the applicant's risk as "standard," "substandard" and "declined." *See e.g.*, Mark A. Rothstein, *Predictive Genetic Testing for Alzheimer's Disease in Long-Term Care Insurance*, 35 GA. L. REV. 707 (2001).

102. Stone, *supra* note 89, at 138; Stone, *supra* note 38, at 293-94.

103. Stone, *supra* note 89, at 138; Stone, *supra* note 38, at 294.

104. Anderson, *supra* note 41, at 1510.

value of genetic information offers new possibilities for discrimination, thus requiring a different legal treatment than regular medical information.

Overall, the arguments fail to establish a significant difference between genetic and regular medical examinations. The arguments fail because they are based primarily on fear without having a real evidentiary basis.[105] Scientifically, genetic information is not fundamentally different, more stigmatizing or more invasive than regular medical examinations.[106] Furthermore, it does not lead to new questions regarding the right to privacy because it is even less intrusive than regular medical examination results. This becomes evident in the difference of a mere prediction based on genetic testing as compared to a diagnosis based on regular medical examinations. The information about the possibility of developing a disease is less intrusive than the information about having a disease. As genetic information is not proved to be fundamentally different from regular medical information, the idea of a genetic exceptionalism has to be abandoned. Therefore, we must conclude from a legal and public policy perspective that genetic discrimination is not clearly distinguishable from discrimination based on regular medical information.

We, therefore, argue that the more convincing case is made by the insurance industry. Genetic information, as well as regular medical information should be available to health insurers. This should become even more evident, if one considers the important social role of private individual health insurance, which provides coverage for over nine percent of the American public. The only means to protect the socially important individual health insurance market from the effects of adverse selection (for example market failure)[107] is to prevent the onset of asymmetric information by granting insurers equal access to genetic information.[108] Nevertheless, as insurers need this information to prevent the onset of adverse selection and for premium calculation, their use of that information should not exceed these purposes. Because the use or disclosure of genetic information other than for risk-classification is unnecessary for actuarial purposes, any use beyond that limitation should be considered to be an "unfair 'genetic' discrimination."

II. POSSIBLE EXAMPLES OF UNFAIR GENETIC DISCRIMINATION IN HEALTH INSURANCE

The next step in our analysis is to describe, where such unfair "genetic discrimination" might actually arise in the health insurance market. In order to do so, we will deal with the group and individual market segments separately.

---

105. Colin S. Diver & Jane Maslow Cohen, *Genophobia: What is Wrong with Genetic Discrimination?*, 149 U. PA. L. REV. 1439, 1448 (2001); Geetter, *supra* note 3, at 57.

106. DFG, *supra* note 5, at 23; Diver & Cohen, *supra* note 105 at 1452.

107. *See generally* Akerlof, *supra* note 43, at 493.

108. Holmes, *supra* note 6, at 545.

A. The Group Market

Genetic discrimination might occur in several stages of health insurance coverage. The first opportunity to use genetic information is provided in the stage prior to enrollment of an individual in a specific plan. Health insurance issuers could establish rules governing the eligibility of individuals to enroll in a plan, such as the requirement of the individual or a family member of that individual, to undergo a genetic test or the request that genetic test results be provided. Furthermore, health insurance issuers could impose preexisting condition exclusion periods on certain gene-associated diseases. Another possibility would be a raise of the insurance premium of an individual for coverage in a group health plan.

At the end of a plan year, the time of policy renewal, insurers could establish rules for policy renewal based on genetic information with respect to an individual or a family member of such individual. Furthermore, an increase of premium rates for an individual or the group as a whole could occur.

In this context, the disclosure of genetic information by the insurer to an employer or to any other "unauthorized individual" would constitute an example of unfair discrimination against the individual.

B. The Individual Market

In the individual market of health insurance, the above mentioned differentiation between fair and unfair discrimination should be applied. Therefore, in the period prior to enrollment, only a complete denial of health insurance coverage would, if based solely on genetic information, constitute unfair discrimination; this is true because the denial of enrollment based on such information is actuarially unnecessary. While the requirement to undergo a genetic test, to disclose genetic information, or to pay a comparably higher premium amount than the average applicant for the same policy would also constitute genetic discrimination, it would not constitute unfair genetic discrimination if all these requirements have an actuarial justification.

At the time of policy renewal, insurers could demand that the enrollee undergo genetic testing. Based on genetic information derived from such tests, insurers could reassess the premium rate of an individual for continuation of coverage under the policy. Furthermore, insurers could discontinue a particular type of coverage based on the information derived from a genetic test concerning the enrollee or completely deny policy renewal. In theory, the insurer could also terminate the whole policy in the region. Similar to the phase prior to policy enrollment, an insurer has to actuarially calculate the proper premium amounts for the enrollment of an individual under a policy at the time of plan renewal. Only a denial of policy renewal solely based on genetic information concerning that individual or a family member of that individual would not be justifiable by business standards. In order to offset the impact of asymmetric information, insurers have to know the same information

as enrollees. This does not change at the time of policy renewal because both parties, enrollee and insurer, are faced with a new business decision. Therefore, it can also not be considered unfairly discriminatory, if the insurer decides to terminate the whole policy or to completely pull out of the market in a region.

Thus, we conclude the only possibility to unfairly discriminate against individuals who either seek individual health insurance coverage or plan-renewal, is by denying the enrollment in a plan for an actuarially fair premium. As in the group market, the disclosure of genetic information to third parties is not dependent on any actuarial necessity and therefore unfairly discriminatory.

III. HOW DO EXISTING LAWS AFFECT GENETIC DISCRIMINATION IN HEALTH INSURANCE?

It is notable that the laws which relate to genetic discrimination in health insurance so far do not offer a constitutive definition of the term "genetic discrimination." However, in the field of employment discrimination, several laws offer a definition of covered discriminatory behavior. In this Article we will concentrate on the provisions offered by the Americans with Disabilities Act of 1990 (ADA)[109] and Executive Order 13,145 of 2000.[110] Though these two laws primarily deal with employment, their examination might help to pinpoint already prohibited behavior in health insurance through an analogous use in employment discrimination.

A. Provisions Under the ADA

The most discussed law dealing with employment discrimination has been the Americans with Disabilities Act of 1990 which was enacted to secure that individuals with disabilities are able to participate more fully in society.

Although the ADA does not offer a conceptual definition as to what precisely constitutes discrimination, it prohibits discrimination in all the "terms, conditions, and privileges of employment."[111] Thus, the ADA offers a broad based view as to what behavior constitutes employment discrimination against the disabled. In its scope of protection against discrimination, the ADA, according to the regulations issued by the Equal Employment Opportunity Commission (EEOC), encompasses a multitude of employment relationships, including fringe benefits available by virtue of the employment relationship, whether or not administered by the employer. This means that health insurance as a fringe benefit is a protected employment relationship under the

109. 42 U.S.C. §§ 12,101-12,213 (2005).
110. 65 Fed. Reg. 6877 (Feb. 8, 2000).
111. Americans with Disabilities Act of 1990, 42 U.S.C. §§ 12,111-12,117 (2005).

statute.[112] Therefore, discriminatory actions, as defined by the ADA, may not be directed against fringe benefits of employees, such as health insurance.

The question is how this protection relates to genetic information. The ADA itself does not mention "genetic discrimination," but the EEOC included genetic information in its interpretation of the term "disability" under the statute.[113] Under the ADA an individual is considered to be "disabled" if the individual either (1) has a physical or mental impairment which substantially limits one or more of that person's major life activities, (2) has a record of such impairment, or (3) is regarded by the employer as having such an impairment.[114] The EEOC stated in its Compliance Manual, Section 902.8,[115] that "[c]overed entities that discriminate against individuals on the basis of [such] genetic information are regarding the individuals as having impairments that substantially limit a major life activity." This means that any discrimination on the basis of genetic information concerning an individual simultaneously would constitute discrimination on the basis of a disability under the third category listed above.

Although this interpretation is highly debatable,[116] there is neither the room nor the necessity to question the EEOC's interpretation of disability in this article, because it offers the only interpretation that includes genetic information as protected under the statute. In the context of this interpretation, the next question is whether the examples of discriminatory action given by the ADA are of any value for use in health insurance.

Under the ADA, prohibited discriminatory behavior includes, among others, the following: "limiting, segregating, or classifying a job applicant or employee with a disability in a manner that adversely affects the opportunities or status of the individual."[117] This example directly prohibits the use of genetic information in employment and thus simultaneously prohibits the use of genetic information in the employment—related group market of health insurance. In the case of health insurance offered as a fringe benefit, the limitation of an employee's access to fringe benefits on the basis of genetic information would constitute discrimination on the basis of a disability. As mentioned above, this interpretation depends on the applicability of the EEOC's

---

112. Geetter, *supra* note 3, at 37.

113. Jared A. Feldman & Richard J. Katz, *Genetic Testing & Discrimination in Employment: Recommending a Uniform Statutory Approach*, 19 HOFSTRA LAB. & EMP. L.J. 389, 404 (2002); Rachinsky, *supra* note 83, at 591; Richard H. Underwood & Ronald G. Cadle, *Genetics, Genetic Testing, and the Specter of Discrimination: A Discussion Using Hypothetical Cases*, 85 KY. L J. 665, 677 (1997).

114. Feldman & Katz, *supra* note 113, at 402-03.

115. U.S. EQUAL EMPLOYMENT OPPORTUNITY COMM., COMPLIANCE MANUAL § 902 (2000), *available at* http://www.eeoc.gov/policy/docs/902cm.html (last modified Feb. 1, 2000).

116. Steinforth, *supra* note 7, at 999 (noting that courts are not obliged to follow the EEOC's interpretation); Diver & Cohen, *supra* note 105, at 1450; Krumm, *supra* note 7, at 517; Rachinsky, *supra* note 83, at 598; Underwood & Cadle, *supra* note 113, at 678.

117. Americans with Disabilities Act of 1990, 42 U.S.C. §§ 12112(a)–(b)(1) (2000).

interpretation of the term "disability." Nevertheless it offers an example of a discriminatory practice, which is applicable in the group market of health insurance. In contrast to the, at least questionable, application of the ADA on genetic discrimination in health insurance, the Federal Executive Order 13,145 directly deals with genetic discrimination in health insurance.

B. Provisions Under Executive Order 13,145

Executive Order 13,145 was signed on February 8, 2000, by former President Clinton.[118] It restricts the use of genetic information by federal departments and agencies in federal employment relationships. Under section 1-101, it is prohibited to discriminate against an employee with respect to the privileges of employment.[119] Section 1-202(a) and (b) prohibits the segregation, limitation, or classification of employees that would deprive them of their employment opportunities.[120] Furthermore, the employing department or agency shall not request, require, purchase, or collect protected genetic information with respect to an employee, or information about a request for or receipt of genetic services by such employee.[121]

Though originally relating to employment, the provision of section 1-202(c) in connection with section 1-301(b)(4) and section 1-202(a) and (b) entirely prohibits the use of genetic information in health care services other than related to medical treatment, which excludes health insurance.[122] Therefore, under this Executive Order, federal departments and agencies are prohibited from using genetic information in any hiring, promotion, or other employment action, such as health insurance as a fringe benefit. Still, the provisions only apply to employment by federal departments and agencies, which cover only a small fraction of the insured American population.[123]

C. Discussion

Limiting the access of an employee to fringe benefits, such as health insurance, could constitute prohibited discrimination in health insurance against a disabled person under Section 1211 (a) of the ADA. This interpretation depends, however, on the ruling of courts on the EEOC's interpretation of the term "disability." The other provision of the ADA is not usable in the health insurance business because it prohibits the use of common and vital risk—classification methods. The effect of Executive Order 13,145 is limited

---

118. Exec. Order No. 13,145, 65 Fed. Reg. 6877 (Feb. 8, 2000).
119. *Id.* at 6877.
120. *Id.* at 6878.
121. Exec. Order No. 13,145 § 1-202 (c), 65 Fed. Reg. 6877, 6878 (Feb. 8, 2000).
122. Exec. Order No. 13,145, 65 Fed. Reg. 6877, 6878-79 (Feb. 8, 2000).
123. J. Andrew Maniko, *Who ShouldKnow?: The Disclosure Debate Over Genetic Information*, 26 SETON HALL LEGIS. J. 151, 178 (2001).

to employment in federal departments and agencies. Nevertheless, it offers federal employees a broad protection against the use of genetic information in the federal employment-related group market of health insurance.

Even though both laws offer protections against genetic discrimination in the market for employment, they both do not address the market for health insurance directly. Therefore, both laws do not reflect the unique market necessities of health insurance, such as fair discrimination among insurance applicants and protections against asymmetric information. As a consequence, the nondiscrimination provisions of both laws are not, by analogy, transferable to the market for health insurance.

IV. PROVISIONS OF HIPAA AND GINA CONCERNING GENETIC TESTS AND GENETIC INFORMATION IN HEATH INSURANCE

The final step in this paper is to analyze the influence of the Health Insurance Portability and Accountability Act of 1996 (HIPAA)[124] and Genetic Information Nondiscrimination Act of 2003 (GINA)[125] provisions on genetic discrimination in health insurance. It is notable that both laws are based on the assumption that genetic discrimination represents a new form of discrimination, based on a new form of information, i.e. genetic information. Although it is shown this assumption is not sufficiently backed by scientific reality, we will analyze HIPAA and GINA in terms of what is specified in the laws and reflected in their definitions of genetic information. However, as we will show, even based on these assumptions, both laws do not offer comprehensive protection against genetic discrimination. The analysis will deal separately with the group and individual markets.

A. Definition of "Genetic Information" Under HIPAA and GINA

In order to analyze the influence of HIPAA and GINA on genetic discrimination in health insurance, it is imperative to establish what HIPAA and GINA include under the term "genetic information." As we have concluded above in Part I, genetic information can be derived from several sources, including family histories, phenotype analysis, cytogenetic tests, and DNA-tests. Many of these sources are currently used in the diagnosis of diseases in regular medical examinations. Furthermore, as we have shown in Part I,[126] genetic information is not distinguishable from regular medical information. However, in this context, it is important to stress to basic assumption underlying the development of HIPAA and GINA.

---

124. Health Insurance Portability and Accountability Act of 1996, Pub. L. No. 104-191, 110 Stat. 1936 (codified as amended in scattered sections of 18, 19, 16, 42 U.S.C.).

125. Genetic Information Nondiscrimination Act of 2003, S. 1053, 108th Cong. (2003).

126. *See* Scientific Appendix *infra* Part VI.

In contrast to the argument presented in this Article, both laws are based on the concept of genetic discrimination as being something "new," which requires special legal attention. This concept is mirrored in the definitions of "genetic information" employed by HIPAA and GINA. As these definitions are the basis for the statutory provisions, the underlying conceptual assumption is directly expressed in the scope of protection offered by each statute.

1. Definition of "Genetic Information" Under HIPAA

Although HIPAA itself does not include a definition of genetic information, the HIPAA regulations offer a definition. The HIPAA regulations define "genetic information" as information about genes, gene products, and inherited characteristics that may derive from the individual or a family member. This includes information regarding carrier status and information derived from laboratory tests that identify mutations in specific genes or chromosomes, physical medical examinations, family histories, and direct analysis of genes or chromosomes.[127] It is interesting to note that in this definition, genetic information includes information derived from physical medical examinations and family histories. As our earlier explanation of genetic testing methods show, a distinctive differentiation between information derived from genetic tests and information derived from regular medical examinations is not justifiable. The difficulty involved with pinpointing what actually constitutes genetic information seems to be reflected in the definition of genetic information offered in the HIPAA regulations. These regulations employ a definition which effectively equals genetic information to health status information derived from physical medical examinations. Because information derived from physical medical examinations is already protected under the "health status" provision of HIPAA, genetic information automatically is included under this protection. Thus, the inclusion of genetic information under HIPAA only has clarifying value.

2. Definition of "Genetic Information" Under GINA

Under GINA, "genetic information" means information derived from an individual's genetic test, the test of a family member, and the occurrence of a defect or disease in family members of the individual, but excludes information about the sex and age of the individual.[128] Let's quote GINA directly: "Genetic test" means "an analysis of human DNA, RNA, chromosomes,

---

127. *See* 26 C.F.R. § 54.9801-2T (2005); 29 C.F.R. § 2590.701-1(2005); 45 C.F.R. 144.103 (2005).

128. Genetic Information Nondiscrimination Act of 2003, S. 1053, 108th Cong. §§ 101(c), 102(a)(3), 103(c) (2003).

proteins, and metabolites, that detects genotypes, mutations or chromosomal changes."[129] Nevertheless, it does not include

> an analysis of proteins or metabolites that does not detect genotypes, mutations, or chromosomal changes, or an analysis of proteins or metabolites that is directly related to a manifested disease, disorder, or pathological condition that could reasonably be detected by a health care professional with appropriate training and expertise in the field of medicine involved.[130]

This means the definition of "genetic information" under GINA does not include health status information that is directly related to an already manifested disease in an individual or information concerning the sex and age of an individual. Thus, GINA clearly exempts individuals with diseases, whether gene-associated or not, from its nondiscrimination provisions.

B. The Group Market

In the group market for health insurance, both HIPAA and GINA provide protections against genetic discrimination. Both laws amend the following statutes relating to health insurance:[131] the Employee Retirement Income Security Act of 1974 (ERISA),[132] the Public Health Service Act (PHSA),[133] the Internal Revenue Code of 1986[134] and Title XVIII Social Security Act relating to medigap.[135]

1. HIPAA Provisions for the Group Market

The Health Insurance Portability and Accountability Act of 1996 offers nondiscrimination provisions concerning the group market, including small employers.[136] We will divide these provisions into provisions for eligibility

---

129. *Id.* §§ 101(c), 102 (a)(3), 103(c).

130. *Id.* §§ 101(c), 102(a)(3), 103(c).

131. In the next sections we will provide citations to HIPAA and GINA where the specific amendments of both laws to ERISA, PHSA, Internal Revenue Code of 1986 and Title XVIII Social Security Act can be found.

132. Employee Retirement Income Security Act of 1974, Pub. L. No. 93-406, 88 Stat. 829 (codified as amended in scattered sections of 26, 29 U.S.C.).

133. Public Health Service Act, ch. 373, 58 Stat. 682 (1944) (codified as amended in scattered sections of 42 U.S.C.)

134. 26 U.S.C. §§ 1-9833 (2004).

135. Social Security Act, ch. 531, 49 Stat. 620 (42 U.S.C. 301 et seq.), Chapter 7, Subchapter XVIII, Part D, § 1395ss.

136. Health Insurance Portability and Accountability Act of 1996, Pub. L. No. 104-191, 110 Stat. 1936 (codified as amended in scattered sections of 18, 19, 16, 42 U.S.C.), Title 1, Subtitle A.

and provisions relating to nondiscrimination in premium contributions, and discuss them accordingly.

a. Rules for eligibility

The provisions for eligibility are entwined with the rules concerning similarly situated individuals. Insurance issuers are still allowed to treat groups of similarly situated individuals differently. However, they are prohibited from treating the similarly situated individuals inside one group of similarly situated individuals differently from one another.[137] The groups of similarly situated individuals may not be established using health status related factors.[138] Likewise, any differentiation among the group members may neither be based on health status related factors (including genetic information), nor be directed at a specific individual's health status.[139]

The purpose of the statute is a provision of health insurance portability.[140] As such a prohibition of discrimination in (or denial of) enrollment against individuals based on their health status, which includes genetic information as defined in the regulations, is part of the statute.[141] The question relating to genetic discrimination is whether the rules concerning the eligibility have a significant effect as nondiscrimination legislation with regard to genetic information as defined by HIPAA. In relation to genetic information, the provisions of HIPAA concerning the eligibility of individuals to enroll in a group health plan can be summarized as follows:

> 1. The insurance issuers have to apply rules for eligibility uniformly at all the plan members.[142] The insurance issuers are neither allowed to establish groups of high risk based on the health status of the individuals, nor can they to impose preexisting condition exclusions on genetic risks, as these are not regarded as preexisting conditions.[143] Thus, they may not use genetic information as a basis for the eligibility of an individual to enroll in a group health plan.

---

137. Health Insurance Portability and Accountability Act of 1996, Pub. L. No. 104-191, 110 Stat. 1936 (codified as amended in scattered sections of 18, 19, 16, 42 U.S.C.), Title 1, Subtitle A, Part 1 §§ 101, 102; Title 4, Subtitle A, § 401.

138. *Id.*

139. *Id.*

140. *See* the preamble to the Health Insurance Portability and Accountability Act of 1996, Pub. L. No. 104-191, 110 Stat. 1936 (codified as amended in scattered sections of 18, 19, 16, 42 U.S.C.).

141. *Id.*

142. *Id.*

143. Health Insurance Portability and Accountability Act of 1996, Pub. L. No. 104-191, 110 Stat. 1936 (codified as amended in scattered sections of 18, 19, 16, 42 U.S.C.), Title 1, Subtitle A, Part 1, §§ 101, 102; Title 4, Subtitle A, § 401.

> 2. Nevertheless, insurance issuers are still able to restrict the amounts of benefits or cancel the coverage for certain medical conditions, if they apply these restrictions uniformly on a group of similarly situated individuals.[144] The only exception might be that a restriction was being directed at specific individual participants in the plan; however, this would have to be established.
> 3. Since there is no mandatory minimum benefit or coverage package provided by the statute or the regulations, insurance issuers are free to place a cap on benefits or coverage relating to genetically identifiable diseases, as long as the benefit or coverage or coverage in question is denied to the whole group.[145]

Although a genetic predisposition to a disease may not be excluded from coverage as a preexisting condition, this provision is open to debate. Since insurers are still allowed to exclude the coverage of certain diseases for the whole group, a preexisting condition exclusion for a specific disorder, which would only be in force for twelve months, seems less favorable for the insurers. Thus, in the context of this provision, the provision against preexisting condition exclusions based on genetic information seems irrational from a clinical or legal perspective. This is even more apparent if the insurance industry is faced with late-onset diseases, such as Alzheimer's, which is unlikely to occur during the first twelve months of coverage. Therefore, a preexisting condition exclusion concerning genetic information can be meaningless, since the insurer could exclude coverage for a specific disorder in a renewal decision, as long as it applies to the whole group.[146] Considering the above findings, the HIPAA provisions relating to the eligibility to enroll provide no safety against genetic discrimination to insurance applicants, other than the guarantee of enrollment in a health insurance plan with uncertain coverage.

b. Nondiscrimination in premium amounts

The provisions relating to the prohibition of discrimination in setting premium rates stipulate that an individual may not be charged more than the other enrollees in the same group of similarly situated individuals based on any

---

144. *Id.*

145. *See* 26 C.F.R. § 54.9802-1T(b)(2)(i)(B) (2005); 29 C.F.R. § 2590.702(b)(2)(i)(B) (2005); 45 C.F.R § 146.121(b)(2)(i)(B) (2005).

146. *See* 26 C.F.R. § 54.9802-1T(b)(2)(i)(B); 29 C.F.R. § 2590.702(b)(2)(i)(B); 45 C.F.R § 146.121(b)(2)(i)(B).

health status related factor of the individual or a dependent of the individual.[147] Furthermore, the HIPAA interim final regulations clarify, that list-billing (billing with separate individual premium rates) practices are prohibited.[148] This serves as a protection of small groups, containing less than twenty-five to fifty employees, against individual insurance underwriting, and as such against the use of genetic information in the underwriting process.

Nevertheless, HIPAA does not restrict the amount an insurance issuer may charge an employer for coverage of a group of similarly situated individuals.[149] Consequently, insurance issuers are still able to charge employers higher premium rates based on the claims-experience of the individuals inside the group. Therefore, the insurance issuers could still use an estimate of genetic predispositions in a group based on a group's claims experience in order to adjust the price an employer has to pay for coverage of a group.

A real protection against higher premium rates based on health factors and especially genetic information is not achieved, because insurance issuers still can reflect a negative claims experience in higher premium amounts, not directed against the "cause," the actual individual requiring expensive treatment, but instead directed against the group as a whole. Thus, HIPAA only shifts the burden of high medical expenses of individuals to the whole group.

Taking the above findings into consideration, it is arguable that no comprehensive protection against discrimination based on genetic information is offered by HIPAA in the group market of health insurance. The prohibition of underwriting for small groups might be regarded as a protection against discrimination in premium amounts. However as we have explained earlier in Part I, small groups with less than twenty-five employees are usually too small to sustain a risk-spreading system and qualify for experience rating. This legal obligation to experience rate in the small group market forces insurance issuers to set premium rates according to the past claims experience of groups, which are too small to sufficiently compensate high medical expenses by the group's premium pool. In order to keep the policies profitable, insurers would either have to raise the group's premium rates, which is still allowed under HIPAA, or discontinue offering health insurance coverage for small groups. Thus, insurance for small groups would become either very expensive or unavailable. Although the prohibition of list-billing in the small group market might be regarded as being a provision against discrimination in premium amounts,

---

147. Health Insurance Portability and Accountability Act of 1996, Pub. L. No. 104-191, 110 Stat. 1936 (codified as amended in scattered sections of 18, 19, 16, 42 U.S.C.), Title 1, Subtitle A, Part 1, §§ 101, 102, Title 4, Subtitle A, § 401.

148. 26 C.F.R. § 54.9802-1(c)(2)(ii) (2005); 29 C.F.R. § 2590.702 (c)(2)(ii) (2005); 45 C.F.R. § 146.121 (c)(2)(ii) (2005).

149. Health Insurance Portability and Accountability Act of 1996, Pub. L. 104-191, 110 Stat. 1936 (codified as amended in scattered sections of 18, 19, 16, 42 U.S.C.), Title 1, Subtitle A, Part 1, §§ 101, 102, Title 4, Subtitle A, § 401.

it should instead be seen as what it is: a menace to the stability of the small group market of health insurance.

2. GINA Provisions for the Group Market

Following basically the same conception of genetic information as HIPAA,[150] GINA enhances the protections given by HIPAA. GINA prohibits employer-sponsored group health plans and health insurance issuers offering health insurance in connection with a group health plan by doing the following:

> (1) Imposing enrollment restrictions based on genetic information (which includes the request for or receipt of genetic services by an individual or a family member of such individual).[151]
>
> (2) Adjusting premiums or contribution amounts for a group on the basis of genetic information of an individual in the group or a family member of such an individual.[152]
>
> (3)Requesting or requiring an individual to undergo a genetic test.[153]

In sum, GINA specifies that even the request for or receipt of genetic services by an individual, which might hint at a predisposition, is considered to be genetic information and is therefore prohibited from serving as a basis for the application of enrollment restrictions. It is safe to conclude the new provision by GINA only clarifies the comprehensiveness of protection of "genetic information," since HIPAA already restricts the eligibility of an individual for group health insurance coverage based on genetic information.

The third provision concerning the request for, or requirement, of a genetic test as a condition for enrollment, is also little more than a clarification. As the request for, or requirement, of a genetic test per definition leads to genetic information,[154] the HIPAA-provision concerning the restriction of eligibility already applies. Although the possibility of a request for, or requirement, of a genetic test might be used by health insurance issuers for other purposes (that is other than the determination of the eligibility of an individual

---

150. *See* discussion *infra* Parts IV.A.1, IV.A.2.

151. Genetic Information Nondiscrimination Act of 2003, S. 1053, 108th Cong., §§ 101(a)(1), 102(a)(1)(A), 103(a)(1) (2003).

152. *Id.* §§ 101(a)(2)(B), 102(a)(1)(B)(ii), 103(a)(2)(B).

153. *Id.* §§ 101(b), 102(a)(2), 103(b).

154. As defined in GINA (*See* discussion *supra*, Part IV.A.2.) and, though slightly different, in the interim final regulations concerning HIPAA (*See* discussion *supra*, Part IV.A.1.).

to enroll in a plan), this remaining interest in genetic tests or the results thereof is already covered by the second provision.

In this second provision, GINA fills a gap left by HIPAA concerning the calculation of premium rates for the groups of similarly situated individuals. Under the HIPAA provisions, the insurance issuers were still allowed to adjust the premium rates for a group as a whole, and to consider all relevant health factors in order to establish aggregate rates for coverage provided under a group health plan. This could lead to unfair discrimination against the healthy group members;[155] consequently GINA prohibits this kind of discrimination against the group. In other words, the only new anti-discrimination provision introduced by GINA in the group market of health insurance is the prohibition of discrimination against a group as a whole based on genetic information concerning an individual in the group.

C. The Individual Market

As is true for the group market, both HIPAA and GINA offer protections against genetic discrimination in the individual health insurance market.

1. HIPAA Provisions for the Individual Market

HIPAA amends the provisions of the Public Health Service Act concerning the individual market of health insurance[156]. We will divide the HIPAA provisions into: (1) provisions for eligible individuals, and (2) provisions for all individuals seeking health insurance coverage in the individual market of health insurance.

a. Provisions for eligible individuals

The central provision of HIPAA concerning the individual health insurance market is a provision for "eligible"[157] individuals only. It states that

---

155. *See* discussion *supra*, Part IV.B.1.

156. Health Insurance Portability and Accountability Act of 1996, Pub. L. 104-191, 110 Stat. 1936 (codified as amended in scattered sections of 18, 19, 16, 42 U.S.C.), Title 1, Subtitle B, § 111.

157. Eligibility:

> The individual is eligible if it had at least eighteen months of creditable coverage and its last coverage was under a group health plan, governmental plan or church plan. Furthermore, the individual may not be eligible for coverage under a group health plan, Medicare or Medicaid, and may not have other health insurance coverage (as defined in the Health Insurance Portability and Accountability Act of 1996, Pub. L. 104-191, 110 Stat. 1936 (codified as amended in scattered sections of 18, 19, 16, 42 U.S.C.), Title 1, Subtitle B, § 111).

Creditable coverage:

> The individual's previous coverage has to be creditable coverage as defined in 45 C.F.R. § 146.113 (2005).

insurance issuers have to offer eligible individuals coverage and enrollment in insurance policies actively marketed in the individual health insurance market.[158]

According to the interpretation in the HIPAA regulations,[159] a health insurance issuer meets these requirements, if it provides information about all available options, enrolls the individual in any coverage the individual selects and if it does not impose preexisting condition exclusions on the individual. Nevertheless, insurance issuers are still free to set premium rates for individual health insurance coverage for eligible individuals.[160] Thus, the basic provision for eligible individuals consists of guaranteed health insurance issuance without the possibility of being subjected to a preexisting condition exclusion.

In the context of genetic testing and genetic information, this means that eligible individuals have to be offered individual health insurance, and be enrolled in a plan of their choice, regardless of their genetic makeup.

Still, the regulations do not restrict the amount an issuer may charge for enrollment under the policy. Furthermore, the regulations only implement a restriction on preexisting condition exclusion periods, which do not apply to genetic information, as genetic information does not constitute a preexisting condition under the statute and the regulations.[161] Nevertheless, genetic testing as a condition for enrollment is prohibited, because that would be contrary to the main purpose of the regulations; which is to guarantee individual health insurance coverage portability through guaranteed access.

Insurance issuers still can base the premium rates for insurance coverage of eligible individuals according to their individual risk, and still are allowed to do so by demanding and using genetic information. This could offer health insurance issuers a possibility to protect themselves from adverse selection.

The most important groups of creditable coverage are the following: coverage under a group health plan as defined in 45 C.F.R. § 144.103 (2005), health insurance coverage as defined in 45 C.F.R. § 144.103, and Medicare and Medicaid. This means that creditable coverage not only is group health coverage but also any health insurance coverage, which is, according to 45 C.F.R. § 144.103, benefits consisting of medical care under any hospital or medical service policy or certificate, hospital or medical service plan contract, or HMO contract offered by a health insurance issuer. Thus, also individually purchased health insurance coverage, which is health insurance coverage not offered in connection with a group health plan, 45 C.F.R. § 144.103, is creditable coverage under 45 C.F.R. § 146.113. Following 45 C.F.R. § 148.103(2) (2005), only the most recent prior creditable coverage of the individual must have been either under a group health plan, governmental plan or church plan (or health insurance coverage offered in connection with any of these).

158. Health Insurance Portability and Accountability Act of 1996, Pub. L. 104-191, 110 Stat. 1936 (codified as amended in scattered sections of 18, 19, 16, 42 U.S.C.), Title 1, Subtitle B, § 111.

159. *See* 45 C.F.R. § 148.120(a)(1) (2005).

160. *See* 45 C.F.R. § 148.120(g)(4).

161. See 45 CFR § 148.120(a)(1) in connection with the Health Insurance Portability and Accountability Act of 1996, Pub. L. 104-191, 110 Stat. 1936 (codified as amended in scattered sections of 18, 19, 16, 42 U.S.C.), Title 1, Subtitle A, Part 1, § 102(a).

Because the regulations do not impose restrictions on the pricing of the policies, this could still be done by the states. There is no preemption of state legislation regarding this dimension.

However, states are allowed to evade the provisions for eligible individuals by implementing an alternative mechanism.[162] This alternative mechanism has to provide for a guaranteed offer of health insurance coverage to all eligible individuals, and for a prohibition of preexisting condition exclusion or affiliation periods for coverage of eligible individuals.[163] Nevertheless, the implementation of an alternative mechanism by a state actually does not limit the possibilities of the insurance issuers to set premium rates according to individual risk.[164]

If a state does not implement such an alternative mechanism, an insurance issuer may elect to limit the coverage required under the regulations[165] if it offers eligible individuals at least two policy forms, which must meet certain requirements.[166] As 45 C.F.R. § 148.120(g)(4) clarifies, issuers making this election are still able to adjust premium rates according to the individual risk, and thus based on genetic information, of eligible individuals.[167] Nevertheless, the premium adjustments have to be consistent with applicable state law.

b. Provisions for all individuals

According to 45 C.F.R. § 148.122, insurance issuers in the individual market are required to renew the health insurance coverage of all individuals at the option of the individual.[168] Thus, a health insurance issuer may only discontinue a particular type of coverage, if it provides notice in writing ninety days prior to the termination of the coverage and offers individuals the option to purchase any other individual health insurance currently offered in the state.[169] Furthermore, the insurance issuers have to act uniformly without regard to health status related factors of the individuals or their covered dependents.[170]

Insurance issuers are still permitted to modify a health insurance coverage policy form at the time of renewal of coverage if it applies the

---

162. This alternative mechanism is described in 45 C.F.R. § 148.128 (2005).

163. *See* 45 C.F.R. § 148.128(a)(1).

164. 45 C.F.R. § 148.120(g)(4).

165. 42 C.F.R. § 148.120(a).

166. *Id.* § 148.120(c). Each policy must be designed for, marketed to and enroll all individuals. The policies must be either the two most popular policy forms as described in § 148.120(c)(2), or representative samples of individual health insurance offered by the issuer in the State, as described in § 148.120(c)(3).

167. *Id.* § 148.120(g)(4).

168. 45 C.F.R. § 148.122 (2005).

169. 45 C.F.R. § 148.122(d)(1), (2).

170. 45 C.F.R. § 148.122(d)(3).

modification uniformly for all individuals with that policy form. In addition, this modification has to be consistent with applicable state law.[171]

This means that all individuals are guaranteed renewal of their individual health insurance coverage, and individual level genetic information may not be used in this decision. Furthermore a discontinuation of coverage may not be based on the individual's, or a covered dependent of the individual's, genetic information. An insurance issuer can still modify the offered coverage based on genetic information, if it acts uniformly for all individuals with that policy form, and the modification is consistent with state law.

As long as a state does not enact laws prohibiting the adjustment of premiums based on genetic information, genetic information and testing may still be used to calculate and adjust premium amounts for non-eligible individuals in that state. Furthermore, genetic testing and information derived from such tests may be used by issuers to restrict and condition enrollment of non-eligible individuals.

2. GINA Provisions for the Individual Market

The possible uses of genetic information allowable under HIPAA, as specified above,[172] are significantly reduced by GINA. GINA stipulates that health insurance issuers offering health insurance coverage in the individual market may not require an individual to undergo a genetic test, may not establish rules for the eligibility or continued eligibility to enroll based on genetic information, and may not adjust premium or contribution amounts based on genetic information.[173] This means eligible individuals, as defined under HIPAA, may not be charged different premiums based on their genetic information (or that of a family member), as was still permitted under HIPAA.[174]

Noneligible individuals may not be subjected to enrollment restrictions based on their genetic information (or that of a family member), or be subjected to premium adjustments based on their genetic information (or that of a family member), and neither may be required to undergo a genetic test prior to enrollment.[175] Therefore noneligible individuals may still be denied enrollment based on other than genetic information (as defined under GINA).

Eligible individuals, as defined under HIPAA, still have guaranteed access to health insurance coverage in the individual market. The pricing of the individuals' coverage, which was still adjustable under HIPAA, may not

---

171. 45 C.F.R. § 148.122(g).

172. See *supra*, Part IV.C.1.a. and b.

173. Genetic Information Nondiscrimination Act of 2003, S. 1053, 108th Cong., Title 1, §§ 102(b)(1)(B), 104(a)(1)(b)(1) (2003).

174. *See supra*, Part IV.C.1.a.

175. Genetic Information Nondiscrimination Act of 2003, S. 1053, 108th Cong., Title 1, §§ 102(b)(1)(B), 104(a)(1)(b)(1) (2003).

be adjusted on the basis of genetic information concerning that individual (or a family member of the individual).[176]

D. Summary of the Offered Protections in Light of Unfair Genetic Discrimination

In summary, the results of the analysis of the provisions of HIPAA and GINA concerning unfair discrimination in the group market of health insurance are:

> - HIPAA guarantees enrollment in group health plans with uncertain coverage;
> - Under HIPAA, insurance issuers can still reflect a negative claims experience of a group in setting higher premium rates for the whole group;
> - No real protection against unfair discrimination based on genetic information is offered by HIPAA for the group market, except for the prohibition of underwriting for small groups, which, as explained above, does not constitute unfair genetic discrimination as underwriting is necessary in the small group market;
> - GINA prohibits the discrimination against a group as a whole based on genetic information concerning an individual in the group.

In summary, the results of the analysis of the provisions of HIPAA and GINA concerning unfair genetic discrimination in the individual market are:

> - HIPAA provides that eligible individuals are guaranteed health insurance coverage, regardless of genetic information concerning them or their family members;
> - Under HIPAA, all individuals are guaranteed an offer of policy renewal;
> - Under HIPAA, insurance issuers are still able to calculate their premiums according to genetic information concerning eligible individuals, as long as the states do not enact contrary legislation and policies are ultimately issued;
> - The implementation of an alternative mechanism through a State only restricts issuers in the premium calculation process, thereby forcing them to subsidize high-risk individuals;

---

176. Genetic Information Nondiscrimination Act of 2003, S. 1053, 108th Cong., Title 1, §§ 102(b)(1)(B), 104(a)(1)(b)(1) (2003).

- The election to limit coverage under 45 C.F.R. § 148.120(c) actually leads to adverse selection problems;
- HIPAA limits the use of genetic information and genetic testing by health insurance issuers to the determination of premium rates and to the determination of the enrollment of noneligible individuals;
- Under HIPAA, the states are left to legislate insurance issuer's use of genetic information;
- Under GINA, noneligible individuals may still be denied enrollment based on factors other than genetic information;
- The pricing of the individual's coverage, under GINA may not be adjusted on the basis of genetic information concerning that individual or a family member of the individual.

V. CONCLUSION

The current legal and public policy approach for dealing with "genetic discrimination" is fundamentally flawed. As we have shown there is no reason to treat genetic information derived from genetic tests different from information derived from regular medical examinations. Moreover, we have shown that the two types of information are not readily distinguishable from each other. Yet the information derived from regular medical examinations has a different legal status.

GINA, in combination with HIPAA, provides broader protection for genetic information than for information derived from regular medical examinations. This effect matches the Greely's proposal, who proposed that the possibility of an individual to sue for genetic discrimination on the ground that a manifested disease provided evidence about a genotype might be prevented.[177] Because a probative basis for this differentiation does not exist, we have to conclude that HIPAA and GINA, as they provide for a differentiation between genetic information and regular medical information, are laws merely reacting to fears in the American public,[178] which were fueled by a biased view of the underlying science.

Taking the above analysis into consideration, we conclude that GINA prohibits the use of genetic information in a market where the availability of such information is crucial for health insurers to offset the impact of asymmetric information. GINA fails to acknowledge the necessity of underwriting through fair discrimination in the individual and small group health insurance

177. Greely, *supra* note 64, at 1503.

178. *See, e.g.*, Epstein, *supra* note 72, at 3 (stating that "[t]he dominant attitude . . . is strongly hostile to genetic discrimination. . . ."); Diver & Cohen, *supra* note 105, at 1448-49.

market. Thus, GINA endangers the stability of these markets,[179] especially in the small group market. Furthermore, the different treatment of presymptomatic and symptomatic conditions is incomprehensible in view of gene-associated diseases. Yet, the current legal approach would allow for such differentiation.

In the individual health insurance market, HIPAA provides guaranteed enrollment for eligible individuals, regardless of their health status. Although under HIPAA, the possibilities to calculate actuarially fair premiums according to genetic information concerning all individuals, as well as to determine the eligibility of noneligible individuals to enroll in a policy according to genetic information concerning these individuals are left to health insurers, both possibilities are prohibited by GINA. Thus, the insurers' possibility to calculate actuarially fair premium rates is limited to information about the health status of individuals that is not derived from genetic information as defined by GINA. According to the definition of genetic information of GINA, this means that insurers are allowed to calculate premiums only based on information about already manifested diseases.

Furthermore, contrary to our findings concerning unfair discrimination in the small group market of health insurance, HIPAA prohibits list billing practices in this market. Thereby, HIPAA prohibits the possibility of insurers to use underwriting in a market where risk-spreading alone is insufficient to keep the group profitable, and thus, insurable. Because of the guaranteed enrollment provision, small groups do not have to fear that insurers would deny them enrollment. Nevertheless, insurers who are confronted with the mandatory enrollment of individuals in unprofitable small group plans will have an incentive to limit the coverage offered in connection with small group health plans in order to keep the plans profitable. This might ultimately lead to a decrease in the level of insurance coverage for small groups, as the extent of coverage is not guaranteed by HIPAA. As mentioned above, both HIPAA and GINA fail to resolve this problem. Instead both statutes prohibit the underwriting practices, which are necessary for this market segment.

HIPAA provides for the states and insurance companies offering health insurance coverage in the individual market the possibility to prevent the application of the requirements and provisions for eligible individuals. However, the offered methods are undesirable for the health insurers. On the one hand, the implementation of an alternative mechanism by a state forces insurers to either subsidize eligible individuals through a subsidization system between different policies or to subsidize each other in order to cover the expectable higher costs. The possibility for the insurance issuers to limit the required coverage leads to adverse selection problems and is therefore also not desirable for them.

---

179. *See, e.g.*, Diver & Cohen, *supra* note 105, at 1457 (fearing that "adverse selection and favorable deselection could literally tear the individual health insurance market apart").

In the group market, the gap left by HIPAA concerning the possibility of health insurance issuers to limit the level of coverage offered for a group health plan remains open. Because insurers are not allowed to react to higher claims experiences of a group by raising premium amounts, they might instead restrict the amounts of benefits offered in connection with that group health plan. GINA tries to differentiate clearly between information about genotypes and information about (gene-associated) manifested diseases. Although the former is protected from insurers use, the latter is not. Because of this differentiation, GINA does not cover the possibility that the genotypes translate into diseases. Despite the protection of genotype related information, GINA fails to prohibit the use of information about the diseases that result from these genotypes. Consequently, GINA fails to protect against the possibility of unfair discrimination because of gene-associated diseases. This documents the failure to employ an all-inclusive definition of genetic information.

Genetic information is inseparably intertwined with information derived from regular medical examinations.[180] Since many diseases are caused by the interaction of genetic predispositions and environmental factors, this leads to difficulties in distinguishing between discrimination which is based only on genetic predispositions and discrimination that is based on diseases which are at least partly caused by such predispositions. Why discrimination based on an individual's genotype should be protected and discrimination based on an individual's (gene-associated) already manifested disease, should not be, is incomprehensible. Because the limitation of coverage for group health plans, based on higher claims for gene-associated diseases constitutes unfair genetic discrimination, this type of discrimination should be prohibited. Unfortunately neither HIPAA nor GINA provides the necessary protection.

Overall, the main flaw of both HIPAA and GINA is the assumption that genetic information and thus genetic discrimination is different from discrimination based on regular medical examinations. First, the methods used in getting the information are not fundamentally different for genetic and regular medical examinations. Second, the quality of information does not differ fundamentally because the only suggested distinguishing feature, the better predictability of gene-associated diseases, is usually unreliable. Discrimination based on the possibility of the development of a disease—as shown—is less intrusive or stigmatizing than discrimination based on already manifest diseases. Third, by prohibiting the use of genetic information, HIPAA and GINA systematically neglect the information requirements of the individual and small group health insurance market and thereby endanger the availability and affordability of these health insurance market segments.

GINA in combination with HIPAA only provides for two protections against genetic discrimination. First, GINA fills the gap left open by HIPAA

---

180. *See* Mark A. Rothstein, *Why Treating Genetic Information Separately is a Bad Idea*, 4 TEX. REV. L. & POL. 33, 34-35 (2000).

concerning the calculation of premiums of the whole group based on genetic information concerning an individual inside the group. Second, in the group market of health insurance, the use of genetic information is effectively banned in enrollment and premium calculation. These two provisions correspond to the view which we adopted earlier in this paper. In the group market of health insurance genetic information serves no actuarial purpose. Therefore, any use of genetic information in this market should be considered to be unfairly discriminatory. Unfortunately, these two protections are the only parts of HIPAA and GINA that are comprehensibly directed against unfair discrimination in health insurance.

Many new technological developments encompassing the Human Genome Project, such as gene-chips and microarrays, will offer great possibilities for diagnosis and treatment of diseases. Newborn screening, somatic cell therapy, and ultimately the field of pharmacogenomics show tremendous progress towards improving health care.

As "pharmacogenomics holds the promise that drugs might one day be tailor-made for individuals and adapted to each person's own genetic makeup,"[181] genetic information about an individual will be crucial in order to reach this goal. Unfortunately, the current legal environment counteracts the achievement of this goal by blocking access to genetic information and by fueling unjust fears of genetic discrimination.

In contrast to our findings relating to the impact of HIPAA and GINA on "genetic discrimination" in health insurance, we think that public policy should welcome this technological progress and promote the offered benefits to the American public instead of focusing on the creation of a negative view of genetics throughout society. Consequently, laws should not be oriented toward blocking society from receiving the benefits these new technological developments offer.

VI. SCIENTIFIC APPENDIX

Blotting Techniques

Until the invention of the Polymerase Chain Reaction, the analysis of DNA sequence polymorphisms relied mostly on "restriction fragment length polymorphisms" (RFLP)[182] combined with the Southern blot technique. The technique uses local changes of the DNA-sequence, so called polymorphisms, which are the product of mutations, such as nucleotide (nucleic acid building

181. Human Genome Project Information, Medicine: Pharmaceuticals, *at* http://www.ornl.gov/sci/techresources/Human_Genome/medicine/pharma.shtml (last visited Apr. 16, 2005) (on file with the Indiana Health Law Review).

182. THOMAS D. GELEHRTER & FRANCIS S. COLLINS, PRINCIPLES OF MEDICAL GENETICS 309 (1990); PETER J. RUSSELL, GENETICS 488 (4th ed. 1996).

block) deletions, insertions or pointmutations.[183] High-molecular-weight DNA is cut with enzymes recognizing a certain nucleotide sequence (restriction enzymes).[184] These enzymes produce DNA fragments of differing lengths, depending on the locations of the different restriction sites on the DNA molecule.[185] These DNA fragments are then transferred onto a membrane filter by blotting and visualized, using radioactive or fluorescent probes.[186] RFLPs will always be visible, if they are located at the restriction sites recognized by the restriction enzymes employed.[187] A disease-associated RFLP allows the direct identification of a particular genetic defect.[188]

A different blotting technique analyzes ribonucleic acid (RNA), instead of DNA molecules. This technique is called Northern blot analysis. In this technique, RNA is extracted from a cell and separated by size through gel electrophoresis.[189] Then, the RNA molecules are transferred and bound to a filter in a procedure which is analogue to Southern blotting.[190] After the hybridization with a labeled probe, bands indicate the locations of RNA species that were complementary to the probe.[191] Northern blot analysis can be used to determine, for example whether a gene is transcribed (copied into RNA), or whether the RNA is correctly spliced (fragmented and pieced together) so as to give rise to an RNA species (messenger RNA) which serves the synthesis of proteins within a cell. Both types of blotting procedures are, however, fairly laborious, and will only be used in special cases for disease gene identification.

Chromosomal Aberrations

Chromosomal aberrations are divided into numerical aberrations, such where the number of chromosomes has changed, and structural aberrations, where the structure of one or more chromosomes is altered.[192] Chromosomal defects usually lead to characteristic clinical symptoms, such as Down-Syndrome,[193] the triple occurrence of chromosome 21 within the genome (the entirety of the genetic information) of an individual.[194]

---

183. RUSSELL, *supra* note 182, at 593.
184. *Id.* at 480.
185. *Id.* at 481.
186. *Id.*
187. *Id.* at 488.
188. *Id.*
189. RUSSELL, *supra* note 182, at 481.
190. *Id.*
191. *Id.*
192. DFG, *supra* note 5, at 16.
193. RONALD W. DUDEK, EMBRYOLOGY 258 (2d. ed. 1998).
194. *Id.*

Cytogenetic Analysis

Examinations on the chromosomal level (cytogenetic analysis) aim at discovering anomalies in the number, size, and form of the human chromosomes.[195] This is done through the light-microscopical analysis of so called karyotypes, a complete set of all the metaphase chromosomes in a cell.[196] Through chromosomal analysis, it is possible to detect chromosomal anomalies not only in an individual but also through prenatal diagnosis performed on embryo-derived cells during pregnancy.[197] Chromosome-analysis helps in diagnosing or verifying diagnoses of different hereditary diseases, such as Down-Syndrome (trisomy 21).

Monogenic Diseases

Monogenic diseases are caused by defects affecting a single gene.[198] Except for mitochondrial mutations,[199] monogenic defects are inherited according to Mendelian rules.[200] Diseases due to a monogenic defect are relatively rare, though differences between ethnic groups may occur.[201] Nevertheless, monogenic diseases can be modulated by other genes or environmental factors.[202] This category of diseases may be divided into "autosomal dominant," "autosomal recessive," "x-chromosomal," "Y-chromosomal," and "mitochondrially inherited" diseases.[203] Examples of monogenic diseases are polydactyly, an autosomal dominant disorder, which is characterized by the presence of extra digits on the hands or feet, and fragile X syndrome, which is an X-linked recessively inherited disease, and characterized by moderate mental retardation.[204]

Phenotype-Analysis

The first level of gene-analysis is the examination of an individual's phenotype.[205] Because the phenotype is the outcome of genetic and exogenous factors, such as environmental circumstances, it is possible to reach conclusions concerning certain genetic traits of an individual by looking at the

---

195. RUSSELL, *supra* note 182, at 593.
196. *Id.* at 328; TAEUSCH ET AL., *supra* note 52, at 104.
197. RUSSELL, *supra* note 182, at 286-87.
198. DFG, *supra* note 5, at 16; Holmes, *supra* note 6, at 527.
199. Andreas Ziegler, *Basic Mechanisms of Monogenic Inheritance*, 40 EPILEPSIA 4, 7 (Supp. 3 1999).
200. *Id.* at 6.
201. *See, e.g.*, Gostin, *supra* note 7, at 137; DFG, *supra* note 5, at 16.
202. DFG, *supra* note 5, at 24.
203. Ziegler, *supra* note 199, at 6.
204. DUDEK, *supra* note 193, at 259-60.
205. GELEHRTER & COLLINS, *supra* note 182, at 307.

physical characteristics, to which the genotype decisively contributes. The analysis itself does not require special techniques other than an examination of an individuals' family history or clinical methods already used in regular medical examinations, such as blood-pressure, blood-sugar, urine, or cholesterol-analysis, as well as ultrasonography and x-ray, which can all serve as highly specific indicators of genetic defects.[206]

Polygenic-Multifactorial Diseases

Polygenic or multifactorial diseases, for example Neural tube defects or cardiovascular anomalies,[207] are caused by an often complex interplay of one or more genetic defects and environmental factors.[208] The importance of genetic factors in the development of multifactorial diseases is usually not fully understood. Nevertheless, it is possible to estimate the contribution of genetic defects through studies using identical twins.[209] The concordance-rates encountered in such studies are a measure for the relative contribution of genetic factors in the development of diseases.[210] Usually the concordance-rates for multifactorial diseases in identical twin studies are between forty and sixty percent.[211] This means that the remaining percentage must be due to nongenetic factors. Furthermore, even with all genetic factors identified that are associated with a multifactorial disease, the susceptibility to that particular disease will only have a probabilistic quality. It is only possible to predict a relative risk for the carrier of the genetic risk factors. Because it is impossible to determine all nongenetic factors, such as environmental influences, the predictability will never be higher than the concordance-rates of identical twin studies.[212] Therefore, the idea of a genetic determinism has to be abandoned.

Polymerase Chain Reaction (PCR)

The PCR-technique, developed by Kary B. Mullis in 1983, quickly advanced to become the single most important technique in genetic sciences because it allows the amplification[213] of small amounts of DNA (one molecule is sufficient) for the subsequent analysis, e.g. by sequencing.[214] But the PCR itself can also be used to detect mutations, such as point mutations, insertions,

206. DFG, *supra* note 5, at 12.
207. DUDEK, *supra* note 193, at 260.
208. Holmes, *supra* note 6, at 528; RUSSELL, *supra* note 182, at 755.
209. RUSSELL, *supra* note 182, at 126.
210. DFG, *supra* note 5, at 19.
211. *Id.*
212. DFG, *supra* note 5, at 21.
213. GELEHRTER & COLLINS, *supra* note 182, at 299.
214. J. Christopher Post & Garth D. Ehrlich, *The Impact of the Polymerase Chain Reaction in Clinical Medicine*, 283 JAMA 1544 (2000).

or deletions in DNA.[215] In PCR, a primer,[216] which may end directly at the location of a suspected mutation, is designed on the basis of a known DNA or RNA nucleotide sequence.[217] First, DNA is denaturated during a heating process into two single strands.[218] Next, the primers are allowed to bind to single strand target DNA in a process called "annealing."[219] Then, in a process called elongation, the primers are extended by an enzyme (Taq DNA polymerase), which synthesizes a DNA strand complementary to that already present.[220] Thus, the DNA fragment is copied for the first time. All three steps are then repeated, usually for about thirty cycles.[221] If the primer is homologous to the unmutated (wild-type) DNA sequence,[222] an amplification of only the wild-type DNA in conjunction with another gene-specific primer on the gene will occur.[223] As the wild-type primer will not amplify mutant DNA, likewise, the mutant primer will only amplify mutant DNA.[224] Using PCR with both sets of primers in separate reactions, the presence of a mutation sequence is now determinable.[225]

Protein-Chemical Analysis

The protein-chemical analysis is an examination method, by which one tries to discover gene-associated diseases linked to the presence, absence or alteration of proteins, which can lead to metabolic defects, as encountered in phenylketonuria.[226] Furthermore, it is possible to prenatally diagnose neural tube defects by the presence of elevated alpha-fetoprotein levels in the maternal amniotic fluid.[227] Though in this type of analysis, it is unnecessary to know the involved gene or chromosome, the use of this technique is restricted by the fact that the proteins which are to be examined, must be available.[228]

---

215. Bruce Korf, *Molecular Medicine: Molecular Diagnosis, Part 2*, 332 NEW ENG. J. MED. 1499 (1995).
216. RUSSELL, *supra* note 182, at 360.
217. Post & Ehrlich, *supra* note 214, at 1545.
218. *Id.* at 1545.
219. *Id.*
220. *Id.*
221. *Id.*
222. RUSSELL, *supra* note 182, at 69-70.
223. Korf, *supra* note 215, at 1499.
224. *Id.*
225. *Id.* at 1500.
226. Greely, *supra* note 64, at 1485.
227. DUDEK, *supra* note 193, at 260.
228. RUSSELL, *supra* note 182, at 286.

Sequencing Techniques

In DNA sequence analysis, cloned DNA fragments are analyzed to determine the nucleotide pair sequence of the DNA.[229] Gene sequencing is useful for identifying and controlling gene sequences in the cloned DNA fragment.[230] Two techniques for the rapid sequencing of DNA molecules were developed in the 1970s, the Maxam-Gilbert sequencing technique and the Sanger sequencing technique.[231] The Maxam-Gilbert method uses chemicals to cleave the DNA chain at specific nucleotides.[232] By contrast, the Sanger method uses a procedure in which the synthesis of new DNA strands is stopped by the incorporation of a labeled dideoxynucleotide.[233] By using four different dideoxy analogs, synthesis of new DNA strands is terminated at differing nucleotide positions, eventually allowing the determination of a complete DNA sequence.[234]

Although sequencing techniques are limited in their scope as they are restricted to the analysis of one or very few genes at a time,[235] the development of DNA-microarrays or gene-chips might enable the large-scale application of these techniques.[236] In short, these gene-chips allow the simultaneous analysis of thousands of genes in one single analytical step.[237]

Despite the promising potential of these chip/array technologies, they still face a number of unsolved problems. In particular, the specificity of the results is questionable: given the possibility that non-target nucleic acids with a seventy-five percent sequence similarity may show cross-hybridization,[238] the results are more or less based on computerized statistical probability estimation.[239] Therefore, the need for conformational studies to assess the relevance of the results still remains an important issue. Until these problems are overcome, DNA-microarrays should not be considered to be of practical relevance in the analysis of genetic defects and disease predispositions.

---

229. *Id.* at 482.
230. *Id.*
231. *Id.*
232. *Id.*
233. *Id.*
234. RUSSELL, *supra* note 182, at 484.
235. Hadley C. King & Animesh A. Sinha, *Gene Expression Profile Analysis by DNA Microarrays*, 286 JAMA 2280 (2001).
236. Jonathan Weems, *A Proposal for a Federal Genetic Privacy Act*, 24 J. LEGAL MED. 109, 111 (2003); Francis S. Collins, *Shattuck Lecture – Medical and Societal Consequences of the Human Genome Project*, 341 NEW ENG. J. MED. 33 (1999).
237. King & Sinha, *supra* note 235, at 2280.
238. *Id.* at 2286.
239. DFG, *supra* note 5, at 20.