A Double-Edged Sword: Recommendations for Healthcare and Business Entities When Using Polygenic Scores

Kathleen J. Foley*

I. INTRODUCTION

Polygenic scores (PGS) provide a quantitative measure to understand the cumulative effects of isolated variants when determining the genetic underpinning of a specific trait.[1] PGS show enormous potential in clinical medicine—especially in early intervention strategies for those with a strong genetic predisposition to a complex medical disease or disorder.[2] PGS for social traits—including intelligence, voting behaviors, and education

* J.D. Candidate, The University of Iowa College of Law, 2024; Master of Science in Molecular Biology, Lehigh University, 2021. I would like to thank my parents and brother for their unending support, Zach Mass, Greg Ludeman, and Professor Anya Prince for their insightful edits, and all my colleagues at the *Journal of Corporation Law*. Finally, thank you to Professor Justin McAlister for being my first research advisor and continued mentor.

1. *Polygenic Score Data (PGS),* HEALTH & RET. STUDY SURV. RSCH. CTR., https://hrsdata.isr.umich.edu/data-products/polygenic-score-data-pgs [https://perma.cc/X8XT-MVV4].

2. For a discussion of PGS use in clinical trials, see Akl C. Fahed, Anthony A. Philippakis & Amit V. Khera, Comment, *The Potential of Polygenic Scores to Improve Cost and Efficiency of Clinical Trials*, NATURE COMMC'NS, May 2022, at 1, 1–2; Cathryn M. Lewis & Evangelos Vassos, *Polygenic Risk Scores: From Research Tools to Clinical Instruments*, GENOME MED., May 2020, at 1, 4.

attainment—show less potential in clinical medicine.[3] The potential for misuse of social PGS is significant.[4]

Today, the two foremost regulatory authorities concerning discrimination and genetic information are the Genetic Information Nondiscrimination Act (GINA) and the Health Insurance Portability and Accountability Act (HIPAA). Both GINA and HIPAA provide a strong foundation for genetic protection, but both congressional acts leave room for private and corporate entities to discriminate based on PGS. With the rapid progression of genetic technology—beginning with the Human Genome Project—HIPAA and GINA are currently ill-suited to address all privacy and ethical concerns that arise with the development and use of PGS.

This Note discusses (1) PGS and their potential uses, then (2) how HIPAA and GINA fail to address foreseeable harms from PGS misuse, and finally (3) recommends securing genetic information, including PGS, to ensure that PGS cannot be used for discriminatory purposes.

II. BACKGROUND

A. Polygenic Risk Scores

Since the turn of the millennia, researchers have sought to understand and decode the human genome. The Human Genome Project, designed to be a "moon-shot" initiative to sequence the entire human genome, commenced in October of 1990.[5] The first draft of the initial sequence of the human genome was subsequently published in February of 2001.[6] Although sequencing the genome was a monumental step in genomic analysis, researchers have continued to identify and annotate new functional regions of the genome.[7]

The mammalian genome consists of just over three billion pairs of molecules known as "base pairs."[8] Base pairs are the most basic building blocks of the genome.[9] Although base pairs are comprised of molecules, complementary base pairs operate as one unit analogous to how words are comprised of letters but sentences are comprised of words.

---

3. *See Beyond the Medical: The ELSI of Polygenic Scores for Social Traits*, UNIV. OF N.C. SCH. OF MED., https://www.med.unc.edu/cgs/beyond-the-medical-the-elsi-of-polygenic-scores-for-social-traits/ [https://perma.cc/2XYY-MELQ] (discussing the use of PGS to determine disease risk as well as social and behavioral traits).

4. *See generally* Aviad Raz & Jusaku Minari, Opinion, *AI-Driven Risk Scores: Should Social Scoring and Polygenic Scores Based on Ethnicity Be Equally Prohibited?*, FRONTIERS IN GENETICS (May 30, 2023), https://www.frontiersin.org/articles/10.3389/fgene.2023.1169580/full [https://perma.cc/WV2D-XSDK] (discussing the risks of social polygenic scores in the context of racial disparities).

5. *Human Genome Project Timeline*, NAT'L HUM. GENOME RSCH. INST., https://www.genome.gov/human-genome-project/timeline [https://perma.cc/7CXU-U26C].

6. *Id.*

7. Laralynne Przybyla & Luke A. Gilbert, *A New Era in Functional Genomics Screens*, 23 NATURE REVS.: GENETICS 89, 89 (2022).

8. Jonathan Henninger, *The 99 Percent . . . of the Human Genome*, HARV. UNIV.: GRADUATE SCH. OF ARTS & SCIS. (Oct. 1, 2012), https://sitn.hms.harvard.edu/flash/2012/issue127a/ [https://perma.cc/2TYB-PGGQ].

9. *Id.*

Base pairs are combined into "coding" and "non-coding" regions.[10] Coding regions refer to areas of the genome that ultimately form proteins.[11] Non-coding regions, formerly known as "junk DNA,"[12] are sequences of DNA that regulate coding regions or play no known role in the biological system.[13] Today, researchers continue to identify isolated "non-coding" regions of the genome that play a significant role in observable traits (phenotypes).[14]

Single nucleotide polymorphisms (SNPs) are isolated, individual variations of one base pair in either coding or non-coding regions.[15] One individual SNP may modify an individual's appearance.[16] For example, a singular SNP is the primary determinant of whether an individual has blue or brown eyes.[17] A collection of SNPs acting together may cause a gradation in the observed phenotype. These observed phenotypes include height, the presence or severity of a genetic disorder, or the propensity of an individual to have a specific personality. Two to ten percent of height variance is thought to be determined by 200 SNPs.[18] Genomicists conduct genome-wide association studies (GWAS) to identify SNPs that correlate with observable traits.[19] Researchers sequence the genomes of a target population with a specific phenotype or observable trait.[20] Using statistical analysis, researchers identify specific genetic markers whose frequencies are significantly more common in a target population as opposed to a control group.[21] If the variants remain significantly more common in the target population after quality control analyses, variation in the genome are thought to contribute to the observed phenotype.[22]

Once researchers correlate variants with a phenotype, one individual's genome (as opposed to tens of thousands in a GWAS) may be tested for the susceptibility of an individual in developing a trait.[23] The quantitative aggregation of risk based on identified SNPs or areas of variation within one individual's genome is known as a PGS or polygenic risk score.[24] This numerical score indicates an individual's risk of developing the

10. Michele Clamp et al., *Distinguishing Protein-Coding and Noncoding Genes in the Human Genome*, 104 PROS. NAT'L ACAD. SCIS. 19428, 19428 (2007).

11. *Id.*

12. Eva Frederick, *So-Called "Junk" DNA Plays a Key Role in Speciation*, MASS. INST. OF TECH.: BIOLOGY (Aug. 23, 2021), https://biology.mit.edu/so-called-junk-dna-plays-a-key-role-in-speciation/ [https://perma.cc/U8GT-NLH3].

13. *Beyond the Medical*, *supra* note 3.

14. Raz & Minari, *supra* note 4.

15. Chris Gunter, *Single Nucleotide Polymorphisms (SNPS)*, NAT'L HUM. GENOME RSCH. INST., https://www.genome.gov/genetics-glossary/Single-Nucleotide-Polymorphisms [https://perma.cc/26T7-V52D].

16. *Id.*

17. Richard A. Sturm et al., *A Single SNP in an Evolutionary Conserved Region Within Intron 86 of the* HERC2 *Gene Determines Human Blue-Brown Eye Color*, 82 AM. J. HUM. GENETICS 424, 424 (2008).

18. Ge Zhang et al., *Extent of Height Variability Explained by Known Height-Associated Genetic Variants in an Isolated Population of the Adriatic Coast of Croatia*, 6 PLOS ONE 1, 1 (2011).

19. Carolyn M. Hutter, *Genome-Wide Association Studies (GWAS)*, NAT'L. HUM. GENOME RSCH. INST., https://www.genome.gov/genetics-glossary/Genome-Wide-Association-Studies [https://perma.cc/L98V-GWRM].

20. *Id.*

21. *Id.*

22. *Id.*

23. *Polygenic Risk Scores*, NAT'L HUM. GENOME RSCH. INST., https://www.genome.gov/Health/Genomics-and-Medicine/Polygenic-risk-scores [https://perma.cc/25LJ-GCJG].

24. *Id.*

phenotype in comparison to the general population; the score does not imply or indicate certainty that an individual will develop the trait.[25] In fact, most PGS represent only a modest increase in susceptibility to a specific phenotype.[26] Additionally, PGS cannot account for environmental influences or gene-gene interactions that may affect an individual's overall risk.[27]

Increasingly, PGS have practical implications for clinical treatments.[28] With whole-genome sequencing costs less than $1000,[29] it is now economically feasible for a middle-class American to sequence their entire genome. With a sequenced genome, medical professionals may now search for SNPs that correlate with a higher risk of medical disorders like heart disease, obesity, diabetes, and breast cancer.[30] In turn, patients may be able to alter their behavior to decrease their overall risk to medical disorder despite a patient's genetic predisposition.[31]

Genomicists now use PGS techniques to measure an individual's predisposition to a non-medical trait. These traits include an individual's likely educational attainment,[32] voting behaviors,[33] intellect,[34] and their likelihood of smoking.[35] The determination of the likelihood of these traits is just as simple and inexpensive as determining an individual's predisposition to a medical disorder. Currently, public and private entities may use PGS in medical and non-medical contexts.[36] Although it is likely PGS will aid providers in administering medical recommendations, the risks of using PGS in non-medical contexts are significant and "ethically dubious."[37]

The field of personal genomics is historically rooted in concepts that contributed to eugenics. Eugenics is the term modern ethicists and researchers use to describe a set of concepts first developed by Francis Galton in the late 1800s.[38] Galton used statistics to claim that disease, social characteristics, and intelligence were intertwined with heredity

---

25. *Id.*

26. Robert Plomin & Sophie von Stumm, *Polygenic Scores: Prediction Versus Explanation*, 27 MOLECULAR PSYCHIATRY 49, 49 (2021).

27. *Id.*

28. *See* Lewis & Vassos, *supra* note 2, at 4 (discussing polygenic risk scores and the tools for clinical treatments).

29. Kris A. Wetterstrand, *The Cost of Sequencing a Human Genome*, NAT'L HUM. GENOME RSCH. INST., https://www.genome.gov/about-genomics/fact-sheets/Sequencing-Human-Genome-cost [https://perma.cc/JS25-XH63].

30. *Polygenic Risk Scores*, *supra* note 23.

31. *Id.*

32. *Id.*

33. Christopher T. Dawes et al., *A Polygenic Score for Educational Attainment Partially Predicts Voter Turnout*, 118 PROCS. NAT'L ACAD. SCIS. 1, 5 (2021).

34. Erhan Genç et al., *Polygenic Scores for Cognitive Abilities and Their Association with Different Aspects of General Intelligence–a Deep Phenotyping Approach*, 58 MOLECULAR NEUROBIOLOGY 4145, 4153 (2021); *see also* Sophie von Stumm & Robert Plomin, *Using DNA to Predict Intelligence*, 86 INTEL. 1, 2–4 (2021).

35. Jacqueline M. Vink, *Polygenic Risk Scores for Smoking: Predictors for Alcohol and Cannabis Use?*, 109 ADDICTION 1141, 1141 (2014).

36. Adebowale Adeyemo et al., *Responsible Use of Polygenic Risk Scores in the Clinic: Potential Benefits, Risks and Gaps*, 27 NATURE MED. 1876, 1879 (2021).

37. *Id.*

38. *Eugenics and Scientific Racism*, NAT'L HUM. GENOME RSCH. INST., https://www.genome.gov/about-genomics/fact-sheets/Eugenics-and-Scientific-Racism [https://perma.cc/33U5-B4B9].

and, subsequently, ethnicity and race.[39] Researchers in the early 1900s perpetuated a (patently false) narrative that certain ethnic and racial groups were inherently more educated, intelligent, successful, and beautiful than others.[40] This research morphed into a social movement, capturing the attention of the public as well as ethicists, scientists, and statisticians in pre-WWII Europe.[41] This Eugenics movement had profound social consequences culminating in the horrific evils of Nazi Germany.[42] Publications in eugenics were used as "scientific" justification for the genocide and mass sterilization of Jewish people, those with disabilities, and members of the LGBTQIA+ community in Nazi Germany.[43]

Eugenics continued in post-WWII America.[44] Notably, eugenics-based research was used to justify the involuntary sterilization of those with intellectual disabilities or "anti-social" behaviors through the 1970s.[45] Over 30 states passed laws allowing involuntary sterilization based on eugenics research and social support.[46]

B. Genetic Information Non-Discrimination Act

GINA was first introduced in 1995 by Representative Louise Slaughter of New York.[47] Representative Slaughter drafted GINA to prevent health insurance companies from setting deductibles based on genetic information and to protect consumers from having their genetic information published without their consent.[48] When GINA finally passed in 2008, almost eight years after the sequencing of the human genome, the final bill flatly prohibited discrimination based on genetic information by health insurance companies.[49] GINA, in its final iteration, prohibited covered entities from requesting or discriminating against employees based on genetic information.[50] Covered entities include employers with 15 or more employees, apprenticeship training programs, labor organizations, and municipal, state, and federal governments.[51]

GINA is now the foremost authority on how private and public entities may use genetic information. Under GINA, covered entities include group health plans sponsored by private employers, issuers in the group and individual health insurance markets, and

39. *Id.*
40. *Id.*
41. *Id.*
42. *Id.*
43. *Eugenics and Scientific Racism*, *supra* note 38.
44. *Id.*
45. *Id.*
46. *Id.*
47. *Timeline of the Genetic Information Nondiscrimination Act (GINA)*, NAT'L HUM. GENOME RSCH. INST., https://www.genome.gov/about-genomics/policy-issues/timeline-genetic-information-nondiscrimination-act-GINA [https://perma.cc/EH5P-YPJE].
48. *Id.*
49. *Id.*
50. *Id.*
51. *Fact Sheet: Genetic Information Nondiscrimination Act*, U.S. EQUAL EMP. OPPORTUNITY COMM'N, https://www.eeoc.gov/laws/guidance/fact-sheet-genetic-information-nondiscrimination [https://perma.cc/95CQ-ARGQ].

issuers of medical supplemental insurance.[52] Covered entities may not use genetic information to make health insurance policy decisions.[53] GINA declines to extend these protections to automobile, home, and life insurance companies.[54] Additionally, GINA is silent on whether other types of private entities may use genetic information in rendering services, including mortgage lenders, credit card companies, and institutions of higher learning.[55] Currently, all those not explicitly listed in GINA are implicitly allowed to incorporate genetic information in non-employment related matters. Today, no legal bar prevents private entities, apart from those listed above, from using PGS as a factor in rendering services.

C. Health Insurance Portability and Accountability Act

The Health Insurance Portability and Accountability Act of 1996 (HIPAA) requires "covered entities" to comply with guidelines aimed at protecting health information stored and used in health-related entities.[56] HIPAA's "Privacy Rule" covers health care providers, but also requires that health plan issuers and health care clearinghouses follow HIPAA regulations.[57] Entities that issue health plans include health insurance companies, health maintenance organizations, company health plans, Medicaid, Medicare, and veterans' health care programs.[58] A health care clearinghouse processes "nonstandard health information" into standardized transactions or data as part of either a public entity or private corporation.[59] For HIPAA, these entities process named sensitive health information into data points for further analysis.[60]

HIPAA extends regulations one step further—to business associates of any covered entity.[61] Any public or private entity that contracts with a covered entity for a stated purpose may be considered a business associate if health information is disclosed to the business associate.[62] Business associates may perform legal, consulting, data aggregation, administrative, management, or financial services.[63]

---

52. Regulations Under the Genetic Information Nondiscrimination Act of 2008, 75 Fed. Reg. 68912, 68912, 68923–24, 68930, 68938 (Nov. 9, 2010) (to be codified at 29 C.F.R. pt. 1635).

53. *Id.*

54. Sarah Zhang, *The Loopholes in the Law Prohibiting Genetic Discrimination*, THE ATLANTIC (Mar. 13, 2017), https://www.theatlantic.com/health/archive/2017/03/genetic-discrimination-law-gina/519216/ [https://perma.cc/79M3-XEJB].

55. Anikka Hoidal, *Genetic Discrimination: Why We Should Expand GINA*, UNIV. OF UTAH: S.J. QUINNEY COLL. OF L. (Apr. 5, 2016), https://www.law.utah.edu/news-articles/genetic-discrimination-why-we-should-expand-gina/ [https://perma.cc/H2CE-P2CW].

56. 45 C.F.R. § 160.10 (2024).

57. *Id.* § 160.102 (2024).

58. *Id.* § 160.103 (2024) (defining "health plan").

59. *Id.* (defining "health care clearinghouse").

60. *Id.*

61. *Business Associates*, U.S. DEP'T OF HEALTH & HUM. SERVS., https://www.hhs.gov/hipaa/for-professionals/privacy/guidance/business-associates/index.html [https://perma.cc/C3V3-N3BX].

62. *Id.*

63. *Id.*

The HITECH Act of 2009 expanded security and privacy requirements for business associates.[64] HIPAA, incorporating the HITECH Act, now mandates that business associates provide "satisfactory assurances" to the covered entity that the entity will comply with HIPAA requirements.[65] Business associates must sign a contract that details the boundaries of prescribed use of health information, requires the business associate to maintain confidentiality, and requires the business associate to use appropriate safeguards in protecting sensitive information.[66] If the business associate violates any term of the contract, the covered entity must terminate the contract and take steps to report the breach to the Department of Health and Human Services (HHS).[67]

Although HIPAA generally mandates a contract for business associates using health information, HIPAA includes several exceptions to the general rule.[68] The act allows medical professionals and other covered entities to disclose health information to researchers without being classified as a business associate.[69] Researchers may gain access to sensitive genetic or health information if the patient consents or, in the absence of patient consent, if the data is part of a deidentified data set.[70] If the researcher is not performing any of the enumerated functions of a business associate, then HIPAA does not regulate researchers under the Act.[71]

Health information, under HIPAA, includes any information that "[r]elates to the past, present, or future physical or mental health or condition of an individual."[72] Initially, HIPAA did not protect an individual's genetic information.[73] Only after GINA was passed in 2008 did Congress reconfigure the definition of protected health information to incorporate genetic information.[74] This newly expanded definition applied to both HIPAA and GINA.[75] In 2013, the HIPAA Omnibus Rule expanded coverage under the HIPAA privacy rule and prohibited health insurance companies from using genetic information in policy underwriting.[76] Today, any information relating to a physical or mental health trait derived from the genome would be considered health information covered by HIPAA.[77]

---

64. *HITECH Act Enforcement Interim Final Rule*, U.S. DEP'T OF HEALTH & HUM. SERVS., https://www.hhs.gov/hipaa/for-professionals/special-topics/hitech-act-enforcement-interim-final-rule/index.html [https://perma.cc/B43Z-9SCF].

65. *Eugenics and Scientific Racism*, *supra* note 38.

66. *Id.*

67. *Id.*

68. *Id.*

69. *Id.*

70. *Eugenics and Scientific Racism*, *supra* note 38.

71. *Business Associates*, *supra* note 61.

72. 45 C.F.R. § 160.103 (2024) (defining "health information").

73. Mavis Asiedu-Frimpong, *HIPAA Turns 25, and It's Adapting Nicely*, UNIV. OF PA. LEONARD DAVIS INST. OF HEALTH ECON. (June 29, 2021), https://ldi.upenn.edu/our-work/research-updates/hipaa-turns-25-and-its-adapting-nicely/ [https://perma.cc/Z8GP-AT9K].

74. *Id.*

75. *Id.*

76. Amanda Gammon & Deborah W. Neklason, *Confidentiality & the Risk of Genetic Discrimination: What Surgeons Need to Know*, 24 SURGICAL ONCOLOGY CLINICS N. AM. 667, 670 (2015).

77. 45 C.F.R. § 160.102 (2024).

Covered entities and business associates must meet HIPAA compliance requirements for both the HIPAA Privacy and HIPAA Security Rule.[78] In summary, the HIPAA Privacy Rule ensures the rights of individuals to share genetic information with named entities and individuals or safeguard the privacy of genetic information.[79] Conversely, achieving HIPAA security compliance while using modern technology is costly and complex for even the most advanced healthcare entities.

The HIPAA Security Rule covers all "electronic protected health information" (e-PHI).[80] All health care plans and covered entities must have achieved Security Rule compliance by 2006 and maintained compliance since that time.[81] If an entity or individual fails to comply, HHS may recommend both civil and criminal penalties on the offending party.[82] The Security Rule sets forth general requirements for maintaining security standards. They include ensuring the confidentiality of e-PHI, protecting against anticipated threats or unpermitted uses, and ensuring compliance by the relevant workforce.[83] Four core elements are mandatory for compliance: risk analysis, administrative safeguards, physical safeguards, and technical safeguards.[84]

Risk analysis must be performed before holding e-PHI to determine the appropriate safeguards to ensure the security of protected information.[85] HHS recommends that the risk analysis process includes an evaluation of the risks to protected information, documentation of security measures (and rationale in adopting those measures), and continued maintenance of appropriate security for e-PHI.[86] Risk analysis should continue after onboarding e-PHI and should evolve while the nature of the information changes.[87]

Administrative safeguards include designating a professional or set of professionals that are responsible for implementing security procedures.[88] These professionals are also responsible for supervising those that work directly with e-PHI.[89] One of the key policies recommended by HHS as an administrative safeguard is to limit the access of employees to e-PHI—allowing only those employees with an articulable need to access e-PHI.[90] If any employee violates the terms of the security policies and procedures, the designated

78. *HIPAA for Professionals*, U.S. DEP'T OF HEALTH & HUM. SERVS., https://www.hhs.gov/hipaa/for-professionals/index.html [https://perma.cc/R7AJ-W3TN].

79. *Id.*

80. *HIPAA Security Rule & Risk Analysis*, AM. MED. ASS'N, https://www.ama-assn.org/practice-management/hipaa/hipaa-security-rule-risk-analysis [https://perma.cc/KP38-XNRW].

81. *Id.*

82. U.S. DEP'T OF HEALTH & HUM. SERVICES, OCR PRIVACY BRIEF: SUMMARY OF THE HIPAA PRIVACY RULE 17–18 (2003).

83. 45 C.F.R. § 164.306(a) (2024).

84. *Summary of the HIPAA Security Rule*, U.S. DEP'T OF HEALTH & HUM. SERVS., https://www.hhs.gov/hipaa/for-professionals/security/laws-regulations/index.html [https://perma.cc/XAX8-VN83].

85. *The Security Rule*, U.S. DEP'T OF HEALTH & HUM. SERVS., https://www.hhs.gov/hipaa/for-professionals/security/index.html [https://perma.cc/538L-WLSG].

86. *Id.*

87. *Id.*

88. *Id.*

89. *Id.*

90. *The Security Rule*, *supra* note 85.

security professional is tasked with enforcing the policy and sanctioning the infringing employee.[91]

Physical safeguards must include storing e-PHI in secure facilities with controlled access.[92] Employees may only be able to access e-PHI on controlled workstations[93] where devices with access to e-PHI have set parameters for transfer, deletion, and overall use of e-PHI.[94]

Finally, information technology professionals must implement technical safeguards.[95] Technical safeguards include electronic-controlled access where only authorized employees may log on to e-PHI access points.[96] Devices with access to e-PHI must implement software that records all activity in the information systems,[97] commonly known as "tracking clicks." Similarly, software on accessible devices must prevent inappropriate alteration or deletion of health-related documents.[98] Finally, devices with e-PHI access must limit the transmission of sensitive information over the internet or other external electronic networks.[99]

III. ANALYSIS

PGS are a double-edged sword; the scores have the potential to revolutionize clinical medicine but also the potential to enable discriminatory practices. Currently, GINA does not protect consumers from discriminatory practices based on genetic information unless a consumer is seeking medical care or health insurance in the context of employment.[100] GINA does not regulate life insurance, long-term care insurance, automobile insurance, car insurance, or healthcare cost-sharing plans. Similarly, HIPAA does not classify unnamed genetic information as protected health information, despite evidence that researchers can identify a specific individual from their genome in less than one day.[101] Both HIPAA and GINA fail to adequately protect consumers from discrimination when entities use PGS or genetic information.

A. Polygenic Scores Have the Potential to Be a Discriminatory Weapon

PGS provide a limited measure of an individual's susceptibility to a particular trait. As genetic studies continue and samples become more diverse, PGS will become more

91. *Id.*
92. *Id.*
93. *Id.*
94. *Id.*
95. *Summary of the HIPAA Security Rule*, *supra* note 84.
96. *Id.*
97. *Id.*
98. *Id.*
99. *Id.*
100. *See* Jessica D. Tenenbaum & Kenneth W. Goodman, *Beyond the Genetic Information Nondiscrimination Act: Ethical and Economic Implications of the Exclusion of Disability, Long-Term Care and Life Insurance*, 14 PERSONALIZED MED. 153, 154 (2017) (advocating that GINA should apply to long-term care and disability insurance).
101. Peter Pitts, Opinion, *The Privacy Delusions of Genetic Testing*, FORBES (Feb. 15, 2017), https://www.forbes.com/sites/realspin/2017/02/15/the-privacy-delusions-of-genetic-testing/ [https://perma.cc/J6BU-29SY].

accurate.[102] As PGS increase their accuracy and precision, PGS will become a valuable tool for clinicians to genetically assess their patients' predisposition to complex diseases and disorders.[103] In clinical medicine, the potential applications of PGS are profound.[104]

Few researchers deny the potential benefits of PGS for medical diagnoses. But PGS for social traits (educational attainment, intelligence, height, etc.) are not regarded as inherently positive.[105] Researchers fear that PGS will become the "new eugenics," where unsavory actors will use PGS to defend discriminatory practices.[106] Comparative genomics, including PGS, has its roots in eugenics.[107]

Today, no reputable journal has published research that correlates a specific race with a *social* trait.[108] The absence of studies comparing ethnicities in the literature may be a symptom of increased recognition of eugenics in the history of comparative genomics. However, it is possible that because data sets are not ethnically diverse, studies are not readily feasible. Genetic samples used in biobanks are typically ancestrally homogenous, with most participants identifying as non-Hispanic Caucasians.[109] As of 2009, those of non-European ancestry account for only four percent of samples.[110] In 2016, the proportion rose to 20% but with predominantly Asian cohorts added.[111] Today, Caucasian cohorts are still disproportionately overrepresented in Biobank samples.

Researchers have conducted studies correlating race with the prevalence of a medical trait using PGS.[112] Researchers compared the polygenic scores of "African American versus White" Americans to determine the incidence of Dementia and Alzheimer's Disease in the respective racial categories.[113] Researchers specifically used "cognition performance PGS" to determine the propensity of populations to cognitive disease.[114] This study, published in 2022 in *Brain Communications*, is currently available on "PubMed."[115] PubMed is one of the foremost databases for medical and life science research maintained by the National Institutes of Health (NIH).[116]

---

102. Alicia R. Martin et al., *Current Clinical Use of Polygenic Scores May Exacerbate Health Disparities*, 51 NATURE GENETICS 584, 584–85 (2019).

103. *Id.*

104. *Id.*

105. Aaron Freedman, *Can DNA Tests Tell Us Who We Are? Only If We're Racists.*, WASH. POST (Feb. 5, 2020), https://www.washingtonpost.com/outlook/can-dna-tests-tell-us-who-we-are-only-if-were-racists/2020/02/05/7adbdcfe-3e12-11ea-baca-eb7ace0a3455_story.html (on file with the *Journal of Corporation Law*).

106. *Id.*

107. Jo C. Phelan, Bruce G. Link & Naumi M. Feldman, *The Genomic Revolution and Beliefs About Essential Racial Differences: A Backdoor to Eugenics?*, 78 AM. SOCIO. REV. 167, 167 (2013).

108. Theresa M. Duello et al., *Race and Genetics Versus 'Race' in Genetics*, 9 EVOLUTION MED. & PUB. HEALTH 232, 232–37 (2021) (discussing race as an outdated concept and advocating for increased ethnic diversity in genetic studies).

109. Megan Prictor, Harriet J. A. Teare & Jane Kaye, *Equitable Participation in Biobanks: The Risks and Benefits of a "Dynamic Consent" Approach*, 6 FRONTIERS PUB. HEALTH 1, 2 (Sept. 5, 2018).

110. *Id.*

111. *Id.*

112. May A. Beydoun et al., *Race, Polygenic Risk and Their Association with Incident Dementia Among Older US Adults*, 4 BRAIN COMMC'NS 1 (Sept. 26, 2022).

113. *Id.* at 1.

114. *Id.* at 12.

115. *Id.*

116. *Id.* at 1.

Private entities now have the option to offer social PGS research in in-vitro fertilization (IVF) services, although the accuracy of the tests is still disputed.[117] In 2019, select clinics offered to calculate PGS for intelligence in embryos.[118] The Genomic Prediction clinic in New Jersey used PGS to "screen[] out embryos with abnormally low IQ."[119] This led to widespread backlash by the public and major media outlets.[120] Esteemed institutions of higher learning criticized the accuracy of the tests and compared the tests to eugenics practices in the 20th century.[121] Today, the clinic no longer advertises these services, at least on the company's public-facing website.[122]

B. GINA Allows Private Entities to Leverage PGS Scores to Determine Whether to Render Services

Much like IVF clinics, life insurance companies see enormous value in PGS.[123] As recently as 2022, researchers found that PGS, although imperfect, could identify a shorter lifespan (by 2.6 years) in the top ten percent of scores.[124] Researchers went one step further, stating: "We conclude that existing genetic risk scores can already improve life insurance underwriting."[125] These same researchers called for increased regulation from governmental agencies and identified key issues with PGS including "a tangible risk of growing information asymmetries that could lead to adverse selection."[126]

---

117. Jocelyn Kaiser, *Screening Embryos for IQ and Other Complex Traits Is Premature, Study Concludes*, SCIENCE (Oct. 24, 2019), https://www.science.org/content/article/screening-embryos-iq-and-other-complex-traits-premature-study-concludes [https://perma.cc/9JZW-N8YZ].

118. *IVF: Choosing the Most Intelligent Embryo*, CTR. FOR REPROD. MED. (May 30, 2019), https://www.ivforlando.com/blog/ivf-choosing-the-most-intelligent-embryo [https://web.archive.org/web/20210420202646/https://www.ivforlando.com/blog/ivf-choosing-the-most-intelligent-embryo] (interviewing Dr. Hsu, the founder of Genomic Prediction); *see also* Kaiser, *supra* note 117 (referencing Genomic Prediction's services in testing for "'intellectual disability' or low IQ").

119. Hannah Devlin, *IVF Couples Could Be Able to Choose the 'Smartest' Embryo*, THE GUARDIAN (May 24, 2019), https://www.theguardian.com/society/2019/may/24/ivf-couples-could-be-able-to-choose-the-smartest-embryo [https://perma.cc/8HWX-L2WF].

120. Dalton Conley, *A New Age of Genetic Screening is Coming–and We Don't Have Any Rules for It*, WASH. POST (June 14, 2021), https://www.washingtonpost.com/outlook/2021/06/14/genetic-screening-ivf-moral-dilemmas/ (on file with the *Journal of Corporation Law*) (assessing the public's attitude towards PGS measuring intelligence or IQ).

121. *See* Antonio Regalado, *Eugenics 2.0: We're at the Dawn of Choosing Embryos by Health, Height, and More*, MASS. INST. TECH. TECH. REV.: BIOTECHNOLOGY & HEALTH (Nov. 1, 2017), https://www.technologyreview.com/2017/11/01/105176/eugenics-20-were-at-the-dawn-of-choosing-embryos-by-health-height-and-more/ [https://perma.cc/PHP7-TEDX] (setting forth ethical questions when testing embryos for intelligence or other non-medical traits); *see also* Carl Zimmer, *Genetic Intelligence Tests Are Next to Worthless*, THE ATLANTIC: SCI. (May 29, 2018), https://www.theatlantic.com/science/archive/2018/05/genetic-intelligence-tests-are-next-to-worthless/561392/ [https://perma.cc/KQ3S-TUZL] (discussing the value of DNA testing using PGS as an adult).

122. *Better Tests, Better Outcomes*, LIFEVIEW: POWERED BY GENOMIC PREDICTION, https://www.lifeview.com [https://perma.cc/P97H-A75D].

123. Richard Karlsson Linnér & Philipp D. Koellinger, *Genetic Risk Scores in Life Insurance Underwriting*, 81 J. HEALTH ECON. 102556, 1–15 (2022).

124. *Id.* at 10.

125. *Id.*

126. *Id.* at 13.

Most life insurance companies determine rates based on the health indicators of applicants including blood pressure and history of risky behaviors.[127] If included, PGS may be a valuable tool for insurance companies when classifying individuals into risk categories.[128] However, PGS, no matter how precise, still only account for a minority of the variation underlying a medical or psychological trait.[129] Environmental effects and personal choices still contribute to the overall propensity of an individual in developing a complex disease or trait.[130]

GINA allows life insurance companies to use genetic information when they determine rates during the underwriting process.[131] Although New York specifically forbids life insurance companies (and other private entities) from using genetic information in underwriting,[132] no federal law bars such practices.[133] Without federal intervention, GINA will not apply to genetically informed underwriting in disability insurance policies, life insurance policies, or long-term care insurance policies.[134]

Feasibly, private entities that participate in healthcare cost-sharing may use PGS to determine who they admit into their funding pools. Entities that do not underwrite or sell health insurance are not covered under GINA[135] and healthcare cost-sharing plans are not regulated as healthcare insurance companies.[136] Although most healthcare cost-sharing funds are religiously based, no federal law bars healthcare-sharing funds from being religiously unaffiliated. Several large healthcare-sharing funds are not religiously based, gearing their services towards contractors or W-2 employees whose employers do not offer health insurance.[137]

Under GINA, employers are explicitly barred from soliciting information about an employee's health or genetic information.[138] However, employers are allowed to solicit from interested applicants a "Voluntary Self-Identification of Disability Form" during the

127. *See* Cameron Huddleston & Jason Metz, *Life Insurance Underwriting Classes Explained*, FORBES ADVISOR (Oct. 27, 2023), https://www.forbes.com/advisor/life-insurance/underwriting-classes/ [https://perma.cc/M4QX-HRJA] (explaining factors that determine rates in life insurance).

128. *Polygenic Risk Scores*, *supra* note 23.

129. Plomin & von Stumm, *supra* note 26.

130. *Id.*; MALCOLM GLADWELL, OUTLIERS 9–10 (discussing research on a region's astonishing lack of heart disease, finding that the "town's social structure" produced an environment that reduced the heart problems).

131. Jessica D. Tenenbaum & Kenneth W. Goodman, *Beyond the Genetic Information Nondiscrimination Act: Ethical and Economic Implications of the Exclusion of Disability, Long-Term Care and Life Insurance*, 14 PERSONALIZED MED. 153, 153–57 (2017).

132. N.Y. INS. LAW § 2615(a) (McKinney 2023); Jarrod O. Anderson, Anna C.F. Lewis & Anya E.R. Prince, *The Problems with Patchwork: State Approaches to Regulating Insurer Use of Genetic Information*, 22 DEPAUL J. HEALTH CARE L., no. 1, 2021, at 1, 12.

133. *Id.* at 4.

134. *See id.* at 38 (discussing how federal legislation could fill the gaps of GINA and create uniformity).

135. 45 C.F.R. § 160.102 (2024).

136. Beth Braverman, *What Is a Medical Cost-Sharing Program?*, GOODRX HEALTH (Oct. 7, 2021), https://www.goodrx.com/insurance/alternative/medical-cost-sharing-program [https://perma.cc/YA6C-Z9DE].

137. *E.g.*, *The Care+ Plan*, HSA FOR AM., https://hsaforamerica.com/the-care-plan [https://perma.cc/KSD7-ZSYT] (explaining how the Care+ plan is "[d]esigned for W2 employees or people not working" and "[s]elf-employed, [b]usiness [o]wners, and [i]ndependent [c]ontracts"); *The HSA Secure Plan*, HSA FOR AM., https://hsaforamerica.com/mpowering-benefits/ [https://perma.cc/HYA9-8LT5] ("HSA Secure is only available to business owners, independent contractors, and self-employed individuals.").

138. 29 C.F.R. § 1635.8(a) (2024).

application period.[139] Although employers are legally barred from discriminating against applicants based on voluntary disclosure of disabilities, domestic and international researchers are now using PGS to correlate a medical diagnosis with a stigmatized social trait. For example, researchers in Finland recently published a cohort study showing a link between those with a high schizophrenia PGS (or schizophrenia traits) with poor performance in the labor market, measured as failure to maintain long-term employment.[140] Summarily, researchers concluded that "genetic liability for schizophrenia may hinder individuals' ability to fully participate in the labour market."[141]

C. Statutory HIPAA Compliance Is Ill-Suited to Protect Genetic Information

HIPAA allows for the sharing and selling of deidentified information to researchers and other public entities.[142] Current, ubiquitous technology allows for the reidentification of information in short timeframes.[143] In 2017, with access to "anonymous" genetic information, one MIT professor was able to identify five randomly selected individuals from a public research database.[144] It took him less than one day to identify the individuals from deidentified genomic data.[145] Similarly, a researcher at Harvard Medical School was able to identify the identities of approximately 80% of the school's internal genetic database based on markers for physical traits.[146] Estimates published in 2018 assert that 60% of Americans with European ancestry can be identified from genomic data available in public databases.[147]

HIPAA provides security benchmarks that health insurance companies and business associates must follow when sharing named, identifiable health information.[148] If business associates fail to follow HIPAA security guidelines, the corporate entities must be reported to HHS by those with specific knowledge—often the covered entity in a contractual agreement with the business associate.[149]

Even if a business associate or health insurance company complies with HIPAA, all genetic information is vulnerable when stored electronically.[150] Risk analysis paired with administrative, physical, and technical safeguards secure vulnerable information better than information not secured by defenses outlined in the HIPAA privacy rule.[151] HHS's

139. Jennifer S. Kiesewetter, *Diversity and Inclusion: Getting Candidates to Self-Identify*, ADP, https://www.adp.com/spark/articles/2019/02/diversity-and-inclusion-getting-candidates-to-self-identify.aspx [https://perma.cc/39E3-2WTE].

140. Jutta Viinikainen et al., *Schizophrenia Polygenic Risk Score and Long-Term Success in the Labour Market: A Cohort Study*, 151 J. PSYCHIATRIC RSCH. 638, 638–40 (2022).

141. *Id.*

142. *Business Associates*, *supra* note 61.

143. Pitts, *supra* note 101.

144. *Id.*

145. *Id.*

146. *Id.*

147. Yaniv Erlich et al., *Identity Inference of Genomic Data Using Long-Range Familial Searches*, 362 SCIENCE 690, 690 (2018).

148. *See* 45 C.F.R. § 160.103 (2024) (defining "health information").

149. *Id.*

150. OFF. OF THE PRIV. COMM'R OF CAN., PRIVACY AND CYBER SECURITY EMPHASIZING PRIVACY PROTECTION IN CYBER SECURITY ACTIVITIES 2–3 (2014).

151. *Summary of the HIPAA Security Rule*, *supra* note 84.

Office for Civil Rights (OCR) published in their quarterly newsletter that most cyber-attacks could be prevented or mitigated if covered entities updated their cyber security practices with the latest HIPAA guidance.[152] The HIPAA statute provides a baseline for privacy and security regulations, but the latest guidance from HHS provides evolving guidance based on current threats.[153] Today, phishing attacks account for most information technology breaches.[154] The latest guidance from HHS instruct covered entities and business associates on how to protect their information from these cyber-attack techniques.[155]

IV. RECOMMENDATIONS

First and foremost, researchers and funding entities should add internal safeguards against using and developing social PGS. As for Policymakers, they should amend pre-existing privacy law. GINA provides that covered entities, including health insurance companies, may not use genetic information when determining rates for coverage or determining employment.[156] GINA should extend the prohibition against genetically informed services to other types of insurance including life, long-term care, automobile, and home insurance providers. GINA should also expand to cover healthcare cost-sharing pools. Additionally, HIPAA should be amended to remove exceptions for researchers and de-identified limited data sets for all genetic information, as "de-identified information" should no longer include genetic sequences or other genetic information, including PGS.

A. Internal Safeguards for Research or Corporate Entities

Increasingly, clinicians and researchers use PGS to decipher the complex web of interactions in the mammalian genome. By comparing an observed trait to the human genome, researchers can ascertain a patient's genetic predisposition to a complex disease. This advance in genomic research provides a method that could save lives through early intervention, but it also provides a method to weaponize genetic information in support of discriminatory activities. The long history of eugenics in the 19th, 20th, and 21st centuries should color how we approach PGS. Economic and social pressures contributed to eugenics proliferating in Nazi Germany.[157] Acknowledging this history of eugenics, private businesses that are not research institutions should be discouraged from developing, funding, and using PGS for non-medical traits.

---

152. *OCR Quarter 1 2022 Cybersecurity Newsletter: Defending Against Common Cyber-Attacks*, U.S. DEP'T OF HEALTH & HUM. SERVS. (Mar. 17, 2022), https://www.hhs.gov/hipaa/for-professionals/security/guidance/cybersecurity-newsletter-first-quarter-2022/index.html [https://perma.cc/E77F-DSEJ].

153. *Id.*

154. Steven Bowcut, *How to Spot and Protect Yourself from a Phishing Attack*, CYBERSECURITY GUIDE, https://cybersecurityguide.org/resources/phishing [https://perma.cc/T5D8-YKR4].

155. Lisa Pino, *Improving the Cybersecurity Posture of Healthcare in 2022*, U.S. DEP'T OF HEALTH & HUM. SERVS. (Feb. 28, 2022), https://www.hhs.gov/blog/2022/02/28/improving-cybersecurity-posture-healthcare-2022.html [https://perma.cc/N22M-8Z8T].

156. *Genetic Information*, U.S. DEP'T OF HEALTH & HUM. SERVS., https://www.hhs.gov/hipaa/for-professionals/special-topics/genetic-information/index.html [https://perma.cc/J2R3-LJ2E].

157. *Eugenics and Scientific Racism*, *supra* note 38.

A flat prohibition on a singular type of research may infringe on free speech and First Amendment protections—although dueling interpretations of the First Amendment prevent clear guidance on the matter.[158] In light of this, policymakers should impose restrictions on funding.[159] Currently, nearly half (42%) of all basic research is funded through government grants.[160] Restricting publicly available funding from projects related to developing social PGS would significantly impede the development of problematic and ethically dubious PGS.

Social pressure against developing and implementing PGS should be encouraged by reputable research institutions and peer-reviewed journals in the form of publications focusing on the ethical, legal, and social implications of such research.[161] These publications should explain the widespread ramifications of using and perpetuating social PGS. Mainstream media outlets should be encouraged to report on the ramifications of social PGS through easy-to-understand press releases, accessible authors, and public speaking engagements. Social pressure and outcry against businesses and corporations who use such scores should send a strong signal to curious corporations and businesses that social PGS come at a steep social and monetary cost.

To recommend against developing PGS for medical traits would disregard the potential benefits of PGS in clinical medicine—especially the opportunity to implement early intervention strategies before disease and disorder symptoms present. The consuming public may also profit from medical PGS outside of clinical medicine whether in IVF services or in early access to programs designed to ameliorate the progress of disease. However, private or corporate entities using PGS should not have unfettered use of PGS. Any entity seeking to use PGS in decisions related to rendering services should not be able to solely rely on medical PGS to discriminate between applicants.

B. Genetic Information Nondiscrimination Act

GINA provides that health insurance companies, and other health providers, may not use genetic information when determining rates for coverage.[162] GINA should extend the prohibition against genetically informed rates to life insurance, long term care insurance, automobile insurance, and home insurance providers. Additionally, GINA should expand to cover healthcare cost-sharing plans.

Medical PGS do not incorporate lifestyle choices, healthy habits, or prior medical history when assessing propensity to disease. Currently, medical PGS capture a modest amount of variation in disease risk. For example, PGS only capture an estimated two percent variance in those with clinically diagnosed depression. Ninety-eight percent of

158. Amedeo Santosuosso, Valentina Sellaroli & Elisabetta Fabio, *What Constitutional Protection For Freedom of Scientific Research?*, 33 J. MED. ETHICS 342, 342 (2007).

159. *See* Jeffrey Mervis, *New White House Rules Restrict Use of Grant Funding to Deal With COVID-19 Impacts*, SCIENCE (June 30, 2020), https://www.science.org/content/article/new-white-house-rules-restrict-use-grant-funding-deal-covid-19-impacts [https://perma.cc/893E-5TXC] (detailing the evolving restrictions on COVID-19 funding since the beginning of the pandemic).

160. NAT'L SCI. BD., THE STATE OF U.S. SCIENCE AND ENGINEERING 2020 (2020).

161. *See* Angelica Ronald, Editorial, *Polygenic Scores in Child and Adolescent Psychiatry—Strengths, Weaknesses, Opportunities and Threats*, 61 J. CHILD PSYCH. & PSYCHIATRY 519, 520 (2020) (explaining that widespread backlash would impede the development and use of PGS).

162. *Genetic Information*, *supra* note 156.

variation stems from the environment, gene-gene interactions, or other unmeasured variables.[163] Assessing an individual's rate based solely, or largely, on PGS disincentivizes the individual from partaking in healthy behaviors and undermines an individual's total risk for developing some of the most common health issues including heart disease, high blood pressure, and high LDL cholesterol.

C. Health Insurance Portability and Accountability Act (HIPAA)

HIPAA specifically articulates exceptions to the HIPAA Privacy Rule where business associates and researchers do not have to abide by the HIPAA Security Rule if the information used by the entities is de-identified in a limited data set.[164] With the advent of new genetic technologies and the proliferation of advanced statistical software, all genetic information should be considered readily identifiable even if names, addresses, and other personal information are redacted from the genetic information. Although it seemed unlikely that genomes could be paired to a specific individual (with no other identifying information) MIT and Harvard University researchers proved it was easy, fast, and cheap to do.[165] As a result, HIPAA should be amended to remove exceptions for researchers and de-identified limited data sets for all genetic information, including PGS and genetic sequences.

As a direct result of reclassifying genetic information as protected health information, private entities, including businesses and corporations, must comply with security measures mandated under the HIPAA Security Rule. The HIPAA Security Rule mandates that all business entities using or harboring protected health information engage with risk analysis and implement administrative, technical, and physical safeguards.[166] By altering the definition of e-PHI, corporations or businesses who use PGS but fail to achieve security compliance under HIPAA would be subject to the same criminal and civil penalties of those that fail to secure e-PHI under current HIPAA regulations.

D. Healthcare Cost-Sharing Plans

Allowing insurance companies and healthcare cost-sharing plans to use PGS as a primary determinant of rates may ultimately foster a consumer marketplace where individuals with a high propensity for common diseases pay more for modern necessities, like life insurance. This striation of rates based on pre-determined genetic data lends itself to a new age of discrimination, one where the "genetically poor" pay more than those with "better genetics." Policymakers should flatly prohibit such a marketplace. Healthcare cost-sharing plans should be included and incorporated under both HIPAA and GINA.

V. CONCLUSION

PGS show enormous potential for clinicians and healthcare providers, especially in the areas of IVF and early intervention clinical management. PGS for social traits (e.g.,

163. Cathryn M. Lewis & Evangelos Vassos, *Polygenic Risk Scores: From Research Tool to Clinical Instruments*, 12 GENOME MED. 1, 6 (2020).

164. *Timeline of the Genetic Information Nondiscrimination Act (GINA)*, *supra* note 47.

165. Plomin & von Stumm, *supra* note 26, at 21.

166. *Summary of the HIPAA Security Rule*, *supra* note 84.

voting behaviors, educational attainment, intelligence) show less potential for clinical medicine and have enormous potential for misuse. Considering eugenics practices in the 19th, 20th, and 21st centuries, private institutions—including corporations and unincorporated businesses—should be discouraged from developing or using PGS scores for social traits. Research institutions should be prohibited from performing social PGS development or utilization studies with government or public funding.

For entities that seek to use PGS for medical traits, business, insurance, and corporate entities should not be able to use PGS for medical traits in decisions related to rates or rendering services. All entities that choose to use or harbor PGS for IVF, early intervention, or health-related services should be classified as "business associates" and should comply with HIPAA regulations for named and identifiable information.

Previously, genetic information without identifying characteristics did not qualify as protected health information under HIPAA. Policymakers should recognize that technology now allows for the re-identification of genetic information using statistical software. Policymakers should remove exceptions under HIPAA that allows the sharing of de-identified genetic information without mandatory compliance with the HIPAA Security Rule. Finally, those who do share or harbor PGS or genetic information must comply with the HIPAA security measures listed in the HIPAA Security Rule—including risk assessment and administrative, technical, and physical safeguards.