

Precision Medicine for the Population—The Hope and Hype of Public Health  
Genomics

Annotated Bibliography

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Public health is the most recent of the biomedical sciences to embrace “precision.” Advocates for “precision public health” (PPH) call for a data-driven, computational approach to public health, leveraging genomic and other data to inform public health decision-making. Yet, like precision medicine, PPH risks overselling the value of genomic data to determine health outcomes, but on a population level. History has shown that over-emphasizing heredity tends to disproportionately harm underserved minorities and disadvantaged communities. Comparing and contrasting current PPH with an earlier attempt at using genetics to inform public health during the Progressive era (1890–1920) highlights some potential risks of genotype-driven preventive public health.

This annotated bibliography expands the Further Reading list in the article titled “Precision Medicine for the Population—The Hope and Hype of Public Health Genomics” published in *CHANCE* magazine, Vol. 36, No. 3, 2023, pp. 25 – 27 (<http://chance.amstat.org>).

1. Collins FS. Medical and Societal Consequences of the Human Genome Project. *The New England Journal of Medicine*. 1999;341(1):28-37. doi: 10.1056/NEJM199907013410106.

This article is written by Francis Collins, the director of the National Human Genome Research Institute (NHGRI) in 1999, who led the publicly funded effort in the Human Genome Project (HGP). Nearly a decade since launching the HGP, Collins lays out a brief history of how the effort materialized, the significance of sequencing the human genome, and predictions on how the human genome will impact medicine and society in 2010. Written before the HGP’s completion,

this article offers insights into the promises and hopes scientists had for the project, becoming a rallying vision for precision medicine.

2. Comfort N. The Overhyping of Precision Medicine.

<https://www.theatlantic.com/health/archive/2016/12/the-peril-of-overhyping-precision-medicine/510326/>. Accessed Nov 16, 2022.

This op-ed historicizes how the biomedical sciences, particularly those concerning genes, tend to over-inflate their promises. The article argues for the importance of science literacy to differentiate the facts from friction.

3. Joyner MJ, Paneth N. Promises, promises, and precision medicine. *The Journal of clinical investigation*. 2019;129(3):946-948. doi: 10.1172/JCI126119.

This commentary retrospectively evaluates Francis Collins's predictions for precision medicine in his 1999 article "Medical and Societal Consequences of the Human Genome Project." Authors Michael Joyner and Nigel Paneth demonstrate how a genomics-driven precision medicine failed to live up to its hype, urging the biomedical research community to reconsider their fixation on the human genome.

4. Khoury MJ. Precision Public Health and Precision Medicine: Two Peas in a Pod. <https://blogs.cdc.gov/genomics/2015/03/02/precision-public/>. Accessed Nov 17, 2022.

This blog post by Muin Khoury, the founding director of the Centers for Disease Control's (CDC's) Office of Genomics and Precision Public Health, is one of the first to define and describe "precision public health" (PPH). Drawing parallels to how genomic technologies promise rapid advancements for precision medicine, Khoury predicts how genomics would likewise make public health precise.

5. Khoury MJ, Holt KE. The impact of genomics on precision public health: beyond the pandemic. *Genome Medicine*. 2021;13(1):67. doi: 10.1186/s13073-021-00886-y.

Formalizing PPH in academic literature, Khoury's journal editorial provides an updated conceptualization of PPH in light of new genomics research resulting from the COVID-19 pandemic. The authors present a strategic vision of how genomics can marry public health to provide "the right intervention to the right *population* at the right time,"

mirroring precision medicine's aim of targeting "the right intervention to the right *patient* at the right time."

6. Oude Munnink BB, Nieuwenhuijse DF, Stein M, et al. Rapid SARS-CoV-2 whole-genome sequencing and analysis for informed public health decision-making in the Netherlands. *Nature Medicine*. 2020;26(9):1405-1410. doi: 10.1038/s41591-020-0997-y.

This article presents peer-reviewed research on how genomics, together with epidemiological data, reliably informed public health decision making about COVID-19 community transmission in the Netherlands. This provides a concrete example for how, under the PPH paradigm, infectious disease genomics complements traditional public health policy making.

7. Vogel MJ, Rosenberg CE. *The Therapeutic Revolution*. Philadelphia: University of Pennsylvania Press; 1979.

This classic collection of essays historicizes the social and cultural contexts of the nineteenth and twentieth centuries in which American medicine, particularly therapeutics, took shape. The authors demonstrate how in American medical history, science, institutions, practitioners, and medical knowledge are intertwined, with the development of therapeutics influencing and being influenced by advances in mental hospitals, nursing, and insurance.

8. Macklin MT. The Role of Heredity in Disease. *Medicine (Baltimore)*. 1935;14(1):1-75. doi: 10.1097/00005792-193502000-00001.

This article by physician, medical geneticist, and eugenicist Madge Macklin was one of the first to systematically examine and unfoundedly expound the hereditary patterns of human disease, a broad term used at the time encompassing prejudiced categorizations such as "feeble-mindedness" or any general intellectual disability. Mathematically gifted, Macklin justified her work by quantitatively analyzing vast amounts of pedigrees.

9. Comfort N. *The Science of Human Perfection: How Genes Became the Heart of American Medicine*. Illustrated ed. New Haven: Yale University Press; 2014.

This book historicizes the emergence of medical genetics and eugenics in nineteenth century America. Portraying the physician, scientific, political, and public health actors, historian of science Nathaniel

Comfort demonstrates how medical genetics emerged from the public health eugenics of the Progressive era. He argues that both eugenics and modern medical genetics encompassed goals of reducing suffering in the short term and improving the population in the long term.

10. Roberts MC, Fohner AE, Landry L, et al. Advancing precision public health using human genomics: examples from the field and future research opportunities. *Genome medicine*. 2021;13(1):97-107. doi: 10.1186/s13073-021-00911-0.

This opinion piece takes a deeper dive into the latest materialized examples of PPH in five facets of public health research: biostatistics, epidemiology, health policy, environmental health, and social and behavioural health.

11. Khoury MJ, Gwinn M, Duggal P. The Public Health Impact of COVID-19: Why Host Genomics?. 2020.

This blog post provides another relevant example of how genomic research catalyzed by the pandemic has sparked more vocal calls for PPH.

12. Fisher I, Fisk EL, Life Extension Institute I. *How to Live: Rules for Healthful Living, Based on Modern Science*. New York: Funk and Wagnalls; 1915.

This book by renowned economics and statistician Irving Fisher captures his theories on how hygiene and eugenics can be justifiable tools to promote preventative health.

13. Bix AS. Experiences and Voices of Eugenics Field-Workers: 'Women's Work' in Biology. *Social Studies of Science*. 1997;27(4):625-68. doi: 10.1177/030631297027004003.

This article offers detailed insights into the fundamental role women played as eugenic "field-workers," who collected hereditary data from families for the Eugenics Record Office at Cold Spring Harbor, in the 1910s–1930s. With most having professional training in science and medicine, women were active and independently thinking agents in the eugenic research process. When the science seemed to undermine rather than support eugenic hypotheses, some women expressed concern but were ultimately shunned for their questioning. Through highlighting women in eugenic America, this article critically evaluates women's role in early twentieth century science.

14. Leavitt JW. "Typhoid Mary" Strikes Back Bacteriological Theory and Practice in Early Twentieth-Century Public Health. *Isis*. 1992;83(4):608-629. doi: 10.1086/356292.

Re-analyzing the archetypal case of the asymptomatic carrier of infectious disease "Typhoid Mary" Mallon, this article argues that bacteriology nudged public health officials' attention away from social determinants of health and toward an ethos of personal responsibility that effectively absolved themselves of duty for the public's health.

15. Andermann A. Taking action on the social determinants of health in clinical practice: a framework for health professionals. *Canadian Medical Association journal (CMAJ)*. 2016;188(17-18):E474-E483. doi: 10.1503/cmaj.160177.

This article offers a concrete framework for how clinicians could identify and intervene with a patient's social determinants of health. Specifically, the article presents evidence for how social determinants of health such as socioeconomic status and education are linked to health outcomes, and how clinicians can make an impact on the patient, practice, and community levels to mitigate patients' social needs.

16. Benjamin R. *Race after technology: abolitionist tools for the new Jim Code*. Cambridge, UK: Polity; 2019.

This book examines the ways in which race is encoded in social algorithms. Benjamin shows how programming choices exacerbate social inequities through intensifying racial hierarchies, overlooking societal divisions, or operating under a false claim to be racially unbiased.

17. Golding N, Burstein R, Longbottom J, et al. Mapping under-5 and neonatal mortality in Africa, 2000–15: a baseline analysis for the Sustainable Development Goals. *Lancet (London, England)*. 2017;390(10108):2171-2182. doi: 10.1016/S0140-6736(17)31758-0.

This article exemplifies how social and environmental data—without the use of genomics—could be successfully harnessed using computational tools to facilitate public health decision-making.

18. Thorpe L, Chunara R, Roberts T, et al. Building Public Health Surveillance 3.0: Emerging Timely Measures of Physical, Economic, and Social

Environmental Conditions Affecting Health. *American journal of public health (1971)*. 2022;112(10):1436-1445. doi: 10.2105/AJPH.2022.306917.

This systematic review is a handy guide to data sources capturing nuanced measures of physical, economic, and social environments that impact health. The authors further describe statistical and computational tools to harness that information, with applications to improving public health surveillance.

19. Khera AV, Emdin CA, Drake I, et al. Genetic Risk, Adherence to a Healthy Lifestyle, and Coronary Disease. *The New England journal of medicine*. 2016;375(24):2349-2358. doi: 10.1056/NEJMoa1605086.

Meta-analysis of four GWAS studies examining the relative influence of genetic and lifestyle factors associated with coronary artery disease. Fifty genetic loci associated with hereditary predisposition to coronary artery disease are known; when combined in a polygenic score, they are highly predictive of disease risk. The authors note, however, that lifestyle factors are also significant in determining risk. Could healthier lifestyle choices mitigate genetic risk? The authors offer a hopeful prognosis.