Chapter 8

Genomics and Public Health: Ethical, Legal, and Social Issues

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Introduction

This article is divided into sections discussing key ethical, legal and social implications of genomic science for public health. It includes resources that may help public health professionals develop an approach for resolving these issues as they take shape now and in the future. Ethical, legal and social are somewhat arbitrary categories because these issues are almost always intertwined. For example, the potential misuse of genetic information for purposes of discrimination and stigmatization raises ethical concerns, points out the need for protective legislation, and describes a significant social problem. Ethical, legal and social issues may be either real or only perceived, but the distinction between reality and perception often does not matter to consumers and policy makers. Examining these issues from a public health perspective can be helpful in either instance.

Ethical, Legal and Social Issues (ELSI)

During the last decade and a half, the National Human Genome Research Institute (NHGRI) has supported an extensive program of scholarly work on the ethical, legal, and social implications of genetics research, known collectively as ELSI. ELSI inquiry examines the values underlying the use of new genetic technology, ideally before it is in use.

Public Health Ethical, Legal and Social Issues (PHELSI)

Since 1999, genetic research has increasingly focused on the discovery of human genetic variations linked to susceptibility to common illnesses, rather than on the rare, **single-gene disorders** that have been the traditional focus of clinical genetics. Efforts to understand and prevent these more widespread conditions, which often involve complex gene-environment interactions, fall under the purview of public health. We have used the term PHELSI to refer to ethical, legal and social implications that arise when genomics is used for the advancement of public health.

Single-Gene Disorder
Refers to a disorder that is determined by a single gene.

Is PHELSI Different From ELSI?

The ethical, legal and social implications of using genetic technology in medicine have been the subject of a rich and growing literature.[1] Most of this scholarship has applied principles of bioethics to the use of genetics in medical research and practice, and emphasizes the individual and the patient-physician relationship.

At the same time, there has been a renaissance in the literature on public health ethics in the past decade. Public health actions are intended for the public good, defined either in terms of groups of individuals or the population as a whole.[2] The public health perspective is at the center of the distinction between PHELSI and ELSI.

Public Health Ethics

While scholars have considered the ethical principles underlying public health practices for many years, more recent scholarship has made a distinction between bioethics and principles more specifically relevant for public health. Lachmann writes about the "conflicts between the priorities of public health and the emphasis of medical ethics on the duty of the doctor to the individual patient".[3] Lane et al. describe the historical identification of bioethics with the rights of the individual, limiting its value to address current issues of public health—especially issues relating to health disparities among different demographic groups.[4]

Rothstein has identified an "ongoing need to reassess [public health's] scientific, ethical, legal, and social underpinnings",[5] and Cole has pointed out that most public health programs require an explicit fundamental justification which can be based upon principles of morality.[6] Other scholars have recently formulated frameworks for the application of ethics in public health,[7,8] and have identified literature uniquely appropriate for considering ethical issues in public health.[9] The American Public Health Association has promulgated a Public Health Code of Ethics.[10]

As genomics is increasingly studied and practiced in the public health context, it is useful to analyze the ensuing ethical, legal and social issues using a public health framework that emphasizes:

- the use of science to further the health of the population, rather than the health of particular individuals,
- the welfare of the collective as well as the autonomy of the individual,
- issues of discrimination and health disparities,
- the historical relationship between public health and distributive justice (the societal obligation to be fair when providing health resources to different groups), and
- balancing the prevention of disease against the curing of illness.[11]

Scrutiny of the Tuskegee syphilis study and the discriminatory sickle cell screening programs of the 1970s has led public health officials to emphasize the avoidance of social harms to particular groups (e.g., African Americans); in addition to the desire to avoid harm, public health ethics are also concerned about treating all groups with fairness. The principle of social justice seen through the lens of public health ethics requires that differences in race/ethnicity, socioeconomic status, and inherited family background should not skew how the benefits of genomic research are distributed.[4,12] Public health policy offers a variety of safeguards against potential inequities, from public education to collective action.

Ethical Issues in Public Health Genomics

As genomic research points out new ways to identify persons at risk, using this knowledge presents new ethical challenges. Most of the literature on ethical issues related to public health genetic screening deals with mandatory newborn screening programs for single-gene disorders (e.g., phenylketonuria and sickle cell disease). Tests for these disorders have high predictive value, and treatment can either eliminate or reduce the severity of symptoms. Tandem mass spectrometry, in contrast, has delivered an expanded list of potential newborn screening tests, for which predictive value and usefulness are less certain.[13] See *Chapter 5, Newborn Screening for MCAD Deficiency*, for more information.

Increasingly, screening programs are being suggested for the identification of individuals at risk for chronic disease (e.g., cystic fibrosis, hemochromatosis, coronary heart disease, and cancer), suggesting a different balance of ethical considerations.[14] An even more divisive ethical area is prenatal screening for conditions without definitive or effective treatment, e.g., beta-thalassemia (Cooley's anemia), Tay-Sachs disease, or serious or fatal **trisomies**. Some have argued that the focus of public health efforts should be on "phenotypic prevention" (the prevention of disease manifestation) rather than "genotypic prevention" (avoiding the birth of individuals with a given genotype).[15] Others have pointed out the benefits of prenatal screening as a public health intervention, given that it provides couples with risk-related information. The informed couple can use this information in their decision-making, and make specialized plans in the case of a decision to deliver an infant with a genetic condition.[16]

Trisomy
The presence of an extra chromosome, resulting in a total of three chromosomes of that particular type instead of the usual pair.

The use of family history to identify individuals at risk for disease has been a traditional tool of medical diagnosis and is now being tested as a potentially useful public health tool for identifying at-risk populations.[17] See *Chapter 6, The Family History Public Health Initiative*, for more information. Similarly, information on averted deaths (e.g., from arrhythmia in long QT syndrome) and on cause of death from death records has been proposed as a basis for identifying family members at risk for the same disease.[18] The use of family history and death records, however, raises the issue of privacy rights of persons alive or dead.[19]

Another area of increasing public health interest is the use of existing biological samples gathered for one purpose, e.g., blood spots from newborn screening programs, or blood collected for the National Health and Nutrition Examination Survey (NHANES), (see *Chapter 1, National Health and Nutrition Examination Survey)*, for other applications, such as epidemiologic research and identifying individuals with similar risk profiles who could benefit from screening.[20, 21]

Each of these current and potential public health activities raises issues of informed consent, confidentiality of genetic information, potential stigmatization and discrimination, and the balancing of individual autonomy against the public health goal of collective action. Appropriate analysis of these issues requires care in order to maintain a clear distinction between activities undertaken for research and those to be implemented in public health practice.[22, 23]

Legal Issues in Public Health Genomics

Each of the ethical issues identified above can also be considered from a legal perspective. In general, laws and policies to guide the use of genomic technology lag far behind its actual application in medical and public health practice. While legal scholars have developed useful models of legislation, summaries of relevant state legislation maintained by the National Conference of State Legislatures disclose diverse policies, variable responses among states to the need for legislation, and lack of consensus on whether federal, state or mixed legislation is most appropriate.[24] Variance also exists within and among states in the spread of protections offered by state public health records privacy laws, further complicated by Health Insurance Portability and Accountability Act (HIPAA) privacy rules on unauthorized disclosures of health information.[25]

A growing area of concern is commercialization arising from the private ownership of genomic technology, and the increasing conflict between financial incentives driving the marketing of biomedical technology and the public health goal of maximizing the public's health through cost-effective interventions.[26, 27]

Social Issues in Public Health Genomics

The incorporation of genomics in public health practice has significant implications for social policy. As we consider the implications of each new genomic intervention in public health, it is essential that we also consider the cumulative impact of genomics on the nature of our society. Two related areas of social concern are fears of a rebirth of eugenics and the potential of genetics to widen health disparities between different demographic groups.

Historians have pointed out the intersections between public health and eugenics during the early 20th century and the danger that new genetic technologies might

be misused to serve goals other than that of preventing disease. Pernick warns that "Past similarities between eugenics and public health serve as an alarm clock for all the health sciences, not as a lullaby for genetics",[28] and Duster has expressed concern that given the discriminatory context of American society, the application of new genetic technologies could lead to a return of eugenics through the "back door".[29] Conversely, Kitcher has described the positive potential of "utopian genetics" to serve public health goals, given adequate public education and equal access to genetic technology.[30]

The "double-edged sword" of genetics pointed out by many commentators can result in either the widening or the narrowing of health disparities among the population. An increasing amount of genetics research is focused on chronic diseases, and highlights group disparities in disease frequency. Disparities in access to the benefits of genomic research, or the distortion of research findings to stigmatize racial and ethnic minorities, could further widen health inequities.[31] If the new tools of genetics are made available to all who could benefit, however, the prevalence of many chronic diseases could be reduced in the American population.

Engagement and Education to Address PHELSI

How can we realize the positive potential of genomics as a tool of public health while avoiding social harms? The literature suggests that the related strategies of public engagement and public education are crucial.

The active engagement of an informed public is essential to ensuring that these new, powerful scientific tools are used in the public interest to achieve improvements in total population health. A large and growing body of literature has developed to define and support a resurgence of civic participation in policy making.[32, 33] The NIH-funded project *Communities of Color and Genetics Policy* has demonstrated a successful process for engaging minorities in policy development to address concerns of special relevance to African-American and Latino citizens.[34] In addition to participation in community policy making, the representation of diverse groups on newborn and chronic disease advisory committees and among key genetics decision-makers should be a major priority of public health.

The most important factor for determining whether genetics will enhance or impede public health goals is the extent to which the public is adequately informed about genetics. Unfortunately, a large share of public knowledge about genetics has been derived from mass media, highlighting presumed genetic breakthroughs, and fostering a sense of genetic determinism. The interplay of genes and environment in most diseases is not widely understood.[35]

Public health leadership should promote citizen education in several ways:

- *Information to the Media:*
  Public health professionals in practice and academia should become providers of accurate information on genetics to the media, in order to counterbalance the more sensational reporting that too often occurs. The public health viewpoint can add depth and social concern to the sources often tapped by the media for information: biomedical researchers and corporate biotech and pharmaceutical firms.

- *Education:*
  Public health practitioners have a role in responding to teaching requests from social and civic organizations, and in providing "information-on-demand" resources relating to genetics.[36, 37] In addition, academically-based public health professionals have the responsibility of assuring that future public health practitioners are knowledgeable about public health genetics and PHELSI issues.[7, 38] A fundamental, long-term educational strategy also includes K-12 education. Since most formal education in genetics is acquired by the end of high school, it is essential that this basic education be accurate and stress the ethical, legal and social implications of genetics as well as the science.[39] If our youth learn about genetics as one of several factors influencing health and disease, and as a growing technology that can be put to beneficial or harmful uses, they will have the intellectual background to interpret and judge other sources of information on genetics that they encounter as adults.

A Genetic Agenda for Public Health

Our brief review of the literature on PHELSI suggests several key roles for public health professionals in public health agencies, academic institutions, or other organizations whose work involves improving community health. In addition to learning about and using genetic tools that can be incorporated in public health practice, public health professionals must address the ethical, legal and social issues that arise. They can carry out this role in their practice and by encouraging public engagement, promoting public education, and becoming effective providers of balanced information. By assuming these responsibilities, public health professionals will help assure that genetic technologies are applied in ways that are ethically, socially, and legally just, and consonant with the values of a diverse society.[2]

References

For an expanded list of references for this chapter, please visit our Web site at http://www.cdc.gov/genomics/activities/ogdp/2003.htm

1. Annas GJ, Elias S, eds. *Gene Mapping: Using Law and Ethics as Guides*. New York: Oxford Univ. Press; 1992.
2. Institute of Medicine, Board on Health Promotion and Disease Prevention. Committee on Assuring the Health of the Public in the 21st Century. The future of the public's health in the 21st century. Washington, DC: National Academies Press; 2003:28-33. Also available at: http://www.iom.edu/report.asp?id=4304. Accessed Feb 16, 2004.
3. Lachmann PJ. Public health and bioethics. *J Med Philos* 1998;23:297-302.
4. Lane SD, Rubinstein RA, Cibula D, et al. Towards a public health approach to bioethics. In: Cantwell A-M, Friedlander E, Trainor ML, eds., Ethics and Anthropology: Facing Future Issues in Human Biology, Globalism, and Cultural Property. *Ann NY Acad Sci* 2000;925:25-36.
5. Rothstein MA. Rethinking the meaning of public health. *J Law Med Ethics* 2002;30: 144-9.
6. Cole P. The moral bases for public health interventions. *Epidemiology* 1995;6:78-83.
7. Callahan D, Jennings B. Ethics and public health: forging a strong relationship. *Am J Pub Hlth* 2002;92:169-76.
8. Kass NE. An ethics framework for public health. *Am J Public Health* 2001;91:1776-82.
9. Beauchamp DE, Steinbock B, eds. New Ethics for the Public's Health. New York: Oxford Univ. Press; 1999.
10. American Public Health Association. Public Health Code of Ethics. Washington, D.C;2001. Web site. Available at: http://www.apha.org/codeofethics. Accessed Jan 11, 2004.
11. Michigan Center for Genomics and Public Health. Ethical, legal, and social issues in public health genetics. Ann Arbor, MI; 2002. Web site. Available at: www.sph.umich.edu/genomics/focus_areas/PHELSI.html. Accessed Jan 11, 2004.
12. Turnock BJ. *Public Health: What It Is and How It Works*. Gaithersburg, MD: Aspen Publishers; 1997.
13. Atkinson K, Zuckerman B, Sharfstein JM, Levin D, Blatt RJ, Koh HK. A public health response to emerging technology: expansion of the Massachusetts newborn screening program. *Pub Health Rep* 2001;116:122-31.
14. Burke W, Coughlin SS, Lee NC, Weed DL, Khoury MJ. Application of population screening principles to genetic screening for adult-onset conditions. *Genet Test* 2001;5:201-11.

15. Khoury MJ. Relationship between medical genetics and public health: changing the paradigm of disease prevention and the definition of a genetic disease. *Am J Med Genet* 1997;71:289-91.

16. Clayton EW. What should be the role of public health in newborn screening and prenatal diagnosis? *Am J Prev Med* 1999;16:111-5.

17. CDC. Family history as a tool for public health and preventive medicine: a public health perspective. Atlanta, GA; 2003. Web site. Available at: http://www.cdc.gov/genomics/info/perspectives/famhistr.htm. Accessed Dec18, 2003.

18. Liebman J. Some legal, social, and ethical issues related to the genetic testing revolution, as exemplified in the long QT syndrome. *J Electrocardiol* 2001;34 Suppl: 183-8.

19. Botkin JR. Protecting the privacy of family members in survey and pedigree research. *JAMA* 2001;285:207-11.

20. McQuillan GM, Porter KS, Agelli M, Kington R. Consent for genetic research in a general population: the NHANES experience. *Genet Med* 2003;5:35-42.

21. Pelias MK, Markward NJ. Newborn screening, informed consent, and future use of archived tissue samples. *Genet Test* 2001;5:179-85.

22. Press N, Clayton EW. Genetics and public health: informed consent beyond the clinical encounter. In: Khoury M, Burke W, Thomson E, eds., *Genetics and Public Health in the 21st Century*. New York: Oxford Univ. Press;2000:505-26.

23. Holtzman NA, Andrews LB. Ethical and legal issues in genetic epidemiology. *Epidemiol Rev* 1997;19:163-74.

24. National Conference of State Legislatures. Genetic Technologies Project. Denver, CO;2003. Web site. Available at: http://www.ncsl.org/programs/health/Genetics/gtp-o.htm. Accessed Jan 11, 2004.

25. Gostin LO. National health information privacy regulations under the Health Insurance Portability and Accountability Act. *JAMA* 2001;285:3015-21.

26, Gollust SE, Hull SC, Wilfond BS. Limitations of direct-to-consumer advertising for clinical genetic testing. *JAMA* 2002;288:1762-7.

27. Caufield T. Genetics services and market forces. *Nat Rev Genet* 2002;3:734.

28. Pernick MS. Eugenics and public health in American history. *Am J Public Health* 1997;87:1767-72.

29. Duster T. *Backdoor to Eugenics*. New York: Routledge; 1990.

30. Kitcher P. *The Lives to Come*. New York: Simon & Schuster; 1996.

31. Lee SS, Mountain J, Koenig BA. The meanings of "race" in the new genomics: implications for health disparities research. *Yale J Health Policy Law Ethics* 2001;1:33-75.

32. Yankelovich D. *The Magic of Dialogue*. New York: Simon & Schuster; 1999.

33. Garland MJ. Experts and the public: a needed partnership for genetic policy. *Public Underst Sci* 1999;8:241-54.

34. Citrin T. Communities of color and genetics policy project: policy reports and recommendations. Ann Arbor, MI:University of Michigan;2001. Web site. Available at: http://www.sph.umich.edu/genpolicy. Accessed Jan 11, 2004.

35. Khoury MJ, Thrasher JF, Burke W. Challenges in communicating genetics: a public health approach. *Genet Med* 2000;2:198-202.

36. EvansGroup. Washington state genetics education plan. Seattle, WA; 1997. Web site. Available at: http://mchneighborhood.ichp.edu/wagenetics/media/wagenetic.pdf. Accessed Jan 11, 2004.

37. Kaye CI, Proud VK. Genetics education: guidelines and resources for the public health. In: Freeman SB, Hinton CF, and Elsas, II, LJ, eds., Genetic Services: Developing Guidelines for the Public's Health. Atlanta, GA: Council of Regional Networks for Genetics Services (CORN); 1996.

38. Modell SM, Citrin T. Ethics instruction in an issues-oriented course on public health genetics. *Hlth Educ Behav* 2002;29:43-60.

39. McInerney JD. The Human Genome Project and biology education. *BioScience* 1995;45:786-91.