

ETHICAL, LEGAL AND SOCIAL ISSUES IN PUBLIC HEALTH GENETICS
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Key Points

- **Population-oriented versus Individualized Focus**
Public health is devoted to the care of groups of people and populations. This mission contrasts with the traditional medical emphasis on the clinical care of the individual patient. Points of intersection exist. As knowledge of individual genetically-associated disease predispositions becomes more refined, it will be possible to selectively test individuals for particular types of alleles, but the goals and protocols followed in widespread genetic screening will be aimed at the population. The public health perspective has implications for the administration and conduct of population-based programs, and the ethical principles that guide them.
- **Collective Welfare versus Individual Autonomy**
The emphasis in genetic counseling, and in medical practice in general over the last two decades, has been on individual autonomy – the right of individuals to decide for themselves on their particular form of care. At times individual autonomy may be in agreement with the welfare of the group to which they belong. Public health often faces situations where the exercise of individual autonomy, e.g., a person's decision not to undergo genetic testing, may affect others. Practitioners and policy makers may adopt additional sets of ethical principles, such as the maximization of social best interests, to protect important public interests. Ethical decisions in public health may also depend on more collective ethical theories - utilitarianism stressing the aggregate good of the population, and communitarianism stressing collaboration among persons to make benefits available for all members of society.
- **Discrimination**
Since the public disclosure of the U.S. Public Health Service-initiated Tuskegee Syphilis Study, in which poor black sharecroppers were denied treatment to follow the natural history of the disease, discussion on the rights of study participants and underserved populations has become more vigorous. The distinctive aspect of public health efforts is that they touch many people at one time. HIV trials in American communities and abroad, patenting of genes on an international scale, population-wide tissue banking, and enrollment of groups in ethnocentric gene studies have continued to test people's sense of fair and just treatment for diverse groups. Legal actions and legislative developments at the state and national levels are leading to new standards to avoid discrimination of the disabled and those with genetic predispositions. Public policymakers continue to develop protections to avoid discrimination in an increasing number of social contexts.
- **Health Disparities**
Research into human genetic variation depends on the study of specific groups to identify and locate significant disease-associated polymorphisms. The results of this research can aid population sub-groups with higher gene frequencies for particular medical conditions or drug sensitivities, but it can also have a stigmatizing effect.

One major question to be asked is: “How can genetics be used to reduce and not widen health disparities?”. If the fruits of genetic discovery are allocated strictly by the marketplace, the expense of genetic technologies can widen disparities in health and usage; reproductive genetic testing could marginalize groups not fitting the stereotypical profile for selecting the “perfect child”; and testing could result in stigmatization and discrimination of the disabled and underserved. Broader sets of ethical guidelines and innovative forms of dialogue are being used to make certain genetic technologies are used to reduce and not widen health disparities between groups.

- **Distributive Justice**

When resources are in short supply or are costly, the option to undergo a particular kind of genetic test or medical intervention will be unavailable to some members of society. Many forms of genetic testing, preimplantation genetic diagnosis which may require up to \$40,000 to yield a successful birth, or BRCA1 testing for familial breast cancer, more than \$2,000 per test, surpass the average person’s budget. Policymakers have proposed that government could provide partial subsidies or insurance could provide partial coverage with co-pays for these services. The adoption of such solutions is a matter of distributive justice regarding the ethical allocation of resources among the various members of society. Distributive justice questions may differ according to the particular type of technology and the perceived societal benefit. Given the increasing ability to detect genetic mutations in almost all persons, recent writings have also begun to look at the issue of whether distributive justice alone is adequate to decide on the allocation of resources, or if more communitarian principles should take priority.

- **Limitations on the Principle of Informed Consent**

Informed consent has been the hallmark of enrollment in research studies and in medical procedures over the last half century. Individuals should be given the opportunity to make informed, educated decisions about their care. In situations where the individual’s capacity to decide is impeded – such as with young children, the mentally impaired, or those with severe physical trauma, a proxy decision maker is often called upon. Public health poses additional challenges for the notion of informed consent, since the intervention is imposed on an entire group, not just one individual. Who is to decide whether to allow a set of researchers to enroll the community in a research study, or to draw samples on the community’s members? Should it be a selected member of the group or should each member for themselves? How would the informing or educating of the entire group take place? Should the community be consulted on the design of the protocol? Informed consent in the public health context is an evolving concept, with new ideas constantly being voiced in the literature.

- **Individual Privacy versus Group Rights**

In expanding beyond the patient-provider relationship to concerns equally faced by the family and community, public health complicates the notion of the individual’s basic right to privacy. Authors have already noted that single gene defects like

Huntington's disease and familial breast cancer impact the entire family and often call for disclosure and shared decision-making between family members. HIV contact tracing has also required a departure from the rule of privacy to stop further spread. Although genetic conditions are not transmitted like infectious diseases, different relatives can be affected by the same inherited condition. Enrollment of relatives in linkage studies and use of newborn blood spots and surveillance results in population frequency studies have also engendered debate among professionals and policy makers on the ethics of sharing confidential information for the public good.

- **Policy Challenges Faced by Public Health Departments**

Due to the scale of their undertakings, community and public health departments experience challenges to institutional policy quite distinct from what other health care institutions, such as hospitals and clinics, might encounter. In many state health departments, newborn blood spots have been accumulating for over twenty years. Appropriate storage policies at the institutional and state government levels are still in flux. There is a great need to protect public health data in general, which has not been evenly met by public health records privacy laws. At the same time, public health departments are in need of reviewing data they have accumulated for population health and quality assurance purposes. With the recent passage of the Health Insurance Portability and Accountability Act (HIPAA) privacy rules, legal challenges are forcing a revisiting of health department privacy policies, which can at best be classified as "works-in-progress."

- **Community-Based Approaches**

When investigators approach whole communities to conduct genetics research studies, they must be attentive to community concerns both to engage community members in the study and to allow the community to benefit from its participation. Community and cultural sensitivity is especially important as genetic studies expand beyond single populations to diverse groups holding different sets of priorities and beliefs. Research institutions are in the process of articulating community-based principles for social and health science research. The principles are not fixed, but are often worked out by the investigative team and community leadership together. Effective ways of engaging communities in genetics studies are continually being updated and assessed in the literature.

- **Public Deliberation**

The establishment of medical guidelines often is an outcome of professional gatherings and meetings of professional societies. Public health efforts – environmental clean-up, introduction of new forms of mass population screening, sharing of newborn blood spot information with third parties – impact the public at large and generally require public input before becoming ingrained policy. Over the last two decades volatile health care decisions, including those involving genetic and reproductive technologies, have been brought to public attention and deliberated in public forums. Committees containing members of the public or seeking public input have also deliberated over the specifics of releasing various forms of genetic testing into the mainstream. New examples of public deliberation in health care are arising

and being written about all the time. The ideal composition and operation of bodies engaged in public deliberation is a topic of continuing discussion.

- **Ethics of Different Prevention Strategies**

Interventions in the health care arena may be classified as primary, secondary, or tertiary. Tertiary interventions act to prevent the complications of disease and further deterioration. Medical and surgical practice have traditionally occupied this phase of health care. Public health is much more geared towards primary prevention – the maintenance of health and avoidance of disease before it manifests, and secondary prevention – early detection and management of disease once it has manifested. The prioritization of these strategies by government and insurance companies is a mixed decision resting on cost-benefit analysis and input from various interests including the public. The decision of whether to implement public health programs at any one of these levels can often be contentious. Decisions on whether to proceed with the supplementation of cereal with folate to prevent neural tube defects, the inclusion of cystic fibrosis screening in newborn genetic screening programs, and population-level screening for hemochromatosis – a disease involving iron overload – are being actively debated. The debates, based on pragmatic and ethical considerations, are not yet settled, with new information and perspectives arising in the literature and in policymaking settings.

- **Societal Benefits and Burdens**

Whatever risks and benefits await the single patient undergoing a medical intervention get multiplied many times over when applied to an entire community or society. Many factors enter the cost-benefit equation – economic, epidemiologic, and intangible value judgments. Political factors have also played a part in the implementation of major genetic screening programs. The ethics of starting a program for the population go considerably beyond the ethics of clinical trials. A decision to allocate services to one group may mean subtracting related services from another. Taking a genetic approach towards a social problem may also influence funding for more environmentally-oriented approaches. These complexities show why ethical considerations are an important ingredient in the institution of society-wide programs, and why each instance differs.

- **The Environment**

Modern public health's purview stretches from the genome central to our cells to the physical environment and occupational settings. Government has passed major protective legislation over the last thirty years setting health standards and exposure limits for food, air, and the workplace. The Presidential / Congressional Commission on Risk Assessment and Risk Management has recommended that inter-individual variations be incorporated into protections of population subgroups. It is a matter of debate as to whether environmental protection implies physical clean-up of the workplace or genetic testing to rule-out chemical sensitivities. Investigation of group and individual genetic susceptibilities under the Environmental Genome Project is compelling a closer look at the psychosocial consequences of learning one's susceptibilities to environmental exposures. Also, environmental groups are

beginning to devote attention to the social implications of genetic testing for socially marginalized groups whose neighborhoods often exist in environmentally risky areas.

- **Mandatoriness versus Voluntariness**

The mandatory nature of many public health programs is what distinguishes them from medical interventions guided by strict patient autonomy. The *parens patriae* doctrine, or protective role of the state, is a hallmark of public health policy. Genetics in public health has been evolving, with the discontinuation of some programs (adult sickle cell screening) that in the past posed social and ethical dilemmas. With other programs, phenylketonuria (PKU) for example, a national consensus is still lacking as to whether newborn screening should be voluntary or mandatory. Sickle cell screening for newborns has become a universal practice. In the last few years, the issue of parental consent for newborn screening as a whole has also arisen. Additional ethical issues involving what level of compulsion should exist for screening of common chronic diseases will continue to challenge people with differing perspectives.

- **Genotypic versus Phenotypic Prevention**

A major issue in deciding what types of genetic tests ought to be performed is whether the goal should be to prevent births of children with a characteristic disease genotype or genetic arrangement, or whether testing and follow-up should seek to alleviate the *manifestation* of the disease – its phenotypic appearance. Decisions in many clinical genetic testing programs are based on the sheer existence of a significant disease gene or chromosomal rearrangement in the fetus. Whether this form of prevention should be the mission of public health programs employing prenatal testing is a continuing source of acrimony, with state programs occupying different positions. Broader still is the question of whether modern molecular genetics can avoid the trap of eugenics when harnessed for population health.