

Health Care Policy Issues as a Result of the Genetic Revolution: Implications for Public Health

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The genetic revolution has spawned 4 distinct issues of universal importance to health care policy and society: genetic privacy, regulation and standardization of genetic tests, gene patenting, and education.

Adequate policy advancements for these 4 areas are lacking. Stringent controls must be placed on individual health records to prevent their misuse. Genetic testing within the clinical setting should undergo thorough evaluation before it is implemented. Regulations are needed to prevent the monopolization of DNA sequences.

Society and health care professionals must be educated about the scope of genetic testing because current trends indicate that genetic and molecular assessments are destined to become a routine component of health care. (*Am J Public Health*. 2005;95:385-388. doi:10.2105/AJPH.2003.026708)

THE SUCCESS OF THE HUMAN

Genome Project (HGP) has led to the emergence of challenging policy issues and unparalleled opportunities of immediate and future concern. Genetic and molecular technology developed during the HGP has already begun to revolutionize medical research, practice, and health care delivery. Advances in the identification of genes involved in hereditary disorders, drug metabolism, and dose response (pharmacogenomics and pharmacogenetics), as well as the identification of single nucleotide polymorphisms, have only begun to make apparent the potential of genetic testing.¹⁻⁸

Ideally, public health use of molecular and genetic tools will allow for population screenings and identification of disease before the onset of clinical symptoms.²⁻⁹ Furthermore, these tools will allow for individualization of drug treatment by establishing effect and dose determination on the basis of epidemiological pharmacogenetics and for implementation of behavioral modification on an individual basis by determining disease risk associated with the expression of high-risk genes.¹⁻³ However, with the potentials of population-based genetics are inherent issues that require continuous assessment to ensure that the technology benefits society and that it is not simply a costly byproduct of scientific evolution.

Although numerous issues have emerged from the genetic

revolution, 4 particular issues are of universal importance to science, business, politics, and society: genetic privacy, regulation and standardization of genetic tests, gene patenting, and education. If unresolved, these issues could undermine and inhibit any potential benefits of genetic technology to society as a whole and could render a potentially invaluable tool as misguided science.

ARRIVAL OF THE SEQUENCED GENOME

When the HGP was initiated by the National Institutes of Health and the US Department of Energy in 1990, planners and futurists were aware of the potential effect its completion might have, although certainly no prediction could have foreseen all pertinent issues. The project's designers did, however, devote 3% to 5% of the total funding for the project to the analysis of ethical, legal, and social implications.^{1,2,10,11} The desire to avert any potentially negative issues and the establishment of specific funds for this purpose was unprecedented in the scientific community. Before the HGP, legal and ethical issues were not evaluated prospectively in scientific endeavors but were discussed primarily in response to developments after the scientific knowledge had made its impact. Since the HGP was declared officially complete by the US Department of Energy in April 2003,¹² unresolved as well

as new issues have emerged as products of the project are now beginning to make a stronger impact on science and society alike.

Genetic Privacy

Genetic testing has caused the debate of patient confidentiality to resurface. Legislation concerning patient privacy is limited to individual state-enforced legislation that varies between states and primarily concerns medical records.^{13,14} However, with the advent of genetic technology, current trends in basic and clinical research are shifting away from direct human involvement to the use of genetic and molecular materials. The shift in focus necessitates greater emphasis on confidentiality because using the genetic profile of an individual has different implications than using a physical being. Genetic profiling often can be more revealing than previous research techniques and has more potential predictive value. The inherent nature of the insurance industry may compel insurers to incorporate genetic information in their actuarial risk analysis. Genetic information obtained this way can be misused by insurance companies to deny or cease coverage.

Regulation and Standardization of Genetic Tests

Genetic testing has already become a prominent tool for establishing paternity and for assessing disorders such as Down syndrome, Huntington's disease,

phenylketonuria, and cystic fibrosis, to list a few.^{4,11,15} Several of these and other genetic tests are used in prenatal and early postnatal care. Given the value and success of existing genetic tests, the current trend in health care is to establish new genetic tests on the basis of the results of the HGP in an effort to arrive at earlier and more accurate diagnoses.

Existing genetic tests and the formulation of new genetic tests raises the issue of testing regulation and standardization. Not all tests will be beneficial for assessment. Many disorders are multifactorial and may involve multiple genes, thus there could be several tests for the same disorder. The validity and reliability of new tests will need to be firmly established before clinical administration.^{5,6} The issue is further complicated by the necessity for profit in an industry in which research and development consumes a heavy portion of finances.¹⁶ Market competition may lead to invalidated tests and a compromise in the quality of testing.

Gene Patenting

At the core of regulating genetic tests is the issue of gene patenting by the companies that create such tests. Biotechnology companies are creating a lucrative industry by patenting specific sequences of DNA that are used within certain tests.^{10,16,17} Essentially, any research or clinical procedure on a patented section of DNA requires payment of a fee associated with its use to the patent owner. Unfortunately, this trend is creating monopolies on genetic data that consequently limits public access to genetic technology as a result of the inflated cost of genetic

tests.^{16,18} Furthermore, gene patenting has the potential to hinder research that involves patented sections of DNA. Considering the issue of polygenic disorders, correlative studies involving various DNA sequences may prove difficult if these sequences are patented to different companies. Although financial profit is not iniquitous, economic motivation has stunted what could be even greater scientific progress because of competition that discourages a steady exchange of scientific data and ideas.^{16,19} Therefore, gene patenting must be immediately addressed to avoid creating further monopolies and additional disenchantment of the scientific community.

Education

Increased use of genetic tests in clinical settings raises the issue of educating the general population and health care professionals regarding the tests, their purposes, and their implications. Currently, the average person is not properly educated about genetic testing because of its novelty.^{3,16} Depending on the outcome of such tests, especially with regard to tests for potentially high-risk genes, prolonged postexamination education on the scope of risk and follow-up must be provided to ensure proper understanding of the results and the implications to personal health. The result of a genetic test also can be a profound psychological burden on the individual and his or her family.^{3,15} On the basis of the potential predictive value of genetic tests, proper follow-up must be established for diseases whose early detection could result in improved treatments and outcomes.

IMPLICATIONS AND DIRECTIONS

Genetic Privacy

Currently, only a few major federal regulations specifically protect an individual's health and research information. Unfortunately, such laws are limited in scope. The Federal Privacy Act of 1974 applies only to federal agencies and is only one of a few limited acts that reaffirm the lack of federal regulation over all pertinent health information.¹³ Fortunately, advances have been made in protecting health information. The recently initiated Health Insurance Portability and Accountability Act (HIPAA) imposes stricter controls on health information by deeming breach of patient confidentiality a punishable offense. HIPAA also prohibits group health plans from using any health status-related factor, including genetic information, as a basis for denying or limiting eligibility for coverage or for charging an individual more for coverage (see <http://cms.hhs.gov/hipaa>).²⁰ No similar law applies to private citizens seeking health insurance in the individual market. Furthermore, the prohibition of discrimination does not extend to employers who obtain genetic information. This situation underscores the necessity for more comprehensive action.^{4-7,13,14}

Regulation and Standardization of Genetic Tests

Regulatory issues concerning genetic testing are quite apparent. However, no proactive, large-scale attempts have been made to remedy the situation. A survey of 35 state health departments across the country revealed an imposing necessity to promote quality assurance programs that evaluate and validate genetic

tests.²¹ Validation of genetic tests is essential because the nature of the US health care system in its current state does not allow for excessive spending on tests with little or no immediate clinical value to the consumer.¹⁵ Furthermore, despite majority agreement that academic research is being stunted by the quest for commercial profit, genetic testing regulations have not received adequate attention.^{1,16} If the trend continues, the development of new genetic tests will be motivated by profit rather than necessity. Acute disorders with low prevalence within the population may be overlooked in favor of mild yet widespread disorders because of a larger consumer base and thus greater profit.

Gene Patenting

The clash of economy and ethics also underlies the issue behind gene patenting. The HGP began as a publicly funded project; however, by 2000, private funding had exceeded public funding because of the race for patenting of genomic sections.¹⁰ Given the number of patents already granted, the ethical issue associated with the patenting of genes has been surpassed by concern for equity of access to patented sequences from a social and scientific perspective.^{17,18,22,23} A widely publicized case between the Canadian government and Myriad Genetics, which owns the sequence for the *BRCA 1* and *BRCA 2* genes (implicated as biomarkers for both breast and ovarian cancer),^{1,6,11,16,22,24} typifies the potential for patent misuse. Myriad mandated that any testing for the genes be conducted within its own facility. Overall, the cost of assessing the genes is approximately \$2500 through Myriad.^{16,18,22,24}

However, Canadian agencies provide an equivalent test for 65% to 80% less than Myriad's cost.^{16,18,22,24} This is only one of the various misuses of patent law fueled by economic profit that could potentially alienate society from the technology available if immediate regulations are not implemented.

To further complicate the issue, recent trends also indicate there is increasing interest in patenting protein products of gene sequences.^{19,23} In contrast to genetic tests, in which the gene sequence is the primary concern, pharmaceutical therapies commonly target the protein products of a gene sequence.²³ However, a particular gene sequence may encode for several different protein products. Therefore, possible situations could develop in which a company owns a patent over a particular sequence, but another company owns the patent for 1 of the protein products of the same sequence. In such an occurrence, clinical and research endeavors could be caught in a quagmire of administrative procedures that involve multiple patents to separate companies.

Education

The education of health care practitioners and society is of equal priority. There should be greater emphasis on education about genetic testing in general, but even more importance should be placed on educating test recipients about behavioral modifications that could be recommended as a result of a genetic test.^{1-3,8,16,25,26} As a reflection of the lack of education currently provided for patients, a study reported that before being administered a genetic test for a hereditary genetic disorder, only 18.6% of the subjects received

counseling.²⁷ This very low percentage is a distressing testament to the lack of emphasis on education about genetic testing and reiterates the necessity for expanded education programs.

RESOLVING THE ISSUES

Genetic Privacy

Although there have been recent advancements in patient privacy and confidentiality, pores still exist within the fabric of privacy that allow third parties to misuse the information. Much more stringent controls must be placed on health records, not only those of patients involved in research, but also those of all health care beneficiaries. Figurative ownership of health care records belongs to the specific individual, and access to those records should be mandated by that person alone. Appropriate measures must be taken to ensure that insurance companies and employers do not have access to a person's genetic profile. Penalties should be levied on companies that abuse the concept of genetic privacy.

Regulation and Standardization of Genetic Tests

Regulation of current and future genetic tests made available for clinical use should undergo thorough evaluation before implementation. Regulatory agencies must take more responsibility for the quality assurance of genetic testing. Such an evaluation process also would encourage the refinement of genetic testing, because it would promote an atmosphere of competition to develop the most valid and reliable test that provides optimum benefit to society. There is also a potential for fraud and diagnostic error by

manufacturers of genetic tests. Both federal and state agencies must be vigilant in reviewing standard procedures in new and established genetic laboratories. Operating licenses must be granted to such laboratories on the basis of successful adherence to strict standards.

Gene Patenting

An estimated 18 174 separate patents on DNA sequences had been filed by the end of 2001.¹⁷ Obviously, any efforts to revoke already granted patents would be met with great resistance from patent owners. Furthermore, the essence of issuing patents is to promote innovation and discovery^{16-18,23}—both noble premises from a scientific and social perspective. However, given the increasing number of genetic patents, regulations are essential to prevent monopolization of DNA sequences.

A distinct regulatory committee should be formed that monitors patent applications on genetic material in an effort to reverse the negative developments of gene patenting. Essentially, the committee would serve as the overseeing authority regarding the uses and abuses of patent applications. The committee would recommend regulations for the scientific community that prevent any form of patent infringement, yet allow and encourage scientific progress. Science and society should implement a system that promotes genomic research, encourages patenting on the basis of innovation and discovery, and benefits society as well as the patent owner.

Education

Conclusively, it is of great importance that society be educated about the scope of genetic testing. Current trends indicate

that genetic and molecular assessments are destined to be a routine component of health care. If society is to benefit from these components, health care practitioners, as well as the rest of society, must be educated about the proper use of genetic information. Education is a necessity because genetic predisposition does not always equate to phenotypic expression. Although an individual may possess a gene implicated in the development of a disorder, gene activation may not occur until triggered by a particular environmental interaction. The environmental interaction may be a single exposure to a triggering event or one that requires repeated exposures over time before clinical manifestation of disease. Therefore, comprehensive education also should serve to inform susceptible people regarding precautionary measures or behavioral modifications to prevent or delay the onset of disease.

Local health departments should play a key role in educating the general public, because public health practitioners have the capacity to conduct community-based educational projects. High schools and junior high schools also must be more willing to introduce genetic and molecular education within their science curriculum to ensure that the upcoming generation is prepared for genetic innovations. Overall, every sector of society must cooperate to use genetic technology to its fullest potential and to provide the benefits for which it is intended. ■

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Contributors

R.P. Ojha developed the ideas for this article. The ideas were reviewed and further refined by R. Thertulien. Both authors performed the research required and contributed to writing the article.

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Human Participant Protection

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