Ethical Concerns About Genetic Testing and Screening

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Many of the ethical concerns raised by genetic testing and screening relate to accuracy, cost, and confidentiality. Perhaps the most serious worry—one that is not without merit—is that the new genomics is a disguised version of the old eugenics. On balance, however, genetic testing and screening seem to be in society's best interests.

Today, people have unprecedented access to genetic information about themselves and, in some instances, others. Companies such as 23andMe (https://www.23andme.com/) give people access to their genetic profile without necessarily providing adequate means to interpret this information. The cost to complete the first sequencing of the human genome in 2003 was estimated at $2.7 billion (in Fiscal Year 1991 dollars) [1]. By June 2013, however, an individual could have his or her entire genome sequenced for $5,000 (a price that included an iPad containing the results), and some experts estimate that the price for whole-genome sequencing will drop to $500 in the near future [2]. People are now able to know a great deal about their present and future health status, but this knowledge is not without problems. Serious ethical questions surround both genetic testing of individuals and genetic screening of populations.

One of the main ethical issues surrounding genetic testing and screening is accuracy. Enormous and devastating consequences can result from receiving either a false-positive result (being told that you have a deleterious genetic condition when you do not) or a false-negative result (being told that you do not have a deleterious genetic condition when you actually do). Also, many people make decisions about whether to have a baby based on knowledge about the genetic condition of the fetus. A couple who are undergoing in vitro fertilization—who may have invested much time, physical and psychic energy, and money in this process—may decide to abort a fetus on what turns out to be a false-positive result for a particular genetic condition [3]. Similarly, it cannot be emphasized enough that women should not assume that they will not get breast cancer simply because they test negative for mutations in the *BRCA1* and *BRCA2* genes—only 5%–10% of all breast cancers are linked to such mutations [4]. The public needs to be repeatedly reminded that health status is not genetically determined in a simplistic way; for the most part, genes merely contribute to overall health. If an individual has the gene for Huntington disease, he or she will almost certainly develop this neurological disorder [5]. In contrast, a person may have multiple genes related to type 2 diabetes, but depending on his or her lifestyle choices, those genes may or may not be activated [6].

The high cost of genetic testing and screening is another source of concern. Genetic screening for the presence of *BRCA1* and *BRCA2* mutations and for conditions such as cystic fibrosis, Tay-Sachs disease, and Down syndrome ranges in cost from less than $100 to more than $2,000 [7]. In June 2013, Myriad Genetics had the exclusive right to test for *BRCA1* and *BRCA2* mutations, and the cost for this test was nearly $4,000 when a related genomic rearrangement test was included in the analysis [8]. Even if the cost of whole-genome sequencing drops from $5,000 to $500, an individual, institution, or organization still must bear the cost, and some people will also want follow-up tests to help their health care providers determine the subsequent course of treatment. Therefore, most health care ethicists recommend that genetic testing and screening be offered only to individuals who are at relatively high risk for a serious genetic disease. Requests for medically unnecessary genetic testing and screening should not be honored by health care professionals, even if patients threaten to get the information from other sources that may be less reliable [9].

In addition to expressing concern about the accuracy and cost of certain genetic tests, many health care ethicists and health professionals worry about the consequences if such tests were to become mandatory. For example, no one wants to repeat the poorly conceived and often misunderstood mandatory screening program for sickle-cell anemia that was instituted in the 1970s, especially given the mistrust that many African Americans already have of the health care system [10]. Sickle-cell disease is especially prevalent in the African American community; thus, when a relatively inexpensive test was developed in the 1970s that could iden-

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tify carriers for sickle-cell disease, laws were passed in many states that made testing mandatory for African American schoolchildren [11]. Unfortunately, some health care insurers rejected health insurance applications from individuals who were carriers of sickle-cell anemia, because they did not want to cover individuals whose children were likely to have the disease [11]. In addition, some employers refused to hire people who were carriers of sickle-cell anemia because they believed that these individuals would be too sick to work and/or that it would be too expensive to provide health care insurance for them [11]; these employers did not understand the difference between being a carrier of the disease and actually having the disease [12].

Maintaining confidentiality in genetic testing and screening is vital. Confidentiality is one of the foundations for a successful patient-provider relationship. Patients who cannot trust their health care providers to safeguard private information are not likely to reveal the information, thereby depriving themselves of good medical care [12]. In addition, patients sometimes ask their health care providers to withhold the results of genetic tests from family members or other intimates for fear of alarming or alienating them. But problems may arise when patients hide their genetic status from loved ones. Sometimes family members cannot make important life or medical decisions without the information that is being withheld from them. For example, a woman might want to know her future husband's Huntington disease status before agreeing to stand by him in sickness as well as in health, and before having children with him [13].

The Genetic Information Nondiscrimination Act of 2008 (GINA) does not provide health care professionals with definitive guidelines for balancing the need for patient confidentiality against the harm that could result from not disclosing the patient's genetic profile to family members and/or other intimates. GINA prohibits health insurers and employers from discriminating based on genetic information [14], but it remains silent about providing information to family members or intimates of the person with a genetic condition.

Perhaps the greatest concern about genetic testing and screening is whether it will lead to a program of eugenics aimed at eliminating those who are "unfit" and allowing only those who are "fit" to reproduce. Some health care ethicists and professionals fear that genomics will replicate the mistakes made by eugenics programs around the world during the first half of the 20th century. The eugenics programs that flourished in the United States from about 1890 to 1940 continued to operate in North Carolina until 1974. Between 1929 and 1974, North Carolina sterilized approximately 7,600 individuals who were deemed "feeble-minded or otherwise undesirable" [15]. Although the state officially apologized to the surviving victims of these involuntary sterilizations in 2003 and promised to make reparations [16], only recently did the state set aside $10 million to compensate these individuals [17]. An amount of $50,000 per victim has been suggested as an amount that would provide adequate compensation [17], but one wonders whether even several million dollars would adequately compensate someone who was denied the ability to procreate, sometimes without even being informed that he or she was being sterilized.

Because society has misunderstood and misapplied genetic information in the past, worries have arisen about the underlying motives for today's genomic medicine, but I believe there are important differences between eugenics programs of the past and today's genomic testing. Eugenics programs were based on very poor scientific evidence; for example, some proponents of eugenics believed that there were genes for criminality and promiscuity [18]. Eugenics also involved forced sterilizations—getting rid of undesirable people and sacrificing the individual for the supposed good of the group. In contrast, genomics is about controlling one's genetic destiny, choosing the kind of children one wants, and being as healthy and happy as possible [12]. Also, with today's genomic testing, every effort is made to be objective and to set aside assumptions about people's race, sex, ethnicity, and wealth.

Genomics enthusiasts often stress that the aim of reproductive genetic testing and screening is simply to inform prospective parents about the genetic health status of their future child, not to prompt prospective parents to select for only the best prodigy possible [19]. Nonetheless, a high percentage of parents do choose to abort a fetus if it tests positive for a serious genetic disease. Although people with Down syndrome can lead meaningful lives and report that they are happy [20], a 2012 analysis of 7 population-based studies and 9 hospital-based studies published between 1995 and 2011 found that 67% to 85% of women ended their pregnancy when they learned that the fetus had Down syndrome [21]. There is also evidence that a relatively high percentage of parents would consider aborting a fetus if it had a minor genetic defect such as myopia [22]; a propensity toward a disease such as obesity, which can be controlled by lifestyle adjustments beginning in early childhood; or Huntington disease, which has its onset quite late in life [23]. Lastly, in countries where there is a marked preference for boy babies over girl babies, some parents will abort a fetus if it is the "wrong" sex (ie, female). In both China and India, the sex ratio at birth is now 1.12 males for every female [24].

Many health care ethicists are troubled by the possibility that reproductive genetic testing could lead to elimination of undesirable fetuses, with prospective parents aiming to replace them with better or preferred children. A society in which prospective parents are under severe pressure to produce a perfect baby is one that probably has less tolerance for and acceptance of people who deviate from whatever is deemed "normal." As health care costs rise, some disability rights activists fear that rather than making reasonable accommodations for people with disabilities, societies will take the inexpensive way out and make it very difficult for

people to produce children who have mental or physical challenges [25]. Michael S. Lagan, a vice president of the National Organization for Rare Disorders, goes so far as to speculate that

> Eventually there will be discrimination against those who look "different" because their genes were not altered. The absence of ethical restraints means crooked noses and teeth, or acne, or baldness, will become the mark of Cain a century from now [12].

Although these problems are worth considering, indications are that better genetic testing and screening will increase rather than decrease people's freedom—that is, their ability to make autonomous decisions about their health and that of their children. Moreover, most people will not overreact after they see the results of their genetic tests. Rather, they will seek the help of health care professionals, who will do more targeted genetic tests and carefully explain to patients about their options [26].

However, one concern is the shortage of genetic counselors. In 2012 there were only 3,000 genetic counselors in the United States [27]. This small number of genetic counselors cannot be expected to answer the many questions people will likely have about their genetic test results, especially if use of such screening increases. Greater emphasis needs to be placed on increasing the number of genetic counselors, which could be accomplished in part by increasing their financial compensation. The few who are employed by genetic testing companies (about 9% of the 3,000) typically earn about $65,000 per year, which is at the high end of the pay scale [27]. Incorrectly interpreted genetic information is potentially harmful; by improving the quality of the interpretation of genetic test results, we are all more likely to benefit from this technology. NCMJ

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