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FOCUS: TRANSLATIONAL MEDICINE

Getting a Head Start: The Importance of Personal Genetics Education in High Schools

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With advances in sequencing technology, widespread and affordable genome sequencing will soon be a reality. However, studies suggest that “genetic literacy” of the general public is inadequate to prepare our society for this unprecedented access to our genetic information. As the current generation of high school students will come of age in an era when personal genetic information is increasingly utilized in health care, it is of vital importance to ensure these students understand the genetic concepts necessary to make informed medical decisions. These concepts include not only basic scientific knowledge, but also considerations of the ethical, legal, and social issues that will arise in the age of personal genomics. In this article, we review the current state of genetics education, highlight issues that we believe need to be addressed in a comprehensive genetics education curriculum, and describe our education efforts at the Harvard Medical School-based Personal Genetics Education Project.

INTRODUCTION

Within a few years, sequencing a human genome is expected to cost less than \$1,000, a benchmark for “personal genome” sequencing to approach widespread clinical feasibility [1]. Since the release of the first draft of the human genome sequence just one decade ago, the post-genomic era has

ushered in improvements to sequencing technologies that have dramatically reduced sequencing costs. The cost of sequencing a single human genome has dropped 10,000-fold over the last decade, from \$100 million in July 2001 to \$10,000 in July 2011 [2]. The most precipitous drop began in 2008, when next-generation (or “next-gen”) sequencing

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†Abbreviations: ASHG, American Society of Human Genetics; DTC, direct-to-consumer; ELSI, ethical, social, and legal implications; GENA, Geneticist-Educator Network Alliance; pgEd, Personal Genetics Education Project; PGP, Personal Genome Project; SNP, single nucleotide polymorphism.

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technology matured, and the rate has since outstripped what one would predict based on Moore's Law, a standard for evaluating technological development in computing [3].

As the cost of genome sequencing declines, personalized genetic testing is increasingly available to the public. There are now a number of companies, such as 23andMe, deCODE, and Athleticcode, to name a few, which offer direct-to-consumer (DTC†) genetic testing. Many of these perform "genome scans," utilizing microarray-based single nucleotide polymorphism (SNP) genotyping to provide information about ancestry, carrier status, and traits ranging from disease risk and drug response to behavior and athletic ability. Personal genetic sequencing is also becoming a reality. In 2009, the provider of a major next-gen sequencing platform, Illumina, launched its own personal whole-genome sequencing service for consumers with physician prescription. As of June 2011, the per-genome cost of this service has dropped to \$9,000 and is even lower for patients with life-threatening illnesses [4]. In addition, more than 1,000 volunteers have signed up to have their genomes sequenced as part of the Personal Genome Project (PGP), which aims to investigate genetic and environmental contributions to human traits through open sharing of participants' genome sequences and personal information [5].

The field of personal genomics is still in its infancy, but there are already a number of examples of use that foreshadow its promise in medicine. In 2008, a man in Vancouver, Canada, who was suffering from a rare, extensively metastasized tongue tumor for which no standard treatment was available, had his cancer genome and transcriptome sequenced. By comparing these datasets with those from the patient's normal cells and other unrelated tumors, scientists identified one gene, *Ret*, that was amplified and over-expressed, and based upon this finding, his physician started him on a targeted drug treatment. His disease was stabilized for several months until, unfortunately, new mutations in other pathways appeared and he passed away [6]. In 2010, a child from Wis-

consin with an undiagnosed inflammatory bowel disease, who had undergone more than 100 surgeries since age 2, had his exome sequenced. Identification of a variant in the *XIAP* gene led to a life-saving bone marrow transplant for this little boy [7]. In 2011, a pair of twins from California, who were previously diagnosed with the genetic disease dopa-responsive dystonia, had their genomes sequenced. Identification of the genetic variant responsible for their conditions allowed neurologists to tailor their treatment with an additional drug that significantly alleviated their symptoms [8].

These successes highlight the potential in utilizing patients' genetic information to improve diagnostics and therapeutics. What role will personal genetics play in routine preventive medicine? As our understanding of the complex relationship between genotype and phenotype advances, the question will be how to integrate genetic information, generally based on population-level data, to predict future health risks for healthy individuals.

As we enter this new age of personal genetics, are we, as individuals and as a society, ready? What decisions will we face, and what challenges lie ahead? How well does each individual need to understand genetics to make informed decisions? In this perspective, we will discuss the types of personal and broad social questions that are likely to arise, our views on the urgent need for public education, and our efforts at the Personal Genetics Education Project (pgEd).

CHALLENGES IN THE AGE OF PERSONAL GENOMES

Imagine that you have been interested in DTC genetic testing for a while. You have talked to your family members about it, and although they are unsure about this new technology, they give you their support. Suppose, now, that your report comes back revealing that you have a 20 percent lifetime risk for developing a particular kind of cancer (4-fold higher than the general population). How should you interpret this information? Would (or should) you change

your daily lifestyle, opt for more frequent screenings, or undergo a preventive surgery that could drastically reduce your likelihood to develop the cancer [9]? In order to make these types of decisions, you would need a basic understanding of concepts including risk, the effect of genes on traits, genetic penetrance, and the role of the environment, in addition to specific information about the disease and available treatment options. If you are not particularly well versed in genetics or biology, should you be required to consult a medical doctor or genetic counselor to help you interpret the genetic data [10], and if so, are doctors themselves sufficiently trained in genetics to perform this role [11]?

The genetic information will not only affect you as an individual, but your family members as well, as it will have implications for their health as well as the potential for conflicts over issues of privacy and autonomy. For example, if you have or plan to have children, would you want to (and should you be allowed to) have your children tested? If you want to disclose this genetic information in an online forum, would you ask for your family members' approval, since their genetic makeup might also be partially disclosed in the process? If, instead, you decide to keep this information private, would a doctor who helped to interpret your results have an obligation (morally, legally, or otherwise) to disclose this information to your relatives [12-14]?

Imagine, in the future, it is recommended that newborn babies have their genomes sequenced at birth, augmenting newborn screening programs that are mandatory across much of the United States [15]. Should parents be allowed to opt out? Who will have access to the information (parents, doctors, teachers/coaches), what information should be shared (e.g., disease risk), and which diseases should be reported (pediatric, adult-onset, or only treatable ones)?

These are all pressing questions that we as a society — patients, doctors, politicians, educators — need to discuss as personal genetic testing becomes increasingly available.

Many of these questions are highly personal, making it all the more important that individuals are well informed and prepared to make their own choices.

A NEED FOR EDUCATION

As technology races ahead, there is a critical educational need to prepare the public for the increasing accessibility of genetic information. However, misunderstanding of basic genetic concepts is well documented. Various studies have found that many segments of the population (whether high school or undergraduate students or genetic counselors) have a generally deterministic view of genetics and often attribute phenotypes to the action of a single gene, not appreciating that complex traits are the results of interactions of several genes with the environment [16-19]. This is particularly troubling, as it has been found that misconceptions in genetics and statistics influence how patients make decisions about testing, treatment, and reproduction [20]. It is important to act quickly to prepare the next generation of young adults, who will come of age in an era when choices about personal genetics will increasingly impact health care and reproduction. While individuals might be assisted by health care practitioners in the interpretation of their genetic information, education of the medical community cannot replace a broader educational initiative that empowers each individual to make informed decisions about his or her own DNA and to have a voice in shaping how personal genomics is integrated into our society.

To highlight the urgent need for education at the high school level, it is instructive to look at a couple of recent studies. Since 2006, the American Society of Human Genetics (ASHG) has sponsored an essay contest for grade 9 to 12 students to coincide with the annual National DNA Day. An analysis of a sample of 500 submitted essays found that at least one significant misconception about basic genetic concepts (such as assuming complex traits to be simple or confusing “genetic” with “hereditary”) could be identified in more than half of the

sampled essays [18]. Beyond misconceptions, a significant number of essays also mentioned some kind of genetic engineering or germline gene therapy to “design” offspring, without awareness of the ethical and social implications. Moreover, a recent assessment of high school science standards in the United States with respect to genetics revealed that topics such as polygenic and complex traits, principles of gene expression, and gene-environment interaction are inadequately covered [21]. Clearly, there is a significant educational need for both the scientific and ethical aspects of genetics.

VISION FOR PUBLIC EDUCATION

The current generation of high school students will be the first to come of age in the era of personal genomics, making choices that will determine how personal genetic information is incorporated into society. Targeting educational efforts toward the existing infrastructure of high schools offers a broad and cost-effective approach to ensure that the majority of this generation has been exposed to key genetic concepts and has had the opportunity to discuss and debate the benefits and risks of personal genetics for individuals and society [22].

An emphasis on fundamental concepts such as genetic non-determinism, paired with a discussion of ethical, social, and legal implications (ELSI) of personal genetics, will be powerful in preparing the public to make informed and personal decisions with genetic information. As individuals, we will need to consider whether we want to get tested, what information we are interested in learning about ourselves, how we will decide to access our own genetic information, and with whom we would share this information. The ultimate goal of engaging students in the discussion of these issues is to foster informed citizens with a lifelong interest in genetics and self-confidence that empowers them to make their personal decisions.

Biologists (and particularly geneticists) at all levels, from graduate students to faculty members, are particularly important for

these education initiatives, given our working knowledge of the most up-to-date genetic science, and our societal responsibilities to the public that funds our research. ASHG has published a *Statement on the Importance of Participation of Scientists in K-12 Science Education*, which urges academic institutions to facilitate faculty outreach in the community to promote public education on genetics and emphasizes that “informed participation in an increasingly genetics-based health care system demand[s] that consumers understand genetics and its importance in health and disease” [23]. In line with this, ASHG ran a National Science Foundation-funded project called Geneticist-Educator Network of Alliances (GENA) between 2007 and 2010 to promote K-12 outreach activities by scientists.

PERSONAL GENETICS EDUCATION PROJECT

pgEd is a group of scientists and educators based at Harvard Medical School that aims to promote public awareness of fundamental genetic concepts and ELSI of personal genetics. Initiated in 2006, we have led interactive workshops for hundreds of high school students and their teachers in Massachusetts and Maine on various issues relating to personal genetics. In addition, we are developing curriculum tools that capture the content from our workshops, which teachers can use to facilitate these important discussions in their classrooms. We integrate this information into detailed lesson plans with a variety of interactive exercises and make these freely available on our website (<http://www.pged.org>) [24]. Our workshops and lesson plans are designed for high school students (grades 9-12) but are also engaging for undergraduate-level and adult audiences.

Our lessons cover topics ranging from genetics, personalized medicine, DTC testing, and reproduction to athletics and crime, while examining the personal, familial, and societal impacts of increased accessibility to genetic information. They require little prior knowledge of genetics and introduce key

concepts, including complex traits and environmental influences that are reinforced throughout the curriculum. For example, the lesson on athletics focuses on currently available DTC genetic tests that purport to evaluate athletic potential and susceptibility to injury, marketed by companies such as Athleticcode and SportsXFactor. Through scenarios to which many students can easily relate, students discuss scientific issues including the predictive value of the tests, the role of environmental and social factors, risk, and pleiotropic effects of a genetic variant linked to concussion recovery and Alzheimer's disease [25]. At the same time, they explore broader issues of personal choice, psychological impact, fairness, autonomy, and privacy.

In our experience, we have found that topics such as these energize classrooms. Students are easily engaged in ethical issues and, as a result, become motivated to learn more about genetics. Our lessons fit naturally into the biology curriculum, either as a hook at the beginning of a unit on genetics to engage students or at the unit's end to discuss social implications of genetic technologies. Moreover, these discussions are not limited to the biology classroom, as they often touch on multiple other fields including psychology, social justice, policy, and religion. Given their interdisciplinary nature, the lessons can also be used in other subjects such as social studies or health education.

Our curriculum at pgEd complements many wonderful resources for high school audiences developed by other education programs that integrate genetics education into bioethics curricula. These include the Northwest Association for Biomedical Research (<http://www.nwabr.org/>), the High School Bioethics Project at the University of Pennsylvania Center for Bioethics (<http://www.highschoolbioethics.org/>), the Genetic Science Learning Center at the University of Utah (<http://teach.genetics.utah.edu/>), and the NIH Curriculum Supplement: Exploring Bioethics (<http://science-education.nih.gov/StateStandards/>). Their lessons explore topics such as the fundamentals of bioethics, genetic testing, personalized medicine, reproductive ge-

netics, newborn screening, and human research, training students to become informed consumers who will be prepared for the complex decisions they will face in the personal genomic era.

The challenge for the future will be to ensure that all these resources are available to teachers and to facilitate their integration into high school curricula across the United States (and the world). Educators and policymakers must take the necessary steps in a timely fashion to anticipate the widespread availability of personal genetic sequencing.

CONCLUSIONS AND OUTLOOK

The advancement of personal genetics has been dubbed an “unstoppable train” [26]. It seems only a matter of time before these technologies will be commonly used for diagnostic and preventive purposes, or even simply to satisfy curiosity. What we can and must do is to make sure our current generation of students will be well informed about the science, benefits, risks, and ethical issues, so they will be prepared to make personal decisions about their own genomes and their own health when widespread application becomes a reality.

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