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Scientific Life

Participatory Genomic Testing as an Educational Experience

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Several institutions have incorporated participatory genomic testing into their curricula to engage students in experiential learning, and this has raised ethical concerns. We summarize strategies for managing these concerns and review evidence of the influence of this experiential approach on student knowledge and attitudes towards genomics.

Genomic Testing Comes to the Classroom

As a result of advances in technology and increased affordability, personal genomic testing (PGT) is widely available on a direct-to-consumer (DTC) basis and is becoming routinely incorporated into medical practice [1]. To equip future physicians with the skills to appropriately apply genomic testing in their practice, it is imperative that they understand not only the genetic and genomic foundations of human disease, but also the risks, benefits, and ethical concerns about these tests. A recent survey of medical genetics educators indicates a shift towards emphasizing genomic approaches and related ethics in their curricula [2], which is supported by national curricular guidelines [3]. Towards this end, some educators have proposed that incorporation of PGT in the classroom can accomplish these goals. However, this approach has not yet achieved widespread acceptance in the educational realm, with only 11% of institutions reporting the use of genomic testing in the classroom and only 7% considering its use (unpublished data from [2]). Reasons reported for hesitation in incorporating classroom PGT include expense, limited resources (including access to genetic counselors), ethical concerns, potential harm from identifying disease-related variants, and concerns about student privacy and consent (unpublished data from [2]). Thus, careful examination of the outcomes of the first wave of courses using PGT or other methods of incorporating genomic data in terms of ethical concerns, trainee perceptions, and knowledge gained will be helpful in assessing the efficacy of these methods. Here we review participatory genomic educational approaches taken at several institutions (summarized in Table 1).

Course Design and Ethical Considerations

While appreciating a need for improved education in genomics among health-care professionals, medical institutions recognize the potential risks of offering PGT for educational purposes, including coercion, data confidentiality, informed decision making, and financial inducement (Box 1) [4–6]. Schools addressed these issues in different ways, with common themes of offering PGT on a voluntary basis, providing anonymous data for students who choose not to undergo PGT, and offering genetic counseling. Schools have also chosen various genotyping strategies that range from the most limited genotyping to whole-genome analysis.

University of California (UC) Berkeley made a big splash in 2010 when, along with the required textbook list, they sent all 5000 incoming freshmen an optional PGT kit to test for common genetic variants in three genes: *LCT* (lactose intolerance), *ALDH2* (alcohol metabolism), and *MTHFR* (folic acid metabolism) [7]. While the intention was to enhance student learning, their approach set off wide-scale criticism from bioethicists and the public [7,8]. In response, UC Berkeley made several changes including providing the students with information and lectures on the ethical and legal implications of genetic testing, reporting only aggregated results during class sessions, switching a contest prize from a full genetic test conducted by a commercial company (which could be perceived as an endorsement) to a cash prize, and providing students with optional private counseling.

Taking a more cautious approach before incorporating PGT into their medical curriculum, Stanford University officials appointed a task force of basic scientists, clinicians, legal experts, genetic counselors, ethicists, and medical students who spent a year addressing concerns about educational PGT. Issues raised included ensuring confidentiality and anonymity, the need for genetic counseling, the impact of student test results on families, conflicts of interest due to faculty ties with testing companies (23andMe and Navigenics), and the potential for coercion [4]. To address these concerns, the course was offered as an elective rather than a required course, genetic testing via DTC personal genome scan was optional with a confidential choice between two companies, the cost was subsidized by the university with students paying $99 to avoid financial inducement, students were given the opportunity to use their own personal genotype data or publicly available data for class exercises, and information derived from the genomic data was presented only in aggregate to the class. In addition, the first three sessions of the course were dedicated to the risks, benefits, uses, and limitations of genomic testing to provide a background for informed consent, and confidential genetic counseling as well as counseling by faculty in the Department of Psychiatry and Behavioral Sciences was offered. The advanced elective was offered to medical and graduate students, postdoctoral fellows, and medical residents/fellows. Institutional Review Board (IRB) approval was obtained to examine differences in educational outcomes as well as to conduct a qualitative study of student attitudes (see Outcomes section below; [9,10]).

Table 1. Approaches to incorporating genomic testing into health care provider training curricula

	Stanford [4,9,10,13]	Mt. Sinai [6,14,15]	Tufts School of Medicine [5]	Temple School of Pharmacy [11]	Temple School of Medicine [12]
Genetic test strategy	SNP[b]-genotype based DTC[a] testing	WGS[d]	SNP[b]-genotype based DTC testing anonymous samples	Genotype single pharmacogenetic SNP[b]	Whole exome sequence of cadavers
Student population	Advanced elective for medical and graduate students, postdoctoral fellows, and medical residents/fellows	Required course for genetic counseling students; Advanced elective for medical and graduate students, pathology fellows and medical genetics residents. Prerequisite introductory course.	First year medical students	Second year PharmD students	First year medical students
Enrollment per class offering	46 students	20 students with personal data and additional students with reference data	200 students	150 students	35 dissection teams of six students each
Course design	Choice of two subsidized DTC[a] tests *or* use anonymous data. In-class exercises focused on analysis.	Free WGS[d] OR anonymous genome. Students received raw data to analyze.	Anonymous samples chosen to include relevant genotypes to illustrate material from classroom sessions	Student genotypes assessed in aggregate; individual data not provided to students	SNVs[c] selected from exome data for analysis by individual dissection teams. Used as diagnostic test for cadaver.
Study design	Perceptions: Longitudinal study, qualitative analysis of interviews at multiple times before and after genotyping; pre- and post-course surveys comparing students who did and did not analyze personal genome Knowledge: Assessed via pre-test and post-test Behavior: Surveys	Perceptions: Longitudinal study, questionnaires at different times before and after course. In-depth interviews 6 months after course. Knowledge: Internally developed test of technical knowledge, student-reported understanding Psychological Impact: Center for Epidemiologic Studies Depression Scale, State-Trait Anxiety Inventory, Multidimensional Impact of Cancer Risk Assessment, Decision Regret Scale, Decisional Conflict Scale	Descriptive report	Descriptive report	Descriptive report

[a]DTC = Direct-to-consumer
[b]SNP = Single Nucleotide Polymorphism
[c]SNV = Single Nucleotide Variant
[d]WGS = Whole Genome Sequencing

Tufts University School of Medicine took a similar approach of appointing a multidisciplinary faculty committee to examine ways to improve genomic education [5].

Box 1. Ethical Issues Regarding Educational Use of PGT

- Anonymity
- Confidentiality
- Coercion
- Ability to make informed decision about PGT
- Need for genetic counseling and psychological support
- Impact of test results on students and their families
- Conflicts of interest among faculty with ties to genomic testing companies
- Financial considerations

As part of their deliberation, they performed a pilot with a small group of seven faculty who underwent PGT and discussed their experience with the committee. Their final decision was to use anonymous DTC data rather than student PGT to teach material on genomic medicine. The medical genetics course was part of the first block of the required first-year curriculum, which made this curriculum available broadly rather than limiting its offering to fewer students with a special interest in genomics. The course content was expanded with an additional lecture on the science and technology of genomic testing, and updated by adapting a pre-existing lecture on ethics to include a substantive discussion of the potential benefits and harms of PGT and the lack of evidence for clinical utility of these approaches [5].

Another approach to avoiding ethical issues was to use limited genotype data from students and to present it only in aggregate. Second-year Temple University pharmacy students genotyped one specific SNP in *NAT2* [11]. The SNP is associated with adverse drug reactions and was selected for its direct relevance to the future pharmacists. The exercise took students from the genotyping activity to a discussion