Genome Medicine

RESEARCH HIGHLIGHT

# Personal genotypes are teachable moments

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**Abstract**

There is an urgent need for effective genomics education for healthcare professionals. Recent analysis of an experimental genomics curriculum showed that medical students' examinations of their own genotypes provide a valuable learning experience. Such experiential learning has a long tradition in medical education and its application to genomics is enabled by increasingly powerful and decreasingly costly genome science and technology. Personal genotyping is an important option to consider when designing educational programs for healthcare professionals.

## Empowered patients outpace unprepared professionals

Rapid changes in technology and cultural behavior are challenging the traditional role of healthcare professionals as 'learned intermediaries' who are responsible for interpreting and translating medical information for patients and the general public [1]. Advances in genomic science and technology are rapidly outpacing the diffusion of this information through the traditional channels of medical education and training [2], a phenomenon that is becoming increasingly apparent even to consumers [3]. Furthermore, studies have repeatedly shown that online sources and social networks have become the primary or even sole sources of health information for patients and their friends and families; healthcare professionals are consulted later, if at all [1].

Direct-to-consumer (DTC) genotyping services have contributed to this disintermediation of physicians and other healthcare professionals [1]. Moreover, the emergence of 'empowered patients' practicing 'participatory medicine' [4] has eroded professional hegemony and created significant challenges, but also new opportunities, for physicians. In addition, the impact of role models in popular culture who are utilizing DNA technologies to address a variety of health issues [5] has interjected another powerful cultural variable into patient-physician dynamics.

To meet some of these challenges, two initiatives were launched in 2009 aimed at medical and graduate students [2] and post-graduate medical trainees [6]. Both of these programs offered voluntary participation in personal genotyping as a pedagogical enhancement to curricula. The article by Vernez *et al.* [2] in this issue of *Genome Medicine* reports on student experiences in a pre-doctoral elective course, GENE 210, on 'Genomics and Personalized Medicine' provided at Stanford University School of Medicine. Here, we consider the content of the course, its foundation in learning theory, student observations and experiences, and some unexpected findings about consultative support in programs of this type.

## Genomics is not genetics and DNA is not destiny

Traditional medical genetics has a specific focus and target population, being primarily the study of inheritance, and is most typically applied to reproductive health issues and pediatric-onset disorders. Genomics is more focused on risk mitigation or on the management of complex, multifactorial diseases, assessing prognosis or individualizing therapies, particularly in adults.

There are two distinct categories of medical genomics [7], a fact that is not often obvious and frequently leads to confusion and co-mingling of medical uses and their ethical, legal and social implications (ELSI). The first category encompasses pre-symptomatic genotyping for disease risk assessment using the results of genome-wide association studies (GWAS). In GWAS, variations (polymorphisms) within the genomic DNA nucleotide sequences of individuals are studied for their statistical associations with various diseases or disease traits. An individual's genotype with respect to these single nucleotide polymorphisms (SNPs) can indicate an increased probability of being affected by, or developing, a certain disease or medical condition. These probabilistic associations are typically rather weak and the contributions of

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these genetic variations to disease are often greatly overshadowed by environmental (lifestyle) factors that contribute more significantly to increasing the risk of diseases such as lung cancer or type II diabetes. This type of SNP genotyping data was made available to pre-doctoral students at Stanford [2] and post-doctoral pathology trainees [6] at Beth Israel Deaconess Medical Center, a teaching hospital of Harvard Medical School.

The second category of medical genomics data results from post-diagnostic genotyping for the purposes of prognostication and/or individualized therapy [8]. For example, information on the genotypes of relevant drug-metabolizing enzymes might be used to plan personalized dose regimens for anti-coagulants, whereas genomic 'subtyping' of cancers might be used to assess prognosis or to select a genotype-targeted anti-cancer drug.

## Experiential learning, clinical utility and consultative support

Experiential learning ('see one, do one, teach one') has a long and very useful tradition in medical education. When medical students are learning how to do physical examinations, for example, they often practice skills like auscultation on fellow students. Likewise, testing to determine a student's own blood type is another commonly used pedagogical technique for pre-clinical medical education.

Conceptually, blood typing differs from genotyping in technology and scope. Blood typing is a phenotypic test (based on antibody-antigen interaction) that allows the inference of genotypes (of the DNA sequences that encode the ABO blood-group proteins). By contrast, SNP genotyping measures DNA sequences directly and on a vastly larger scale (millions of gene-associated DNA variations). Genotypes allow the prediction of phenotypes such as the ability to metabolize certain drugs and the susceptibility to certain diseases.

According to experts in medical curriculum development [9], 'reflection on … new experiences built into the curriculum is a key component of experiential learning'. In this formulation, personal genotyping by Stanford students was the new experience and the fact that they were using their own DNA and genotype data triggered deeper and more sustained reflection on and increased motivation to learn the course material. Thus, students' personal genotyping amounts to an engineered teachable moment [10] (Figure 1) that is based on a direct, personal life experience that students concluded would help them better relate to future patients who might undergo a similar test [2].

The Stanford students were skeptical and even dismissive of genotyping results related to their risk prediction for complex disease and cognitive-behavioral abilities [2]. In our view, this reaction was appropriate given the state of the science and the generally weak, probabilistic nature of most GWAS associations. Results pertaining to drug metabolism and reproductive issues (that is, carrier testing) were considered more relevant and valuable, although not imminently important to the healthy, young individuals in this cohort. Because the Stanford course focused on pre-symptomatic genotyping for disease risk assessment, it precluded full coverage of more medically 'actionable' genotyping for acute, post-diagnostic applications, such as cancer prognosis and tumor subtyping for targeted therapy.

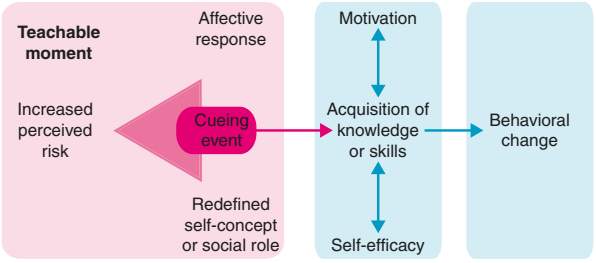


**Figure 1. Teachable moments are distinguished by a cueing event (also called a triggering or sentinel event) that increases perception of risk, elicits an emotional response, and/or represents a life experience that changes an individual's self-concept or one of their social roles [10].** In the case of the Stanford GENE 210 curriculum, the cueing event was the experience of personal genotyping. The students who were surveyed overwhelmingly felt that having their personal data motivated them to acquire new knowledge (the course material) and skills (in bioinformatics) [2]. Students also expressed intentions to make modest behavioral changes [2]. Figure adapted with permission from [5].

Although genetic counseling services were designed as an essential, supportive component of the Stanford course, few students availed themselves of consultations and none felt that such counseling should be required of students enrolled in the class [2]. The students believed that, because they were health professionals-in-training, they possessed the background and skills to interpret the results properly. Many were motivated and interested to use bioinformatics tools to reinterpret their raw data, although some felt the need for more individualized assistance and consultative support for this analytic 'data mining' activity [2].

One area of education that seemed curiously lacking in the experience provided to students was an understanding of the consultative support role of clinical laboratory professionals and the regulated environments in which they operate. The course designers seem to have taken for granted the integrity of sample tracking, analytic validity of the genotyping assays and the accuracy of the results, despite well-publicized errors that have been made by some commercial laboratories. Pathologists are the physician-custodians of laboratory testing and important consultants on diagnostic tests that determine significant

medical interventions [7]. We suggest that, for future programs, these specialists in the conduct and interpretation of clinical laboratory tests should be included as members of multi-disciplinary curriculum development and delivery teams.

## Conclusions

The work of Vernez *et al*. [2] provides evidence of the benefits of personal genotyping as a pedagogical tool for teaching medical genomics. This experiential education approach is grounded in well-established learning theory and practice and, despite potential ELSI issues [2], can be conducted in a manner that minimizes risk. More work is needed in the future to develop broader and more nuanced course content and to refine consultative support.

### Abbreviations

DTC, direct-to-consumer; ELSI, ethical, legal and social implications; GWAS, genome-wide association studies; SNP, single-nucleotide polymorphism.

### Competing Interests

MSB is on leave from Harvard Medical School and Beth Israel Deaconess Medical Center and is currently serving as Chief Medical Officer of Genome Health Solutions, Inc.; he is also a minority partner in Celebrity Diagnosis LLC. MRB is the co-founder and managing partner of Celebrity Diagnosis LLC. RMB has no competing interests.

### Authors' contributions

All authors participated in writing the manuscript

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### References

1. Boguski MS: **Online health information retrieval by consumers and the challenge of personal genomics.** In *Essentials of Genomic and Personalized Medicine*. Edited by Ginsburg GS, Willard HF. London: Academic Press; 2010:151-160.
2. Vernez S, Salari K, OrmondKE, Lee SS: **Personal genome testing in medical education: student experiences with genotyping in the classroom.** *Genome Med* 2013, **5:**24.
3. Weinstock CP: **When Genetic Testing Can Be Dangerous to Your Health.** *The Oprah Magazine*, March 2013, 95-97 [http://www.oprah.com/health/Genetic-Testing-Dangers-DNA-Research-Breakthroughs#ixzz2Mr4avd6V]
4. Gruman J, Smith CW: **Why the Journal of Participatory Medicine?** *J Particip Med* 2009, **1:**e2.
5. **Celebrity Diagnosis Magazine** [www.CelebrityDiagnosis.com]
6. Haspel RL, Arnaout R, Briere L, Kantarci S, Marchand K, Tonellato P, Connolly J, Boguski MS, Saffitz JE: **A call to action: training pathology residents in genomics and personalized medicine.** *Am J Clin Pathol* 2010, **133:**832-834.
7. Saffitz JE: **Genomic pathology: a disruptive innovation.** *Per Med* 2012, **9:**237-239.
8. Boguski MS: **Pathologists and the third wave of medical genomics.** *Per Med* 2012, **9:**241-242.
9. Kern DE, Thomas PA, Hughes MT: *Curriculum Development for Medical Education: A Six-Step Approach*. 2nd edition. Baltimore: The Johns Hopkins University Press; 2009.
10. Lawson PJ, Flocke SA: **Teachable moments for health behavior change: a concept analysis.** *Patient Educ Couns* 2009, **76:**25-30.

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