

Online Supplement

A Curriculum in Genomics and Personalized Medicine for Pathology Residents

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We have taken a structured approach to develop a practical curriculum in genomics and personalized medicine that would also generate resident enthusiasm and interest in the subject.^{1,2}

Developing Objectives and Educational Strategies

Recognizing the need for resident training in genomics and personalized medicine, a group of pathology faculty and genetic counselors with clinical and research interests in genomics, the residency directors, and the chair of the pathology department at Beth Israel Deaconess Medical Center, Boston, MA, collaborated on establishing the objectives of a new curriculum. The variety of objectives necessitated using several different educational strategies ■ **Table 1** and ■ **Figure 1**.

To achieve knowledge-based goals, the residents attend 3 one-hour lectures on personalized genomics, high-throughput sequencing, and genetic counseling (lectures available at <http://www.genomicmedicineinitiative.org>). During the first lecture, entitled “Clinical and Educational Genomics: Personal Experience With Direct to Consumer Genotyping,” the lecturer compares the 3 direct-to-consumer (DTC) personalized genomics companies and explains personal experience of ordering the kits and being tested. The lecturer also contrasts the often incorrect perceptions of the lay public regarding personalized genomics with the true promise these genomic technologies hold for customized medical care and the role pathologists should have in developing new testing paradigms.

The second lecture, entitled “Next-Generation Sequencing,” includes the limitations of conventional (Sanger) sequencing and the concept of next-generation sequencing. The lecturer reviews the advantages and disadvantages of the main next-generation sequencing technologies in the marketplace today and the effect this technology will have on the practice of pathology.

The third lecture, entitled “Genetic Counseling and Personalized Genomics,” reviews the use of personal and family medical history and traditional test results (ie, biochemical, cytogenetic, and single-gene) for risk assessment in genetic counseling. The lecturer then explores how addition of results from currently available DTC genomic testing might influence risk assessment and patient care.

For adult learners, the “need to know” is a powerful motivator.² As such, in the month following the lectures, residents are offered personal genotyping kits from Navigenics (Foster City, CA), a company that directly markets genetic testing to consumers. By following the instructions in the kit, a saliva specimen is submitted by each participating resident. In 2 to 3 weeks, the company provides a report on genetic “risk” for 28 conditions. The process is the same for people who order the kits directly from the company. Participation is completely voluntary, the results are completely confidential, and genetic counselors are provided by the company to discuss any results. Although the idea of using one’s own samples for resident education in clinical pathology is not new, it is of particular importance for this novel testing.³ Aside from adding personal relevance, we believe offering the testing will also promote learning by allowing residents to become “consumers,” enabling a better understanding of the implications for patients in the era of genomic medicine. An open forum is

Table 1
Curriculum Objectives

Teaching Modality/Objectives

Lecture I: Clinical and educational genomics: personal experiences with direct-to-consumer genotyping
 Describe how personalized genomics might improve the understanding and management of human health and disease
 Describe standards of scientific evidence applied to gene-disease associations and different approaches to risk assessment
 List the educational and informational resources for personalized genomics and its potential
 Define the issues related to evaluation of direct-to-consumer genotyping services
 Interpret genomic information and its correlation with related personal medical and health information

Lecture II: Next-generation sequencing (NGS)
 Describe the automated applications of NGS technology
 Describe the current and near-future diagnostic applications of NGS
 Interpret the key metrics and parameters that govern an NGS sequencing project

Lecture III: Genetic counseling and personalized genomics
 Define genetic counseling
 Critically analyze evidence for association of SNPs with disease risk
 Interpret implications of genomic testing results in the context of other health information
 Describe how the lay consumer's understanding of the implications of their direct-to-consumer test results may differ from that of a health care professional with experience interpreting such results
 Discuss the ethical concerns related to personal genomic testing and direct-to-consumer testing

Offering testing to residents
 Discuss the implications of genomic testing for the patient and physician
 Discuss the implications of direct-to-consumer test marketing for patients and physicians

Resident presentations
 Describe types of GWAS
 Critically evaluate and identify common problems with GWAS
 Describe the technology used in GWAS
 Present data from GWAS regarding the genotype-phenotype correlation for a tested risk factor

GWAS, genome-wide association studies; SNP, single nucleotide polymorphism.

also held with the residents to discuss the implications of the Navigenics testing and DTC testing in general for physicians and patients.⁴

In the final component of the curriculum, the residents are given the list of risk factors that are screened for by 3 personalized genomics companies (Navigenics; 23andMe, Mountain View, CA; and deCODEme, Reykjavik, Iceland)

and asked to select one to discuss with a faculty mentor. The mentor and the resident review the literature on the strength of the genotype-phenotype correlation and then present their findings in a 10-minute session to the other residents and course faculty. The residents have the opportunity to apply what they have learned during the previous components of the curriculum, and, by interpreting genome-wide association

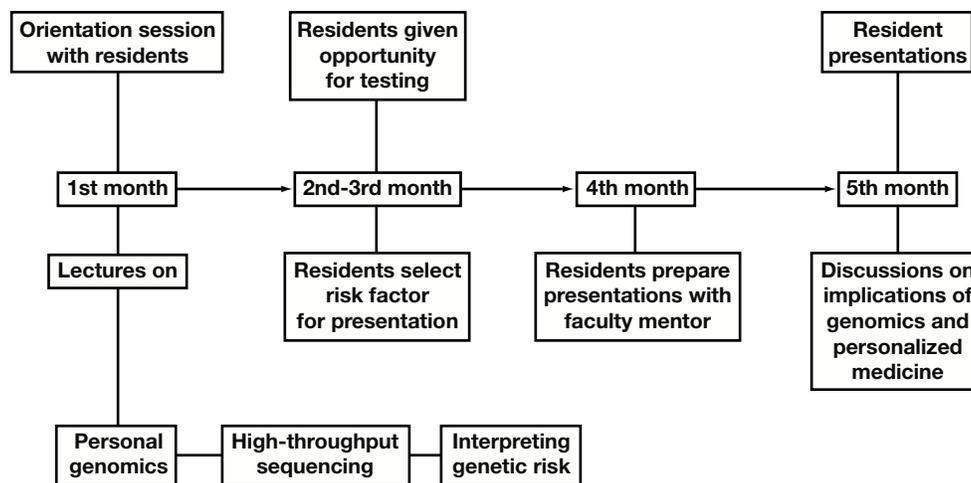


Figure 1 Structure of a pathology resident curriculum in genomics and personalized medicine.

studies for specific risk factors, they also can better understand the potential usefulness and pitfalls of genomic testing. The goal is to create a practical, self-directed exercise because all pathologists will need these skills in the near future.²

Implementation

The course faculty also tried to anticipate potential barriers to implementation of the curriculum.¹ Recently, Stanford University, Palo Alto, CA, considered offering similar testing to its medical students during an advanced genetics course.⁵ This proposal led to a “sometimes heated” debate and, ultimately, a temporary moratorium on this testing. Concerns included access to genetic counseling, potential stress from unwanted results from genetic testing, and concern about peer pressure leading to forced testing. By being aware of the Stanford moratorium, we undertook specific steps to mitigate these issues: (1) The residents were well informed of the technology and the types of results they would be receiving. (2) Genetic counseling would be available to the residents through the company providing the testing. (3) The residents were able to keep their results confidential. (4) The testing was completely voluntary, with no requirement to be tested to participate in the other aspects of the curriculum.

At Stanford, there was also concern about need for institutional review board (IRB) approval and possible conflicts of interest given faculty interactions with testing companies. In regard to IRB review, we consulted with the Committee on Clinical Investigations at Beth Israel Deaconess Medical Center. Given this is an educational initiative with no intent to “study” our residents, the consensus was that IRB approval was not required.

In regard to conflicts of interest, we obtain test kits for the residents from Navigenics. Navigenics is the preferred company for this curriculum because, unlike most major DTC genomic testing companies, it has on-staff genetic counselors available to address questions or concerns that customers might have about their results. Although Navigenics performs the genetic testing for residents who choose to have it, the company has no involvement in the planning or execution of the curriculum. In addition, none of the program’s faculty members have a financial relationship with Navigenics.

The course faculty also sought to promote internal support for the new curriculum.¹ We introduced the concept to the residents through an e-mail message describing the new curriculum, followed several days later by a discussion session. This session, held with the curriculum faculty, residency

directors, and the chair of the pathology department at Beth Israel Deaconess Medical Center, reiterated the rationale behind creating the curriculum and the overall goals and objectives. We emphasized the voluntary and confidential nature of the testing being offered.

We also chose to integrate the curriculum into the second year of pathology training because we believe these residents have the greatest motivation to participate. For anatomic pathology (AP)/clinical pathology (CP) residents, our program is structured with the first year being entirely AP and the second year entirely CP; the third and fourth years are for reinforcement of learned concepts. Because CP is focused on laboratory diagnostics, the second year seemed ideal for introducing a curriculum in genomic medicine. Second-year residents are also not under the acclimation pressures of first-year residents.

Curriculum Assessment and Future Goals

The first iteration of this curriculum began in October 2009. We plan on evaluating the curriculum by having residents participate in a feedback session with the course faculty. They will also be given anonymous evaluation forms to share their thoughts about various components of the curriculum and a general impression of the usefulness of the program.

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