commentary

Genetics of common disease: a primary care priority aligned with a teachable moment?

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We do indeed live in interesting times. Through genome-wide association studies, the Human Genome Project and the related HapMap project are finally yielding dividends related to the genetics of common disorders with great public health import. Not surprisingly, the rush is on to capitalize on these discoveries. Over the last several months, three companies (Navigenics, Inc., deCODE genetics and 23andMe, Inc.) have each announced the availability, direct to consumers, of reasonably low-cost pan-genome testing for markers thought to be predictive of traits and disorders ranging from earwax type to coronary artery disease. Each company has taken a slightly different approach to enhancing their customer's understanding of their genome. Navigenics, Inc., for example presents a more "medical" service, focusing on disease risk estimates, whereas 23andMe, Inc., appears to be taking a more "recreational" tack, with an emphasis on ancestry and the ability to follow traits in users' families. A fair amount of ambiguity exists in the science underlying many of the trait-marker associations that serve as a basis for each company's services, especially in diverse racial and ethnic populations. Perhaps more importantly, there is a virtual abyss regarding how (or whether) such information should be applied in clinical medicine. Each of these applications has a default suggestion when ambiguity rears its ugly head: ask your health care provider for more information.

There is substantial reason to believe that the health care providers fielding such questions will be primary care providers. According to Centers for Disease Control and Prevention data from 2004, approximately 48% of the 1.1 billion ambulatory care visits that occurred in the United States in 2004 were in a primary care setting. This contrasts to about 18% occurring in medical specialty settings, of which visits to medical geneticists likely represent a very small percentage.[1] In fact, medical geneticists represent 0.18% of the 700,000 physicians in the United States.[2] Further, there are data suggesting that the American public expects their primary care providers to know something about genetics, with 72% of 1000 individuals in the United States surveyed in 1998 saying that they would turn first to their primary care provider with a question about a genetic disorder.[3] When patients query their primary care provider regarding the genetics of common disease, they are likely to be disappointed in the answer. Evidence suggests that, on average, primary care providers are ill-equipped to deal with topics in genetics and genomics both from the standpoint of time and education.[4,5] Some within both the primary care and genetics communities, recognizing that primary care would be faced with this challenge, have been calling for increased attention to genetic literacy among providers for some time.[6,7] This call has met, and continues to meet, with ambivalence in segments of both primary care and genetic communities.[8,9]

This issue of *Genetics in Medicine* contains a pair of articles from a group of European investigators that suggest the tide of interest in genetics may be turning in the primary care community.[10,11] The first of these articles relates the validation of a quantitative survey instrument designed to measure the educational priorities of practicing health care providers regarding genetics and genomics topics. The second examines how 1168 general practitioners in five European countries (France, Germany, Netherlands, Sweden, and the United Kingdom) responded to the same instrument. Despite reasonably dramatic differences in survey response rates and provider demographics across the health care systems, "Genetics of Common Disease" was the highest priority of six genetic education topic areas for each country, with an aggregate rating of importance of approximately 3.5 on a 5-point Likert scale. Coming in second and third were "Approaching Genetic Risk Assessment in Clinical Practice" and "Psychosocial and Counseling Issues," respectively. The fact that these priorities were held in common across providers in five distinct health care systems and countries suggests that similar results might be obtained from deploying the survey in the United States primary care provider population. Unfortunately, the survey instrument did not ask the providers to rank topics in genetics and genomics relative to other topic areas outside of genetics and genomics (chronic disease management, childhood vaccinations, etc.), so we have little idea how they would rate "Genetics of Common Disease" in the overall scheme of their educational priorities. This lack of a sense of relative priority ascribed to genetics education in comparison with the other challenges facing primary care providers demands attention. Competing educational priorities may be even more problematic to achieving genetic literacy among primary care providers in the United States, where the lack of a national health care system results in multiple additional pressures affecting how primary care providers select educational options.

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That new information on the causation of common chronic disease is of interest to primary care practitioners should come as no surprise, as it represents an alignment of discoveries in genetics with a topic area central to the culture of primary care.[12] "Genetics of Common Disease" seems a target of opportunity for increased dialogue between the genetics and primary care communities. In many ways, the topic represents an ideal area for interface. The clinical genetics community as a whole is not experienced in screening for, diagnosing, and managing common, chronic, adult-onset conditions. On the other hand, the primary care community is unfamiliar with interpreting the complex issues in population genomics and gene-environment interactions which are critical to the appropriate application of genetic testing for risk of common disorders. Both sides come to the table with a knowledge base and skill set that the other can learn from. Further, there is clearly no basis for competition between medical specialties given the shear volume of potential work. The recent efforts of Navigenics, Inc., deCODE genetics, and 23andMe, Inc., should serve to focus the dialogue, which has previously been hampered by a paucity of concrete clinical applications.

How might the dialogue be advanced? In the United Kingdom, which has a national health care system, the model has been to establish the National Genetics Education and Development Centre (see www.geneticseducation.nhs.uk). Led by Professor Peter Farndon, and deriving funding from the UK National Health Service, this organization has begun an ambitious process to engage and educate health care providers. One would suspect that the genetics of common disease will likely become a priority for them. In the United States, there is no equivalent governmental organization. However, in 1996, the National Coalition for Health Professional Education in Genetics (NCHPEG; see www.nchpeg.org) was established in the United States as a public-private partnership providing a forum for interactions among partners in the health care community with an interest in genetics education. The membership of NCHPEG includes over 80 health professional organizations, representing a broad spectrum of primary care groups as well as genetic service providers. NCHPEG's track record is distinguished by examples of the successful development of synergistic cross-cutting relationships among professional organizations. These relationships have yielded concrete educational products such as the collaboratively created web tool "Genetics in the Physician Assistant's Practice" (see: http://pa.nchpeg.org/).

Personalized genomics in its current form represents a potentially disruptive technology that will both empower and imperil consumers who are early adopters. Depending on the level of public uptake of these services, the downstream consequences could be profound. At a minimum, the wash of data will stress the capacity of the current model for the provision of genetic services and place pressure on primary care practitioners to react to the informational barrage in a vacuum of solid evidence-based guidelines. Given the overall health care environment, mismanagement of this type of genomic data could worsen the fiscal crisis and widen health care disparities. A coordinated approach to personalized genomic testing by the primary care and genetics communities might help the health care system to avoid potential pitfalls, and realize the potential of this technology. The articles contained in this issue of *Genetics in Medicine* suggest that the primary care community (at least in Europe) is ripe for engagement regarding the genetics of common disease. The time for effective action by both communities, perhaps through NCHPEG, is now.

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