COMMENTARY | Genetics inMedicine

# Primary-care physicians' access to genetic specialists: an impediment to the routine use of genomic medicine?

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New genetic and genome-based testing strategies have ushered in the era of genomic medicine. Genetic testing services are playing an increasingly larger role in multiple practice domains. In particular, primary-care physicians (PCPs) are poised to play a major role in the delivery of genomic medicine, given their focus on disease prevention and screening.[1] At this early stage of test adoption, the safe and appropriate use of new genetic tools will require a combination of new educational resources, for both patients and health professionals, point-of-care guidance, and, at least initially, ready access to genetics experts. As tests become more standard, delivery models will evolve with increasing physician knowledge. Arguably, the best practice standards will emerge from a collaboration between medical genetics and other clinical specialties.

Genetics experts—board-certified genetic counselors and doctorate-level professionals—are specially trained in the use, interpretation, communication, and implementation of genetic test results. There are a total of 2,394 diplomates from the American Board of Medical Genetics, with at least one in each state; 1,066 are certified as clinical geneticist physicians, although presumably fewer are practicing as a result of administrative or other nonclinical roles, retirement, and death. According to the American Board of Genetic Counseling, there are more than 3,000 board-certified genetic counselors, with wide variability in their geographic distribution, ranging from zero in West Virginia to a high of 466 in California. Although the number of genetic specialists is relatively low as compared with other specialties, it is not clear if or how many more will be needed to provide care, given the increased use and number of genetic and genomic applications.

Wide variability in physician knowledge about genetics has been associated with their likelihood of using these tools.[2] If PCPs do not have access to geneticists, this lack of access may pose a major barrier for PCPs in the practice of genomic medicine. In a national survey of PCPs, we asked about their access to genetic specialists (refer to Haga et al., 2012[3] for further information). More than half of respondents (53.4 ± 2.24%) indicated they do not have access to genetics expertise. When parsed by practice setting, 74.9% of PCPs based in hospitals (±7.65%) indicated they had no access to genetics expertise if needed, followed by 52.9 (±2.49%) of community-based PCPs, and 17.7% of PCPs based in academic medical centers (±7.17%). No association was detected between medical specialty (family medicine versus internal medicine) and access to genetics expertise ($P = 0.91$). A positive association was observed between access to a genetics specialist and use of genetic testing for disease diagnosis or susceptibility ($P = 0.0001$). Of the PCPs who indicated they had access to a genetics specialist, 32.5 (±3.55%) reported referring patients to a specialist to order genetic testing 1–2 times per year and 40.0 (±3.76%) 3–10 times a year; 16.1 (±2.88%) reported that they have never made a referral to a genetic specialist. Internists were less likely to have ever made a referral to a genetics specialist as compared with family-medicine physicians (63.9 vs. 76.9%) ($P < 0.001$). The majority of respondents strongly or somewhat agreed (91.6 ± 1.23%) that they would refer a patient to a genetic specialist if he or she had questions about how test results or incidental findings may affect family members.

Given the high density of geneticists in academic medical centers, it is no surprise that PCPs outside of these centers have limited access to geneticists. But Klitzman et al.[4] reported that about half of internists surveyed at two academic medical centers did not know of a geneticist/genetic counselor. Therefore, regardless of geographic proximity, access for some PCPs may be effectively limited if they are unfamiliar with these experts or have not had any clinical occasion to consult them. Although options for direct referral to genetic experts may be limited, other modes of consultation are beginning to be routinely used, particularly by genetic counselors, as a result of the demand for their services and their limited numbers. Counseling by phone has become more common, particularly for counseling about genetic risk for diseases such as breast cancer and does not appear to adversely affect patient understanding or anxiety and may even increase knowledge of risk and motivate intention to change.[5] Similarly, telemedicine is being increasingly used and shows high levels of patient satisfaction.[6] Some companies offering testing directly to consumers, such as 23andMe and, formerly, Navigenics, also provide the option to speak with a genetic counselor by phone. In addition, some companies have been established to provide genetic counseling services by phone, including InformedDNA and Genetic Counseling Services.

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Submitted 24 September 2012; accepted 15 November 2012; advance online publication 10 January 2013. doi:10.1038/gim.2012.168

Another strategy for optimizing the number of trained genetics specialists is to collaboratively develop educational tools, training programs, core competencies, and practice guidelines. A number of programs and initiatives have been developed between geneticists and PCPs,[7] but broader dissemination and utilization are needed to increase outreach and access to these resources. Although there appears to be consensus on the need for greater education about genetics and genomics,[8] this process is gradual and must be complemented with multiple and more immediate resources to increase awareness. For example, if a genetics specialist is not available, what is a PCP to do for patients presenting results of a genome screen ordered from a direct-to-consumer company or asking about whether they should have a test that they heard about on the news? Clinical decision supports or point-of-care resources may be particularly valuable for tests urgently needed for treatment decision making, such as pharmacogenetic testing to predict drug response.

Perhaps geneticists have inadvertently slowed the use of some genetic tests by health-care practitioners not formally trained in genetics. As with many areas of medicine, some types of genetic tests may be relatively straightforward and easily integrated into the workflow of PCPs. However, other types of tests may be very complex. By emphasizing the complexity of test interpretation, informed consent, determination of clinical criteria for patient eligibility, and related ethical, legal, and social issues, geneticists may have decreased PCPs' willingness to manage these issues themselves.[9] However, these efforts are important to educate test users about the appropriate use of tests and to minimize patient harms. Nevertheless, some PCPs may prefer to counsel patients on their own, believing that a genetics consultation would be of no benefit to the patient[10] or that the physician has responsibility for the counseling about genetic testing, although their own knowledge may be poor.

With the development of new genomic tools intended for a more general patient population to assess risk for diseases such as heart disease and diabetes, and response for commonly prescribed medications, practitioners and educators are trying to catch up to a rapidly moving field. A collaborative approach to the development of practice guidelines and educational tools could have multiple benefits. In addition to building on genetics expertise in test interpretation and diagnosis of rare diseases, jointly developed approaches could use primary-care expertise in screening, prevention, and disease management. Both PCPs and geneticists might also benefit from sharing the communication skills developed in their respective disciplines. In addition, the increased visibility of genetic and genomic testing may attract more physicians to the discipline, promote greater awareness, and provide an opportunity to educate other physicians about the use of such services.

Genetics and primary care have much to learn from each other.[7] Results from new genomic tests will require careful assessment to ensure that accurate inferences are made about their clinical meaning, both for the patient tested and for family members. Some of the clinical implications will relate to uncommon inherited conditions typically encountered in medical genetics. Increasingly, however, genomic tests will also yield information about health issues related to common multifactorial diseases, risk states, and medications, for which primary-care expertise is needed. Collaborative assessment of clinician needs, and development of the tools and guidance to address those needs, will help to address the limitations in the availability of genetics professionals. A hybrid model of education and support for PCPs and access to specialist consultation when needed may address some of the challenges posed by new genomic applications. Such strategies will allow for enhanced approaches that draw on the complementary expertise of genetics and primary care.

## ACKNOWLEDGMENT

This work was supported by the National Institutes of Health (R01 GM081416-01A1).

## DISCLOSURE

The authors declare no conflict of interest.

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