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What Is the Role of Nongeneticist Physicians, and Are They Prepared for It?

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You are a specialist physician, and someone comes into your office for a new patient evaluation—your differential diagnostic possibilities include several conditions for which a genetic test is available. Or you are a primary care physician, and, in taking your new patient's family history, you realize that this 30-year-old woman has a family history of breast and ovarian cancer that suggests she may be at increased risk herself. Or finally, an established patient comes into your office with results from direct-to-consumer genetic testing and would like for you to explain the meaning of the SNP test results. Now what?

Do you feel prepared to handle your patient's genetic histories, order appropriate testing, interpret the results, and discuss their implications for the patient and family? If not, you are not alone. Genetic-testing technologies are moving at a fast pace, and even physicians in a single specialty area can have difficulty keeping up-to-date on what tests are available, which laboratories are offering the most sensitive test, and the most effective way to proceed with testing. Moreover, most of these technologies had not been discovered when the majority of practicing physicians received their medical genetics training (often through a single course in the first year of medical school), and, for them, genetics and genomics are even more challenging.

A number of studies have assessed what nongeneticist physicians know about genetic testing, their ability to interpret test results, and their comfort with handling genetics cases. In one study, only 10 percent of respondents felt they knew "all [I] need to know about genetics for my job" [1]. Baars et al. suggested low levels of genetics knowledge in various physician groups, and another study suggested that the knowledge of carriers for hereditary nonpolyposis colon-cancer mutation equaled that of their physicians [2, 3]. Other studies document that a high percentage of physicians misinterpreted genetic test results and that they did not use published screening criteria appropriately [4, 5]. Studies like these consistently suggest that both primary care and specialist physicians feel uncomfortable with their ability to manage genetics-related scenarios that arise in their clinical practice. So what do doctors need to know to practice medicine in the age of genetics?

There is no uniformly accepted answer to this question. Medical genetics is considered a primary specialty by the AMA, but the American College of Medical Genetics estimates that there are only about 1,100 board-certified medical geneticists working across the United States, primarily at tertiary medical centers [6]. For the past 40 years, genetic counselors (now certified by the American Board of Genetic

Counseling and, in a handful of states, licensed) have assisted in providing genetic care. There are approximately 3,000 genetic counselors working in the United States with both medical geneticists and other physician specialists—primarily in oncology, cardiology, and neurology, but the number of areas in which genetic counselors work is continuously expanding. There is a smaller number of nurse geneticists who assist in caring for patients with genetic conditions. Given the relatively small numbers of genetic specialists and the fact that they are located primarily at academic medical centers, it is unlikely that physicians will be able to refer all their genetics-related patient issues to specialists. As summarized by Korf et al.,

> …genetics is expected to be incorporated into routine care across all of medicine, and whereas a board-certified physician geneticist will not be involved in every medical decision based on family history information or interpretation of a genetic test, a physician geneticist will be understood to be the expert in these areas [7].

So what should the primary care and nongenetics specialist physician know about genetics? Several publications have presented competencies for offering appropriate genetic and genomic health care [8, 9].

What Physicians Should Know about Genetics

From a practical standpoint, all physicians should have a solid understanding of modes of Mendelian inheritance, which is critical to accurate risk assessment. Specifically, physicians should be fluent in the concept of penetrance (whether or not a genetic mutation results in symptoms in a patient—nonpenetrance is the genetic explanation for conditions "skipping a generation") and variable expression (that a single genetic mutation can result in different features and age of onset even within the same family). Physicians should also understand the concepts of polygenic inheritance (meaning that multiple genes, often with low penetrance as demonstrated by small odds ratios in genetic association studies, contribute to the occurrence of a complex disorder) and multifactorial inheritance (meaning that some combination of genes and environment leads to the phenotype).

With regard to genetic testing, it is key for physicians to understand that the sensitivity of genetic tests varies dramatically based on the type of testing performed and that, at present, no single genetic test detects 100 percent of mutations in a given condition. Variants of uncertain significance may be detected that require additional family studies or laboratory reclassification as deleterious or benign. Initially, testing an affected family member remains the most efficient way to determine the usefulness of a genetic test within a family. For terminally ill affected family members who are not found to carry a genetic mutation through currently available testing, DNA banking for future testing can be suggested, given how rapidly our knowledge in this area is evolving.

All physicians should be able to take a comprehensive three-generation family history to assess the risk to various family members, provide counseling to their patients about which family members may be at risk for the condition, and encourage

patients to inform relevant family members about the risks. Physicians should also be able to recognize the red flags of common adult-onset conditions that have a strong underlying genetic basis (e.g., earlier onset than is typical, multiple affected family members, occurrence in the less-frequent sex, or bilateral occurrence). Finally, physicians should be aware of how and when to make referrals to genetic specialists or other specialist physicians who manage genetic conditions and should have local or regional contacts available for such referrals.

Physicians should be cognizant that genetic conditions, by definition, involve not only their individual patient but the family and may raise issues concerning confidentiality and disclosure. Most professional societies do not recommend genetic testing of children unless test results will change medical management for the child in the near future; in particular, predictive testing for adult-onset conditions is discouraged in order to preserve the child's future right to decide whether he or she wants to be tested. Physicians should also be aware of state laws on protection of genetic information and the federal Genetic Information Nondiscrimination Act which protects against discrimination by health insurance companies and employers on the basis of genetic information, including family history.

Beyond the above general issues, specialty physicians should be aware of the current status of genetic testing for conditions within their specialty, including medical management of individuals with specific mutations. For symptomatic patients, genetic testing may refine the diagnosis (particularly when testing has ruled out other probable diagnoses), and mutation status may become increasingly relevant in clinical trials or treatment protocols. The identification of a mutation in an affected person may suggest the need for predictive testing of relatives. Identified mutation carriers can undergo earlier surveillance or treatment to minimize or delay the onset of symptoms, and those who test negative can avoid unnecessary future screening. Predictive testing also gives individuals information on which to base long-term life decisions, including reproductive planning.

Nevertheless, many do not want such knowledge, particularly if no medical surveillance or treatment is available for the condition. As a result, physicians should work closely with genetics experts to ensure they (the physicians) are securing informed consent (particularly for predictive testing) and that they are ordering and interpreting genetic test results accurately. In some settings, this may mean the incorporation of a genetic counselor or geneticist into a multidisciplinary clinic setting. In others, it may mean that the specialist develops expertise in this area, making referrals to genetics specialists for more complicated cases.

Teaming Up to Provide Genetic Information

How can physicians gain the genetic knowledge they need to provide their patients with the best care? Teamwork among genetic specialists and other health care professionals is a start. Primary care and nongenetics specialists share the advantage of having longer-term relationships with their patients than do genetic specialists, who function primarily in a consultant role. When combined with their ability to

facilitate medical decisions and discuss probabilistic outcomes, primary care and specialist physicians are well positioned to play a role in the provision of genomic-based health care. Genetics professional organizations such as the American Society of Human Genetics, American College of Medical Genetics, and National Society of Genetic Counselors can be resources for genomic information, educational programs, and referrals. Physicians can also make use of web resources such as genereviews.org, genetests.org, and OMIM to obtain information about the genetic basis of various conditions and current status of genetic testing.

Few articles document what is currently being taught in medical schools, in either the preclinical and clinical curriculum years, or in a GME setting [10-13]. Most medical schools teach genetics primarily through a first-year course that includes a combination of basic science and clinical information. It appears that a minority of clerkships include required medical genetics training, but this has been poorly documented. Basic genetics knowledge such as Mendelian and non-Mendelian inheritance, principles of risk assessment, and the skill of accurately taking and interpreting a family history should be taught in the preclinical years and reinforced during clinical training and residency. General clinical principles, including medical-test interpretation (e.g., clinical and analytic validity and utility) and the ability to critically read and evaluate current literature remain central, given the fast pace of genetic technology and testing, and these skills should also be reinforced in the clinical setting as they apply to genetic cases. Finally, since data supports the need to reinforce genetics knowledge and skills during the clinical training years of medical school and residency, the use of genetics objective structured clinical exams (OSCE) cases and practical reinforcement of cases in rounds and on the wards will be critical, but will also require clinical educators to be informed and comfortable with the material to teach and reinforce it in their own clinical settings [11, 13].

The promise of genetics and genomics goes beyond the potential for testing our patients: increased knowledge about genomics allows us to better understand the underlying pathophysiology of disease, modifiers that influence disease onset and progress (both genetic and environmental), and treatments including pharmacogenomic therapies. If physicians don't feel comfortable with the technology, the promise will not be fully realized. Physicians should find ways to educate themselves about the implications of genomic health care and identify genetic specialists who can serve as their partners in providing high-quality care to patients.

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