

Tests for more than 1300 genetic conditions are available to assist individuals and health care providers with management decisions. Genetic tests are used to diagnose disease, predict risk of future disease, inform reproductive decision-making, and manage patient care. In the past, most genetic tests were performed in academic settings, ordered by medical geneticists familiar with the nuances of testing for and interpreting results for rare, so-called “single gene,” genetic disorders. But as knowledge about the genetic underpinnings of common, complex diseases such as diabetes, cancer, and heart disease expands, so will the use of genetic information in routine health care. The practice of genetic medicine is transitioning rapidly from the academic-based genetics clinic into other medical specialty offices and, increasingly, to the community-based primary care provider’s office.

It is the responsibility of health care providers to correctly use and interpret genetic tests. Clinician must know who to test, when testing is appropriate, the correct test to order, what information the test can provide, the limitations of the test, how to interpret positive and negative results in light of a patient’s medical or family history, and the medical management options available. The rapid pace of advances in genetics, the ease with which tests move into the marketplace, and the increasing number of tests available directly to consumers challenge busy clinicians to stay abreast of a fast-moving field. Studies continue to show that many health care providers are ill prepared to use genetic tests in clinical practice.

Professional health care organizations play an important role by developing clinical practice guidelines to aid clinical decision-making. Clinical practice guidelines are “systematically developed statements to assist practitioner and patient decisions about appropriate health care for specific clinical circumstances.” Practice guidelines include an assessment of the quality of the available evidence for a clinical scenario and provide recommendations for the clinician and patient based on that evidence. When effectively disseminated and utilized, guidelines have been shown to improve clinical outcomes. Despite their utility in aiding clinical decision-making, relatively few guidelines for genetic testing have been developed.

Developing robust practice guidelines that will positively impact clinical care is not trivial; it is time consuming and expensive. The rigor of the methodology used to review the available evidence and make recommendations is critical. Organizations frequently rely on busy professionals to volunteer their time. It is not unusual for guidelines to take two years from the time of conception to dissemination to the membership. And guidelines must get to clinicians at the point of care and be utilized to be effective.

In addition to the difficulties organizations face in developing any guideline, there are a number of challenges specific to genetic testing guidelines. There may be limited

genetics expertise to draw upon within the membership to develop the guidelines. Additionally, the clinical validity and utility of a genetic test rarely will have been studied in multiple, randomized clinical trials - considered the gold standard of evidence.

Many health care provider organizations look to outside groups to develop the evidence needed to make clinical recommendations. The U.S. Preventative Services Task Force (USPSTF) is a recognized source of evidence –based reviews and clinical recommendations. However, USPSTF has only taken on reviews for two genetic tests, BRCA1/2 testing to assess the risk for heritable breast cancer and HFE testing for hereditary hemochromatosis. More recently, the Evaluation of Genomics Applications in Practice and Prevention (EGAPP) Initiative of the Centers for Disease Control and Management was established to develop and test a systematic process for evidence-based assessment of genetic tests and other applications of genomic technology. Several reviews and recommendations have been disseminated widely by EGAPP for use by health care provider organizations.

The current piecemeal approach of developing genetic testing guidelines will not be sustainable in the long run; testing will quickly outpace the efforts of individual organizations. In order to fill the gap for clinical decision making tools, the efforts of groups such as USPSTF and EGAPP must be supported and expanded upon. Standard evaluation methodologies must be disseminated, and other groups must be supported in a systemic approach to developing and distributing genetic testing clinical guidelines.

*Last updated by Shawna Williams, August 2008*