

Tests for more than 1000 genetic conditions are available to assist health care providers with patient 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 testing was for rare, so-called “single gene,” genetic disorders. They were ordered by geneticists familiar with the nuances of genetic testing and test interpretation and performed in academic laboratories with which the geneticist had an established relationship. But as the genetic contribution to numerous disorders becomes known, genetics increasingly is being practiced in other medical specialties. And as knowledge about the genetic underpinnings of common, complex diseases – diabetes, cancer, heart disease, and the like – expands, so will the use of genetic information in routine health care. Thus, the practice of genetic medicine is transitioning 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 the health care provider to correctly use and interpret genetic tests. The clinician must know when it is appropriate to test, 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 the patient’s medical or family history, and the medical management options available. However, studies continue to show that many health care providers are ill prepared to use genetic tests in clinical practice.

Professional health care organizations can play an important role in 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. The guidelines that exist were developed by those professional health care organizations whose practices were impacted first by the availability of genetic tests – those practicing medical genetics, pediatrics, reproductive medicine, and more recently, oncology. Yet it is inevitable that as genetic testing moves into the medical mainstream, a wide variety of professional health care organizations will need to develop guidelines.

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 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. The rapid pace of advances in genetics and ease with which tests move into the market also is challenging. 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. Additionally, the minimal level of governmental oversight of the laboratories performing genetic testing and the increasing number of tests available "direct to the consumer" over the Internet puts an added burden on clinicians and those developing genetic testing guidelines.

A piecemeal approach of developing genetic testing guidelines will not be sustainable in the long run; testing will quickly outpace the efforts of individual organizations. A mechanism to support health professional organizations to develop, disseminate, and monitor the implementation of genetic testing guidelines would improve the number and quality of genetic testing guidelines. Federal agencies whose mission is to translate research finding into clinical practice could be a source of this support.

A sustainable source of financial support to which organizations could apply would enhance the number and quality of genetic testing guidelines available and address many of the challenges of developing guidelines in this rapidly expanding area of medicine.

The availability of funding would raise the priority of genetic testing guidelines in organizations where genetic testing must compete with other clinical practice areas for organizational attention and resources. Organizations could devote resources to developing, testing, and disseminating supplemental educational materials for patients and providers. Additionally, criteria could be established for who can apply and what should be included in the guideline. Applicants could be required to document their expertise in a particular topic area and the methods by which they will review the evidence and make recommendations; detail plans for provider and patient education; outline how the guideline will be disseminated and monitored for its usefulness in practice, and revised or update as needed. Encouraging multidisciplinary applications would utilize resources more efficiently, reduce the redundancy of several organizations covering the same topic, and ensure a broader dissemination and implementation plan.

A sustainable funding source would provide standards to ensure that the translation of new genetic discoveries into the clinic occurs in a timely and safe fashion.

*Compiled by Joan Scott
March 2006
Updated by Shawna Williams
September 2006*