Opportunity Knocks

The Pathologist as Laboratory Genetics Consultant

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GENETICS AND MEDICINE

To paraphrase comments used by Francis Collins, MD, PhD, Director of the National Human Genome Research Institute, to challenge audiences in the 1990s, “Everything in medicine is genetic—or influenced by genetics—with the possible exception of trauma; and given disorders like osteogenesis imperfecta, I’m not entirely sure about that.” Advances elucidating the role of genetic factors in human disease and behavior, accelerated by the availability of the human genome sequence, are making a profound impact on medicine. The discovery of genes underlying Mendelian disorders is a routine occurrence, and much current research is directed at multifaceted disorders such as hypertension and cardiovascular disease. The emerging field of pharmacogenetics promises to exploit an individual’s genetic makeup to guide selection of pharmaceuticals, predict risk for disease, and more.1,2 DNA-based tests follow gene discovery. They offer improved (even presymptomatic) diagnosis of common and rare disorders, and are often important components of monitoring and preventing algorithms for inherited cancers. Clinical laboratories are experiencing significant increases in requests for routine genetic tests and newer assays of increased breadth and complexity. Development of Food and Drug Administration-cleared and analyte specific reagent assays has begun to make genetic tests available to nonspecialty laboratories. Vendors are working toward automated platforms for the performance of genetic assays. Public awareness of new genetic tests and direct-to-consumer marketing of tests will add to the demand for services.3

RESOURCES TO PROVIDE GENETIC TESTING SERVICES TO PATIENTS

Despite this revolution in DNA-based knowledge related to genetics, the number of entrants into medical genetics training programs in the United States has steadily declined.4 From 1997 to 2002, only 26 board-certified medical geneticists entered the field annually. Some states have few or no geneticists.4 One recent study described the outlook for the medical genetics workforce as critical.4 Given these realities, authorities predict that primary care physicians in the United States will order an increasing proportion of genetic testing to be performed.2,3 Generalists and physicians with limited genetics backgrounds will likely be called on to interpret and explain the results of genetic tests to patients. In some instances, this may require discussion of probabilistic results, risk estimates, or issues such as penetrance and variable expressivity. There is a substantial body of evidence suggesting that physicians are inadequately prepared to incorporate genetic information into their practices.3,5 Demands on primary care physicians leave little time to acquire this knowledge, or even to obtain detailed family histories.3,5 Specialists in narrowly defined fields will also not be comfortable with, or appropriate providers for, the diagnosis and management of many inherited disorders. The modest number of genetic counselors who graduate annually will help, but few are available in rural areas.6 Moreover, genetic counselors have limited medical backgrounds.

OPPORTUNITY/RESPONSIBILITY FOR THE PATHOLOGIST AS LABORATORY GENETICS CONSULTANT

The pathologist has long served as a consultant for testing of common and rare disorders. Pathologists oversee clinical laboratories, advise clinicians, choose laboratories to which send-out tests are referred, and decide when to bring referral testing in-house. With increasing demand for genetic tests, there is an opportunity—and perhaps a responsibility—for pathologists to expand their involvement in genetic services. Although genetics has not been an area of focus for most pathologists, molecular pathology (including genetics) exposure has been a requirement of residency programs for some time. Educational materials are also available through professional, government, and private Web sites.7 National pathology meetings offer molecular pathology courses that include material on genetic testing; an online course in molecular pathology is available through the College of American Pathologists. Molecular Genetic Pathology fellowship programs provide advanced training that includes laboratory and clinical genetics.

ECONOMIC FACTORS

Molecular genetic testing is billed using Current Procedural Terminology (CPT) codes 83890 to 83914 of the clinical laboratory fee schedule. Several codes—and often multiples of individual codes—are billed on the basis of the number of amplifications, enzyme digests, probes, sep-
arations, or other steps in an assay. Thus, even though payment for individual codes may be modest, technical component reimbursement can cover the per-test cost of reagents, personnel, and overhead. Moreover, because genetic tests are rarely urgent, samples can be batched for cost-effective testing. Add savings from expensive send-out testing, and the equation for the laboratory can be favorable.

There are also 3 Current Procedural Terminology codes for professional effort devoted to molecular genetic testing. Code 83912 (molecular diagnostics, interpretation and report) is billed with a -26 modifier for the interpretation of genetic tests. Multiples of 83912 are justified when several independent interpretations are made. Much of what pathologists will do as laboratory genetics consultants falls under consultation codes 80500 (clinical pathology consultation; limited) and 80502 (clinical pathology consultation; comprehensive). The Medicare criteria to bill these codes include a consultation request from a physician, interpretation that relates to a test result outside the clinically significant normal or expected range in view of the patient’s condition, a narrative report for the patient’s medical record, and the exercise of medical judgment. These requirements are integral to laboratory-based genetic consultation. Even consult requests that involve negative genetic test results may meet criteria of the Centers for Medicare and Medicaid Services because complete evaluation will often include consideration of other abnormal laboratory values and diagnostic studies.

Review of patient and family histories is important for proper interpretation of many tests. Appropriate narrative reports are essential to explain the significance of findings, risk implications, and indications for further genetic counseling. Given the extraordinary demands on contemporary physicians, consultation from a knowledgeable pathologist will likely be welcomed for many patients. After reviewing clinical and laboratory information, the pathologist can provide patient-specific risk assessments or alternative diagnoses, and suggest or discourage additional molecular or nonmolecular testing. The success of interpretative laboratory consultation programs can serve as a model for genetics services.

SUMMARY

One need only read a newspaper or magazine, or listen to television news to realize the important role genetics will play in the future of medicine. This area offers both opportunity and responsibility for pathologists to complement scarce resources and provide valuable laboratory consultative services to patients and primary care physicians.

References