Issues in Genetic Testing

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The Clinical Chemistry Forums of the American Association for Clinical Chemistry are designed to air controversial issues in the broad field of clinical chemistry. The Eighth Annual Forum was held on November 3 and 4, 1998. This Forum differed from previous meetings in the series in that it was sponsored cooperatively by the Association for Molecular Pathology. The topic of the meeting was “Issues in Genetic Testing”. The Forum examined the legislative, regulatory, professional, and scientific issues involved in genetic testing. The Clinical Chemistry Forums of the AACC are oriented toward the regulatory or administrative aspects of clinical chemistry, thus complementing the organization’s Oak Ridge Conferences on innovative technology and Arnold O. Beckman Conferences on clinical applications of clinical chemistry.

We publish here, in their entirety, some of the presentations from the 1998 meeting (1–4). Another article from the 1998 Forum has already appeared in these pages (5). A summary of the remaining presentations will be published in a future issue of Clinical Chemistry. The meeting topics ranged from an overview of genetic testing from its past through its current status to its possible future applications (1). Various aspects of current and pending legislation, in both the House and the Senate, were discussed. The most critical issues are related to the privacy of information and health insurance coverage for genetic testing. The ethical concerns associated with genetic testing are very real because of its potential to predict an individual’s susceptibility to certain diseases and even his or her response to various forms of treatment. The Chair of the NIH Task Force on Genetic Testing presented the group’s recommendations for regulating genetic testing (2). The current status and probable future direction of CLIA regulations (3) on genetic testing and the FDA’s position (4) with regard to regulation of genetic testing also were discussed. The legal responsibility for a laboratory to protect genetic data and the ways in which these differ from traditional data are important considerations for a clinical laboratory performing genetic tests.

Many laboratories that have embarked on genetic testing have found that it is not worthwhile economically and have instead referred the work to reference laboratories. This lack of fiscal viability is attributable to the high cost of what are basically labor-intensive procedures and to the inadequately developed systems to reimburse laboratories for performing the tests. The audience heard how it is possible with aggression and persistence to obtain a reasonable level of reimbursement for some of the genetic tests. Because molecular testing is the area of clinical laboratory medicine that is likely to grow most rapidly in the future, reimbursement is of primary concern.

For clinical laboratories to prosper in the future, an investment in molecular pathology will become essential. The Human Genome Project is evolving faster than envisioned, but one outcome that is causing great concern to clinical laboratory diagnosticians is the patenting of disease-associated genes (5), which may restrict the availability of such assays, while at the same time, through reduction of competition, increase the cost of the tests to patients. Conceivably, a worst-case scenario could arise if only a handful of large financially strong laboratories were able to offer such tests.

As with previous Clinical Chemistry Forums, the topic had a timely message. Genetic testing is evolving rapidly; however, not all the safeguards to protect patients are in place today, and there is a real need to orchestrate responses to the very diverse issues in such a way that society reaps the maximum benefit of this challenging new direction in clinical laboratory testing.

References