Promoting Quality Laboratory Testing for Rare Diseases: Key to Ensuring Quality Genetic Testing

May 20-21, 2004
Atlanta, GA

Executive Summary

Background

Rare diseases collectively affect an estimated 25 million, or more than 1 in 12 individuals in the United States alone. The majority of the 6,000 rare diseases known today are considered genetic conditions, making genetic testing an essential element of the diagnosis and management of these patients and their families. However, genetic testing is currently available for only a small portion of rare diseases. Additionally, for many conditions testing may be available from only one or a few laboratories worldwide, from a single foreign laboratory, or from laboratories primarily conducting research studies. Currently, there is no established process to move potential tests from the research phase to a clinical laboratory setting.

Conference

In May 2004, the Centers for Disease Control and Prevention (CDC), Emory University, the Office of Rare Diseases (ORD) of the National Institutes of Health (NIH), the American Society for Human Genetics (ASHG), the American College of Medical Genetics (ACMG), the Health Resources and Services Administration (HRSA), and the Genetic Alliance, organized a "**Promoting Quality Laboratory Testing for Rare Diseases: Keys to Ensuring Quality Genetic Testing**" conference, in Atlanta, GA. Participants of the conference included more than 50 experts from government, academic institutions, professional organizations, laboratories, industry, healthcare payers, and patient advocacy groups. The goals of the conference included: 1) review of the current rare disease testing landscape; 2) discussion of problems and concerns regarding the quality, availability, access, and resources for rare disease testing; 3) identification of the needs and barriers to quality testing; 4) exploration of potential approaches to promoting quality laboratory testing; and 5) development of specific recommendations and action items for improving availability of and access to quality laboratory testing for rare diseases.

Recommendations

The conference included plenary presentations with brief discussion, two focused discussion sessions with concurrent breakout groups, and a summary session with breakout group reports and additional input from participants. Deliberations of the breakout groups led to both broad and specific recommendations for initiatives in six major areas:

- **Education** to promote quality translation of research findings into clinical testing and to advance understanding of quality standards for patient testing. Appropriate strategies and teaching materials should be developed for the research community; institutional review boards (IRBs), providers and users

of laboratory services, healthcare payers, patients, research participants, and advocacy groups, to minimize adverse impact on access to testing.

- **Guidance, strategies, and criteria** for evaluating the clinical readiness of potential tests. Issues needing further exploration include how newly developed rare disease tests should be validated, and how analytic validity, clinical validity, and clinical utility should be established for rare disease tests.

- Reasonable and achievable **quality assurance strategies** for clinical genetic testing for rare diseases.

- Mechanisms and strategies to promote **quality data collection** during each step of test development through clinical application.

- **Partnership and networks** to improve and facilitate research translation, data sharing, clinical availability, and quality assurance.

- **Infrastructure** to provide momentum and enable development of activities needed, including facilitating the translation process, assuring the quality of testing services, and improving access to testing.

Immediate Outcomes

The conference concluded with the following immediate outcomes and next steps:

- The North American Rare Disease Laboratory Network was reinstated and expanded to consist of six reference laboratories.
- The American Society of Human Genetics and other professional organizations agreed to organize educational activities and develop guidance for rare disease genetic testing.
- The Office for Human Research Protections (OHRP) committed to providing education to IRBs regarding their role in safeguarding the release of individual test results in clinical research.
- A follow-up conference, proposed to be an “Integration Conference”, will be held in six months to convert the recommendations into projects and action items, and to develop additional recommendations for issues needing further consideration.