When asked whether health-related genetic information has important social, emotional and psychological implications for individuals and families, 100% of pediatric primary care providers responding to a survey agreed that it does. However, only half of these respondents feel competent to provide genetic- and genomic-related health care to their patients, according to information presented during the webinar, “Integrating Genetics into Primary Care: Why does it Matter?” held on March 20, 2012. The webinar, which featured Timothy Geleske, MD, FAAP, and Bruce Korf, MD, PhD, FAAP, FACMG, was part of the Time Out for Genetics webinar series hosted by the Genetics in Primary Care Institute (GPCI).

The Important Role of the Pediatric Primary Care Provider in Genetic Medicine

- The primary care provider plays an important role in genetic screening by educating families, explaining outcomes and what to expect, coordinating care with subspecialists, and providing counsel as needed.
- Pediatric primary care providers are in a unique position to consider a patient’s family health history – a valuable genetic screening tool – in its entirety.
- Due to a shortage of clinical geneticists, primary care providers are encouraged to gain a fundamental knowledge of genetic and genomic medicine to be able to answer parents’ questions about testing, diagnoses and care and to help patients obtain adequate care.
- As genetic-based medicine continues to grow, primary care providers can expect to encounter patients with genetic conditions more often.
- A family-centered medical home can help manage chronic conditions as well as acute care.

Primary Care Providers’ Perceived Competencies

A needs assessment survey conducted by the GPCI surveyed members of the American Academy of Pediatrics (AAP) Quality Improvement and Innovation Networks not only questioned respondents on their opinions about the importance of genetic medicine implications and their ability to provide genetic health care to their patients, but it also asked questions related to family health history, genetic testing and genetic medicine professional education.

- 100% of respondents indicated that taking a family health history is important, yet only 32% of respondents obtain a three-generation family history.
- 15% of respondents order genetic testing at least monthly, while 85% order less than three genetic tests a year. In addition, 60% of respondents do not feel comfortable ordering a DNA test, and 65% of respondents are uncomfortable ordering metabolic genetic testing.
- Nearly 97% of respondents indicated that the health care they provide to patients would be enhanced by professional education on genetics and genomics.
How Genetic Medicine has Changed over the Years

The prevention, diagnosis and treatment of genetic diseases have expanded over recent decades and are likely to continue to advance, leading to more genetic disorder diagnoses. Patients whose genetic testing failed to result in a diagnosis only a few years ago could probably benefit from being retested due to recent advances in testing.

• Introduced in the 1960s, the premise of newborn screening was to offer prompt interventions for genetic condition diagnoses in hopes of improving outcomes. Now, tandem mass spectrometry is used for newborn screening, which continues to vary from state to state in terms of requirements. However, efforts are being made by the American College of Medical Genetics and Genomics to standardize newborn screening across the nation.

• Chromosome analysis was one of the first methods to diagnose genetic conditions, but current testing, including array comparative genomic hybridization testing, now makes it possible to detect genetic anomalies or intellectual disabilities that were undetectable with earlier testing methods.

• The cumulative pace of disease gene discovery has increased exponentially since DNA testing was introduced to the public in more than 30 years ago. Now, thousands of genetic diseases can be detected with genetic tests.

Exome vs. Genome Sequencing

Whole genome and exome sequencing may allow patients to avoid the diagnostic odyssey of tests that examine single genes.

• Genome sequencing examines most of an individual’s DNA. It is expensive, but the cost is decreasing.

• Exome sequencing studies the part of the genome that contains genes. Most mutations so far known to be responsible for genetic conditions reside in exomes. Exome sequencing is less expensive than whole genome sequencing.

Therapy

In the past, most genetic conditions were untreatable, but this is no longer true. Therapeutics is now as much a part of genetic medicine as diagnosis. What’s more, genetics is important in the treatment of common as well as rare disorders. Pharmacogenetics is now being used to study an individual’s response to drug therapy. Pharmacogenetics may make it possible in the future to customize a patient’s drug therapy based on their genetic make-up.

About the Presenters

Dr Geleske is a general pediatrician practicing in Arlington Heights, IL, and an assistant professor in clinical pediatrics at Northwestern University Feinberg School of Medicine. He is also a member of the AAP Project Advisory Committees for GPCI, the National Center for Medical Home Implementation, the Quality Improvement Innovation Network, and Education in Quality Improvement in Pediatric Practice.

Dr Korf is a medical geneticist, director of the Heflin Center for Human Genetics, and chair of the Department of Genetics at the University of Alabama at Birmingham. He is also president of the American College of Medical Genetics and Genomics Foundation for Genomic Medicine.

About GPCI

The GPCI was established to increase primary care providers’ knowledge and skills in the provision of genetic-based services. The GPCI is a cooperative agreement between the US Department of Health and Human Services, the Health Resources & Services Administration, the Maternal & Child Health Bureau and the American Academy of Pediatrics.

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