

# Genetics Evaluation, Referrals and More— What to Do Next



The medical home should consider genetics consultation for all patients with abnormalities of growth or development, with congenital anomalies or with signs or symptoms of inborn error of metabolism. This was the topic during the webinar, “Genetics Evaluation, Referrals, and More – What to Do Next?” held on June 28, 2012. The webinar was presented by Carol L Greene, MD, FAAP, FACMG and John B Moeschler, MD, MS, FAAP, FACMG, as part of the *Time Out for Genetics* webinar series hosted by the Genetics in Primary Care Institute (GPCI).

## When a Genetics Referral is Warranted

There are certain signs and symptoms that should prompt a referral to Genetics:

- Developmental disorders: global development delays, intellectual disabilities, autism spectrum disorders and other unexplainable neuro-developmental symptoms.
- Growth disorders: either disproportionate or proportionate short stature or somatic overgrowth.
- Structural disorders: any anomalies, but especially infants or children with dysmorphic features, particularly if short stature and/or neuro-developmental issues are also present.
- Signs and symptoms of inborn error of metabolism: Abnormal newborn screen, altered consciousness, recurrent or persistent vomiting, abnormal tone, altered function of single or multiple organ systems.

Genetics referral is also warranted when family history is positive for a genetic condition for which your patient could be at risk.

## Benefits of a Genetics Consult

Parents want their concerns about their child addressed immediately and completely every time. Parents also want a provider who is family-centered, culturally competent, wise, empathetic, reliable and safe. A primary care provider can help families meet these needs by referring a patient to a genetics specialist for an evaluation when there is a suspected genetic condition or unusual presentation. In addition to obtaining an etiological diagnosis and developing a treatment plan, there are many benefits to confirming a genetic diagnosis.

### Expected Benefits from Medical Genetics

For child/patient	For families
Improved health outcomes	Understanding that comes from genetic counseling including family planning discussion
Improved health care and surveillance	Health care changes (for some)
Improved understanding of condition	Diagnostic testing for other family members, if warranted.
Improved educational planning	Understanding of condition
End unwarranted medical testing and treatments	Social support and peer networking

### Inborn Errors of Metabolism – Under-Recognized Genetic Conditions

Inborn errors of metabolism (IEM), also known as congenital metabolic diseases or inherited metabolic diseases, are quite common and often go undiagnosed. It is estimated that the frequency of IEM ranges from 1 out of 1,000 to 1 out of 100,000 people. Phenylketonuria (PKU) is probably the most well-known IEM, while one of the most common is mitochondrial disease, affecting 1 out of 1,000 individuals. Patients with severe or unusual health problems are more likely to have an IEM.

Like other genetic disorders, IEM can cause disturbance of development, growth or structure. There are some signs and symptoms that are more specifically linked to IEM that would lead the genetics specialist to focus on metabolic disorder screening, such as specific blood and urine tests. However, it is important to remember that proof of one condition does not exclude another condition, especially when it comes to genetic disorders. Patients with one genetic condition are more likely to have another genetic condition. It is especially important to consider IEM as a diagnosis because there are specific therapies to treat metabolic conditions. Symptoms of IEM can be as subtle as mood swings and limited exercise tolerance, and as obvious as coma. IEM can affect any organ individually (e.g. cardiomyopathy) or multiple organs. Sanfilippo can present as autism with a 25% recurrence risk and a progressive degenerative course.

### What Primary Care Providers can do Before the Consult

Parents are more satisfied with care when they understand the role of the specialist in the care of their child. Primary care providers can help families achieve this by explaining what they can expect before, during and after the consult with a genetics specialist. If families are prepared, then they are more satisfied with the experience. In certain circumstances, a medical home can begin basic laboratory testing prior to a referral. Consulting a genetics specialist can help primary care providers decide which tests to order.

### What to Expect from a Genetics Evaluation

During a genetics evaluation, a genetics specialist will obtain a family and clinical history from the patient and parent and perform a physical and neurological examination. Based on the findings, specific diagnostic tests, such as a chromosome microarray, Fragile X molecular testing, metabolic testing or MRI brain imaging, may be performed to diagnose a genetic condition and plan care.

### A Genetic Diagnosis – Now What?

The medical home's involvement in the care of a patient diagnosed with a genetic condition should continue after the genetics referral, evaluation and diagnosis. A team approach with care coordinated and co-managed by the medical home, genetics specialist and family – with clear definitions of responsibility by each party – can be the most effective way to achieve satisfactory outcomes for the patient. In addition, plans for education and counseling should be made. Even if testing does not result in a diagnosis, the medical home and genetics specialist can continue to work together to monitor the patient for any changes.

#### About the Presenters

Dr Moeschler is a professor of pediatrics at Geisel School of Medicine at Dartmouth and director of the Clinical Genetics Program at Dartmouth-Hitchcock Medical Center. He is also chair of the Committee on Professional Practice and Guidelines for the American College of Medical Genetics and Genomics.

Dr Greene is a professor of pediatrics, director of the Pediatric Genetics Clinic and co-director of the Adult Genetics Clinic at University of Maryland Medical Center. She is also president of the Society for Inherited Metabolic Disorders.

#### 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.

**For additional information on the GPCI, contact Natalie Mikat-Stevens, MPH, Manager, Genetics in Primary Care Institute, Division of Children with Special Needs, AAP, at 847/434-4738.**