INITIAL GENETIC COUNSELING APPOINTMENTS

Julie Jesiolowski
Barbara Wedehase

It can be beneficial to start preparing for your pediatric genetic counseling appointment a few weeks in advance. This will allow you to gather all the materials and information that may be needed.

What to bring:

- Any information about your family's health history
- Medical records for the child and other family members who may be similarly affected
- Pictures of the child as a baby
- Questions and concerns you may have
- Support person to assist in gathering information and listening
- Notebook to write down information

Pediatric genetic counseling sessions typically last one hour or more. Some parents will choose to bring toys or snacks for their children while they are waiting. The genetic counselor will ask many questions about family history and draw a three-generation pedigree. This is a paper representation of your "family tree." You will also complete an intake form with the genetic counselor, covering information all about your child's different body systems and behavior.

The geneticist will examine your child and take many different measurements. This is called a dysmorphology exam. Although it may look different than other exams completed at a doctor's offices due to how thorough it is, it is not painful and helps answer many questions.

The doctor will discuss with you any relevant testing that is available and allow you to decide if you would like to pursue these options. Based upon the medical exam and the information you provided about your child, the geneticist may request x-rays to look for a specific pattern of radiological changes seen in MPS diseases, called dysostosis multiplex. Obtaining a urine sample to screen for the presence of glycosaminoglycans (GAGs) is a first step in the diagnostic process. Most, but not all, individuals with MPS have GAG levels in their urine that are higher than those of individuals without MPS. The doctor may request a sample of blood from your child at the end of your time together if you decide on further testing. The definitive diagnostic test for an MPS disease measures enzyme activity and is performed on blood cells, or occasionally on skin cells.

The results from the blood test may take one to two weeks; your geneticist or genetic counselor will discuss how they will inform you about the results. This can be a very stressful time. It is helpful to remember that the diagnosis does not change your child but puts a name to his or her symptoms.

Time may also be spent during your appointment explaining the natural history of MPS, management, progression, treatment options, and risk of the condition in future pregnancies. If the diagnosis is confirmed, your geneticist may recommend a series of specialty evaluations

such as ophthalmology, pulmonology, ENT, cardiology and developmental testing. The genetic counselor can provide referral service information and recommend support resources.

The clinic will coordinate with you when to expect follow-up. Some children are followed yearly or every two years. Most genetic counseling sessions are covered by insurance, but check with your insurance provider for more information. In many cases, health insurance will cover the costs of genetic testing when it is recommended by a doctor.

Additional information about genetic counseling can be found on the National Society of Genetic Counselors' website, http://www.nsgc.org/.