Learning that a child has a genetic disorder, or is at risk for one, can be an overwhelming and stressful time for a parent. A genetic counselor is a trained healthcare professional that can provide knowledgeable guidance to a family not only at the time of testing or diagnosis but throughout the patient’s life, according to information presented during the webinar, “Heard About Genetic Counseling? What Does it Mean for You, Patients and Families?” held on August 30, 2012. The webinar, which featured Karin Dent, MS, LCGC, a certified and licensed genetic counselor, was part of the Time Out for Genetics webinar series hosted by the Genetics in Primary Care Institute (GPCI).

What is Genetic Counseling?
Genetic counseling is typically provided by a genetic counselor who is trained to provide counseling services specifically related to genetics and genomics. However, genetic counseling also can be offered by other health care providers, including pediatricians, family practice physicians, geneticists, obstetricians/gynecologists, oncologists, neurologists and nurses. Genetic counseling usually includes:

- Reviewing family and medical histories to determine a patient’s risk for a genetic disorder
- Teaching families about a detected risk, its pattern in the family and options for dealing with the risk
- Educating families about genetic testing, prevention, diagnosis, resources and research
- Explaining a genetic condition if one is diagnosed
- Counseling families to help them adapt to a genetic condition risk or diagnosis and to give them guidance, support, and information to make informed choices that are best for them

Different needs may be addressed at various points in a patient's life.

Who is a Genetic Counselor and What is Their Role?
Genetic counselors combine their specialized training in science, medical genetics and counseling theory with skills in risk assessment, education, interpersonal communications and counseling. They may act as patient advocates, serve as educators and resources for other health care professionals, and engage in research activities. Genetic counselors can partner with health care providers to help bridge the gap between medical care and science, interpret complex genetic terminology and information for patients, and help resolve ethical dilemmas. Approximately 77% of genetic counselors provide patient care and 15% perform non-clinical work.
The Impact of a Genetic Diagnosis
Approximately 3%-7% of the US population will be diagnosed with a genetic condition in their lifetime. In addition to this public health impact, a genetic diagnosis can personally affect patients and parents in many ways:

- It is a permanent, chronic condition, usually without a cure
- There are often long-term issues regarding independence and care
- There may be a sense of guilt or responsibility if it is a condition that runs in the family
- There is worry about age dependence and life stages
- The perception about the severity of a disorder can be different than the reality
- There are concerns about insurance coverage and employment
- There is apprehension about the possibility of stigma associated with the condition

What Can Patients Expect at a Pediatric Genetic Counseling Appointment?
It may be helpful for primary care providers to explain that patients can expect some of the following to occur at a pediatric genetic counseling appointment:

- The medical and family histories and the reason for the visit will be reviewed
- A diagnosis may be made by a medical geneticist who will perform a physical exam of the patient
- The patient’s risk to have a genetic condition will be assessed based on pedigree, genetic testing, risk of a known condition recurrence or risk of empiric recurrence
- A diagnosis will be explained and information will be provided to help families make decisions
- Psychosocial issues will be assessed and counseling will be provided as needed
- Follow-up appointments may be scheduled throughout the patient’s lifetime
- Reproductive options may be discussed when the patient is older

Will There be Psychosocial Issues?
A new diagnosis of a genetic condition can impact families psychosocially in many different ways. For example, if a genetic condition is anticipated to significantly impact a child’s life, then families may need time to grieve and adjust expectations for their child in terms of physical and cognitive development. A genetic diagnosis also may cause stress on a marriage as well as other relationships, and members of a couple may not be in the same place at the same time in coping with and understanding the diagnosis. In addition, siblings should be monitored for psychosocial issues. Other ways parents may react to a new diagnosis include:

- Grief-like reactions, such as denial, fear, sadness, loneliness or anger
- Confusion about what to tell people about the diagnosis
- Concern about the stigma associated with certain genetic conditions

Genetic counselors can help families address and work through psychosocial issues related to the diagnosis.

Genetic Testing
If a mutation causing a genetic condition is known to run in a family, testing other at-risk individuals for the condition requires testing only for the specific mutation already identified. Appropriate genetic testing can result in significant cost savings. Genetic counselors not only help to direct and coordinate appropriate testing but also play a key role in genetic testing by:

- Discussing medical and social concerns
- Reviewing test results and implications
- Explaining next steps
- Providing psychosocial support for both diagnoses and unknown results

About the Presenter
Ms Dent is a certified and licensed genetic counselor for the Medical Genetics and Genetic Counseling Clinics at Primary Children’s Medical Center, in Salt Lake City, UT. Ms Dent is also an assistant professor of pediatrics at the University of Utah, associate director of the University of Utah graduate program in genetic counseling, a member of the GPCI editorial advisory board and past president of the National Society of Genetic Counselors.

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.

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