When it comes to utilizing genetics to assess risk, diagnose, and care for a patient, primary care providers (PCPs) are not alone. Genetic counselors are specially trained to work with patients and guide them through the process of understanding, utilizing, interpreting, and moving forward from genetic testing. This information was presented during the webinar, “Genetic Counseling in Primary Care,” held January 2013. The webinar, which featured Emily Edelman, MS, CGC, was part of the Integrating Genetics into your Practice webinar series hosted by the Genetics in Primary Care Institute (GPCI).

**What is Genetic Counseling?**

Genetic counseling is the process of helping people understand and adapt to the medical, psychological, and familial implications of genetic contributions to disease. This process integrates

- **Interpretation**: examine family and medical histories to assess the chance of disease occurrence or recurrence
- **Education**: about inheritance, testing, management, prevention, resources and research to help patients understand the process and make informed decisions
- **Counseling**: about concerns, findings and choices to promote informed choices and adaptation to the risk or condition

The purpose of genetic counseling is to

- Educate patients about how heredity contributes to genetic disease in terms of the specific conditions and patterns of inheritance and his/her individual risk of developing genetic disease
- Help patients understand options for dealing with increased risk
- Provide guidance about what a diagnosis means
- Provide psychosocial support
- Aid patients in choosing a course of action that is personally appropriate

Genetic counseling may be provided by many healthcare providers, including physicians, physicians’ assistants, advanced practice nurses, nurses, and genetic counselors.
What is a Genetic Counselor?

Genetic counselors are health professionals with specialized graduate degrees and experience in the areas of basic science, medical genetics, epidemiology, and counseling theory. They apply specialized skill in risk assessment, education, and interpersonal communication and counseling to provide services to clients and their families for a diverse set of genetic and genomic indications.

They are members of a healthcare team who provide information and support to families of patients with birth defects or genetic disorders and to families who may be at risk for a variety of inherited conditions.

Many genetic counselors work in large medical centers or hospitals. Others work in genetic testing laboratories, private practices, HMOs, research, policy, advocacy, education, and more.

Genetic counselors often work with medical geneticists and perform many of the same functions, including risk assessment, creating a differential and evaluation plan, and ordering and interpreting genetic testing. However, medical geneticists provide medical evaluation whereas counselors focus on counseling and related support. Depending on their needs, families may see both types of professionals. Genetics offices can often help triage patients and schedule appointments with geneticists and/or counselors, depending on the patient's situation.

Finding a Genetic Counselor

There are many resources to help you locate a genetic counselor. Some of these include

- Medical genetics department within your institution
- National Society of Genetic Counselors (www.nsgc.org)
- American College of Medical Genetics (www.acmg.net)
- Telemedicine
  - More and more genetic counseling is being conducted by phone and may be a good option if you do not have genetics professional in your area.
- Regional Genetics Collaboratives (www.nccrcg.org)

Resources for Primary Care Pediatricians and Your Patients

There are many resources available to help PCPs provide basic genetic counseling and resources to their patients. The following sites are a good place to start:

- National Society of Genetic Counselors www.nsgc.org
- American Board of Genetic Counseling www.abgc.net
- National Coalition for Health Professional Education in Genetics (NCHPEG) www.nchpeg.org

January 2014
Genetic Counseling in Action: Two Case Scenarios

<table>
<thead>
<tr>
<th>Case Scenario: Sara</th>
<th>Case Scenario: Marcus</th>
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<tbody>
<tr>
<td>• 10-month-old healthy female presenting for well child visit</td>
<td>• 45-year-old healthy male</td>
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<tr>
<td>• 4-year-old brother has Duchenne Muscular Dystrophy (DMD)</td>
<td>• Family history of breast cancer</td>
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<tr>
<td>• Parents have questions about her risk of developing DMD and risk if they have</td>
<td>• Genetic counseling process includes:</td>
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<tr>
<td>another child</td>
<td>o Assessment of risk through comprehensive family history;</td>
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<tr>
<td>• PCP provides risk assessment and determines family is appropriate for genetic</td>
<td>o There is an increased risk of HBOC</td>
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<tr>
<td>counseling</td>
<td>o Development of a family testing plan: most cost efficient to have the aunt or</td>
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<tr>
<td>o Refers to genetics for DMD genetic counseling</td>
<td>cousin who have had breast cancer tested</td>
</tr>
<tr>
<td>o Provides anticipatory guidance regarding inheritance and recurrence risk</td>
<td>o Pre-test counseling and informed consent of aunt</td>
</tr>
<tr>
<td>• Genetic Counselor meets with family</td>
<td>o Coordination of genetic testing: confirms increased HBOC risk</td>
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<tr>
<td>o Educates family about condition</td>
<td>o Communication of cancer risks in HBOC, as well as guidelines and management</td>
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<tr>
<td>o Explains inheritance and recurrence risks</td>
<td>options - including whether and when to test Marcus' two young daughters</td>
</tr>
<tr>
<td>o Provides psychosocial support/aid in decision-making</td>
<td>o Explanation of inheritance</td>
</tr>
<tr>
<td>o Coordinates genetic testing</td>
<td>o Psychosocial support and counseling</td>
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<tr>
<td>o Provides resources</td>
<td>o Resources</td>
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<tr>
<td>o Discusses options for future pregnancies</td>
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</table>

Conclusions

- Genetic professionals have specialized skills in managing genetic risks that can benefit your patients.
- PCPs play a variety of roles in genetic counseling, including: direct referral, collaborative care, and PCP genetic evaluation and counseling.

About the Presenters
Ms Edelman is the Associate Director of Clinical & Continuing Education at The Jackson Laboratory. She is a board certified genetic counselor with a background in genetics education for health professionals, and clinical genetic counseling. In her previous position at the National Coalition for Health Professional Education in Genetics (NCHPEG), Ms Edelman directed the development of targeted education programs and clinical tools in genetics for healthcare providers. She also worked a genetic counselor at the Genomic Medicine Institute at Cleveland Clinic, where she had roles in cancer genetic counseling, cancer research, family health history, and personalized medicine initiatives.

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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