Family History (FH) is a tool used between providers and families to better understand a patient’s genetic predispositions and assess their risk of certain conditions. FH information may provide insights that will help clinicians prevent or lessen negative outcomes and provide guidance about whether to do genetic testing.

While 96% of Americans believe that FH is important to health, only around 30% have tried to collect and organize theirs. There is potential for clinicians to increase the use and benefit of FH as a formal health screening tool, according to information presented during the webinar, “Family History in Primary Care,” held in September 2013. The webinar, which featured Joann Bodurtha, MD, MPH, FAAP, FACMG, was part of the Integrating Genetics into Your Practice webinar series hosted by the Genetics in Primary Care Institute (GPCI).

**Why in an era of genetic testing do we still need to do Family History?**

FH is the starting point for understanding the genetic or environmental context for a patient’s health. It helps clinicians interpret genotypes and phenotypes, classify patient risks, and customize interventions, prevention plans or treatments. In fact, according to Khoury (2003), family history may be one of the most effective public health screening tools for some conditions, including familial cancer.

Research shows that FH provided by patients is generally accurate and reliable and that subsequent disease predictions are also generally accurate and reliable. The utility of FH depends, however, on a variety of factors including the condition and life stage of the patient.

**Collecting family history**

FH collection can be a powerful screening tool if it is collected systemically and updated on an ongoing basis. From a pediatrician’s perspective, the way to maximize this potential is to create an effective system to pay attention to, sort, and address current and potential conditions. During this planning process a pediatrician or practice should ask

- How much do I/we know about the FH of our patients?
- Where am I/are we on the genetic learning curve and what do we need to learn?
- How can I/we sort and tier FH risk?

Effective FH collection systems involve family-centered communication that maximizes patient understanding of the importance of accurate and complete FH tracking. FH is also recorded for long-term reference and updating, and they are intended to be shared with specialists, as needed, in the long-term care of the patient.
In short, clinicians need to:

There are many ways to systematically collect systematic FH, including paper-based or electronic checklists in the clinic or patient portals prior to the visit. For examples of FH tools to use in your practice, visit [www.geneticsinprimarycare.org](http://www.geneticsinprimarycare.org). In whatever format, effective tools are

- Broad and open-ended
- Checklist format
- Condition or guideline-focused
- Modular to ask questions about each individual family member
- Helpful in the construction of pedigrees
- Effective for your practice

**Family history as a genetic screening tool**

A recent Quality Improvement Project conducted by the GPCI to improve the identification of genetic disease in the primary care setting recommends collecting the following information:

- **Histories** of parents, brothers, sisters, aunts, uncles, first cousins, grandparents, nephews/nieces
- **Medical conditions** that run in the family (≥2 relatives)
- **Ethnicity** of the patient
- **Consanguinity** (ie, Are the child’s parents related by blood?)
- **Parental or patient concerns** regarding FH
- **Incomplete knowledge** (ie, adoption, estrangement, etc)
- **Any FH of the following conditions**: structural or sensory birth defects; cancer before age 50; carrier of genetic condition; clotting, bleeding or blood disorders; seizures; developmental delay, intellectual or learning disability, autism spectrum disorder, received special education services; early, sudden, unexplained, or unexpected death before age 50; heart attack (>55 in men or 65 in women); known genetic conditions; or multiple miscarriages/stillbirths

**Looking at family history collection in terms of quality improvement**

To maximize the opportunity provided by using the collecting of FH as a systemic screening tool, it is important for practices to review their process from a quality improvement standpoint. Some questions to ask include

- What are we doing?
- What is working or not working?
- Are people using the system?
- What changes are needed?
- What prompts can be created to increase family or team feedback?
One common concern is the time required to collect FH information. Some ways to increase efficiency while maintaining quality collection include

- Developing tools families can fill out at home or in the waiting room
- Using streamlined tools such as checklists
- Adding family history questions to existing tracking tools
- Planning for routine family history updates
- Developing a referral/testing process

**Using genetic information to tier patient risk**

The goal in using family history with patients is to tier their risk in order to better prevent or treat genetic conditions. For example, family history has been shown to be the strongest risk factor for croup and its recurrence. Likewise, prevention guidelines regarding spina bifida and sudden cardiac death include FH. In general, there are three stages to patient tiering.

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<tr>
<th>Assessment</th>
<th>Risk Classification</th>
<th>Intervention</th>
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<tr>
<td>Family History</td>
<td>Average</td>
<td>Standard prevention recommendations</td>
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<tr>
<td></td>
<td>Moderate /&quot;Familial&quot;</td>
<td>Personalized prevention recommendations</td>
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<td>High/Genetic</td>
<td>Referral for genetic evaluation with personalized prevention recommendations</td>
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Risk stratification involves looking at family history in terms of the incidence of conditions, including the number of people affected, their level of genetic relation to the patient, their age of onset, and whether the condition is present on one or both sides of the family. Additional factors in recognizing family risk include sudden cardiac death in a person who seemed healthy, multifocal or bilateral occurrence in paired organs and ethnic predisposition to certain genetic disorders.

Referrals for genetic testing should be considered when there is

- Significant neurological/developmental problem
- Congenital anomaly
- Growth problem
- Strong positive family history for risk stratification and recurrence
- Other indications such as abnormal skin findings, nonviral cardiomyopathy, clotting abnormalities, bilateral-multifocal malignancies, multiple miscarriages or infertility

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Strategies for improved patient/family communication

Because accurate and complete information is essential to the success of family history collection, strong patient and family communication is a must. Some tips for successful communication include:

- Define family relationships using commonly understood terms, such as “brothers” and “sisters” instead of siblings, or by identifying the relationship to the child or parent such as “mom’s sister” instead of “aunt”
- Notice clues to potentially inaccurate or missing health information, such as having no reported cases of cancer in the family
- If the patient seems doubtful of the accuracy of their knowledge, recommend they communicate with their family to obtain accurate information
- Exercise compassion with the words and phrasing you use

About the Presenter
Dr Bodurtha is the Co-Director of the New York Mid-Atlantic Collaborative on Newborn Screening and Genetic Services as well as a professor at the Johns Hopkins Institute of Genetic Medicine within the Department of Pediatrics and Oncology. At Johns Hopkins, Dr Bodurtha serves on the IRB as well as the JH-NHGRI Genetic Counseling program advisory committee while also seeing genetics patients and attending general pediatrics.

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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