Family history is an important low-tech tool that can be useful for high-tech genetic medicine, according to information presented during the webinar, “Building an Accurate Family History, Constructing a Pedigree – An Overview for Primary Care” held on April 26, 2012. The webinar, which featured Emily Chen, MD, PhD, FAAP, FACMG, and Robert Saul, MD, FAAP, FACMG, was part of the Time Out for Genetics webinar series hosted by the Genetics in Primary Care Institute (GPCI).

**Why a Family History?**

Family history is a critical part of genetic medicine because it can help to:
- Identify hereditary disorders
- Determine inheritance patterns
- Determine recurrence risks
- Identify at-risk individuals
- Identify individuals who are not at risk

Family history, which should be updated annually, can help clinicians improve, prevent or delay adverse outcomes for patients who are identified as being at high risk. Family history is not only crucial to the assessment and management of genetic conditions and related adverse outcomes, but it also serves as an adjunct to the prevention, diagnosis and treatment of most childhood diseases and disorders.

**SCREEN Mnemonic**

The SCREEN Mnemonic can be useful for remembering the important components of a family history:
- **S**: Some Concerns about diseases that run in the family
- **R**: Reproduction problems
- **E**: a history of Early disease, death or disability in the family
- **E**: Ethnicity of the patient
- **N**: Non-genetic risk factors or medical conditions that run in the family

**Correct Documentation of Family History**

To ensure that a complete, informative family history is obtained, there are key facts to document about the patient’s family:
- Correct diagnosis or disease
- Age of onset
- Cause of death
- Relationships between family members
- Whether there is consanguinity
- Ethnic background of both sides of the family

Additional information may be needed, which can be obtained at subsequent visits, by a trained office staff member or at a genetic consultation.

**Barriers to Family History**

According to the GPCI Quality Improvement Network Needs Assessment Survey, all responding clinicians indicated that “taking a family history is an important part of the assessment of an individual’s predisposition to disease.” However, there can be barriers to obtaining a complete family history, including:
- A lack of time
- Incomplete records
Building an Accurate Family History, Constructing a Pedigree—An Overview for Primary Care

- Inaccessible family members
- Incorrect or vague diagnosis
- Blame and guilt
- Multiple caretakers for the child
- Poor follow-through on questions
- Fear of discrimination and stigmatization
- Lack of physician reimbursement
- Difficulty finding family history in the patient’s medical records
- Difficulty entering and updating comprehensive information into the Electronic Medical Record (EMR)

**Red Flags**

Family history questions that result in answers using the descriptors “too” or “two,” i.e. too tall, too short, too early, too many, two tumors, two generations or two birth defects, among others, may indicate a genetic condition.

Other common “red flags” from a family history are:

- Reproductive or prenatal history issues
- Early or unexpected death
- Cognitive and/or behavior issues
- Growth and stature disorders
- Sensory organ deficits

**Pedigree**

Taking the family history a step further is the pedigree, a tool that illustrates generations of family and genetic conditions that run in a family. At minimum, a three-generation pedigree is ideal, but only 33% of clinicians who responded to the GPCI Quality Improvement Network Needs Assessment Survey reported completing a three-generation pedigree for all patients.

**How to Construct a Pedigree**

A family pedigree can be constructed by following these steps:

1. Start with the core family
2. Add aunts and uncles
3. Add cousins
4. Add grandparents and their siblings
5. Identify individuals with genetic conditions to better understand disorders in the family

---

**Pedigree**

*Step 1: Core family*

---

**Pedigree**

*Step 2: Adding aunts and uncles*
### Common Inheritance Patterns

Because the pedigree is a visual tool, it can be valuable for identifying a family inheritance pattern. The most common inheritance patterns are autosomal dominant, autosomal recessive, x-linked recessive, and multifactorial.

- **Autosomal dominant**: often seen in consecutive generations in both females and males. The presentation of the genetic disorder can vary among those family members who are affected. Male-to-male transmission implies autosomal dominant transmission.
- **Autosomal recessive**: usually not found in every generation of the family and sometimes affects only one member of an entire extended family. It can be found in both females and males.
- **X-linked recessive**: if only males in the extended family are affected by a genetic condition, or the condition is more severe in males than in females, then an x-linked recessive condition should be considered.
- **Multifactorial**: a set pattern is not evident, so multiple individuals can be affected in multiple generations. Multifactorial conditions can be caused by a combination of genetic and environmental factors.

### About the Presenters

**Dr. Chen** is a clinical geneticist at Kaiser Permanente San Francisco Medical Center. She is also co-director of the Kaiser Permanente Regional Molecular Laboratory and a clinical professor of pediatrics at the University of California at San Francisco.

**Dr. Saul** is a pediatrician, clinical geneticist, and medical director of General Pediatrics at the Children’s Hospital, Greenville Health System, Greenville, SC. He is also co-medical director of the GPCI, and chair of the American Academy of Pediatrics (AAP) Committee on Genetics.

### 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.