Building an Accurate Family History, Constructing a Pedigree
An Overview for Primary Care

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Thursday, April 26
12:00 - 12:30pm Central

Time Out for Genetics Webinar Series
Presented by the Genetics in Primary Care Institute

Learning Objectives

1. State the importance of the family history in the primary care setting for prevention, diagnosis, and management.

2. Demonstrate how to properly collect and interpret a meaningful family history using the pedigree and other tools.

3. Review the importance of engaging patients and families as active partners in the prevention and/or treatment of their condition.
Acknowledgments

Saal H, Chen E. “Family History and Pedigree Construction.” Medical Genetics in Pediatric Practice

(Scheduled for publication in early 2013)

Family History

“Low” technology tool offering “high” technology medicine!
Family History

• Not just for genetic disorders

• Important in medical management of patients and families
  – Diagnosis
  – Specific management
  – Anticipatory guidance
  – Knowledge of risk factors

Family History

• Birth defects
  – Incidence remains 3%
  – Often linked to genetics
  – Can present later in infancy, childhood, or adulthood

• Approximately 20% of infant deaths are due to genetic causes

• Approximately 12% of pediatric hospitalizations are due to genetic causes, and 71% of these pediatric hospitalizations are due to a significant genetic component
Family History

- Although 96% of Americans feel that knowing their family history is important to their health, only 1/3 have ever tried to gather and organize their families’ health histories (Morbidity and Mortality Weekly Report 53: 1044-1047, 2004)

- On the GPCI Quality Improvement Innovation Network Needs Assessment Survey administered earlier this year:
  - 100% of respondents agreed that “taking a family health history is an important part of the assessment of an individual’s predisposition to disease”
  - Only 33% of respondents gathered a minimum of a three-generation family health history for all patients

Family History – Why?

- Identify hereditary disorders
- Determine inheritance patterns
- Determine recurrence risks
- Identify at-risk individuals
- Identify individuals not at risk
Family History – Why?

• Adjunct to the prevention, diagnosis, and treatment of almost all childhood diseases
  – Rare genetic disorders (such as those detected in newborn screening)
  – Common childhood diseases (eg, asthma)
  – Common societal disorders (eg, obesity, diabetes, hypertension)

• Provides the context of genetics in primary care medicine

Importance for Clinicians

• Use of the family history allows clinicians to classify individuals into different risk categories:
  – Average risk
  – Moderate risk
  – High risk

• Early identification of individuals at highest risk can improve, delay, or prevent adverse outcomes in many cases.
Family History Tools

- Family History for Prenatal Providers, NCHPEG

- My Family Health Portrait, HHS
  https://familyhistory.hhs.gov/fhh-web/home.action

- Bright Futures Tool and Resource Kit
  http://brightfutures.aap.org/tool_and_resource-kit.html

- GPCI (to be developed)

Family History and Pedigree

- Provides a graphic reminder in the chart or EHR of the patient’s genetic relationships (the context) to other disorders in the family

- Displays the inter-relatedness of these relationships and their potential pertinence to the patient’s ongoing care

- These depictions might affect the management of the acute and/or chronic primary care for the patient
Pedigree

Step 1: Core family

Pedigree

Step 2: Adding aunts and uncles
Pedigree

Step 3: Adding cousins

Pedigree

Step 4: Adding grandparents and their siblings
• The exhaustive pedigree demonstrated here is only useful in unusual circumstances (and usually in the context of a genetics evaluation)

• Attempt to obtain a three-generation pedigree

• Variations of the pedigree shown will be useful and allow the benefits previously mentioned
Common Inheritance Patterns

- Autosomal Dominant (AD)
- Autosomal Recessive (AR)
- X-linked Recessive (XLR)
- Multifactorial

Autosomal Dominant

![Autosomal Dominant Pedigree Diagram]

- Affected
- Unaffected

I

II

III

IV
Barriers to Collecting Family History

- Lack of time for pediatrician or other primary care provider
- Incomplete records
- Inaccessible family members
- Incorrect or vague diagnosis
- Blame and guilt
- Multiple caretakers for a child
Barriers to Collecting Family History
(Part 2 of 2)

- Poor follow-through on questions related to family history
- Fear of discrimination and stigmatization
- Lack of reimbursement for family history collection for pediatricians
- Difficulty finding family history in EHR or medical chart

Asking the Right Questions
“The Rule of Two/Too”

- TOO tall
- TOO short
- TOO early
- TOO many
- TOO young
- TOO different
- TWO tumors
- TWO generations
- TWO in the family
- TWO birth defects
Family History **SCREEN** Mnemonic

- **SC** – some concerns re: diseases in family
  “Do you have any questions about diseases or conditions that run in your family?”
- **R** – reproduction problems
  “Have there been any problems with pregnancy, infertility, or birth defects in your family?”
- **E** – early disease, death, or disability
  “Have any members of your family become sick or died at an early age?”
- **E** – ethnicity
  “How would you describe your ancestry?” OR “What countries do your families originate from?”
- **N** – non-genetic factors
  “Are there any other risk factors or nonmedical conditions that run in your family?”

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Common “Red Flags” on Family History

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

- Correct diagnosis or disease
- Age of onset
- Cause of death
- Relationships between members
- Whether consanguinity is present
- Ethnic background of both sides of family
- May need additional documentation after reviewing records or inquiring further

Examples of Genetic Disorders Seen in Specific Ethnic and Racial Groups

<table>
<thead>
<tr>
<th>Race/Ethnicity</th>
<th>Genetic Disorder</th>
<th>Inheritance Pattern</th>
</tr>
</thead>
<tbody>
<tr>
<td>African American</td>
<td>Sickle cell disease, G6PD</td>
<td>AR, XLR</td>
</tr>
<tr>
<td>Amish/Mennonite</td>
<td>MSUD, EVC, CHH, GA1</td>
<td>All AR</td>
</tr>
<tr>
<td>Ashkenazi Jewish</td>
<td>TSS, Canavan, Gaucher Type 1, BRCA1/2, FD, NP</td>
<td>AR and AD (BRCA1/2)</td>
</tr>
<tr>
<td>Finnish</td>
<td>Hered nephrosis, CHH, infantile neuronal CL</td>
<td>AR</td>
</tr>
<tr>
<td>French Canadian</td>
<td>Tyrosinemia, TSS, cystinosis</td>
<td>AR</td>
</tr>
<tr>
<td>Mediterranean</td>
<td>Beta thal, G6PD, sickle cell</td>
<td>AR and XLR (G6PD)</td>
</tr>
<tr>
<td>Middle Eastern</td>
<td>Beta thal, FMF</td>
<td>AR</td>
</tr>
<tr>
<td>Puerto Rican</td>
<td>Hermansky-Pudlak syndrome</td>
<td>AR</td>
</tr>
<tr>
<td>Southeast Asian</td>
<td>Alpha and Beta thal</td>
<td>AR</td>
</tr>
<tr>
<td>Portuguese</td>
<td>MJD</td>
<td>AR</td>
</tr>
</tbody>
</table>
In Summary...

• Pediatricians and other primary care providers are front-line providers who have a key role to play in obtaining the family history

• Construction of a pedigree can help to:
  – identify hereditary conditions
  – determine recurrence risks, identify individuals at risk and not at risk
  – serve as an adjunct to the prevention, diagnosis, and treatment of almost all childhood diseases

• Once family history is obtained, it should be updated annually

• Geneticists and genetic counselors are available for questions about how to draw the family pedigree, interpret it, and explain its relevance

Questions?
Thank you for your participation!

For more information, please contact
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www.medicalhomeinfo.org/GPCI.aspx

Time Out for Genetics

Registration is now open for
“Ordering the Right Tests – Genetics in Primary Care”

Thursday, May 31
12:00 - 12:30pm Central

https://www2.gotomeeting.com/register/621262434