American Medical Association

Physicians dedicated to the health of America

Family Medical History in Disease Prevention

What is the value in obtaining a family medical history?

- An accurate family history is a well-established method to recognize genetic disorders and susceptibilities that may pose risks for future health problems. It remains one of the most powerful "genetic tests" to identify individuals at risk for inheritable disorders when laboratory tests are not available.
- The family history is an essential first step before discussing genetic testing with your patient. It can help to target services for patients with a strong family history of disease, formulate genetic testing strategies, customize preventive treatments, and identify carriers of a deleterious gene who have not yet manifested the specific disorder.
- The family history can be used to identify a single gene disorder or chromosomal abnormality that affect multiple family members. These disorders can be common (breast or colon cancer) or rare (cystic fibrosis).
- More frequently, the family history will identify families with increased susceptibility to disorders such as diabetes or hypertension. These diseases are rarely caused by a single gene mutation and are often influenced by environmental conditions and lifestyle choices.

How can knowledge of family history improve patient care?

- Early identification of families with increased risk for chronic diseases such as heart disease, diabetes, and certain cancers can often improve, delay, or even prevent adverse health outcomes to individual members.
- Risk assessment is unique for each disease and requires periodic re-evaluation based on changes in medical history and lifestyle choices.
- Personalized prevention programs for treatable disorders should be based on individual risk assessment.
- Examples of prevention and treatment options include increased surveillance, lifestyle changes, prophylactic medical measures, surgical intervention, or genetic testing.
- Encouraging patients to generate and update their family history promotes a sense of responsibility and partnership in health care management.

What are some examples of "red flags" in family history suggesting genetic condition or inherited susceptibility to a common disease?[1]

- Several closely related individuals affected with the same or related conditions. Some examples of related conditions include
  - Breast and ovarian cancer

Colon and endometrial cancer
Diabetes, heart disease, and hypertension
Thyroid cancer and colon polyps

- A common disorder with earlier age of onset than typical, especially if it occurs in multiple family members. Some examples include
  - Breast cancer: < age 45-50 years (premenopausal)
  - Colon cancer: < age 45-50 years
  - Prostate cancer: < age 45-60 years
  - Vision loss: < age 55 years
  - Hearing loss: < age 50-60 years
  - Dementia: < age 60 years
  - Heart disease: < age 40-60 years
  - Stroke: < age 60 years
- Sudden death in someone who seemed healthy.
- Individual or couple with 3 or more pregnancy losses (e.g. miscarriages, stillbirths).
- Medical problems in children of parents who are closely related (e.g. second cousins or closer).
- Within an individual, look for
  - A medical condition and dysmorphic features
  - Developmental delay with dysmorphic features and/or physical birth anomalies
  - Learning disabilities or behavioral problems
  - Unexplained movement disorders, hypotonia, ataxia
  - Unexplained seizures
  - Congenital/juvenile deafness, blindness, or cataracts
  - Disproportionate short stature or proportionate short stature with dysmorphic features
  - Unexplained infertility

What is the best way to record a family history and what types of information should be included?

- The most informative family history includes details on first, second, and third degree relatives. Organizing this information into a detailed family tree or pedigree helps to visualize how traits are clustering within families and moving through generations.
- For each family member, include such information as
  - Current Age (for deceased members record age at death)
  - Ethnicity (some genetic diseases are more common in certain ethnic groups)
  - Relevant medical conditions (see section above) and age of onset

- Record date, name of person recording pedigree, and whether information is medically documented.
- Remind your patient that the family history is a lifetime document that should be transferred to new physicians as they move through the health care system.
- Accuracy of information relating to family medical history tends to decrease with decreasing relatedness (i.e. information on immediate family members such as parents and siblings tends to be more accurate than that pertaining to uncles, aunts, and cousins).
- Remember that the personal nature of this information require questions be asked in open-ended and non-judgmental manner. Families that have medical conditions appearing in multiple relatives need to be reassured that no one is to blame when genetic conditions are passed on to future generations.

How do I go about drawing a family history tree?

- Use the pedigree sample and standard symbol legend featured in the attached pocket guide to assist in generating a family tree[2].
- Photocopy the pocket guide and distribute to patients to assist them in drawing their family tree.
- Encourage your patient to gather the necessary information and draw a draft of their own family tree that you can review with them on a subsequent visit.

How should the family history tree be interpreted?

- If a medical condition seems to run in the family, consult with a genetic professional (medical geneticist, genetic counselor, or genetic nurse) to ensure correct interpretation.
- When appropriate, refer to a genetic professional for counseling to help understand disease risk, the availability of confirmatory tests, and types of interventions.
- Remind your patient that patterns often indicate increased risk and do not necessarily predict certainty of developing a medical condition.

How do I locate a genetic professional in my area?

- Many hospitals and university medical centers have board-certified medical geneticists, certified genetic counselors (CGC), and advanced practical nurses in genetics (APNG) on staff.
- A fully searchable, international directory of genetic clinics and laboratories is available at the GeneTests Web site (**www.geneclinics.org**). This site is a publicly funded, peer-reviewed medical genetics information resource that provides current information on inherited disorders and the use of genetic testing in diagnosis, management, and genetic counseling.
- A directory of medical geneticists certified by the American Board of Medical Genetics is available on their Web site (**www.abmg.org**) and on the Web site

of the American College of Medical Genetics (**www.acmg.net**)

- An index of genetic counselors belonging to the National Society of Genetic Counselors is available on their Web site (**www.nsgc.org/resourcelink.asp**) that can be searched by state, city, counselor's name, institution, or area of specialization.

Are there any potential nonmedical concerns associated with a family history?

- The personal nature of information needed for family history can raise concerns about discriminatory practices (work or insurance), confidentiality, and changes in family dynamics.
- There is also potential for psychological, social, and economic consequences of labeling an individual at risk for disease. Within a family these might include parental guilt, knowledge of paternity, and unwanted medical disclosures.
- Specific federal legislation is being considered and certain states already have laws in place to protect patients from discrimination based on genetic information. More information can be found at the National Human Genome Research Institute Web site (**www.nhgri.nih.gov**).
- Current regulations in the Health Insurance Portability and Accountability Act (HIPAA) provide some protection from discrimination based on genetic information. For example, genetic information (e.g. family history) cannot be used by group health plans to deny or cancel coverage or to apply a preexisting condition exclusion to coverage. A preexisting condition exclusion might be applied to group health coverage, however, if genetic information contributes to diagnosis of a medical condition.
- Equal Employment Opportunity Commission (EEOC) has established rules within the American Disability Act (ADA) that protects people with nonsymptomatic genetic diseases from job discrimination.

What skills should a physician master before advising on genetic issues?[3]

- Be able to explain importance of disease prediction and prevention.
- Apply appropriate techniques for conveying difficult medical information to patient.
- Recognize importance of patient confidentiality and be aware of dilemmas imposed by confidentiality when relatives are found to be at risk.
- Appreciate implications that information on genetic background can have on person's self-image, family relationships, and social status and that reactions may differ depending on gender, age, culture, and education.
- Be aware of need for appropriate referrals to genetic and community support groups.

- Recognize your limitations and seek consultation when necessary.
- Undertake program of life-long learning.

Where can I access more information on generating a family medical history?

- Bennett RL. The Practical Guide to the Genetic Family History. New York, NY: Wiley-Liss, Inc; 1999. This book reviews general concepts and specific information on drawing a pedigree and the use of family history in assessing health risk.
- The Centers for Disease Control and Prevention has started a Family History Public Health Initiative. Useful information on this topic can be found at their Web site: **www.cdc.gov/genomics/activities/famhx.htm**.
- The March of Dimes has generated a clinical genetics program that contains a module on family history. This program is entitled Genetics & Your Practice and is available at **www.marchofdimes.com/professionals/682_3989.asp**.
- A Family History Newsletter is available at the National Coalition for Health Professional Education in Genetics (NCHPEG) Web site: **www.nchpeg.org**.
- A national awareness campaign on the importance of family history information has been initiated by the American Society for Human Genetics (**www.ashg.org**), the Genetic Alliance (**www.geneticalliance.org**), and the National Society for Genetic Counselors (**www.nsgc.org**). Instructions for drawing a family tree can be found on each of their Web sites.
- The Cincinnati Children's Hospital Medical Center's Genetics Education Program for Nurses has developed a continuing educational program entitled *Interpreting Family History Information* that is available at **www.gepn.cchmc.org**.
- Additional family history tools can be found at the Genetics & Molecular Medicine Web site of the American Medical Association: **www.ama-assn.org/go/genetics**.

References

1 Bennett RL. The Practical Guide to the Genetic Family History. New York, NY: Wiley-Liss, Inc; 1999.

2 Pedigree based on Family History Tool developed by the American Society for Human Genetics, the Genetic Alliance, and the National Society for Genetic Counselors.

3 Association of Professors of Human and Medical Genetics (APHMG)/American Society of Human Genetics (ASHG) Medical School Core Curriculum in Genetics (2001) **http://genetics.faseb.org/genetics/aphmg/aphmg1.htm.**

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Family Medical History Pocket Information Card

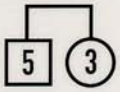
To Draw Your Family Tree

- Use this sample family tree (on reverse side of this card) as a guide to draw your own tree listing your family's medical history.
- Write your name at the top of your paper and the date you drew your family tree.
- In place of the words father, mother etc., write the names or initials of your family members.
- When possible, draw your brothers and sisters and your parents' brothers and sisters starting with the oldest to the youngest, going from left to right across the paper.
- If ages are not known, guess (e.g. 50's, late 60's). If a family member is deceased, record age and cause of death.
- Record disorders such as those listed below. Be sure and indicate the age at diagnosis and whether information was medically documented. Note these are examples and not a complete listing of all inherited disorders. Consult with your physician for more information.

Arthritis
Birth defects
Chronic respiratory disease (asthma)
Depression
Diabetes
Cancer (list specific type such as breast, colon, ovarian, prostate, etc if known)
Hearing loss
Heart disease
High cholesterol
Hypertension (high blood pressure)
Infertility
Mental retardation
Multiple miscarriages
Obesity
Osteoporosis
Physical abnormalities
Stroke
Vision loss

What if there is limited information about family members?

If you do not know names and ages of family members, but do know the number of boys and girls, you can do this:



Example: This shows that there are 5 boys and 3 girls.

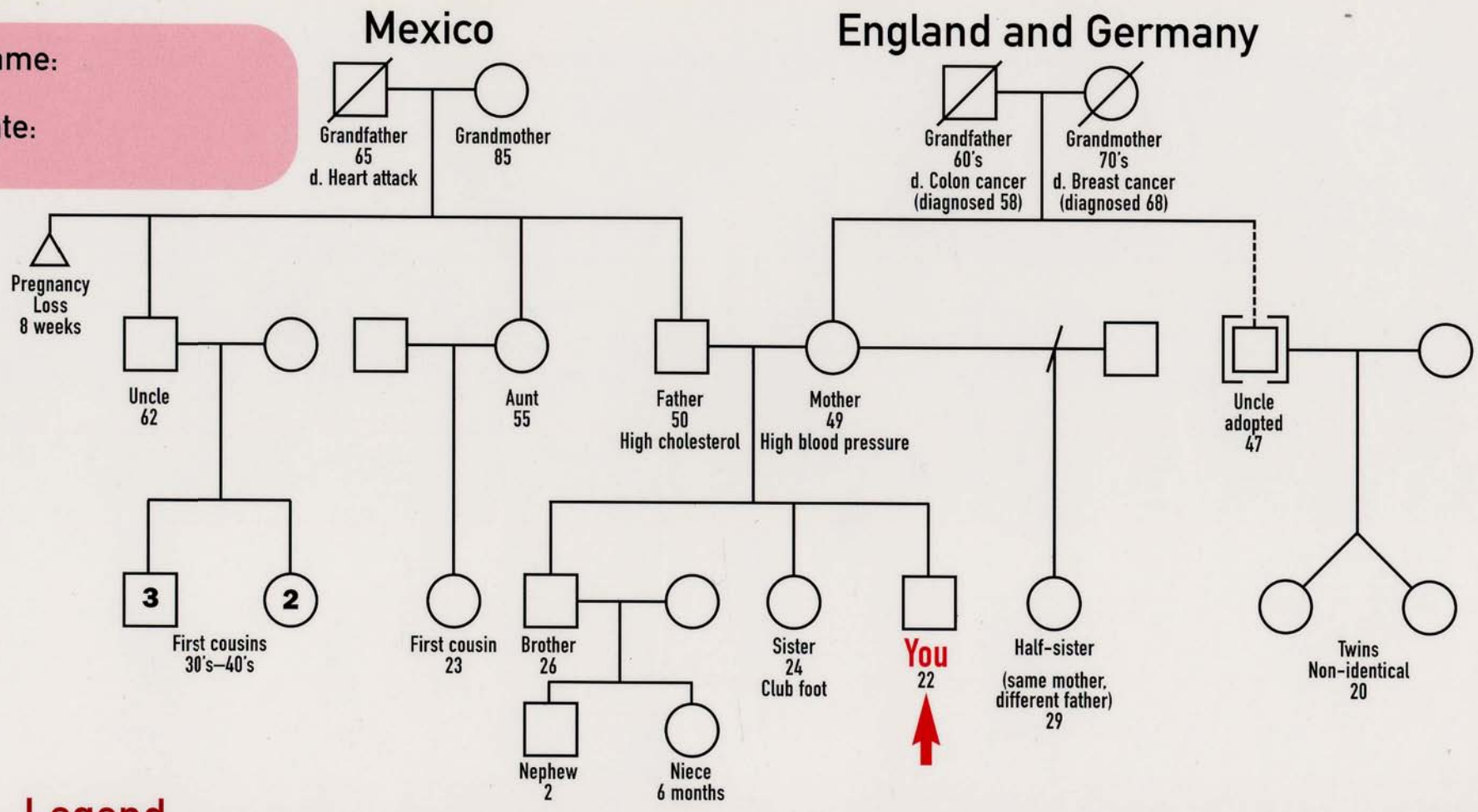
If you do not know the number of boys and girls, use diamond with number inside (if total is known) or "?".

Example: This shows that there are 8 children.

www. ama-assn.org/go/familyhistory

Name:

Date:



Legend

- Male / boy
- Female / girl
- Adopted into a family
- Diagonal line indicates death
- Pregnancy number of weeks, if known
- SB stands for stillbirth. Include number of weeks, if known.
- Diagonal line indicates divorced / separated parents