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Women's Health Care Physicians

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Committee on Genetics

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Family History as a Risk Assessment Tool

ABSTRACT: Family history plays a critical role in assessing the risk of inherited medical conditions and single gene disorders. Several methods have been established to obtain family medical histories, including the family history questionnaire or checklist and the pedigree. The screening tool selected should be tailored to the practice setting and patient population. It is recommended that all women receive a family history evaluation as a screening tool for inherited risk. Family history information should be reviewed and updated regularly, especially when there are significant changes to family history. Where appropriate, further evaluation should be considered for positive responses, with referral to genetic testing and counseling as needed.

Family history plays a critical role in assessing the risk of inherited medical conditions and single gene disorders. Certain types of cancer, such as breast cancer and colon cancer, appear more frequently in some families, as do some adverse birth outcomes. Coronary artery disease, type 2 diabetes mellitus, depression, and thrombophilias also have familial tendencies. The U.S. Surgeon General's Family History Initiative was launched in 2004. The goal of this initiative is to educate both health care providers and patients about the value of collecting a family history as a screening tool and to increase its use and effectiveness in clinical care by simplifying the collection process and analysis of the family history (1). Over the past 20 years, the Human Genome Project has afforded us a better understanding of the effect of genetic variation on health and disease. This has furthered research in identifying genotype–phenotype correlations and enhanced the ability to predict those at risk of developing inherited medical conditions. With increased awareness of the importance of using family history as a screening tool and of the value of preventive measures and increased surveillance, there is hope for improved outcomes.

Tools for Collecting the Family History

Several methods have been established to obtain family medical histories, each with its own advantages and disadvantages. A common tool used in general practice is the family history questionnaire or checklist. Having the patient complete the questionnaire at home allows extra

time for the patient to contact family members and provide more accurate information. Direct patient questioning permits clarification of medical terminology that may be unclear to the patient. Any positive responses on the questionnaire should be followed up by the health care provider to obtain more detail, including the relationship of the affected family member(s) to the patient, exact diagnosis, age of onset, and severity of disease (2).

Another family history assessment tool, commonly used by genetics professionals, is the pedigree. The health care provider may decide to complete a detailed pedigree or refer the patient to a genetics professional for further evaluation. A pedigree ideally shows at least three generations and involves the use of standardized symbols, which clearly mark individuals affected with a specific diagnosis to allow for easy identification (see Fig. 1). The pedigree may visibly assist in determining the size of the family and the mode of inheritance of a specific condition, and it may facilitate identification of members at increased risk of developing the condition (see Box 1). A pedigree should indicate the age of individuals; if deceased, the age and cause of death; and any relevant health history, illnesses, and age of onset. If any genetic testing has been performed on family members, the results should be indicated on the pedigree. The ethnic background of each grandparent should be listed as well as any known consanguinity (3). A general inquiry about the more distant relatives should be made in case there is a possible X-linked disorder or autosomal dominant disorder with reduced penetrance (4).

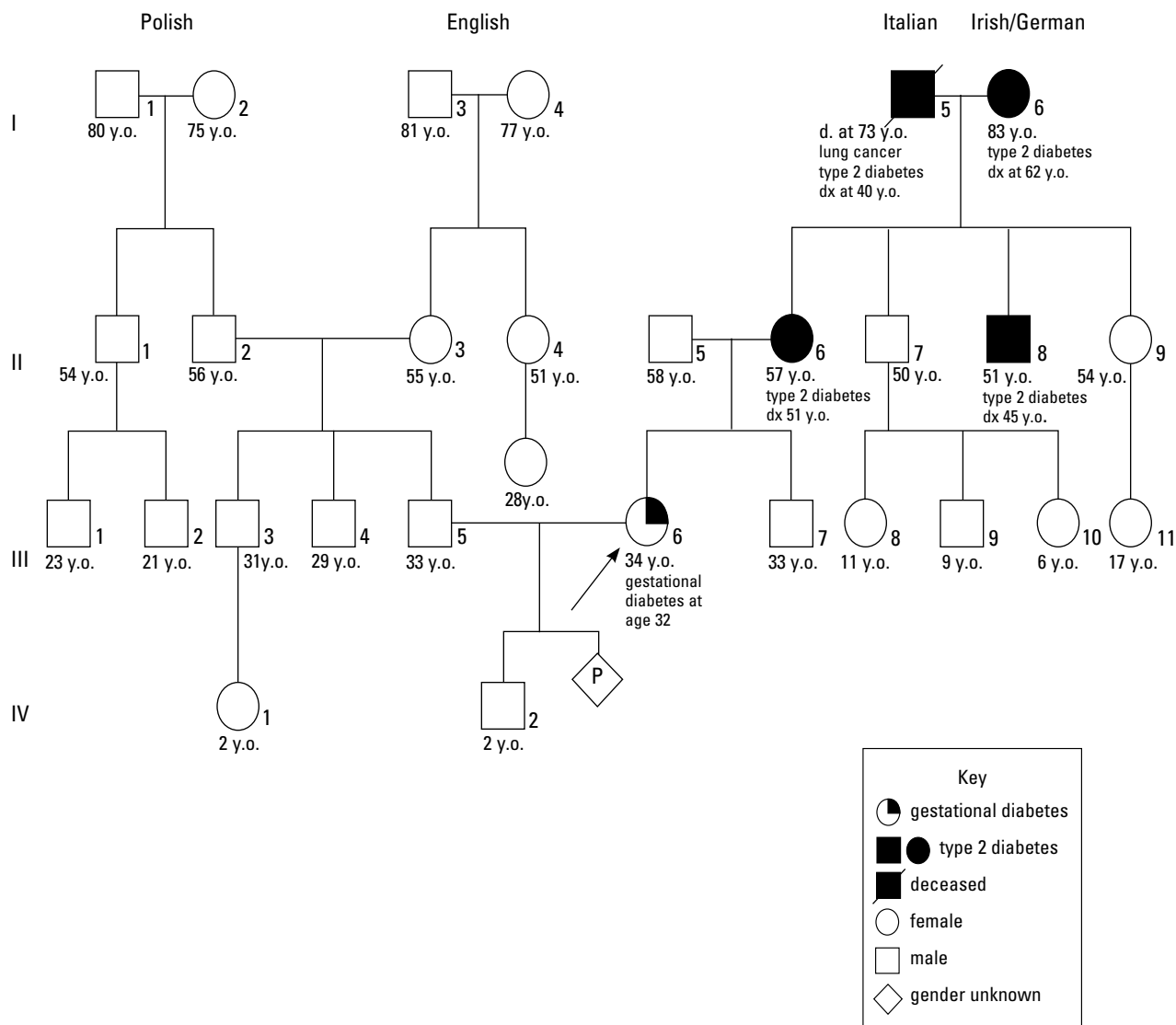


Figure 1. Example of a nonconsanguineous pedigree demonstrating a familial tendency for type 2 diabetes. Abbreviations: d, deceased; dx, diagnosed; P, pregnant; y.o., years old.

The screening tool selected should be tailored to the practice setting and patient population, taking into consideration patient education level and cultural competence. Whether the pedigree or questionnaire is used, it is important to review and update the family history periodically for new diagnoses within the family and throughout pregnancy as appropriate. A family history screening tool will allow the health care provider to stratify levels of risk (5). Moreover, the use of a family history screening tool (pedigree or questionnaire) has been shown to increase the likelihood of detecting a patient at high risk of developing an inherited medical condition by 20% compared with medical record review alone (7).

Reproductive Planning: The Preconception Period

Women often discuss their pregnancy plans with their obstetrician–gynecologist before conception. The preconception period is an ideal time to provide personalized recommendations based on family history. The preconception consultation is also an optimal time to review family history and discuss with a couple the option of undergoing carrier screening for genetic conditions. It is also an opportunity to address any medication concerns before pregnancy (eg, the importance of taking a folic acid supplement and avoiding medications such as

Box 1. Red Flags for Genetic Conditions

- Family history of a known or suspected genetic condition
- Ethnic predisposition to certain genetic disorders
- Consanguinity (blood relationship of parents)
- Multiple affected family members with the same or related disorders
- Earlier than expected age of onset of disease
- Diagnosis in less-often-affected sex
- Multifocal or bilateral occurrence of disease (often cancer) in paired organs
- Disease in the absence of risk factors or after application of preventive measures
- One or more major malformations
- Developmental delays or mental retardation
- Abnormalities in growth (growth restriction, asymmetric growth, or excessive growth)
- Recurrent pregnancy losses (two or more)

Modified from the National Coalition for Health Professional Education in Genetics (NCHPEG). Genetic Red Flags: Quick Tips for Risk Assessment. Available online at: <http://www.nchpeg.org/>. Retrieved on September 3, 2010.

angiotensin-converting enzyme inhibitors) and to ensure that medical conditions are being carefully evaluated. It is important to obtain the family and medical history of both the patient and her partner, including their ethnic backgrounds, any adverse pregnancy outcomes as a couple or with other partners, and any known causes of infertility if applicable. Positive responses will need to be followed up by performance of appropriate risk assessment, testing, and genetic counseling if needed.

Any genetic counseling and testing that can be completed before conception is beneficial to the couple, allowing a broader array of options and more time for decision making. Couples may decide not to conceive, or they may consider using a gamete donor or obtaining a preimplantation genetic diagnosis if available.

A patient who has had a past adverse pregnancy outcome or has a family history of other adverse pregnancy outcomes, such as miscarriage, preterm birth, a newborn screening test result indicating an abnormality, or birth defects, might be at increased risk of these disorders. Because both genetic and environmental factors may contribute to these outcomes, advising a patient that she is at increased risk of an adverse pregnancy outcome based on family history might motivate her to reduce her environmental risk by, for example, stopping smoking or achieving a healthy weight (8).

Common Diseases of Adult Onset and Significance of Family History

Many common adult-onset disorders demonstrate familial tendencies. However, these disorders often have complex genetic–environmental interactions for which environmental modifications can improve the outcome or delay the onset of symptoms (5). For example, diet changes, weight loss, exercise, and glucose monitoring may improve the outcome for an individual with a family history of type 2 diabetes mellitus. Similarly, individuals at risk of cardiovascular disease can benefit from environmental modifications, such as achieving normal blood pressure and cholesterol levels, and those at risk of osteoporosis can undertake calcium supplementation, weight bearing exercise, and bone density screening to improve their long-term bone health.

Some family histories show obvious evidence of cancer risk, such as a family in which there are several members with early-onset breast cancer or colon cancer. In assessing family history of cancer risk, it is important to check for evidence of cancer that might be linked to a single underlying genetic cause, such as Lynch syndrome, in which colon, endometrial, ovarian, urinary, or gastrointestinal cancer may be associated with a single familial gene mutation.

The pedigree in Figure 1 demonstrates a clear familial tendency for type 2 diabetes. The proband, or patient noted in generation III-6, is at increased risk of developing type 2 diabetes not only because of her family history of the disease but also because of her development of gestational diabetes in her previous pregnancy. This patient should be counseled not only about maintaining an appropriate diet and exercise routine to lower her risk but also about obtaining an earlier glucose screening in her current pregnancy.

Limitations

Adoption and limited family size might trigger a lower threshold in detecting family history.

Recommendations

- All women should have a family history evaluation as a screening tool for inherited risk.
- Family history information should be reviewed and updated regularly, especially when there are significant changes to family history.
- Where appropriate, further evaluation should be considered for positive responses, with referral to genetic services as needed.

Resources

The following resources are for information purposes only. Referral to these resources and web sites does not imply the endorsement of the American College of Obstetricians and Gynecologists. Further, the American

College of Obstetricians and Gynecologists does not endorse any commercial products that may be advertised or available from these organizations or on these web sites. This list is not meant to be comprehensive. The exclusion of a source or web site does not reflect the quality of that source or web site. Please note that web sites and URLs are subject to change without notice.

National Society of Genetic Counselors:

Your Family History
<http://www.nsgc.org/About/FamilyHistoryTool/tabid/226/Default.aspx>

Surgeon General's Family Health History Initiative
<http://www.hhs.gov/familyhistory>

March of Dimes: Your Family Health History
http://www.marchofdimes.com/pnhec/4439_1109.asp

American Medical Association: Family Medical History
<http://www.ama-assn.org/ama/pub/physician-resources/medical-science/genetics-molecular-medicine/family-history.shtml>

U.S. National Library of Medicine: Genetics Home Reference: Why is it important to know my family medical history?

http://ghr.nlm.nih.gov/info=inheritance/show/family_history

Cincinnati Children's Hospital Medical Center:
Genetics Education Program for Nurses (GEPN)
Independent Self-Paced Modules
<http://www.cincinnatichildrens.org/ed/clinical/gpnf/ce/skill/default.htm>

Centers for Disease Control and Prevention:
Pediatric Genetics
<http://www.cdc.gov/ncbddd/pediatricgenetics>

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