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The Genetic Family History in Primary Care

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Why take the time to draw a pedigree?

Are you too busy to draw a pedigree in your daily practice? Think again! A pedigree, or a picture of a medical family history using symbols, can be a time-saving, inexpensive diagnostic and screening tool. Many clinicians ask about family illnesses as part of a client's medical evaluation, writing out the history in textural form. However, once a clinician is used to taking a pedigree, it usually requires less time than writing out text, is easier to review later, and is often more concise and specific. For example, the narrative, "Linda's grandmother and two aunts died of breast cancer," does not indicate whether the cancer occurred in the client's maternal or paternal grandmother, nor does it state if the aunts are the sisters of Linda's mother or father. The exact relationship of these affected relatives to Linda, their ages at death, and if the breast cancer was unilateral or bilateral, can make a big difference in your assessment of Linda's risk of developing breast cancer. This same information can be recorded quickly in a pedigree.

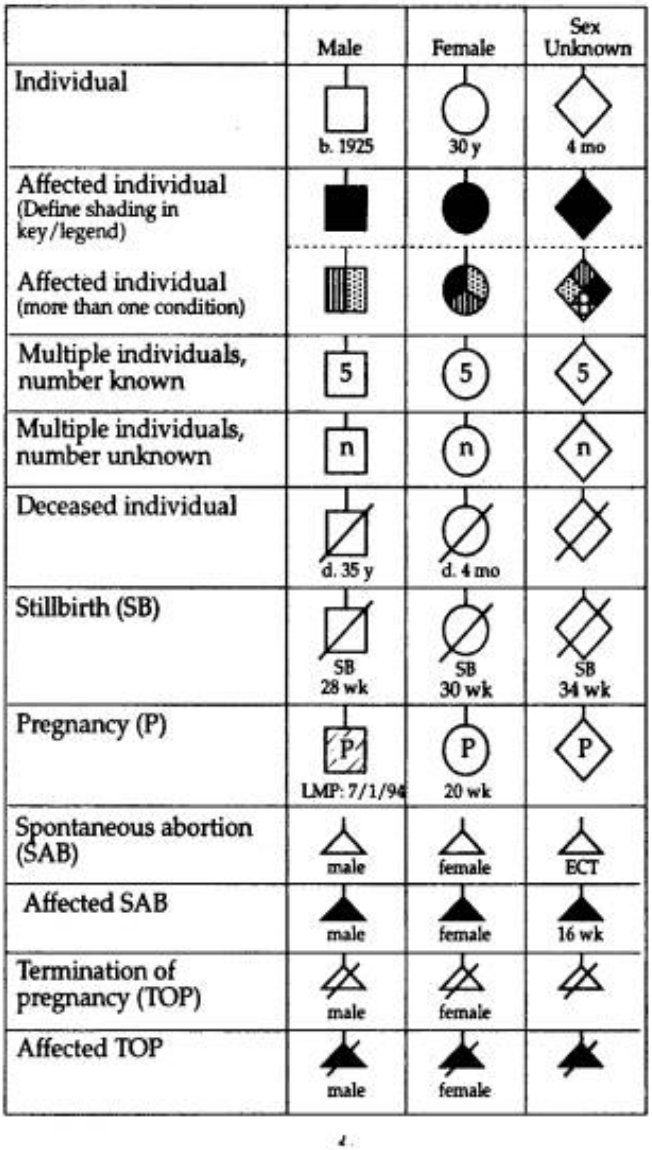
An accurate pedigree can be just as useful in determining a condition is not genetic, as it is in establishing that a condition is inherited in a family. It is also extremely useful in assessing the hereditary component of common cancers such as breast and colon cancer. If you identify several women in a family with pre-menopausal, bilateral primary breast cancer, you are more likely to be concerned that this is a familial breast cancer syndrome, than if two maternal great aunts develop unilateral breast cancer in their 70's. A pedigree can also identify genetic/medical-screening needs for an otherwise healthy individual. For example, Tay-Sachs carrier screening can be offered to someone of Ashkenazi Jewish ancestry; or cholesterol screening can be considered for someone with a strong family history of coronary artery disease.

The process of taking a pedigree also provides an excellent opportunity to establish rapport with a client. The pedigree can serve as a clear picture of family relationships (i.e. divorce, adoption, pregnancy conceived by assisted reproductive technology, deaths, etc.), so that health care professionals and social service providers can be sensitive to issues that might be of concern to the patient or client.

Pedigree symbols are Greek to me!

A pedigree's value is limited if the symbols and abbreviations cannot be interpreted by others. Recent surveys of pedigrees recorded in clinical practice and in professional publications have demonstrated wide variation in pedigree nomenclature. A survey of members of the National Society of Genetic Counselors (NSGC) showed discrepancies even in common symbols used to record a genetic family history (i.e. miscarriage, abortion, termination of pregnancy, adoption, etc.) (Bennett et al, 1993). Similar inconsistencies in pedigree construction were found in a review of current medical genetic textbooks and human genetics journals (Steinhaus et al, 1995). Historical studies have shown incongruencies in pedigree symbols throughout the twentieth century (Resta, 1993). The Pedigree Standardization Task Force (PSTF) was formed through the NSGC to develop peer reviewed recommendations for standardized human pedigree nomenclature. This project was funded by the NSGC, the Pacific Northwest Regional Genetics Group (PacNoRGG) and the Washington State Department of Health, Maternal Infant Health and Genetics. The PSTF's recommendations for standardized pedigree nomenclature were published in the March 1995 edition of the American Journal of Human Genetics (Bennett et al, 1995). A summary of common pedigree symbols and nomenclature are included in Tables 1, 2, and 3.

Table 1. Common pedigree symbols, definitions and abbreviations
(Adapted from Bennett et al., 1995)



What information should be recorded in a pedigree?

A concise pedigree provides both critical medical information and biological relationships at a glance. Table 4 lists some of the critical health information to include in a pedigree. It is also important to note on the pedigree the name and professional background of the person who recorded the information (i.e. genetic counselor, medical geneticist, physician assistant, registered nurse), and the date the information was obtained. This way, when the pedigree is reviewed by others, they will know how long ago the information was recorded, and if the pedigree should be updated. Recording the name of the historian (the person providing the history) can be helpful (i.e. grandparent, spouse, foster parent, etc.). Indicating the date or year of birth of family members makes it simple to determine a person's age at a later date, and avoids the necessity of changing the ages on the pedigree when the pedigree is reviewed at some future date.

Since a pedigree is a visual tool, it is useful to include health information on the pedigree that is specific to the diagnosis in question. For example, if a child is evaluated for short stature, parental and sibling heights may be relevant. In contrast, heights might not be recorded on a pedigree where the consultand (the person seeking genetic information), has a family history of Huntington disease.

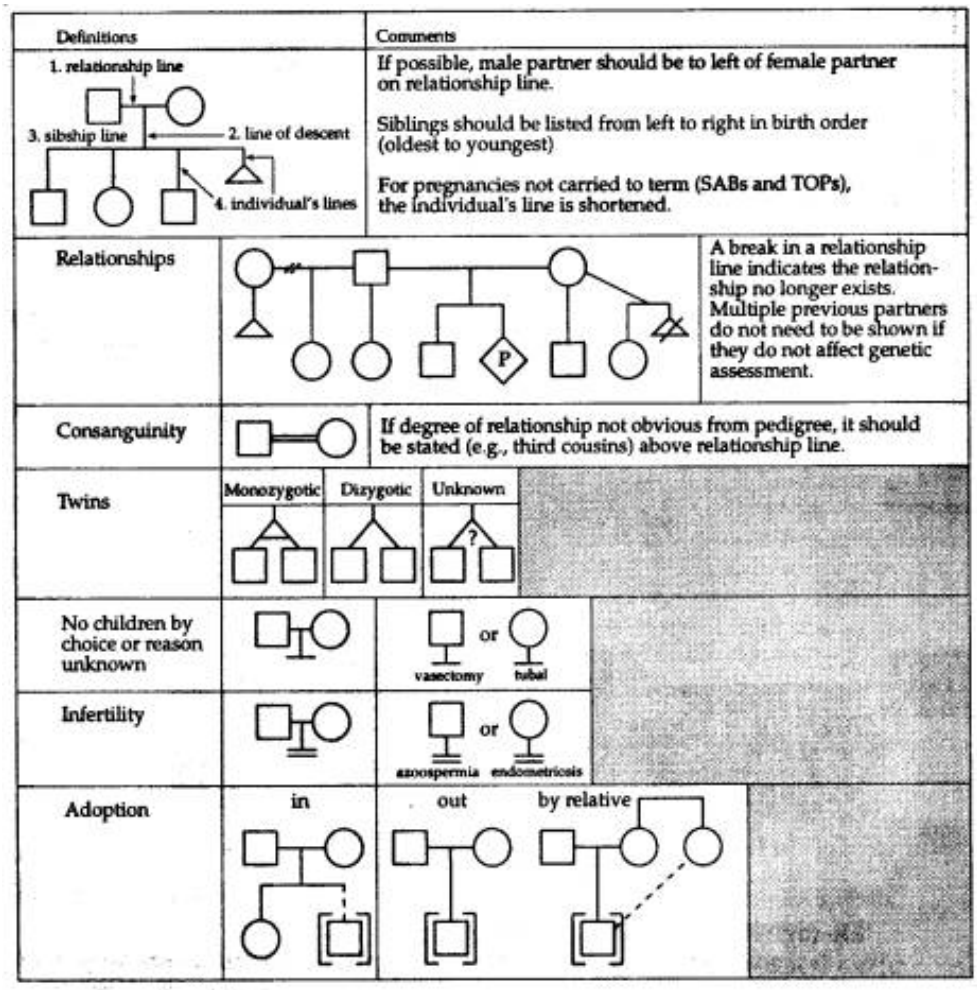
The accuracy of a diagnosis is critical to pedigree analysis. Therefore, a pedigree should indicate if diagnoses were established by record review, physical examination or patient report. It is not uncommon for a "mis-diagnosis" to be unknowingly "handed-down" in the medical records of multiple family members because a medical practitioner did not verify a diagnosis. Documenting who is unaffected in the family is equally important. Table 3 shows how to record a "documented examination" on a pedigree.

Typically, a three-generation pedigree from the consultand (the person requesting medical or genetic information) is recorded.

For example, for an infant being evaluated for dysmorphic features or a pregnancy evaluated due to abnormal ultrasound findings, the pedigree should include siblings, parents, aunts/uncles, nieces/nephews and grandparents. The evaluation of an adult for a genetic condition might require a more extensive history, since children and grandchildren might also be included. When a condition is suspected of being genetic, it is important to extend the history back as many generations as possible to include any additional affected relatives. Excluding individuals who might not contribute to the genetic evaluation (for example each spouse/partner of every relative) can help keep the pedigree concise. Inquire specifically as to whether relatives are full siblings or half-siblings, since this information can certainly affect your genetic assessment. Often in a family of "yours, mine and ours," the historian may fail to make a distinction between half, step, full and adopted siblings and simply refer to them as "my brothers and sisters".

Traditionally, ethnic background is recorded to identify healthy individuals at risk to have children with autosomal recessive conditions with a high incidence in specific populations (for example, Tay-Sachs disease among individuals of Ashkenazi Jewish ancestry) who should be offered genetic screening. Furthermore, knowing the ethnic ancestry can be helpful in a genetic evaluation, since a genetic condition with multiple mutations may have certain mutations associated more commonly with particular ethnic groups. For example, over 400 mutations have been described in the cystic fibrosis transmembrane conductance regulator (CFTR) gene. The major CF mutation, named deltaF508, accounts for about 70 percent of mutant CF chromosomes worldwide, with a higher frequency observed in Northern Europeans in comparison to Southern Europeans (for example, 30-35% in the Ashkenazi Jewish population versus 87% in Denmark) (Welsh et al, 1995).

Table 2. Pedigree line definitions
(Adapted from Bennett et al., 1995)



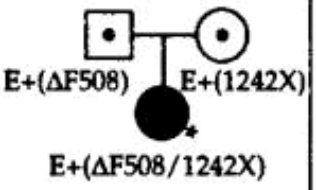
It is also important to ask about consanguinity. Actually writing on the pedigree "consanguinity denied" or "consanguinity as noted" is useful. Identifying consanguinity in a family can increase your suspicion of an autosomal recessive condition. Consanguinity will also affect recurrence risks for multifactorial conditions. Patients are often very sensitive to questions about consanguinity since, in general, there is a stigmatization in many societies about relationships between "blood relatives" (Bennett, 1987).

Ask questions about the family history in a sensitive manner.

Before you begin to take a family history, it is helpful to let your client/patient know that you will be asking many personal questions about his or her family. If your client/patient realizes that this information is essential for your medical assessment, he or she will more likely openly answer questions. Questions should be framed in an open-ended, non-judgmental manner. For example, it is better to ask, "How often do you drink alcohol" rather than "Do you drink a lot?" Acknowledging sensitive issues such as death, suicide, divorce, and pregnancy loss with a phrase such as "I'm sorry about your loss" or "That must have been a difficult time," indicates that you are caring and also leaves an opportunity for the client to give more details.

Table 3. Pedigree symbols for testing/evaluation information
(Adapted from Bennett, et al., 1995)

Instructions:
— Evaluation (E) is used to represent clinical and/or test information on the pedigree.
  a. E is to be defined in key/legend.
  b. If more than one evaluation, use subscript (E_1, E_2, E_3) and define in key. May be written side by side or below each other depending on available space.
  c. Test results should be put in parentheses or defined in key/legend.
  d. If results of exam/family study/testing not documented or unavailable, may use a question mark (e.g., E?).
— Documented evaluation (*)
  a. Asterisk is placed next to lower right edge of symbol.
  b. Use only if examined/evaluated by you or your research/clinical team or if the outside evaluation has been personally reviewed and verified.
— A symbol is shaded only when an individual is clinically symptomatic.

Definition	Symbol	Scenario	Example
1. Documented evaluation (+)		Woman with normal physical exam and negative fragile X chromosome study (normal phenotype and negative test result).	 E-
2. Obligate carrier (will not manifest disease).		Woman with normal physical exam and premutation for fragile X (normal phenotype and positive test result).	 E+(100n/35n)
3. Affected individual with positive evaluation (E+)	 E+	Individual with cystic fibrosis and positive mutation study.	 E+(ΔF508) E+(1242X) E+(ΔF508/1242X)
		18 week male fetus with abnormalities on ultrasound and a trisomy 13 karyotype.	 18 wk E+(tri 13)

Ethical issues in recording a genetic family history.

The personal nature of information recorded on a pedigree brings up several issues surrounding the protection of privacy and confidentiality. For example, recording a termination of pregnancy on a pedigree may be useful for evaluating a woman for infertility since it documents that she has been pregnant. However, her current partner may be unaware of this information. Other commonly recorded yet sensitive information includes: same-sex relationships, non-paternity, pregnancies conceived by assisted reproductive technologies, suicide, alcoholism, and HIV status. This information may be helpful in establishing or excluding a diagnosis, or in helping the health care or social service professional be aware of family issues. However, this information if released to a third party (employer, insurer), may provide the opportunity for discrimination i.e., loss of insurance or a job. Issues of privacy are also raised when a pedigree is exchanged between health care professionals or social service providers involved with different members of an extended family. The pedigree may contain information previously unknown to other relatives i.e., presymptomatic test results, pregnancy terminations, etc. Patient confidentiality should be carefully weighed against clinical and genetic relevance when choosing which information to record on a pedigree. Establishing ethical guidelines to follow in recording a genetic family history both for clinical practice and for research publications can help avoid

these problems.

TABLE 4 Factual and health information to include in a pedigree

- Age/birth date or year of birth
- Age of death
- Cause of death
- Pregnancy with gestational age (LMP) or estimated date of delivery (EDD)
- Pregnancy complications with gestational age noted (i.e. 6 wk, 34 wk)
  miscarriage (SAB)
  stillbirth (SB)
  pregnancy termination (TOP)
- Infertility vs. no children by choice
- Relevant health information (i.e. height, weight)
- Affected/unaffected status (define shading of symbol in key/legend)
- Testing status ("E" is used for evaluation on pedigree and defined in key/legend)
- Ethnic background
- Consanguinity (note degree of relationship if not implicit in pedigree)
- Date pedigree taken or updated
- Name of person who took pedigree and credentials (MD, RN, MSW, CGC)
- Key/legend

The language of the pedigree.

Any new language or dialect is awkward at first. With a little practice, the pedigree can become a valuable tool to incorporate into your daily practice. Including an accurate pedigree in the patient/client's medical record can make the task of providing quality services in the complex world of evolving medical-genetic technology more efficient and complete. Making the initial effort to record a detailed family history in a pedigree format can ultimately save time and prevent errors. It may also improve the quality of communication between health professionals when reviewing a complex medical history and ultimately improve patient care.

http://www.ama-assn.org/ama/pub/category/4901.html

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