



## CHAPTER 3 : PEDIGREE AND FAMILY HISTORY TAKING

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Health care professionals have known for a long time that common diseases—like heart disease, cancer, and diabetes—and even rare diseases—like hemophilia, cystic fibrosis, and sickle cell anemia—can run in families. If one generation of a family has high blood pressure, it is not unusual for the next generation to have similarly high blood pressure. Therefore, family history can be a powerful screening tool and has often been referred to as the best “genetic test.”

Both common diseases and rare diseases can run in families. Therefore, family history can be a powerful screening tool. Family history should be updated on each visit and patients should be made aware of its significance to their health.

### 3.1 IMPORTANCE OF FAMILY HISTORY

Family history holds important information about an individual's past and future life. Family history can be used as a diagnostic tool and help guide decisions about genetic testing for the patient and at-risk family members. If a family is affected by a disease, an accurate family history will be important to establish a pattern of transmission. In addition, a family history can even help to exclude genetic diseases, particularly for common diseases in which lifestyle and environment play strong roles. Lastly, a family history can identify potential health problems that an individual may be at increased risk for in the future. Early identification of increased risk can allow the individual and health professional to take steps to reduce risk by implementing lifestyle changes and increasing disease surveillance.



While many of the well-known genetic disorders are of childhood onset, many complex, adult-onset conditions can also run in families. For example, about 5 to 10 percent of all breast cancers are hereditary. These cancers may be caused by mutations in particular genes, such as BRCA1 or BRCA2. An individual may be at high risk of hereditary breast cancer and genetic testing should be considered if her family history includes more than one first-degree (mother, sister, or daughter) or second-degree relative (aunt, grandmother, or niece) with breast or ovarian cancer, particularly if the diagnosis of breast or ovarian cancer in those relatives was at a young age (50 or younger).

Another example of an adult-onset disease that can be inherited is Alzheimer's disease. Although most of Alzheimer's disease cases are sporadic, a small number are hereditary. Hereditary Alzheimer's disease is an extremely aggressive form of the disease and typically manifests before the age of 65. Three genes that cause early-onset Alzheimer's disease have been identified.

Notwithstanding the importance of family history to help define occurrence of a genetic disorder within a family, it should be noted that some genetic diseases are caused by spontaneous mutations, such as for single gene disorders like Duchenne muscular dystrophy and hemophilia A, as well as for most cases of Down syndrome, chromosomal deletion syndromes, and other chromosomal disorders. Therefore, a genetic disorder cannot be ruled out in the absence of a family history.



## 3.2 HOW TO TAKE A FAMILY MEDICAL HISTORY

A basic family history should include three generations. To begin taking a family history, start by asking the patient about his/her health history and then ask about siblings and parents.

Questions should include:

1. General information such as names and birthdates
2. Family's origin or racial/ethnic background
3. Health status
4. Age at death and cause of death of each family member
5. Pregnancy outcomes of the patient and genetically-related relatives

It may be easier to list all the members of the nuclear family first and then go back and ask about the health status of each one. After you have taken the family history of the patient's closest relatives, go back one generation at a time and ask about aunts, uncles, grandparents, and first cousins.

## 3.3 PEDIGREES

One way to record a family history is by drawing a family tree called a "pedigree." A pedigree represents family members and relationships using standardized symbols (see below). As patients relate information to you about their family history, a pedigree can be drawn much quicker than recording the information in writing and allows patterns of disease to emerge as the pedigree is drawn. Since the family history is continually changing, the pedigree can be easily updated on future visits. Patients should be encouraged to record information and update their family history regularly.

### PEDIGREE SYMBOLS



**Male**



**Female**



**Adopted**



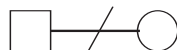
**Deceased**  
Diagonal line used to show that a person has died.



**Pregnancy loss**  
Include the number of weeks if known.



**Still birth**  
Include the number of weeks if known.



**Divorced/not together**  
Diagonal line used to show parents are divorced or not together.

### WHAT IF THERE IS LIMITED INFORMATION ABOUT FAMILY MEMBERS?

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



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

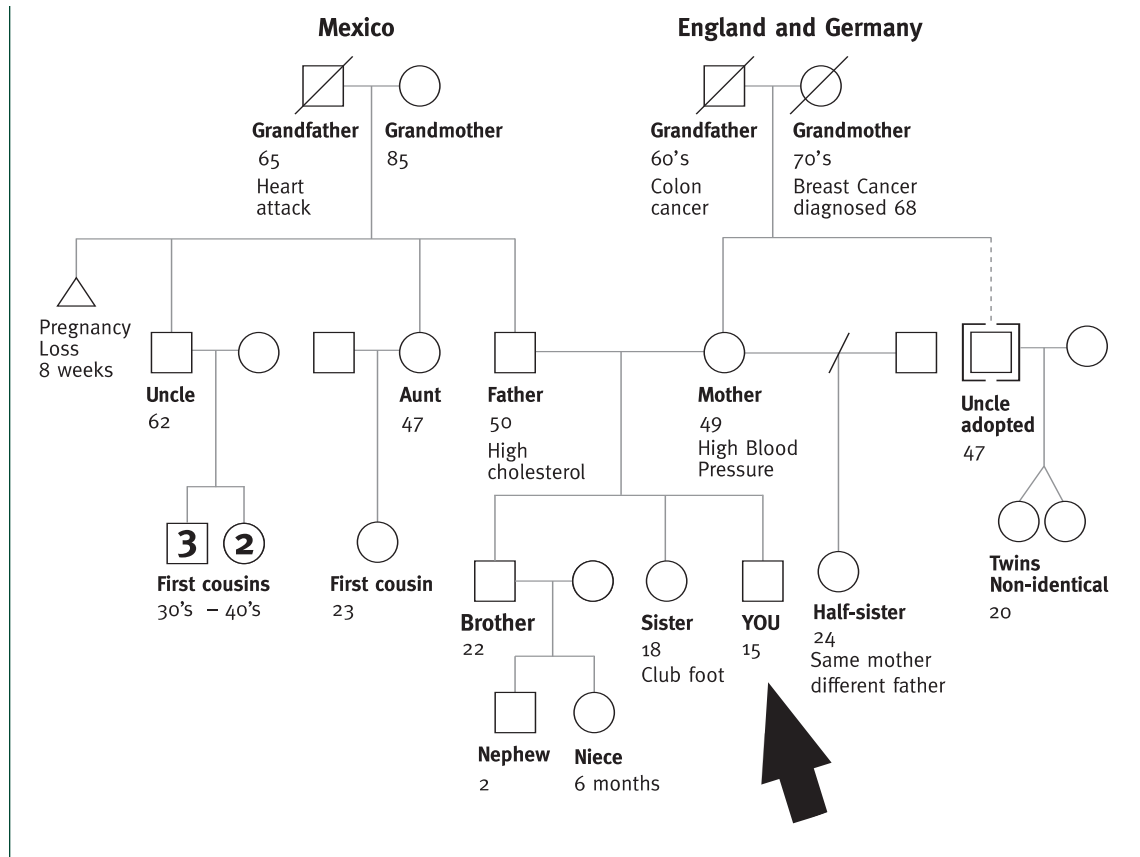
2. If you do not know the number of boys and the number of girls, use diamond with number inside it (if total is known) or "?".



**Example:** This shows that there are 8 children.

The sample pedigree below contains information such as age or date of birth (and, for all deceased family members, age at death and cause of death), major medical problems with age of onset, birth defects, learning problems and mental retardation, and vision loss/hearing loss at a young age. For family members with known medical problems, ask whether they smoke, what their diet and exercise habits are if known, and if they are overweight.

### SAMPLE PEDIGREE



### SELECTED REFERENCES

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