Healthcare professionals have long known that common diseases (e.g., heart disease, cancer, and diabetes) and rare diseases (e.g., hemophilia, cystic fibrosis, and sickle cell anemia) can run in families. For example, if one generation of a family has high blood pressure, it is not unusual for the next generation to have similarly high blood pressure. Family history can be a powerful screening tool and has often been referred to as the best “genetic test.” Family history should be updated on each visit, and patients should be made aware of its significance to their health. (See Appendix D for the Healthcare Provider Card.)
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. A family history can also identify potential health problems such as heart disease, diabetes, or cancer that an individual may be at increased risk for in the future. Early identification of increased risk may allow the individual and health professional to take steps to reduce risk by implementing lifestyle changes, introducing medical interventions, and/or increasing disease surveillance.

Although providers may be familiar with childhood-onset genetic conditions, 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. The U.S. Preventive Services Task Force (USPSTF) recommends that doctors and patients be aware of family history patterns associated with an increased risk for BRCA mutations.

Another example of an adult-onset disease that can be inherited is Alzheimer’s disease. Although most Alzheimer’s disease cases are not seen in many consecutive generations, a small number of cases 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 to-date.

Despite the importance of family history in helping define occurrence of a genetic disorder within a family, it should be noted that some genetic diseases—such as single-gene disorders like Duchenne muscular dystrophy and hemophilia A, as well as most cases of Down syndrome, chromosomal deletion syndromes, and other chromosomal disorders—are caused by spontaneous mutations. 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, healthcare professionals 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, including medical conditions and ages at diagnoses
4. Age at death and cause of death of each deceased 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, 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 can record a family history in several ways, including charts, checklists, forms, and drawings of a family tree or “pedigree.” Pedigrees are sometimes the preferred method of collecting family history information because a pedigree can be drawn more quickly than the information can be written and allows patterns of disease to emerge as it is drawn. A pedigree represents family members and relationships using standardized symbols (see Pedigree Symbols below). Because the family history continually changes, the pedigree can be updated easily on future visits. Patients should be encouraged to record information and update their family histories 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
- **Stillbirth**: ◊
  - Include the number of weeks if known
- **Divorced/not together**: □—○
  - Diagonal line used to show parents are divorced or not together

**What if Information About Family Members Is Limited?**

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 a diamond with number inside it (if total is known) or a “?”
   - Example: This shows that there are 8 children.
The sample pedigree below contains information such as age or date of birth (and age at death and cause of death for all deceased family members), major medical problems (with age of onset), birth defects, learning problems and mental retardation, and vision or hearing loss at a young age. For family members with known medical problems, ask if they smoke, what their diet and exercise habits are, and if they are overweight.

**SAMPLE PEDIGREE**

**Mexico**
- Grandfather: 65 years, Heart attack
- Grandmother: 85 years

- Pregnancy loss: 8 weeks
- Uncle: 62 years
- Aunt: 47 years
- Brother: 22 years
- Nephew: 2 years
- Niece: 6 months
- First cousin: 30s – 40s
- First cousin: 23 years

**England and Germany**
- Grandfather: 60 years, Colon cancer
- Grandmother: 70 years, Breast cancer diagnosed 68 years

- Father: 50 years, High cholesterol
- Mother: 49 years, High blood pressure
- Sister: 18 years, Club foot
- YOU: 15 years
- Half-sister: 24 years, Same mother, different father
- Uncle : Adopted 47 years
- Twins: Non-identical: 20 years

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**SELECTED REFERENCES**


My Family Health Portrait
[familyhistory.hhs.gov](http://familyhistory.hhs.gov)