

The “[Analyzing Pedigrees](#)” activity uses the 2022 guidelines for reading pedigrees from the National Society of Genetic Counselors (NSGC) ([Bennett et al. 2022](#)). The information students need to interpret the pedigrees in the activity is included in the “Student Handout.” This “Additional Background” document provides additional information from the guidelines.

Symbols

Pedigree symbols represent an individual’s **gender identity**, their internal sense of self and how they fit into the world from the perspective of gender.

- **Squares** indicate men/boys.
- **Circles** indicate women/girls.
- **Diamonds** indicate individuals whose gender is unknown or who do not identify as a particular gender.

An individual’s gender identity may differ from their **assigned sex at birth**, which is based on physical attributes (e.g., the appearance of genitalia). In these cases, the assigned sex at birth may be written underneath the symbol, as shown in Figure 1. Some abbreviations are:

- **AFAB:** assigned female sex at birth
- **AMAB:** assigned male sex at birth
- **UAAB:** unassigned any sex at birth

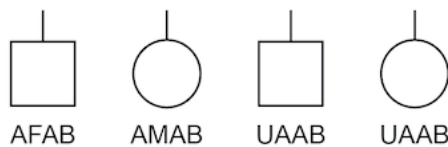


Figure 1. How pedigrees indicate assigned sex at birth if it is different from an individual’s gender identity. Assigned sex at birth is not indicated if it is the same as the individual’s gender identity. (Adapted from Bennett et al. 2022.)

If any genetic testing was conducted, genetic counselors may include additional information from the testing (e.g., whether an individual is XX, XY, or another combination of allosomes) by the pedigree symbols.

Connecting Lines

Pedigrees connect symbols, which represent individuals, with lines that represent how the individuals are related. Since pedigrees are tools for examining how genetic information is passed down from one generation to the next, many pedigrees include only genetic relatives.

The different types of lines and their meanings are shown in Figure 2.

- **Relationship line:** A horizontal line that connects two symbols and represents a romantic and/or sexual relationship between individuals. However, it does not necessarily represent a marriage or even sexual activity.
 - If these individuals are shown to have children (via a line of descent), it is assumed that the children have combined genetic material from these two individuals — unless the pedigree shows an adoption (Figure 4) or an egg or sperm donor (Figure 5).
- **Line of descent:** A vertical line that represents genetic material being transferred to offspring. The symbols at the top of the line are individuals from whom the offspring inherited genetic material. This does not

necessarily represent parenting, caregiving, legal guardianship, or any social relationship. The focus of a pedigree is primarily to show patterns of genetic inheritance.

- **Sibship line:** A horizontal line that represents siblings who received genetic material from the same individuals.

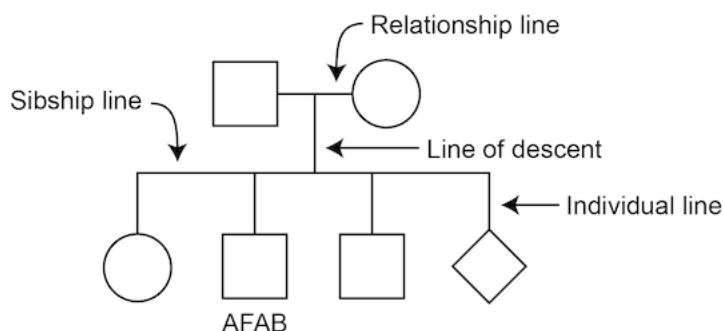


Figure 2. The meanings of lines connecting symbols in a pedigree. AFAB means assigned female sex at birth. (Adapted from Bennett et al. 2022.)

Twins are connected by a line that splits into two (Figure 3).

- **Monozygotic twins** (also called identical twins) result from the fertilization of a single egg by a single sperm. They share the same genomes and are the same assigned sex at birth.
- **Dizygotic twins** result from the fertilization of two separate eggs with two different sperm during the same pregnancy. They share about half of their genomes, just like any other siblings, and may be the same or different assigned sex at birth.

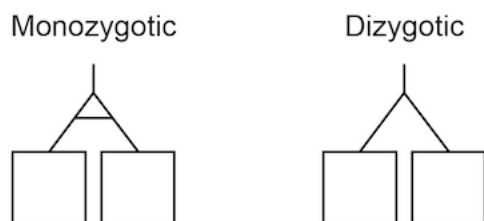


Figure 3. How pedigrees indicate individuals who are twins. (Adapted from Bennett et al. 2022.)

Additional Notation

Genetic counselors use many other types of notations in pedigrees.

For example, adopted individuals (if included) are indicated with square brackets, as shown in Figure 4. The dashed line indicates that there is no genetic relationship between individuals.

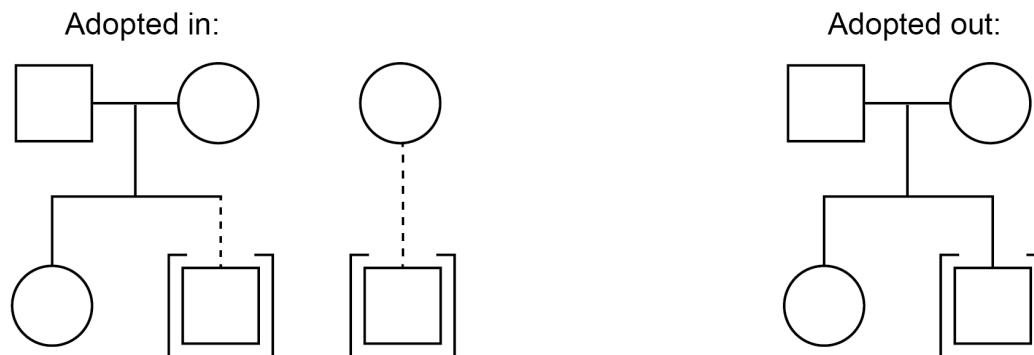


Figure 4. How pedigrees indicate individuals who were adopted. **Adopted in** means an individual was adopted by another individual or individuals in a relationship. **Adopted out** means an individual was given for adoption. (Adapted from Bennett et al. 2022.)

Pedigrees may also indicate sperm donors and egg donors. Figure 5 provides some examples, but there are many other possibilities.

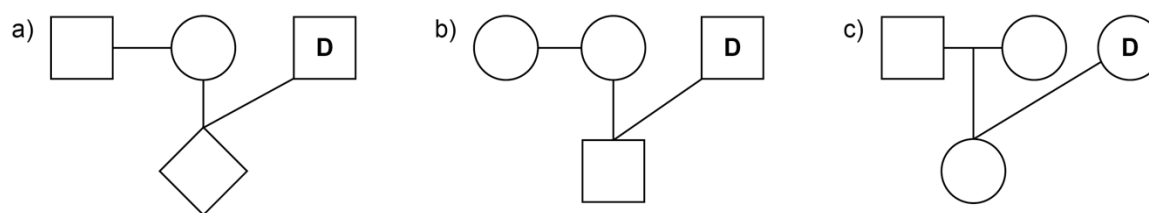


Figure 5. How pedigrees may indicate assistive reproductive technologies. **Sperm donors** are represented by a square with a D inside, and **egg donors** are indicated by a circle with a D inside. In both a) and b), the woman had a child conceived with a donor sperm. In c), the woman had a child conceived with a donor egg and a partner's sperm. (Adapted from Bennett et al. 2022.)