

## Constructing and using a Pedigree

Duchenne muscular dystrophy (DMD) is a severe, recessive, X-linked form of muscular dystrophy is characterized by rapid progression of muscle degeneration, eventually leading to loss of ambulation and death. It affects one in 3500 males, making it the most prevalent of muscular dystrophies. In general, only males are affected, though females can be carriers. Females may be affected if the father is affected and the mother is also a carrier or affected, but such a pairing would be very rare. Symptoms usually appear in male children before age 5 and may be visible in early infancy. Muscle weakness of the legs and pelvis spreads to the arms, neck, and other areas. By age 10, leg braces may be required to aid in walking but most patients are wheelchair dependent by age 12. Due to progressive deterioration of muscle, loss of movement occurs eventually leading to paralysis. The average life expectancy for patients afflicted with DMD varies from late teens to mid-twenties. Knowing how this disease is inherited through family members suspected of carrying the allele will be helpful in genetic counseling and decision making.

### Procedure

1. Consider the following information gathered from four generations of a family:
  - ▶ two people marry and have four children ( $F_1$ ) in the following order: daughter, son, daughter, daughter
  - ▶ the first daughter ( $F_1$ ) marries and has three children ( $F_2$ ): a son, a daughter, and another daughter. Her first daughter does not marry. The second daughter marries and produces a son, who develops DMD, and two daughters.
  - ▶ the son ( $F_1$ ) develops DMD and dies.
  - ▶ the second daughter ( $F_1$ ) marries, but produces no children.
  - ▶ the third daughter ( $F_1$ ) marries and has two children: a son who develops DMD and dies, and a second son. Her second son marries and has a daughter and then a son.
2. Construct a pedigree chart to represent the family history given.

### Questions

- a) If the original parents did not exhibit DMD, how could it have appeared in one of their children?
- b) Is there any indication from the pedigree chart that DMD is sex-linked? Explain.
- c) Are any of the females in  $F_2$  definitely carriers of DMD? Explain.
- d) In the family line of the first daughter from  $F_1$ , could the husband of the married daughter have been responsible for passing the DMD allele to his son? Explain.
- e) Is it possible for a sex-linked disease to pass from one generation to the next without appearing in any offspring? Explain.
- f) In the family line of the third daughter from  $F_1$ , what is the probability of her grandson ( $F_3$ ) developing DMD? Show your work.
- g) In  $F_1$ , concerned by the appearance of DMD in her brother, the second daughter and her husband decide to visit a genetic counselor before having any children. Their decision was to adopt. Suggest the reasoning they might have used to make this decision.