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

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.

1. [SP 2] Construct a pedigree chart to represent the family history given.

2. [SP 1, SP 6] Explain how one of the children of the original parents had DMD even though neither of those parents did.

3. [SP 2, SP 6] Provide evidence from the pedigree to support the claim that DMD is sex-linked.

4. [SP 2, SP 6] Provide evidence from the pedigree to support the claim that the females in F_2 are carriers of DMD.

5. [SP 2, SP 6] 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.

6. [SP 6] Provide reasoning to support or refute the claim that it is possible for a sex-linked disease to pass from one generation to the next without appearing in any offspring.

7. [SP 5] Calculate the probability of the third daughter from F_1 having a grandson (F_3) with DMD.

8. [SP 2, SP 6] 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. Provide reasoning for their decision.