

Sex-Linked Pedigrees

Purpose - To develop an understanding of sex-linked traits and how they may be traced through pedigree charts.

INTRODUCTION

A sex-linked characteristic is determined by an allele that is carried only on the X chromosome. The shorter Y chromosome does not carry the allele for the sex-linked trait. Most sex-linked traits are recessive. Since there is only one X in his genotype, XY, a male who carries a particular recessive allele on the X chromosome will have the sex-linked condition. A female who carries a recessive allele in one X chromosome will not show the condition if there is a dominant allele on her other X chromosome. She will express the recessive condition only if she inherits two recessive alleles – one from each parent. Her chances from inheriting the condition are thus greatly reduced. Red-green color blindness and hemophilia are classic examples of sex-linked traits. Most people who have these traits are men.

Hemophilia – Pedigree Analysis

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1. Study the pedigree for hemophilia found in **Figure I**. In a pedigree squares represent males, circles represent females, colored squares represent males with the trait and colored circles represent females with the condition.

Q1 - How many males in this pedigree have hemophilia?

Q2 - How many males are normal?

Q3 - How many females have hemophilia?

Q4 - How many are normal?

2. A mating or marriage is indicated by a horizontal line connecting a circle to a square. A line perpendicular to a mating line indicates an offspring. If the line ends with either a circle or square, the couple had only one child. However, if the line is connected to another horizontal line, then several children were produced, each indicated by a short vertical line connected to the horizontal line. The first child appears to the left and the last child to the right. Each level represents a generation.

Q5 - How many matings/marriages are indicated in this pedigree ?

Q6 - How many children did the first couple have ? How many boys ? How many girls ?

Q7 - How many generations are represented in this pedigree chart ?

3. The genotypes of both males are easy to determine because the male either has the trait or he doesn't. The gene for normal blood clotting is represented by **N** and the hemophilia gene is represented by **n**. Since these are only on the X chromosome the male will only have one of these letters. The hemophiliac male genotype is **nY** and the

normal male is NY . The Y represents the empty Y chromosome. **Label all the males according to their genotype.**

4. Females who have hemophilia are an easy genotype to identify. They are nn because they must have both recessive alleles. If one dominant were present the female would be normal. Females who do not show the trait for hemophilia may be homozygous normal (NN) or heterozygous (Nn). A heterozygous female is called a carrier. Examination of the offspring can often determine which genotype the parents have. If any child has hemophilia, then the mother must be heterozygous (Nn). If her son has hemophilia, he is nY and he inherited the Y chromosome from dad and the hemophilia carrying X chromosome from mom. If the daughter has hemophilia (nn), she inherited the hemophilia gene from each of her parents. This would make the genotype of the normal mother be Nn , a normal carrier. **Label all females Nn that have children with hemophilia.**

Q8 - How many women have the genotype nn in this pedigree ?

Q9 - What is the genotype of the female in the first generation ?

5. Females who have more than four sons, with none exhibiting hemophilia, are likely to have the genotype NN . If she has had four or fewer sons her genotype is less certain. In such cases her genotype is labelled $N_$. **LABEL THE REST OF THE FEMALES AS EITHER $N?$ OR NN .**

Q10 - What is the probability of parents 1 and 2 having a hemophiliac child ?

Q11 - Is there any chance of parents 3 and 4 having a hemophiliac son ? Explain.

Figure I - Hemophilia Pedigree

