

V. PROBABILITY AND GENETIC COUNSELING

In many genetic counseling situations the counselor will prepare a pedigree for the family or families seeking advice. Insofar as possible, the counselor will determine the phenotype and the genotype for each person in the pedigree. The counselor can then apply probability principles to determine the probability that a child having a particular abnormality will be produced among the offspring of a certain marriage.

To illustrate the application of these concepts, consider the pedigree shown in Figure 3.1. For the purposes of this problem your instructor will indicate which members of the pedigree express the genetic abnormality. Unless there is evidence to the contrary, assume that individuals who have married into this family do not carry the recessive gene for the trait.

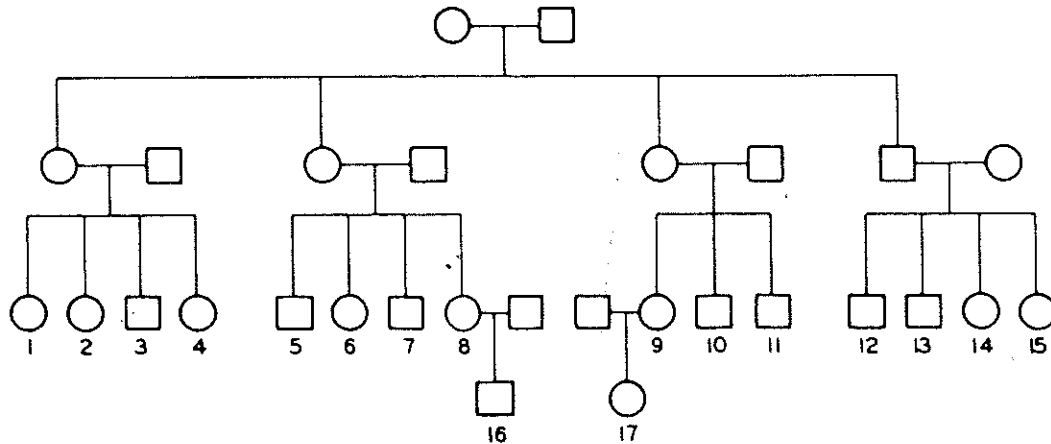


Figure 3.1. Human pedigree showing four generations. Circles represent females and squares represent males.

For a specific example, you might assume that four members of this pedigree are **albinos**: the woman in the first generation, her third daughter (the mother of individuals 9, 10, and 11), and individuals 6 and 12 in the third generation. Now assume that you are a genetic counselor and that individuals 1 and 10 in the third generation of this pedigree come to you and ask, "What is the probability that if we marry and have a family, an albino child will be born to us?"

First, the counselor must determine the probability that individuals 1 and 10 are heterozygous carriers of the recessive gene for albinism. The counselor must also consider the probability of two heterozygous carriers producing a homozygous recessive child. First of all, the mother of individual 1 **must** be heterozygous (Cc) because **her** mother is an albino (cc). One can further assume, because there is no evidence to the contrary, that the father of individual 1 is homozygous normal (CC). Thus the probability is $\frac{1}{2}$ that individual 1 is heterozygous. The probability that individual 10 is heterozygous is 1 (i.e., 100%), because the mother of individual 10 is an albino (cc). Furthermore, if two heterozygous carriers marry ($Cc \times Cc$), the probability of their having a recessive albino child (cc) is $\frac{1}{4}$.

Finally, the genetic counselor may advise individuals 1 and 10 that if they have children, the probability of their having an albino child will be $\frac{1}{2} \times 1 \times \frac{1}{4} = \frac{1}{8}$. This is the probability that the three independent events will occur simultaneously.

A slightly more confusing situation arises in determining the probability that an unaffected child of two known heterozygous parents is itself heterozygous. For example, in the pedigree shown, we know with certainty that the parents of individual 12 must both be heterozygous. The question, then, is, "What is the probability that one of their phenotypically normal children is heterozygous?" We know that in the mating $Cc \times Cc$ the expected ratio of offspring is $1CC : 2Cc : 1cc$; thus, of the expected three normal ($1CC + 2Cc$) offspring, two should be heterozygous. Thus, the probability is $\frac{2}{3}$ that the normal offspring of heterozygous parents will themselves be heterozygous.

Continue to assume that the same four individuals in the pedigree are albinos, and calculate the probability of the albino trait appearing in the offspring if the following cousins and second cousins should marry and reproduce (show your calculations):

- 1×12 _____
- 4×10 _____

- c. 6×13 _____
- d. 7×15 _____
- e. 2×5 _____
- f. 16×17 _____

REFERENCES

- Bishop, O. N. 1983. *Statistics for biology: A practical guide for the experimental biologist*, 4th ed. London: Longman Group.
- Koosis, D. J., and A. P. Coladarci. 1973. *Probability*. Self-teaching guide. New York: John Wiley & Sons.
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- Strait, P. T. 1983. *Probability and statistics with applications*. New York: Harcourt Brace Jovanovich.
- Note:* All references cited in Investigation 4 are also pertinent to this investigation.