

Human Genetics Problem Set

Problem 1: Inheritance of an X-linked recessive trait

Red-green color blindness is X-linked in humans. If a male is red-green color blind, and both parents have normal color vision, which of the male's grandparents is most likely to be red-green color blind?

- A. [maternal grandmother](#)
- B. [maternal grandfather](#)
- C. [paternal grandmother](#)
- D. [paternal grandfather](#)
- E. [either grandfather is equally likely](#)

Problem 2: Using RFLP-analysis to hunt for human genes

When RFLP analysis is used to search for a human gene, the strategy is to first locate?

- A. a known gene on the same chromosome
- B. an homozygous individual with a simple RFLP pattern
- C. a DNA sequence anywhere on the same chromosome
- D. any DNA marker co-inherited with the genetic trait of interest
- E. an exon of the disease gene

Problem 3: Human Genome Project Objectives

Which of the following is not one of the objectives of the Human Genome Project?

- A. Create a detailed genetic map of every human chromosome, with an average of 2-5% recombination frequency between markers.
- B. Obtain a detailed physical map of every human chromosome, based on overlapping recombinant DNA molecules cloned as yeast artificial chromosomes.
- C. Clone human beings.
- D. Determine the sequence of all expressed human genes by cDNA cloning and sequencing.
- E. Determine the complete DNA sequence of each human chromosome.

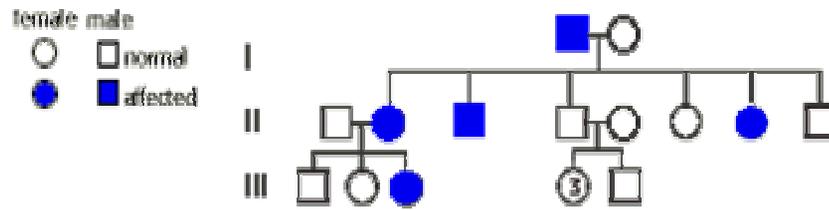
Problem 4: Inheritance pattern for Tay Sachs disease

A couple has a female child with Tay Sachs disease, and three unaffected children. Neither parent nor any of the four biological grandparents of the affected child has had this disease. The most likely genetic explanation is that Tay Sachs disease is inherited as a(n) _____ disease.

- A. [autosomal dominant](#)
- B. [autosomal recessive](#)
- C. [sex-linked recessive](#)
- D. [sex-linked dominant](#)
- E. [cannot make a reasonable guess from this information](#)

Problem 5: Interpreting a pedigree chart

A human geneticist determined the pedigree shown in the diagram with filled symbols showing the affected individuals. How is this pattern of inheritance described?



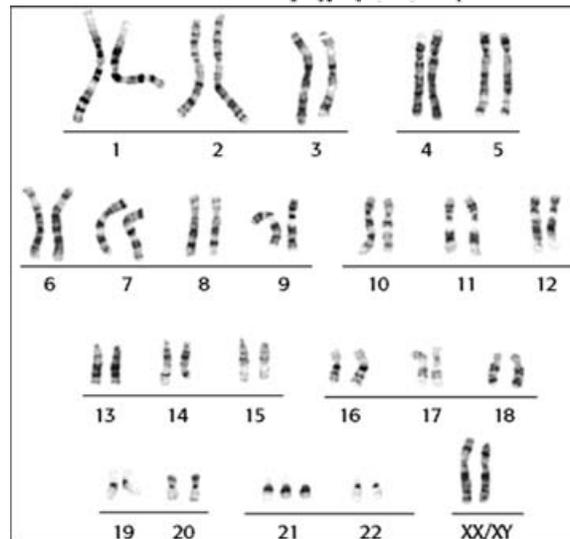
- A. [autosomal dominant](#)
- B. [autosomal recessive](#)
- C. [sex-linked recessive](#)
- D. [sex-linked dominant](#)
- E. [none of these](#)

Problem 6: Diagnosis of aneuploidy

Diagnosis of chromosome aneuploidy of unborn children is normally done by a combination of amniocentesis, cell culture, and _____.

- A. [enzyme assay.](#)
- B. [RFLP analysis.](#)
- C. [pedigree analysis.](#)
- D. [karyotyping.](#)
- E. [somatic cell fusion.](#)

Human Female Karyotype (47, XX, +21)



Problem 7: Down's Syndrome

Which statement about Down's syndrome is false?

- A. The frequency increases dramatically in mothers over the age of 40.
- B. The cause is a non-disjunction when chromosomes do not separate during the first meiotic division.
- C. Affected individuals have an extra autosome.
- D. The long time lag between onset of meiosis in ovarian tissue (during fetal development) and its completion (at ovulation) is most likely the reason for increased incidence in older mothers.
- E. None, all statements are true.

Problem 8: X-linked traits

A human male carrying an allele for a trait on the X chromosome is:

- A. [heterozygous](#)
- B. [homozygous](#)
- C. [hemizygous](#)
- D. [monozygous](#)
- E. [holozygous](#)

Problem 9: Sex determination in humans

Why would you predict that half of the human babies born will be males and half will be females?

- A. Because of the segregation of the X and Y chromosomes during male meiosis.
- B. Because of the segregation of the X chromosomes during female meiosis.
- C. Because all eggs contain an X chromosome.
- D. Because, on average, one-half of all eggs produce females.
- E. Because of the formation of the Barr body early during embryonic development.

Problem 10: Predicting inheritance pattern from pedigree analysis

A man who had purple ears came to the attention of a human geneticist. The human geneticist did a pedigree analysis and made the following observations:

In this family, purple ears proved to be an inherited trait due to a single genetic locus. The man's mother and one sister also had purple ears, but his father, his brother, and two other sisters had normal ears. The man and his normal eared wife had seven children, including four boys and three girls. Two girls and two boys had purple ears. The purple-ear trait is most probably:

- A. [autosomal, dominant](#)
- B. [autosomal, recessive](#)
- C. [sex-linked, dominant](#)
- D. [sex-linked, recessive](#)
- E. [cannot be determined from this information](#)

Problem 11: Who is NOT the father of my grandson?

I am wondering how to find out blood type of my grandson...my daughter is A+.. my grandson is type B+, we do not know the types of the two gentlemen in question...my question is what are the types that the fathers would have to be in order for him to be a B+?

- A. B or O
- B. A, B, AB or O
- C. AB or B
- D. A or B
- E. A, B, or AB