

## Practice problems (with answers)

This is the degree of difficulty of the questions that will be on the test. This is not a practice test because I did not consider how long it would take to finish these problems. It also does not have a matching section, which I will include on the test.

1. DNA polymorphisms on the Y chromosome and on mtDNA have been used extensively to study the history in humans. Describe some of the advantages that these chromosomes have over autosomes for these types of studies.

1. **No recombination**
2. **paternal or maternal inheritance**
3. **high copy number for mtDNA**

2. Androgen insensitivity is an X-linked trait that results from the inability to respond to testosterone and dihydroxytestosterone. Describe the karyotypic sex (which sex chromosomes they have) and the sexual phenotypes of the gonads and external genitalia of individuals with this trait.

### **XY karyotype**

**Gonad male**

**External genitalia female**

3. Two human populations have been isolated on islands since their ancestors first arrived. The mtDNAs of the people on one of the islands is more varied in sequence than the other. From this information which island would you predict was populated earlier and why?

**The more varied population is older because the mtDNA has had more time to accumulate mutations.**

4. In the 18<sup>th</sup> century, a young boy suffered from a skin condition that included thickening of the skin and the formation of loose spines that were periodically sloughed off. This “porcupine man” married and had six sons, all of whom had the same condition. He also had several daughters, all of whom were unaffected. What might you theorize about the location of the abnormal gene?

**It could be on the Y chromosome, because like the Y it is transmitted from fathers to sons.**

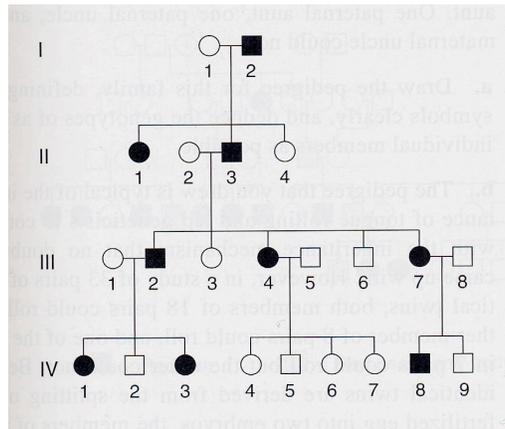
5. Mrs. Smith (40 years old) and her husband have an amniocentesis for advanced maternal age. They already have four healthy children. They receive results indicating a 47,XXY karyotype. What is the phenotypic sex of the fetus?

How many Barr bodies will be found in each somatic cell?

**Answer is male.**

**Number of Barr bodies is 1.**

5. Below is a pedigree of a rare human skin disease.



Inheritance of the disease by the II-3 male from his father rules out what type of inheritance. Explain your reasoning.

**X-linked inheritance because he received the disease from his father, but his X chromosome from his mother.**

Who in the pedigree has the same Y chromosome as the II-3 male? Give all correct answers.

**I-2, III-2, III-6, IV-2**

Who in the pedigree has the same mtDNA as the II-3 male? Give all correct answers.

**I-1, II-1, II-4**

6. 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?

**The maternal grandparents**

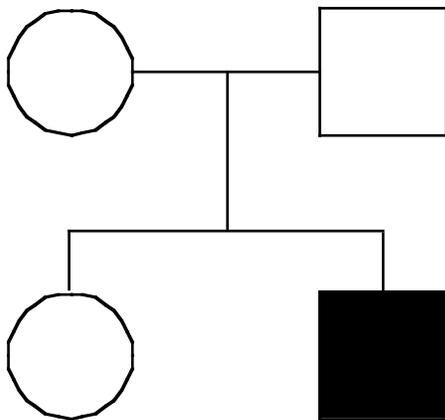
7. A couple comes to a genetic counselor concerned about their chances of having a baby with Tay Sachs disease. The husband had a sibling die of the disease, which is inherited as an autosomal recessive trait. What are the chances that he is a carrier? (This is a little tricky.)

**2/3 His parents' children had a 25% of having an affected child, a 50% chance of having a carrier and a 25% non carrier. Since the husband didn't die of Tay Sachs, he is in one of the latter two categories, which should be produced in a 2:1 ratio.**

PCR analysis showed that both the husband and wife are carriers for Tay Sachs disease. They decide to have IVF and blastomere testing. Describe what will happen.

**The wife will be treated with hormones to cause her to ovulate. The eggs will be fertilized with the husband's sperm in a culture dish. Blastomeres from the embryos will be removed and tested by PCR for the Tay Sachs mutation carried by the parents. Embryos that are not homozygous for the disease will be implanted in the wife.**

8. Below is a pedigree for a neurological disease. The son is affected (solid square)



If the disease is caused by a mutation in a gene on the X chromosome, is the mutation recessive or dominant? Assume this for the remaining questions.

**Recessive**

From which parent did the son inherit the disease gene?

**Mother**

The identity of the disease gene is known. By PCR you amplify part of the disease gene from the affected son and get a 300 base pair PCR product. From the father's DNA, you get a 330 base pair product; from the mother

you get PCR products of 300 and 330 base pairs; and from the sister, you get PCR products of 300 and 330 base pairs. Is the sister a carrier? Explain your reasoning.

**Yes. The disease gene is represented by the 330 base pair DNA, which the sister has.**

The mother becomes pregnant. Amniocentesis shows that the fetus has a Y chromosome. The parents want to know whether the child will be affected. You conduct a PCR analysis of the disease gene of cells from the fetus? What are the possible results, and what will you tell the parents?

**If the child has the 300 base pair product, he will not be affected. If he has the 330 base pair product, he will be affected.**

9. A list of propositions appears below. There is something wrong with each one. Briefly describe what is wrong.

The fact that mitochondrial DNA from African populations is more diverse than mitochondrial DNA from other populations suggests that African populations arose more recently than the less diverse groups.

**The higher diversity indicates that the African populations are earlier.**

The defect in Phenylketonuria (PKU) is caused by a lack of the enzyme phenylalanine hydroxylase, which converts the amino acid phenylalanine into the amino acid tyrosine. If untreated, PKU infants will develop severe mental retardation, but if the infants are fed tyrosine they develop normally.

**Infants with PKU are fed less phenylalanine.**

The nucleus is an organelle that produces energy for the cell.

**The nucleus contains chromosomes--mitochondria produce the cell's energy.**

Women normally have 22 autosomes and one X chromosome.

**Women have 22 autosomes and two X chromosomes.**

Because we can infer from their paternal (male line) descendants that President Thomas Jefferson's maternal uncle and Eston Hemings Jefferson had the same Y chromosome, we can conclude that President Jefferson could have fathered Sally Heming's son Eston.

**We can't conclude anything about President Thomas Jefferson's Y chromosome from the Y chromosome of his maternal uncle.**