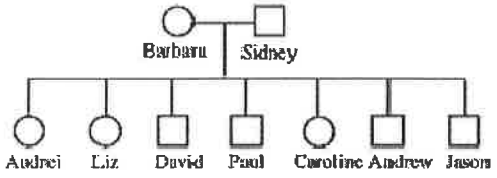
Color Blindness Problem Set

Sex-Linked Inheritance Patterns

This problem set is based on a question received from a woman named Audrei. Audrei is red-green color blind and so are other members of her family. She wanted to know if we could help her understand how she inherited her color blindness.

Audrei's family

There are 7 children in Audrei's family, three girls and four boys. Two of the girls, Audrei and Liz, are red-green color blind. Caroline has normal color vision. Only two of the boys have been tested. Paul is color blind and David has normal color perception. Andrew and Jason, who have not been tested, may or may not have normal color perception.



Barbara, the mother of the seven children, has normal color vision, but Sidney, the father, has the red-green color perception defect. Audrei also has a half brother Stephan. Audrei and Stephan have the same mother, but a different father. Stephan is also red green color blind.

Red-green color blindness

Red-green color blindness is an X-linked, recessive trait. In this problem set we will establish the pedigree of Audrei's family and see how the color perception defect is passed on from one generation to the next, but first let's look at a brief introduction to sex-linked inheritance. For more complete information about sex-linked inheritance review the Sex-linked Inheritance Problem Set in the Mendelian Genetics section.

Brief Introduction to Sex-Linked Inheritance

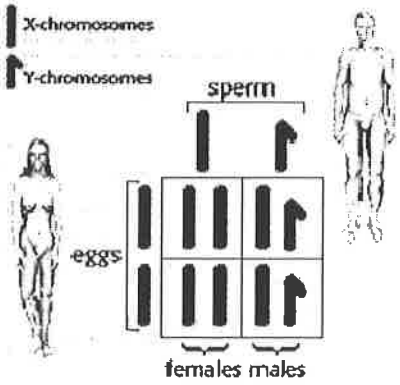
Fusion of egg and sperm

Each normal human egg carries a single X chromosome while each normal human sperm carries either an X or a Y chromosome.

Eggs fertilized by sperm with an X chromosome develop into females.

Eggs fertilized by sperm with a Y chromosome become males.

For more information about working with a Punnett square see the Tutorial of Problem 1 in the Monohybrid Cross section of Mendelian Genetics.



Red-green color blindness

The most common type of red-green color perception defect is due to a mutation on the X-chromosome (i.e. a red-green color blind allele). X-linked red-color blindness is a recessive trait. Females heterozygous for this

trait have normal vision. The color perception defect manifests itself in females only when it is inherited from both parents. By contrast, males inherit their single X-chromosome from their mothers and become red green color blind if this X-chromosome has the color perception defect. Different genotypes for this trait are illustrated below.

The dominant X chromosome is represented as X^R.
The recessive chromosome is represented as X^r.

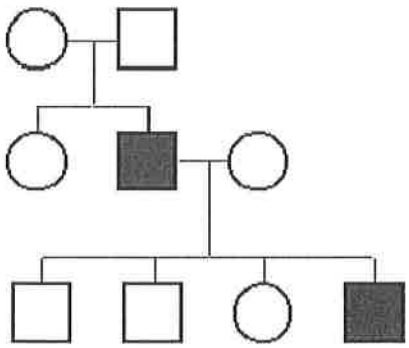
Since males have only one X-chromosome, if this chromosome has the red-green color blind allele, the males will have the color perception defect.

Females have 2 X-chromosomes. Both X-chromosomes must carry the mutant allele for the females to be color blind. Red-green color blind females are homozygous for the recessive allele.

Females with one mutant allele and one normal allele are heterozygous "carriers". They are not color blind, but they can pass the color blindness to their children.

Pedigree

A pedigree is a diagram of family relationships that uses symbols to represent people and lines to represent genetic relationships. These diagrams make it easier to visualize relationships within families, particularly large extended families. Pedigrees are often used to determine the mode of inheritance (dominant, recessive, etc.) of genetic diseases. A sample pedigree is below.

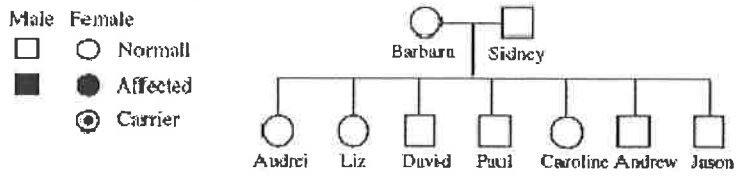


In a pedigree, squares represent males and circles represent females. Horizontal lines connecting a male and female represent mating. Vertical lines extending downward from a couple represent their children. Subsequent generations are therefore written underneath the parental generations and the oldest individuals are found at the top of the pedigree.

If the purpose of a pedigree is to analyze the pattern of inheritance of a particular trait, it is customary to shade in the symbol of all individuals that possess this trait.

In the pedigree above, the grandparents had two children, a son and a daughter. The son had the trait in question. One of his four children also had the trait.

The following pedigree will be used throughout the problem set to record the genetic makeup of each individual of Audrei's family.



Color Blindness Problem Set

Problem 1: Audrei's genotype

Audrei is the family member who contacted us. She and her father Sydney are color blind, but her mother, Barbara, has normal vision. What is Audrei's genotype?

Problem 2: Sidney's genotype

Now that we determined that Audrei is homozygous recessive for the red-green color blind allele. What is her father's genotype?

Problem 3: Barbara's genotype

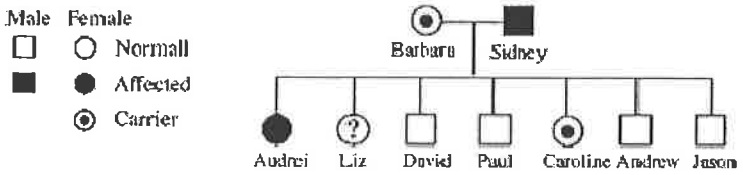
We have determined that Audrei is homozygous recessive for the red-green color blind allele and that her father is hemizygous with a recessive allele. What is the mother's genotype? Remember that Audrei's mother does not have a color perception defect.

Problem 4: Genotype of Audrei's sister Caroline

What is the genotype of Audrei's sister Caroline, who has normal vision?

Problem 5: Pedigree representation for Liz

In her message, Audrei also tells us that her sister Liz and brother Paul are also color blind, but that her brother David has normal vision. How will Liz be represented on the family pedigree?



Problem 6: Pedigree representation for Paul

Paul, like Liz, is color blind. How will he be represented in the family pedigree?

Problem 7: Andrew and Jason

Andrew and Jason have not been tested. What is the possibility that they will have the color perception defect?

Problem 8: Grandparents

We don't know if any of Audrei's grandparents were red-green color blind, but we can make some educated guesses. Is it possible that all of Audrei's grandparents have normal vision?

Problem 9: Stephan's Father

Would we predict that Stephan's father is color blind?

Problem 10: Audrei's sons

What is the probability that Audrei's sons will be color blind?

Problem 11: Audrei's daughters

What is the probability that Audrei's daughters will be color blind?

Sex-Linked Inheritance Problem Set

1. Crossing a white-eyed female and red-eyed male fly

In a cross between a white-eyed female fruit fly and red-eyed male, what percent of the female offspring will have white eyes? (White eyes are X-linked, recessive)

2. Test cross of a red-eyed female fly

A female Drosophila of unknown genotype was crossed with a white-eyed male fly, of genotype $X^w Y$(w = white eye allele is recessive, w+= red-eye allele is dominant.) Half of the male and half of the female offspring were red-eyed, and half of the male and half of the female offspring were white-eyed. What was the genotype of the female fly?

3. Predicting the offspring of a homozygous red-eyed female fly

In a cross between a pure bred, red-eyed female fruit fly and a white-eyed male, what percent of the male offspring will have white eyes? (white eyes are X-linked, recessive)

4. Predicting genotype when phenotype is known

What is the genotype of a red-eyed, yellow-bodied female fruit fly who is homozygous for the eye color allele?
Red eyes (w+) and tan bodies (y+) are the dominant alleles. (Both traits are X chromosome linked).

5. Another white-eyed female x red-eyed male fly cross

A white-eyed female fruit fly is crossed with a red-eyed male. Red eyes are dominant, and X-linked. What are the expected phenotypes of the offspring?

6. Hemophilia in humans

Hemophilia in humans is due to an X-chromosome mutation. What will be the results of mating between a normal (non-carrier) female and a hemophilac male?

7. Red-green color blindness in humans

A human female "carrier" who is heterozygous for the recessive, sex-linked trait causing red-green color blindness (or alternatively, hemophilia), marries a normal male. What proportion of their male progeny will have red-green color blindness (or alternatively, will be hemophiliac)?

8. Tracing the inheritance of the human Y chromosome

Women have sex chromosomes of **XX**, and men have sex chromosomes of **XY**. Which of a man's grandparents could not be the source of any of the genes on his **Y**-chromosome?

9. Tracing the inheritance of the human X chromosome

Women have sex chromosomes of **XX**, and men have sex chromosomes of **XY**. Which of a women's grandparents could not be the source of any of the genes on either of her **X**-chromosomes?

10. Offspring of human females who are carriers for X-linked traits

A human female "carrier" who is heterozygous for the recessive, sex-linked trait red color blindness, marries a normal male. What proportion of their female progeny (offspring) will show the trait?