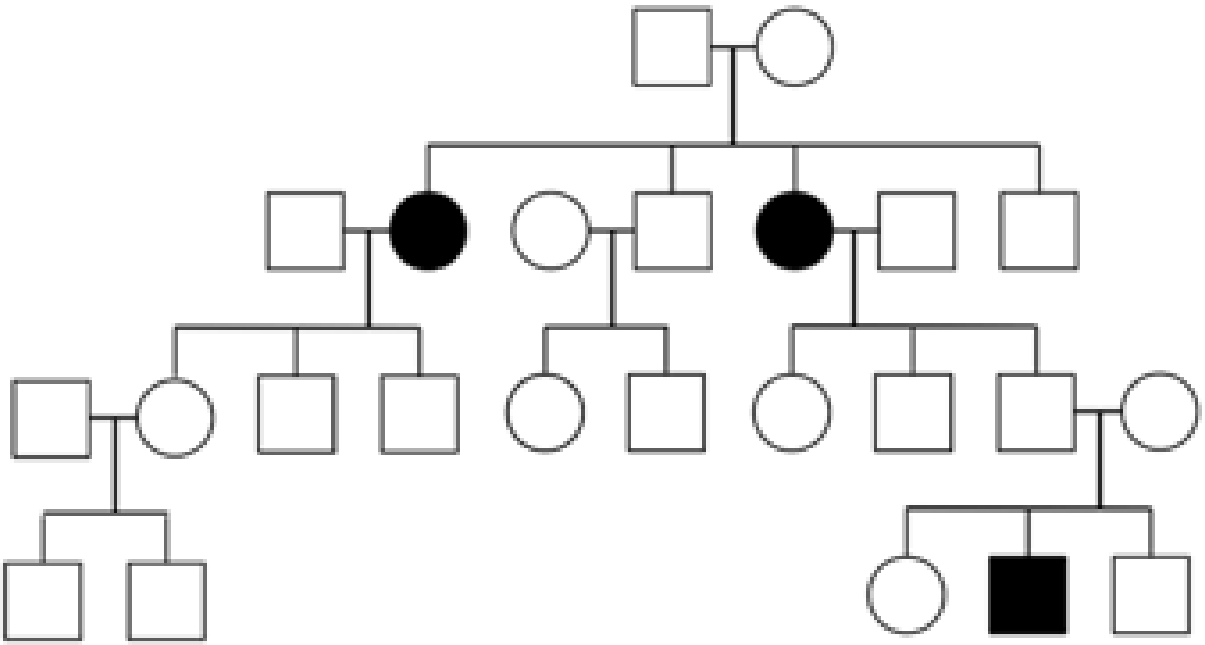
Name _____ **PEDIGREE ANALYSIS**

Pedigree #1 – What's That Smell?

Trimethylaminuria is a genetic disorder that causes a pungent smell. Individuals with this condition cannot break down a compound known as trimethylamine, whose smell has been described as rotting eggs, rotten fish, garbage, or even urine. Trimethylamine builds up in urine, breath, and sweat creating a strong smell. About 200,000 people in the US have trimethylaminuria.

The pedigree below shows how trimethylaminuria is inherited in one family. Shaded individuals are affected by a genetic condition. Could trimethylaminuria be inherited as:

- Autosomal dominant?
- Autosomal recessive?
- Sex-linked?



Label the generations using Roman numerals to the left of the pedigree. How many generations in this pedigree? _____

How many marriages are shown here? _____

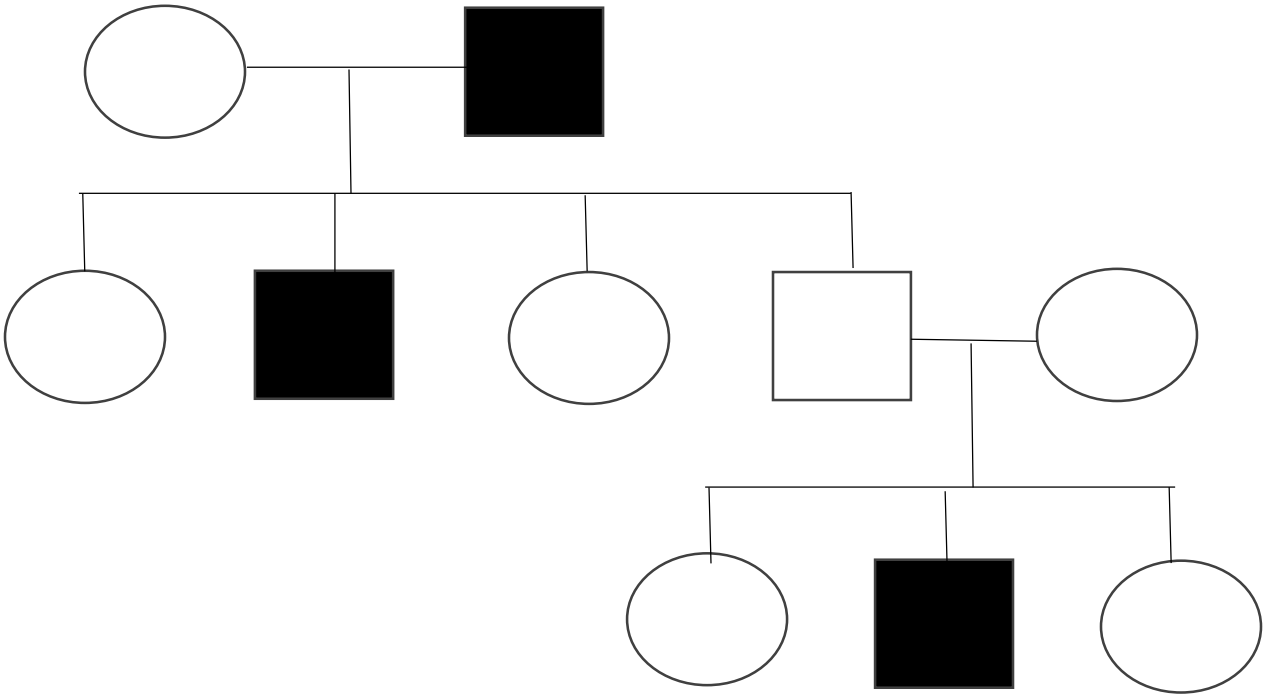
How many children did couple II-5 and II-6 have? _____

Use a Punnett square to determine the chances of a man with trimethylaminuria and a woman who is normal (TT) having a child with trimethylaminuria? _____

Pedigree #2 – Origins of the Werewolf Myth

Hypertrichosis, also known as "werewolf syndrome", is a genetic condition that causes excessive body hair growth. Those that have this condition exhibit excessive hair on the face, shoulders, and ears. There are 2 types of hypertrichosis – the most severe form is *hypertichosis lanuginose* which is autosomal dominant, while generalized hypertrichosis is less severe and is sex-linked recessive.

The pedigree below depicts how hypertrichosis is inherited in one family. Which form of the disease does this family have? _____



A mother is heterozygous for *hypertrichosis lanuginose*. What are the chances of her having a child with hypertrichosis is the father does not have the condition? _____

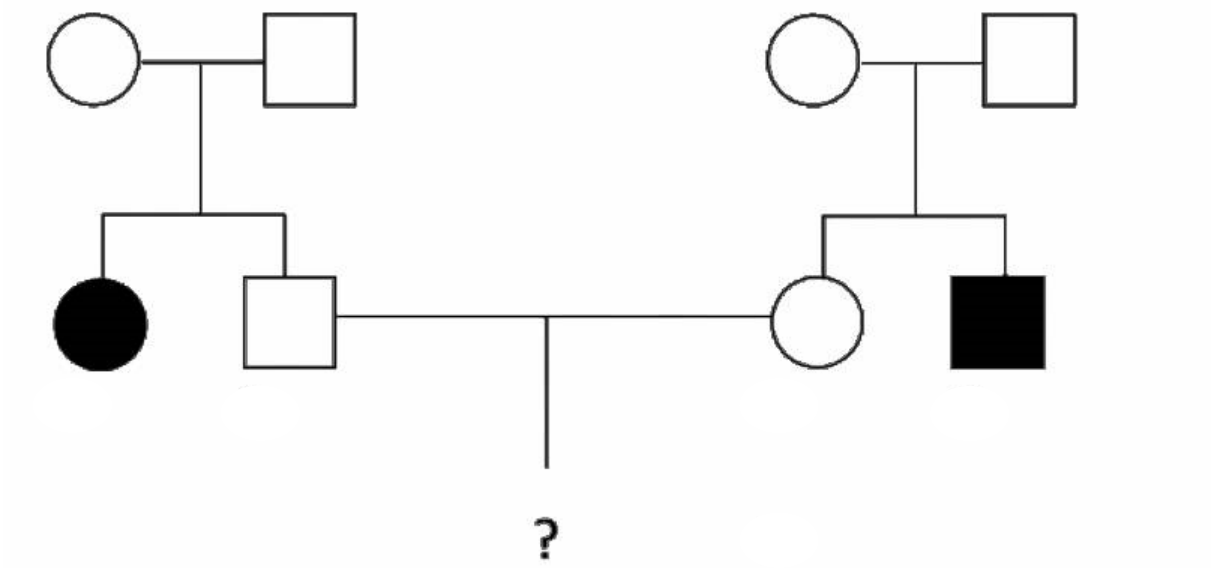
A man has generalized hypertrichosis and has married a woman who does not carry the trait. What are the chances of them having children with generalized hypertrichosis? _____

Pedigree #3 – Backwards or Forwards?

Uner Tan Syndrome is an extremely rare genetic disorder in which individuals travel on all fours, speak primitively, and have varying levels of mental retardation. It was only recently discovered and is localized in western Asia. While there have been multiple accounts of Uner Tan Syndrome, currently only a single family in Turkey is exhibiting symptoms.

In the pedigree below, shaded individuals are affected by Uner Tan Syndrome. Could the disorder be inherited as:

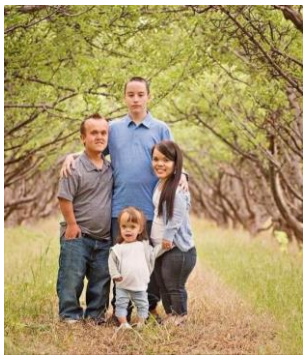
- Autosomal dominant?
- Autosomal recessive?
- X-linked dominant?
- X-linked recessive?
- Y-linked?



What are the chances of the child at **?** has Uner Tan? (hint: there is a range of % depending upon the genotype of the parents. Genetic counselors give these odds all the time)

_____

Pedigree #4 – A New Dwarfism

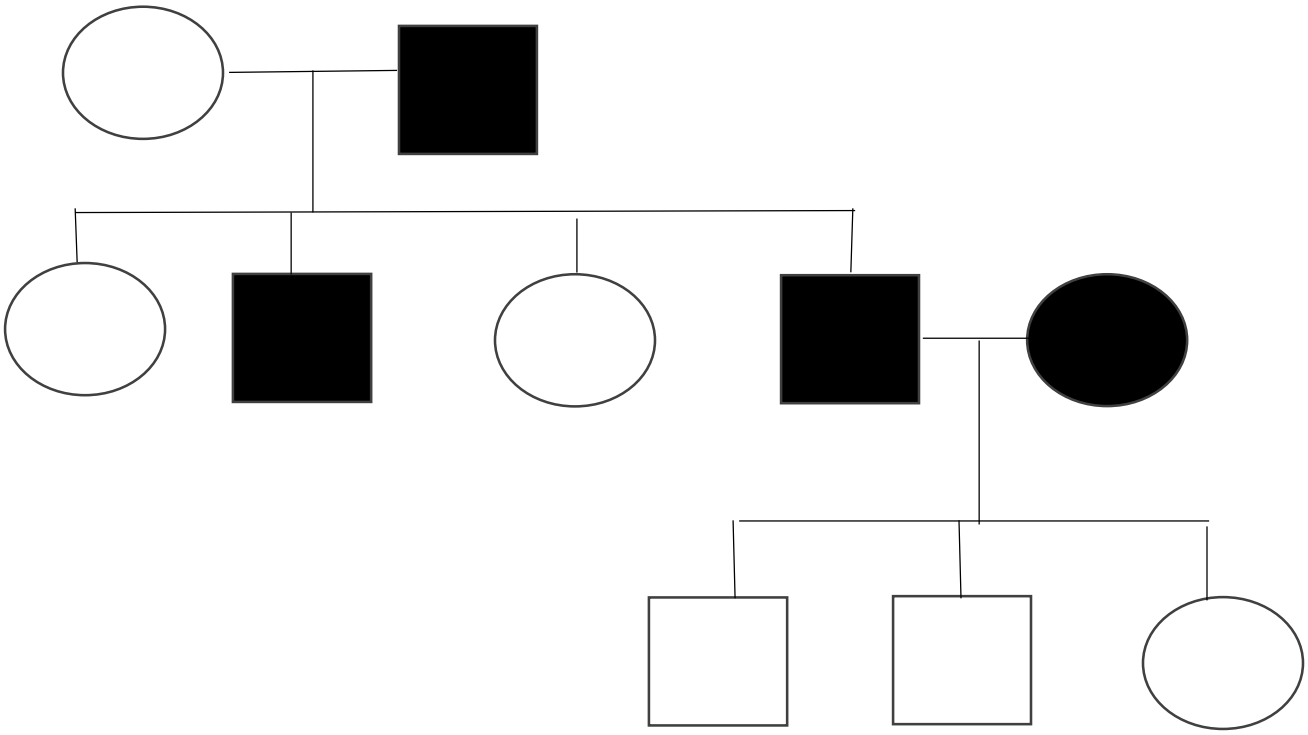


Dwarfism is a condition characterized by individuals who are short in stature caused by a mutation that slows or delays growth. Technically, dwarfism is defined as a height of less than 147 cm (4 feet 9 inches). There are actually different dwarfism mutations. Achondroplasia is autosomal dominant, while diastrophic dysplasia (DTD) is an autosomal recessive condition. Approximately 70% of all dwarfism is caused by achondroplasia.

The same mutation is common in other species as well.

A new form of dwarfism has been discovered in a remote area of Australia. Out of a population of 980, researchers counted 72 individuals with dwarfism. The pedigree below shows one Australian family with the new dwarfism form.

Use the pedigree below to determine if the condition is dominant or recessive. _____
How do you know? _____



Pedigree #5- Hemophilia and the Royals

Hemophilia is a genetic condition in which individuals are missing or have low levels of clotting factors in the blood. Clotting factors and platelets are needed in order to stop bleeding when a cut or bruise occurs. A lock of clotting factors causes uncontrolled external and internal bleeding following an injury, and ca lead to death. Hemophilia spread through the royal families of Europe and Russia and played an important role in the Russian Revolution in 1917.

Use the pedigree on the next page to determine the mode of inheritance for hemophilia. Then answer the questions.

- Autosomal dominant?
- Autosomal recessive?
- X-linked dominant?
- X-linked recessive?
- Y-linked?

Besides being married, how are Prince Phillip and Queen Elizabeth related?

_____

Did any females in Queen Victoria's line inherit hemophilia? _____

Did any males in Queen Victoria's line inherit hemophilia? _____

Why is there a difference in the number of females/males that inherited this trait? _____

_____

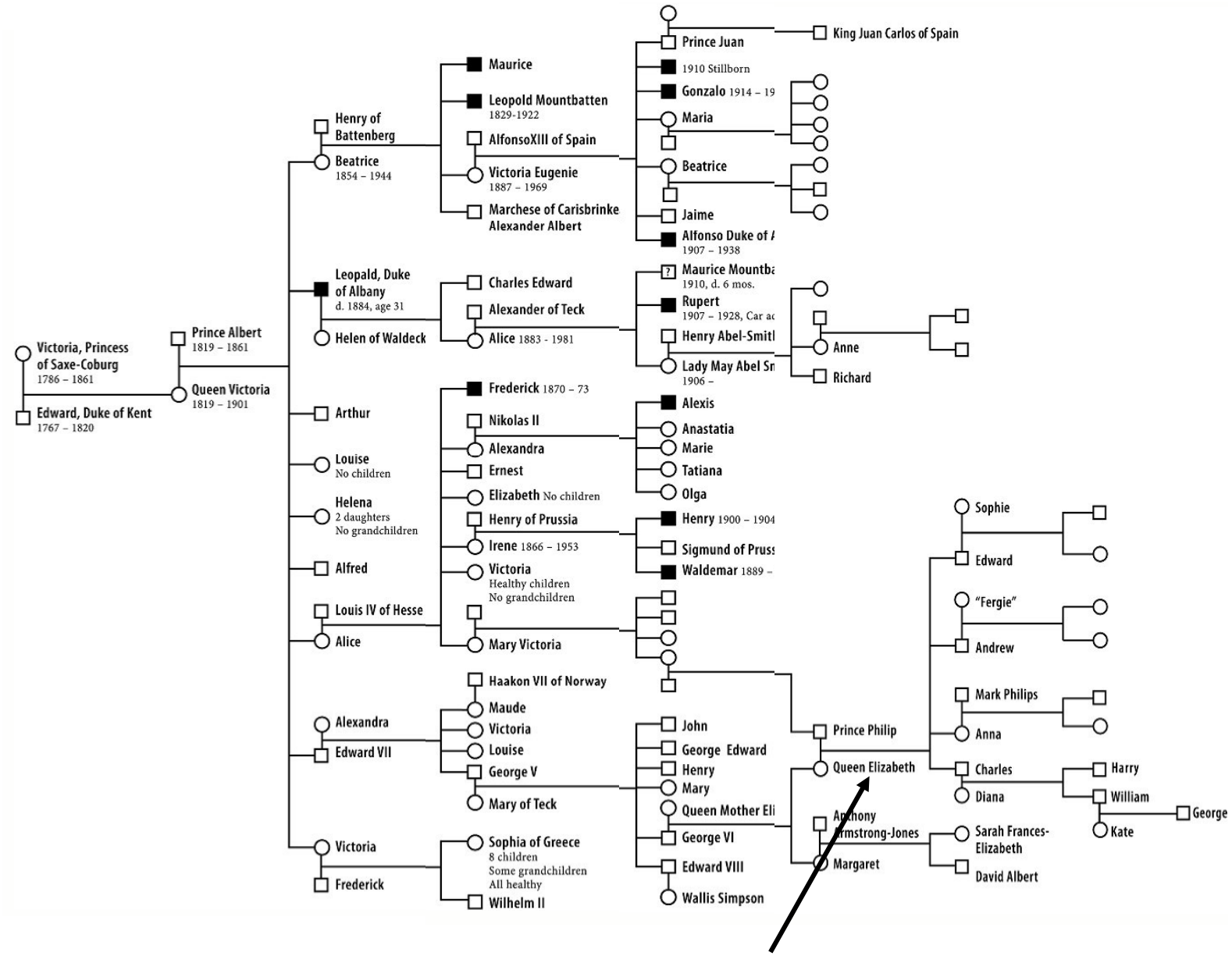
If Maurice or Leopold were to marry a normal female (no hemophilia), what are the chances their children would inherit hemophilia? _____ Could their grandchildren inherit hemophilia? Explain.

_____

Carriers of the disorder are often shaded half-way on a pedigree. Shade in all the known carriers appropriately. There are 7.

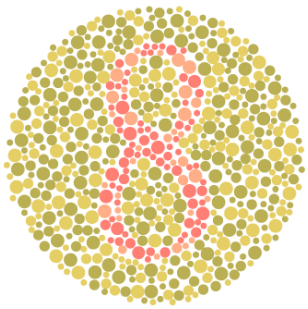
Can a male be a carrier of hemophilia? Explain. _____

_____



Current Queen of England

Pedigree #6 – Seeing Colors



Color vision deficiency (sometimes called color blindness) represents a group of conditions that affect the perception of color. Red-green color vision defects are the most common form of color vision deficiency. Affected individuals have trouble distinguishing between some shades of red, yellow, and green. Blue-yellow color vision defects, which are rarer, cause problems with differentiating shades of blue and green and cause difficulty distinguishing dark blue from black. These two forms of color vision deficiency disrupt color perception but do not affect the sharpness of vision (visual acuity).

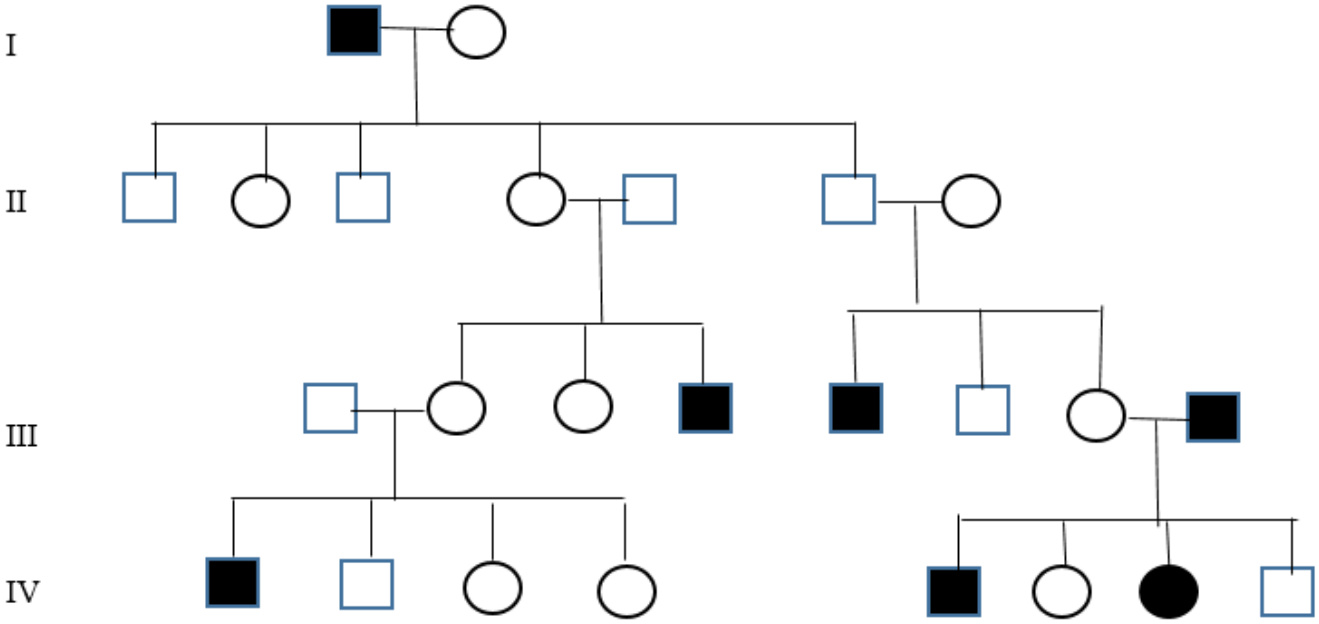
The pedigree below shows a family's pedigree for red-green colorblindness.

1. How is this trait inherited? _____
2. Which gender can be carriers of colorblindness and not have the disorder?_____
3. Why does individual IV-7 have colorblindness? Show the Punnett square of her parents to show how she inherited colorblindness. Circle IV-7 in the Punnett square:

4. Why must all the daughters of generation II carry the colorblind gene? _____

   _____

5. Write the genotypes of all individuals in the pedigree underneath their box/circle.



6. If individual IV-6 married a man with normal eyesight, what is the probability that their daughters and sons will have colorblindness? Do a Punnett square to illustrate your answer.