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Aim

To study the prepared pedigree chart of genetic trait such as colour blindness and haemophilia.

Theory

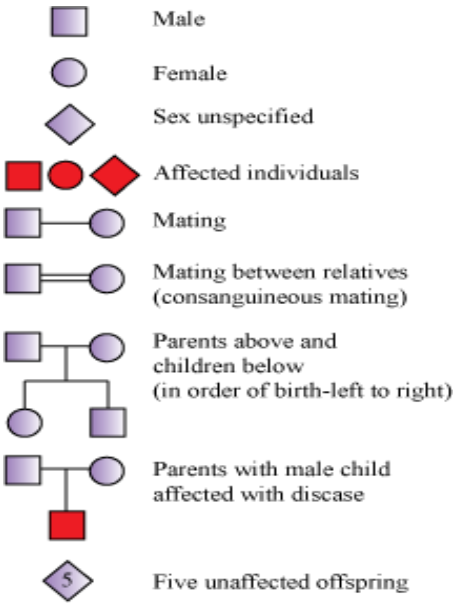
The principle of inheritance traits which was given by Mendel were applicable to plants, animals and human beings.

But the type of crossing done on plants and animals cannot be performed on humans.

So, a record of inheritance of certain genetic traits for two or more generations in the form of a diagram or a family tree called pedigree chart is prepared.

The Mendelian concept of dominance of genes and segregation of characters in subsequent generation can be studied by this method.

Few internationally approved symbols used in this analytic al study are as follows:



Requirements:

A family with genetic disorder for more than one generation, pedigree chart.

Procedure:

1. A family selected with a monogenetic trait, such as colour blindness and haemophilia.
2. Questions were asked to the family members.
3. Trait examined among the surviving individuals.
4. The information made available was the basis for the preparation of pedigree chart using appropriate symbols.
5. The careful examination of chart would suggest the gene for the character is X-linked recessive trait.

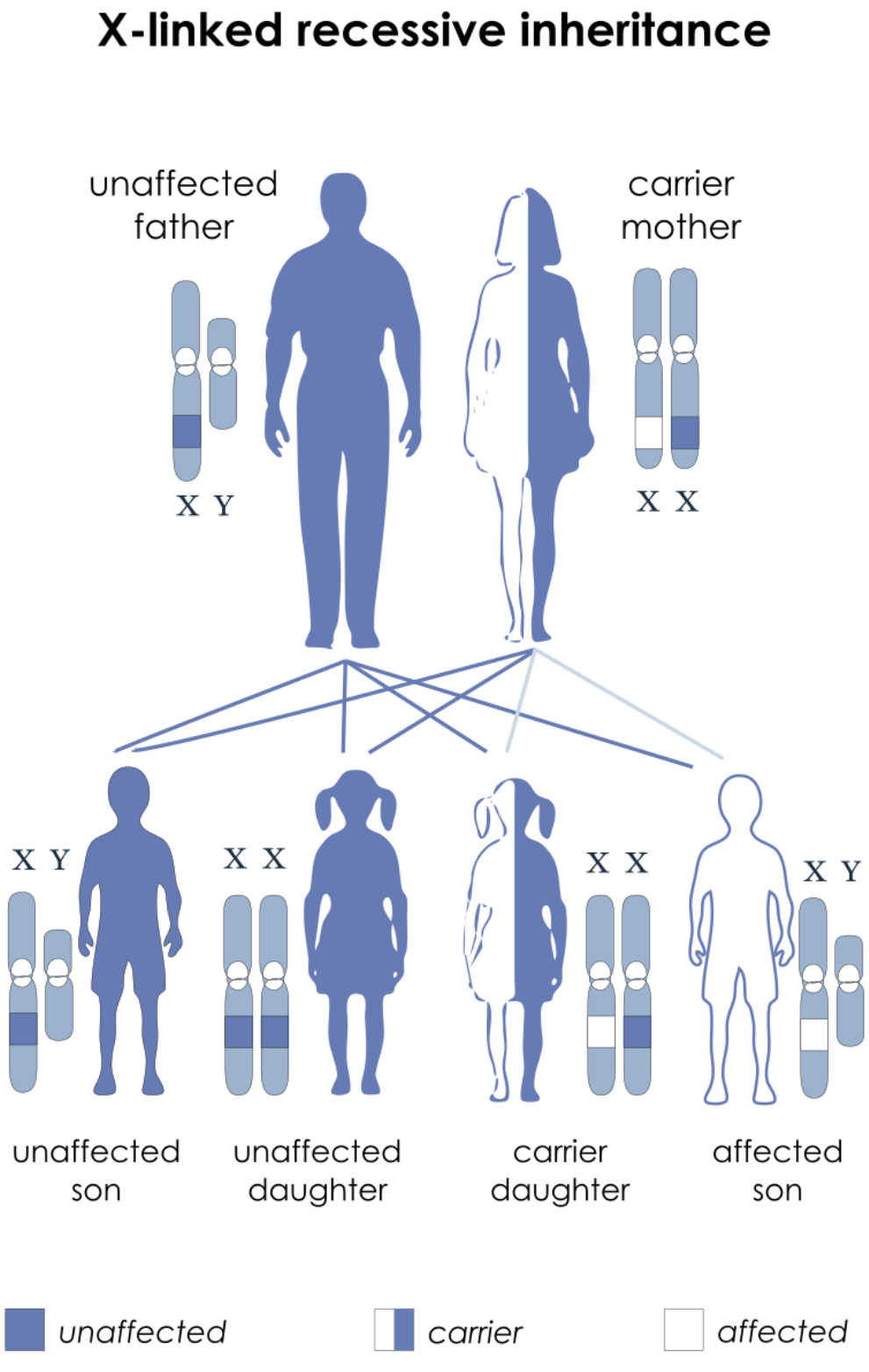
Explanation Of Traits

- Genes are inherited from our biological parents in specific ways. One of the basic patterns of inheritance of our genes is called X-linked recessive inheritance.

What is X-linked inheritance?

- X-linked inheritance means that the gene causing the trait or the disorder is located on the X chromosome.
- Females have two X chromosomes; males have one X and one Y. Genes on the X chromosome can be recessive or dominant.
- Their expression in females and males is not the same.
- Genes on the Y chromosome do not exactly pair up with the genes on the X chromosome.
- X-linked recessive genes are expressed in females only if there are two copies of the gene (one on each X chromosome).
- However, for males, there needs to be only one copy of an X-linked recessive gene in order for the trait or disorder to be expressed.
- For example, a woman can carry a recessive gene on one of the X chromosomes unknowingly, and pass it on to a son, who will express the trait: There is a 50 percent chance that daughters carry the gene and can pass it to the next generation.
- There is a 50 percent chance that a daughter will not carry the gene and, therefore, cannot pass it on.
- There is a 50 percent chance that sons do not have the gene and will be healthy.

- However, there is a 50 percent chance that a son will have inherited the gene and will express the trait or disorder



Examples of X-linked recessive conditions include red-green color blindness and hemophilia A:

COLOUR BLINDNESS

- Color blindness, also known as color vision deficiency, is the decreased ability to see color or differences in color.
- Simple tasks such as selecting ripe fruit, choosing clothing, and reading traffic lights can be more challenging.
- Color blindness may also make some educational activities more difficult.
- However, problems are generally minor, and most people find that they can adapt. People with total color blindness(achromatopsia) may also have decreased visual acuity and be uncomfortable in bright environments.
- The most common cause of color blindness is an inherited problem in the development of one or more of the three sets of color-sensing cones in the eye.
- Males are more likely to be color blind than females, as the genes responsible for the most common forms of color blindness are on the X chromosome.
- As females have two X chromosomes, a defect in one is typically compensated for by the other, while males only have one X chromosome.
- Color blindness can also result from physical or chemical damage to the eye, optic nerve or parts of the brain.
- Diagnosis is typically with the Ishihara color test; however, a number of other testing methods, including genetic testing, also exist.

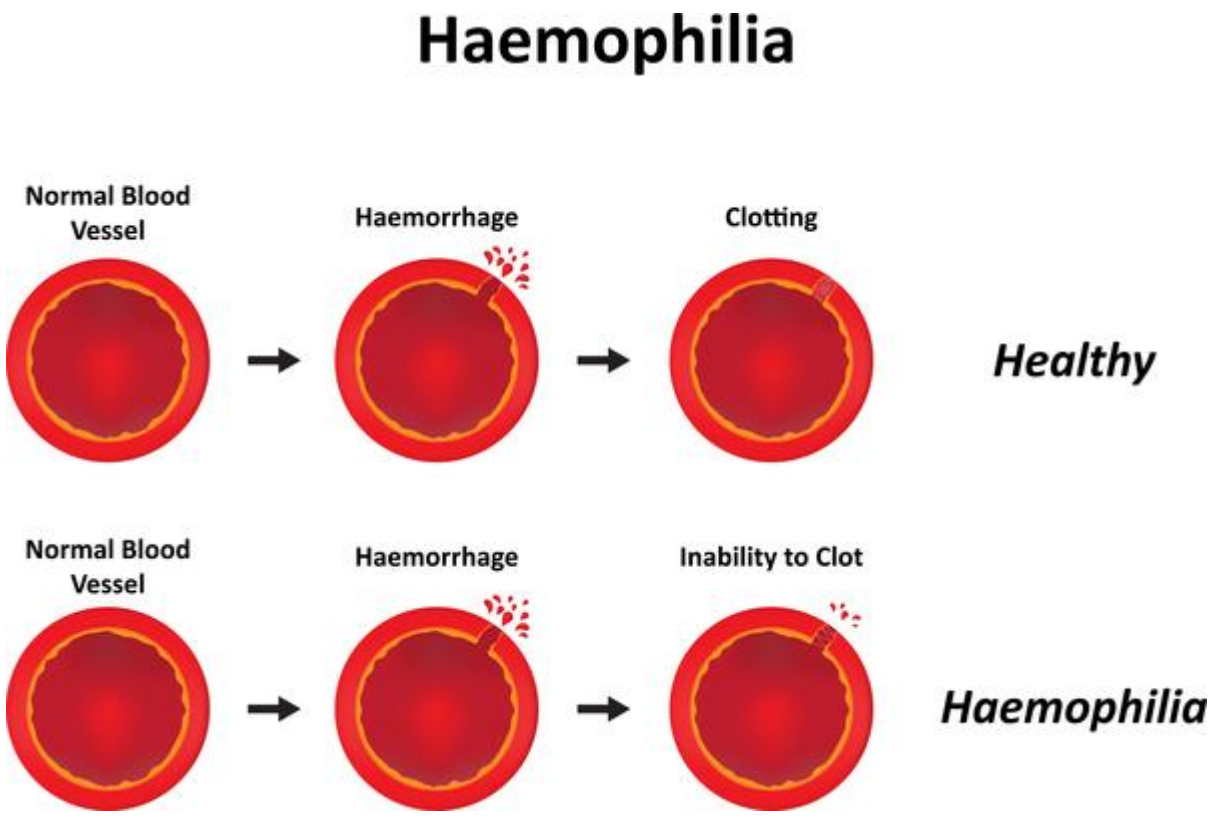
- Red-green color blindness is the most common form, followed by blue-yellow color blindness and total color blindness.
- Red-green color blindness affects up to 8% of males and 0.5% of females of Northern European descent. The ability to see color also decreases in old age.
- Being color blind may make people ineligible for certain jobs in certain countries. This may include being a pilot, train driver, crane operator, and working in the armed forces.
- The effect of color blindness on artistic ability, however, is controversial.
- The ability to draw appears to be unchanged, and a number of famous artists are believed to have been color blindness.

HAEMOPHILIA

- **Haemophilia** is a mostly inherited genetic disorder that impairs the body's ability to make blood clots, a process needed to stop bleeding.
- This results in people bleeding longer after an injury, easy bruising, and an increased risk of bleeding inside joints or the brain.
- Those with a mild case of the disease may have symptoms only after an accident or during surgery.

- Bleeding into a joint can result in permanent damage while bleeding in the brain can result in long term headaches, seizures, or a decreased level of consciousness.
- There are two main types of haemophilia: haemophilia A, which occurs due to not enough clotting factor VIII, and haemophilia B, which occurs due to not enough clotting factor IX.
- They are typically inherited from one's parents through an X chromosome with a nonfunctional gene.
- Rarely a new mutation may occur during early development or haemophilia may develop later in life due to antibodies forming against a clotting factor.
- Prevention may occur by removing an egg, fertilizing it, and testing the embryo before transferring it to the uterus.
- Treatment is by replacing the missing blood clotting factors. This may be done on a regular basis or during bleeding episodes.
- Replacement may take place at home or in hospital.
- The clotting factors are made either from human blood or by recombinant methods. Up to 20% of people develop antibodies to the clotting factors which makes treatment more difficult.
- The medication desmopressin may be used in those with mild haemophilia A. Studies of gene therapy are in early human trials.
- Haemophilia A affects about 1 in 5,000–10,000, while haemophilia B affects about 1 in 40,000, males at birth. As haemophilia A and B are both X-linked recessive disorders, females are rarely severely affected.

- Some females with a nonfunctional gene on one of the X chromosomes may be mildly symptomatic.
- Haemophilia C occurs equally in both sexes and is mostly found in Ashkenazi Jews.
- In the 1800s haemophilia B was common within the royal families of Europe.
- The difference between haemophilia A and B was determined in 1952.



Observation:

- The interrogated families showed the characteristics symptoms of the X- linked recessive disorder inferred in the studies undertaken for the pedigree analysis.

Result:

- The families studied showed the transmission of traits responsible for X-linked recessive disease in the successive generations in each category.

Precautions:

1. The prior knowledge of genetic disorder is must to identify the presence of a particular disorder in pedigree analysis
2. Family history of atleast 3-4 generations must be known.