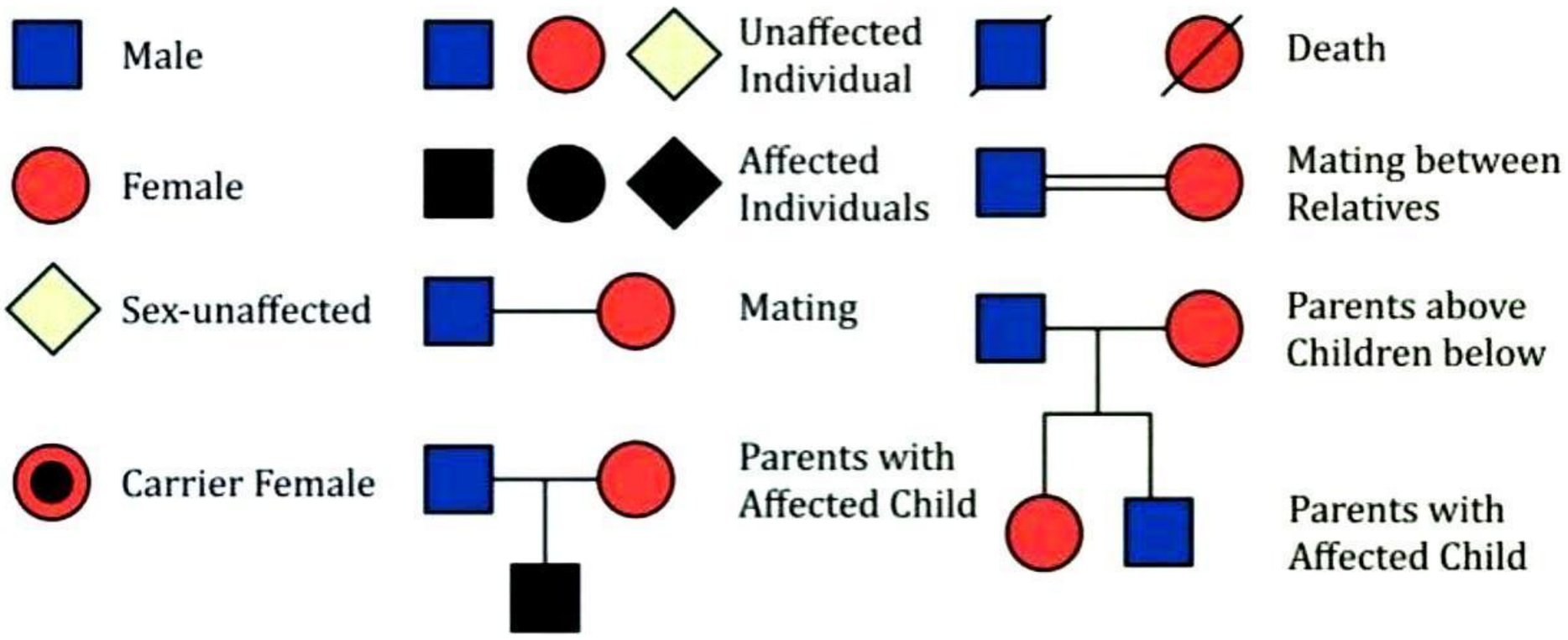
EXPERIMENT

AIM

To study of prepared pedigree charts of genetic traits such as rolling of tongue, blood groups, widow's peach and colour blindness.

THEORY

The principle of inheritance of 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 analytical study are as follows:



REQUIREMENTS

Questionnaire about a particular disorder, a family with genetic disorder for more than one generation, paper, pencil, etc.

PROCEDURE

1. Select a family with a monogenetic trait, such as rolling tongue, widow's peak, colour blindness, blood group, hitch-hiker's thumb, hypertrichosis and of ear dimpling of cheeks, etc.
2. Ask persons exhibiting the trait to tell in which of his/her parents, grandparents, their children and grandchildren, the trait in question is present.
3. Among the surviving individuals the trait may also be examined
4. The information made available is the basis for the preparation of pedigree chant using appropriate symbols.

5. The careful examination of chart would suggest whether the gene for the character is
   (i) Autosome-linked dominant or recessive.
   (ii) X-chromosome linked dominant or recessive.
   (iii) Y-chromosome linked or not.

Explanation of Each Trait

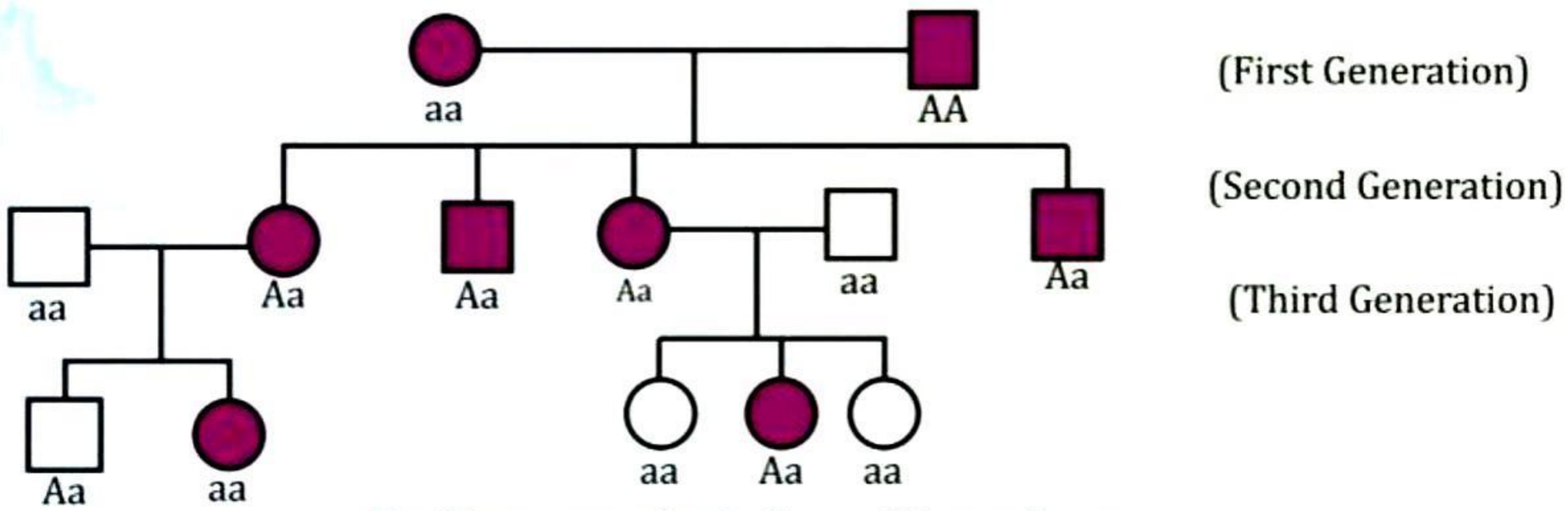
Autosome Linked Dominant Traits

1. The encoding gene of this trait is present on any one of the autosomes.
2. The mutant allele is dominant and the wild type allele is recessive for such traits.
3. In this pedigree chart, the female being interviewed is exhibiting the trait and is indicated by an arrow mark in the chart.
4. Transmission of trait occurs from any of the parent.
5. Male and temale are equally affected.
6. The trait is marked to be present in each of the generation, i.e. pedigree chart is vertical.
7. Multiple generations are characteristically affected.
8. Common examples are brachydactyly, polydactyly, widow's peak and dimple in the cheek.

Widow's Peak

It is a ' *V* '-shaped hairline across the forehead. It is a dominant trait. Thus, both homozygous dominart (AA) and heterozygous (Aa) individuals have widow's peak, while homozygous recessive (az) individuals have a straight hairline.

The outline of pedigree chart is shown below:



Pedigree analysis for rolling of tongue

Autosome Recessive Traits

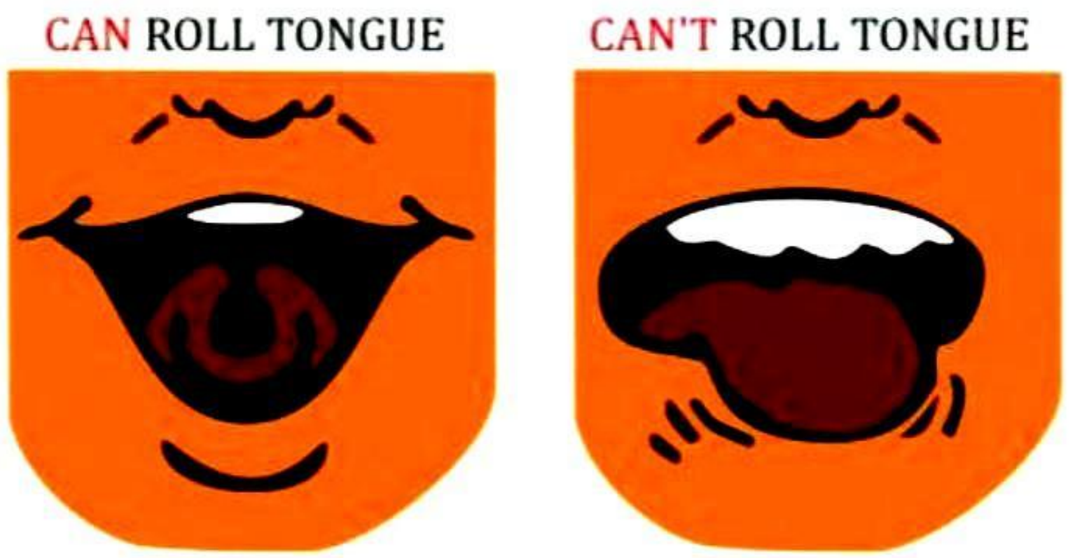
1. In this, mutant allele is recessive to its wild type allele.
2. These traits occur in equal proportions in male and female siblings, whose parents are carriers of the

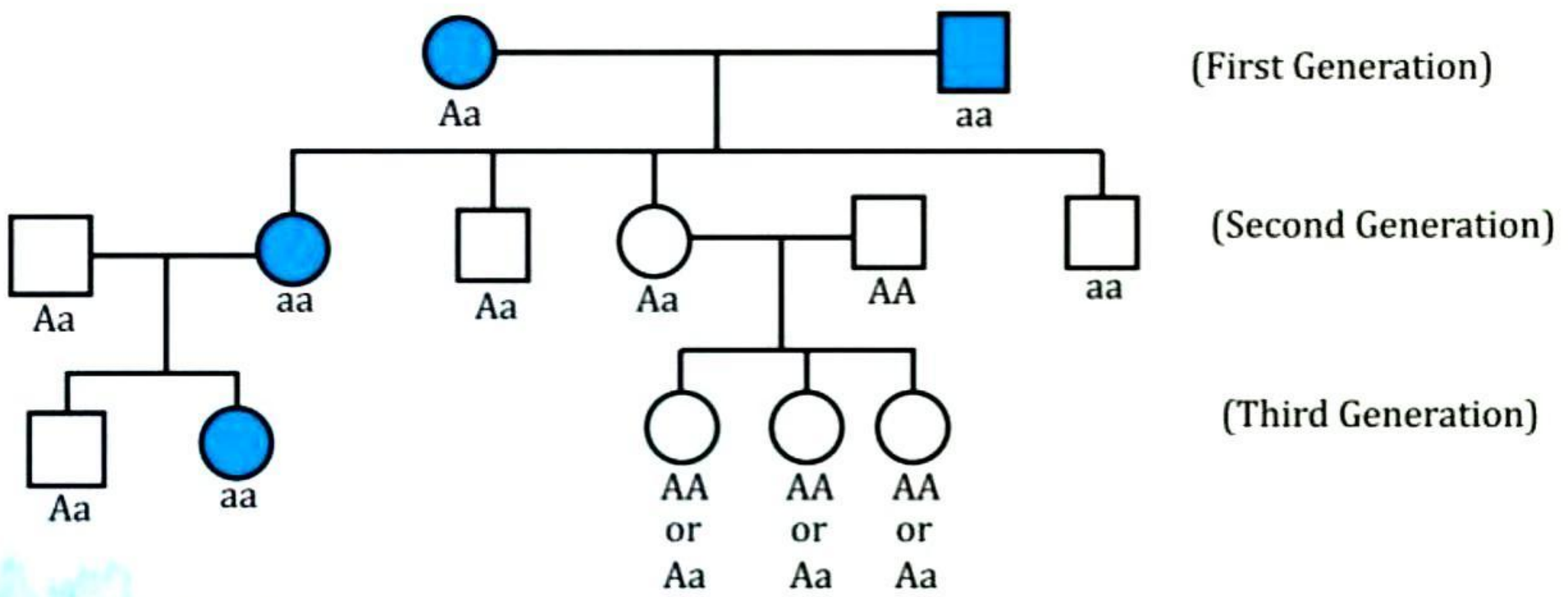
allele, i.e. they have only a single copy of the allele and therefore they are normal.

3. The siblings are homozygous for the defective allele, but their parents, though some may appear normal are obviously heterozygous, i.e. are merely carriers of the trait.
4. The marriage between man and woman genetically related with each other, occasionly results in the appearance of such traits, e.g. albinism and rolling of tongue.

The Rolling of Tongue

Some persons are able to roll their tongue in U-shape. The inability to roll the tongue is caused by an autosomal recessive allele 'a'. Thus, both homozygous dominant (AA) and heterozygous (Aa) individuals are able to roll the tongue, while homozygous recessive (aa) individuals are unable to roll their tongue in this way.





Note: Indicates that these females may either be homozygous dominant or heterozygous for the gene controlling the ability of an individual to roll his/her tongue. In both the cases, these three siblings would be able to roll their tongues.

X-Linked Dominant Traits (Muscular Dystrophy)

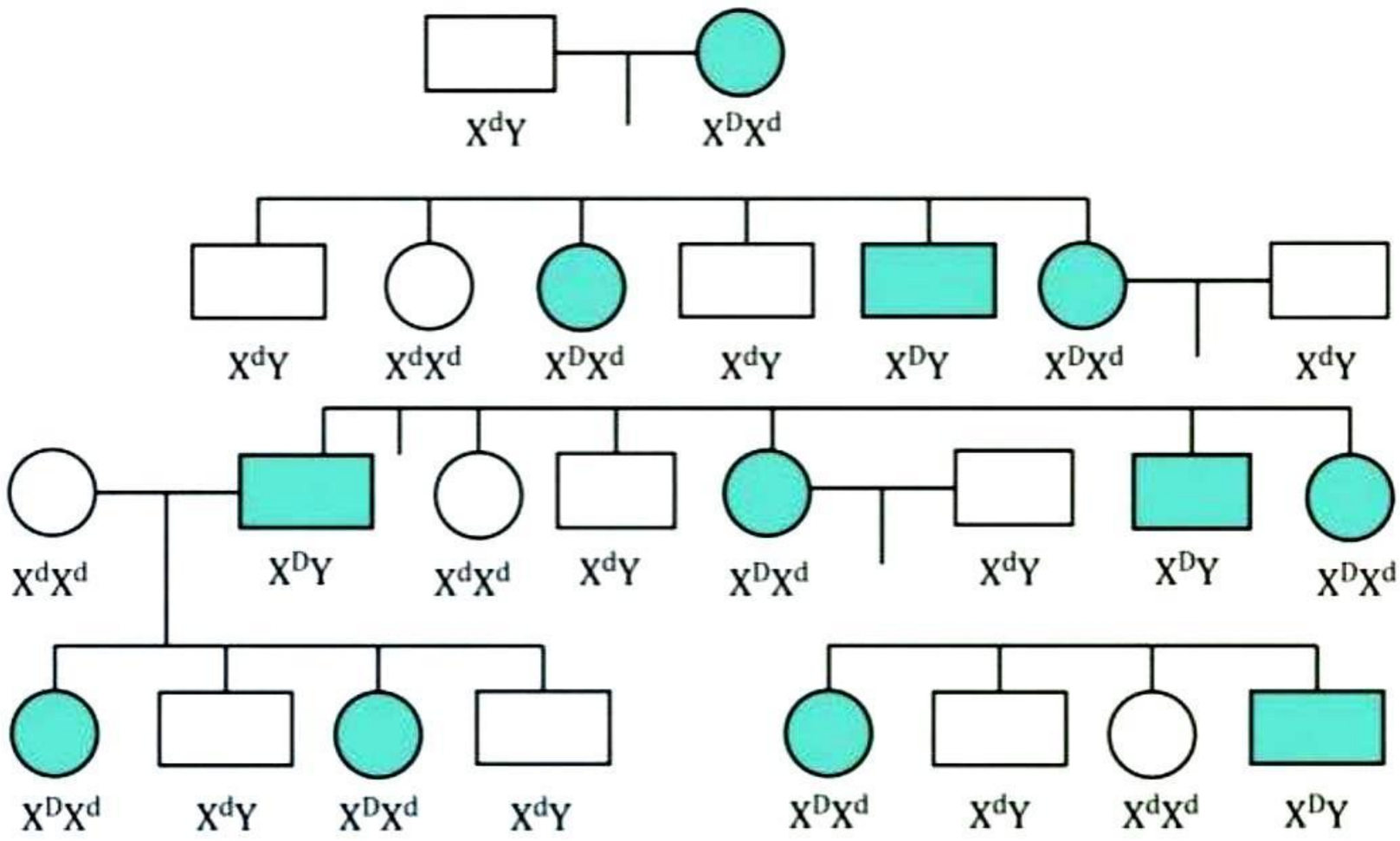
1. These are the traits whose encoding genes are present on the X-chromosome, and the mutant allele of which is dominant over its wild type allele.
2. Such traits are very rare in a given population sample.
3. In this case, the trait appears in almost all the generations and the inheritance is vertical.
4. If the female is affected, then about half of her sons are affected.
5. If the male is affected then half of his daughters would be affected, but none of his sons would be affected.
6. In short, the pedigree resembles the pattern of inheritance of autosomal dominant, except that there is

no male-to-male transmission, e.g. Duchene muscular dystrophy.

Duchene Muscular Dystrophy

Pedigree analysis of muscular dystrophy

1. This results in the absence of teeth and bifid tongue associated with mental retardation,
2. The possible genotypes of affected persons are shown in the following pedigree chart. Here ' D ' is the dominant mutant allele and the recessive wild allele is represented try ' d '.
3. Here, it is important to remember that human females possess two X-chromosomes (XX) and the human males possess one X and one Y chromosome. Males receive their X-chromosomes from their mother only, whereas females received one of her X-chromosome from her mother and the other X-chromosome from her father. This forms the basis of inheritance for X-linked dominarn traits.



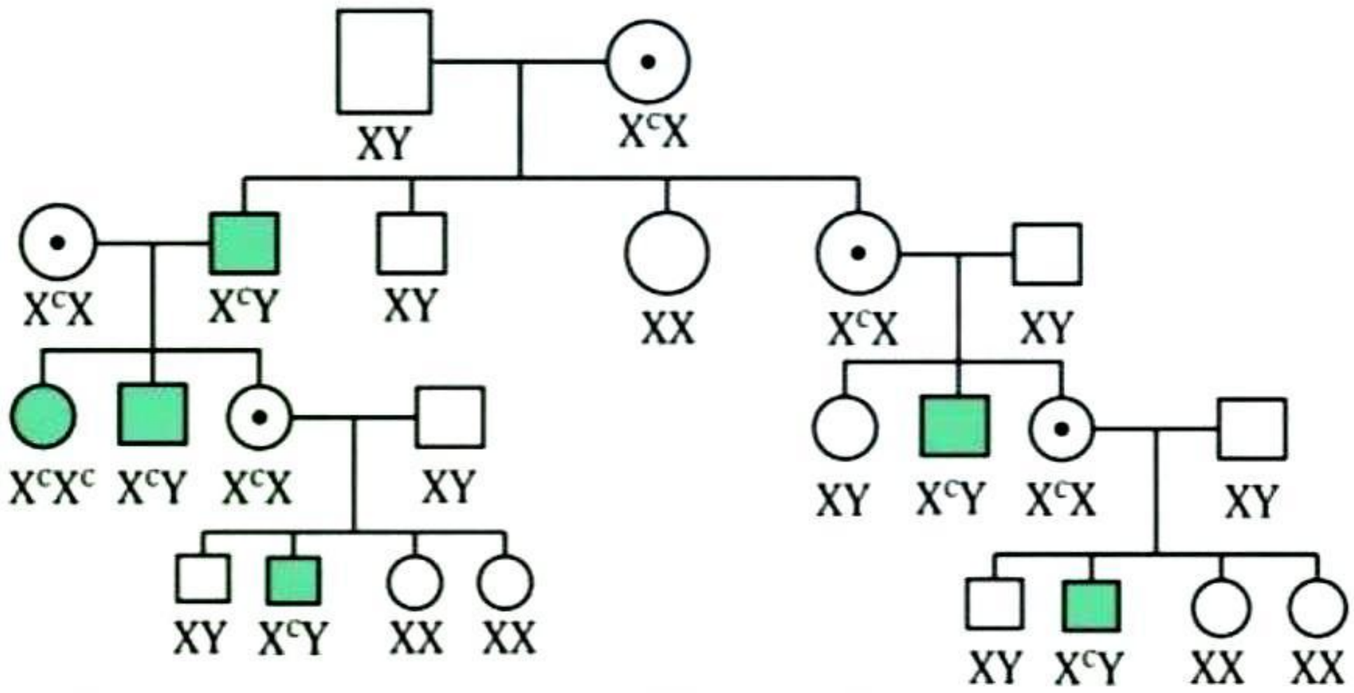
Genotypes of Individuals

X-Linked Recessive Traits

1. These are the traits whose encoding gene is present on the X-chromosome and its mutant allele is recessive to its wild type allele.
2. The females expresses the trait only when they are homozygous for the mutant allele whereas males does so even when they are hemizygous for it.
3. About half of the sons of the carrier (heterozygous for the trait) females are affected.
4. In case of homozygous females showing the trait, 50% of her daughters and all of her sons $a^{2\%}$ likely to be affected. Therefore, it can be concluded that males are most affected population in th: category.
5. Affected persons are related to one another through the maternal side of their family.
6. Any evidence of male to male transmission of the trait rules out the X-linked inheritance.
7. Examples are haemophilia and red-green colour blindness.

Colour blindness

The colour blindness is a sex-linked recessive disorder of humans. In this, the affected individuals unable to differentiate between red and green colours. It ressults in the absence or malfunction in? of one or more of the three types of cone cell responsible for colour vision. Pedigree analysis of colour blindness



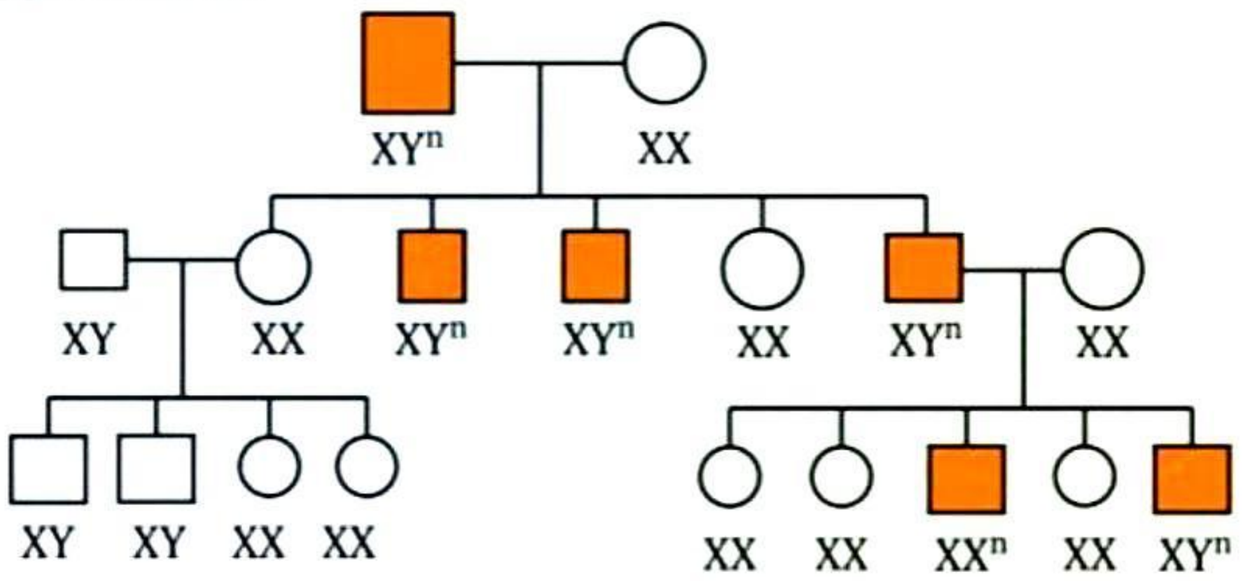
Inheritance pattern of X-linked recessive traits

Y-Linked Traits

1. These are the traits whose gene is present on the *Y*-chromosome.
2. Any trait linked to the *Y*-chromosome thus, is present only in males and certainly not in any of the females (because no female can have *Y*-chromosome).
3. This is why these traits are also called male sex limited traits.
4. All the sons of the affected male would express the trait whereas none of the daughter would do so.
5. Example is hypertrichosis of the ear (the presence of hairs on pinna) is one most common example of such a trait.

Hypertrichosis

Pedigree analysis of hypertrichosis



Inheritance pattern of Y-linked traits

Since, the trait is present on Y-chromosome of males (XY), the females are normal for such traits as they have (*XX*) chromosomes and the trait does not affect the *X*-chromosome. So, all the males in each generation will show the hairs on the pinna in this kind of family tree.

OBSERVATIONS

The interrogated families showed the characteristic symptoms of the genetic disorders infered in the studies undertaken for the pedigree analysis.

RESULT

The families studied showed the transmission of traits responsible for genetic disease in the successive generations in each category.

PRECAUTION

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.

VIVA VOCE

Q1. How do we study inheritance of a character in a family?

Ans. We make a diagrammatic record of inheritance of a particular trait/traits over two or more generations in a family tree. It is called pedigree chart.

Q2. What is meant by pedigree analysis?

Ans. When we analyse the record of inheritance of a particular trait over two or more generations through diagram. it is called pedigree analysis.

Q3. How does pedigree analysis help in genetic studies?

Ans. It helps in knowing the possibilities of the presence of a trait in homozygous or heterozygous state in a particular offspring. It gives the probability that a certain genetic disorder may be present in the offspring from parents having the disorder in their family history.

Q4. What does the horizontal line represent in a pedigree chart?

Ans. The horizontal line represents the parents.

Q5. Who discovered blood groups?

Ans. Landsteiner discovered the blood groups A, B, AB and O.

Q6. Is colour blindness a genetic disorder?

Ans. Yes, it is a genetic disorder in an individual where he/she is not able to distinguish between green and red colours. It is an example of sex-linked inheritance.

Q7. What is the basis of blood groups?

Ans. Blood group inheritance is based on the multiple alleles, i.e. 1^A, 1^D and 1^B.

Q8. Is widow's peak a sex-linked or autosomal trait?

Ans. It is an autosomal recessive trait.

Q9. Discuss the difference in the patterns of autosome linked recessive and sex chromosome linked pedigree.

Ans.

Autosome disorder	Sex chromosome disorder
These arise by gene mutations in autosomal chromosomes.	These arise in sex chromosomes (allosomes) mostly X-chromosomes.
These disorders affect mostly both the sexes.	These affect mostly males.