

Exercise 11

Aim: Preparation and analysis of Pedigree Charts

Principle: The Mendelian concept of dominance and segregation can also be studied in humans by preparing and then analysing the pedigree charts. The internationally approved symbols for indicating males and females, marriages, various generations (I, II, III), etc., are given below.

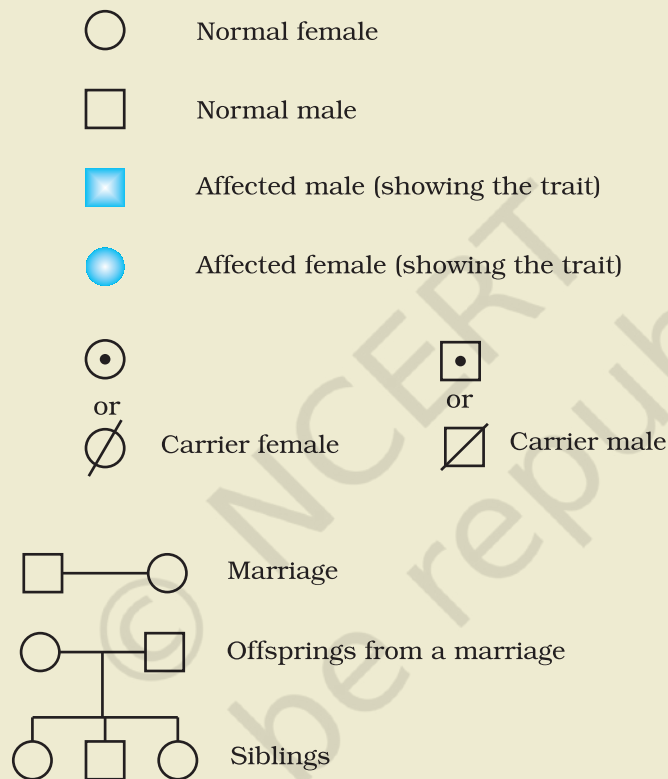


Fig.11.1 Symbols used for preparation of pedigree chart

Requirement: Information about characters/traits in a family for more than one generation

Procedure

Select a family in which any one of the monogenic traits such as tongue rolling, widow's peak, blood groups, red-green colour blindness, dimple in

the cheek, hypertrichosis of ear, hitch-hiker's thumb, etc., is found. Ask the person exhibiting the trait to tell in which of his/her parents, grand parents (both maternal and paternal), their children and grand children the trait in question is present. Among surviving individuals the trait may also be examined. The information made available is the basis for the preparation of pedigree chart using the appropriate symbols. A careful examination of the pedigree chart would suggest whether the gene for the character is autosome-linked dominant or recessive, X - chromosome linked dominant or recessive, Y- chromosome linked or not.

Explanation

1. **Autosome Linked Dominant traits:** These are the traits whose encoding gene is present on any one of the autosomes, and the wild-type allele is recessive to its mutant allele, i.e., the mutant allele is dominant.

The pedigree-chart can be of the undernoted pattern (Fig. 11.2), where the female being interviewed is exhibiting the trait, and is indicated by an arrow-mark in the chart.

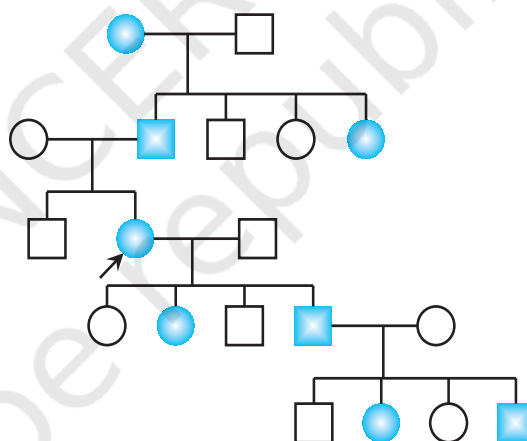


Fig.11.2 Inheritance of Autosome liked dominant traits

The characteristic features of inheritance of such type of traits are:

- (a) Transmission of traits occurs from parents of either sex.
- (b) Males and females are equally affected.
- (c) The pedigree is vertical, i.e., the trait is marked to be present in each of the generations.
- (d) Multiple generations are characteristically affected.

Brachydactyly, polydactyly, dimple in the cheek are some of the common traits of this type.

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2. **Autosomal Recessive trait:** These are the traits whose mutant allele is recessive to its wild type allele.

The pedigree chart can be more or less of the pattern given below (Fig. 11.3), where the lady (marked by the arrow) is showing the trait. The bar

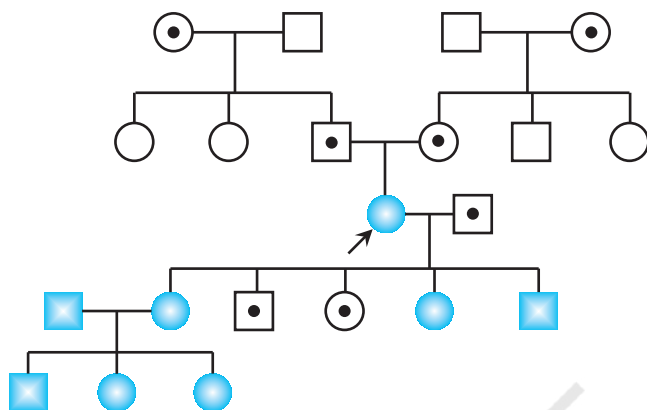


Fig.11.3 Inheritance of Autosome linked recessive traits

in the example represents the presence of corresponding dominant or recessive allele for the specific trait.

Suppose the given trait is **albinism**. Denote its dominant allele as 'A' that produces pigments, and the recessive allele as 'a' that fails to synthesise the pigment, melanin. The female (our subject in generation III) is therefore of genotype aa. She must have received each of her 'a' allele from both the parents (generation-II), who are therefore themselves normal but are definitely of genotype Aa, and are carriers of the trait. The allele a must also have been present in her grand parents too, of course in heterozygous condition also to make them carriers (generation-I)

Albinism in the subject's children (generation-IV) suggests her husband too to be of genotype Aa, a carrier. Marriage of her albino daughter to an albino man is bound to produce all her grand-children albino (gen-V).

The following are the salient features of the inheritance of such type of traits.

- Occur in equal proportions in multiple male and female siblings, whose parents are normal but carriers;
- 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.
- Consanguinity (marriage between man and woman genetically related to each other, such as cousins) occasionally results in the appearance of such traits.

3. **X-Linked Dominant traits:** These are the traits whose encoding gene is present on the X- chromosome, and the mutant allele of which is dominant over its wild-type allele.

Such traits are very rare, and are almost difficult to find in the population. One example is oral-facial-digital syndrome (Duchene Muscular Dystrophy), which results in absence of teeth, cleft (bifid) tongue associated with mental retardation. The pedigree chart may appear as follows (Fig. 11.4):

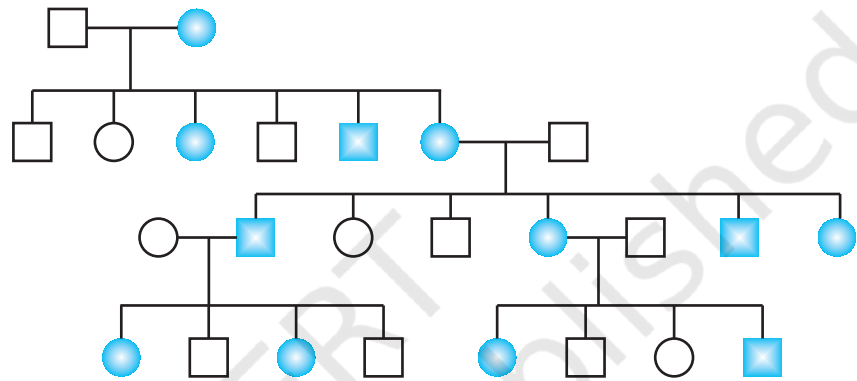


Fig. 11.4 Inheritance pattern of X-linked dominant traits

The possible genotypes of the above pedigree can be written as follows (Fig 11.5):

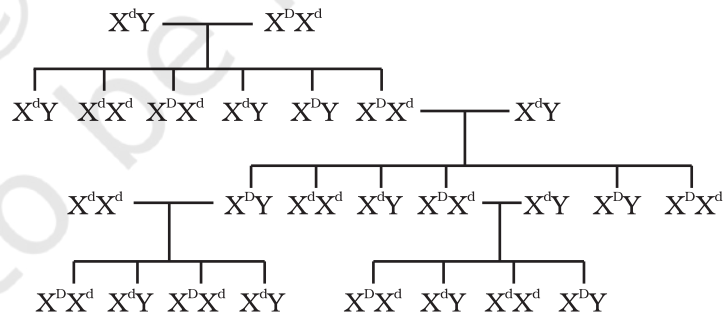


Fig. 11.5 Genotypes of individuals shown in Fig. 11.4

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Here, the dominant mutant allele is denoted by 'D', and its recessive wild type allele is denoted by 'd'. Remember that human females have two X-chromosomes (XX), and the males have only one X and one Y chromosome. Males receive their lone X-chromosome from their mother, and the Y-chromosomes from their father, whereas females receives one of her X-chromosome from her mother, and the other X from her father.

The characteristics of such inheritance are:

- The trait appears in almost all the generations, and the inheritance is vertical.
 - If the female is affected, then about half of her sons are affected.
 - If the male is affected then all of his daughters would be affected, but none of his sons are affected.
 - In short, the pedigree resembles the pattern of inheritance of autosomal dominants, except that there is no male-to-male transmission.
4. **X-linked Recessive traits:** These are the traits whose encoding gene is present on the X-chromosome and its mutant allele is recessive to its wild-type allele.

Red-green colour blindness and hemophilia, are some of its well known examples. The characteristic features of such inheritance are:

- Females express the trait only when they are homozygous for the mutant allele, whereas the males do so even when they are hemizygous for it.

The pedigree chart would appear as the following one (Fig. 11.6):

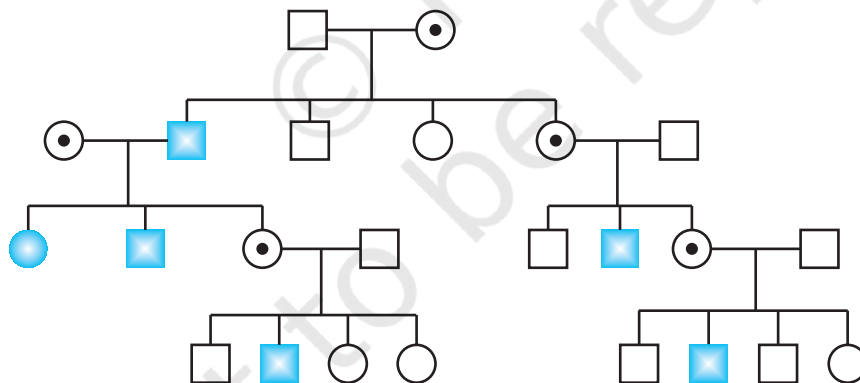


Fig. 11.6 Inheritance pattern of X-linked recessive traits.

- (b) About half of the sons of the carrier (heterozygous for the trait) females are affected. In case of homozygous females showing the trait, fifty percent of her daughters and all of her sons are likely to be affected. Therefore, the males are most affected in the population.
- (c) Affected persons are related to one another through the maternal side of their family.
- (d) Any evidence of male-to-male transmission of the trait rules out the X-linked inheritance.
5. **Y-chromosome linked traits:** These are the traits whose gene is present on the Y-chromosome. The females do not have any Y-chromosome, whereas all the males must have a Y-chromosome to be a male, and this Y-chromosome they get from their father. Therefore, any trait linked to the Y-chromosome must be present only in males, and certainly not in any of the females. This is why these traits are also called male-sex limited traits. All the sons of the affected male would express the trait whereas none of his daughters would do so.

The pattern of the pedigree chart would be as follows (Fig 11.7):

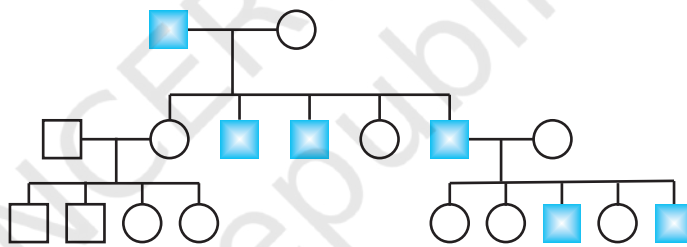


Fig.11.7 Inheritance pattern of Y-linked traits

Hypertrichosis of the ear (presence of hairs on pinna) is one most common example of such traits.

Note: Students may be asked to prepare the pedigree-chart from given data and analyse the pattern of inheritance. The work may be done as a project.

Questions

1. How will you differentiate between autosome linked dominant and sex chromosome linked dominant pedigree chart? Explain.
2. Discuss the differences in the patterns of autosome linked recessive and sex-chromosome linked pedigree.