08-12-21

EXPERIMENT NO. 1

AIM OF THE EXPERIMENT - Pedigree analysis from the given chart.

THEORY -

The Mendelian concept of dominance and segregation can also be studied in humans by preparing and then analysing the pedigree chart.

The intertionally approved symbols for indicating males and females, marriage, various generation (I, II, III) etc are given below.

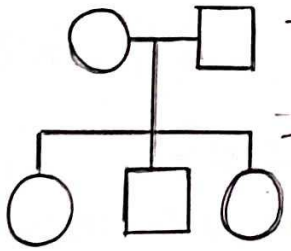
 — Normal female

 — Normal male

 — Affected male (showing the trait)

 — Affected female (showing the trait)

 — Marriage

 — Offspring from a marriage.

— Siblings.

MATERIALS - Pencil, Pen, Notebook, Given Pedigree chart, etc.

1. Autosomal linked Dominant trait -

Observation -

(i) For the particular character the analysis starts by identifying the affected individuals. If both males and females are affected in the chart, the character can't be Y-linked because this chromosome is absent in females, which is the case with the present chart.

(ii) Now, the inheritance pattern is observed. Every affected individual have at least one affected parent and no where in the chart normal parents have affected progeny. This is called vertical transmission which indicates the character/disease to be dominant, which is the case with present chart.

(iii) Now we look at inheritance of the disease/character from the male patient (II-4). If it is transmitted from male parent to both sons (III-12) and daughter (III-11) it can't be an X-linked disease because X-chromosome is not contributed by male parent (II-4) to sons (III-12).

Inference - The disease, identified to be dominant and neither Y-linked nor X-linked, therefore is autosomal.

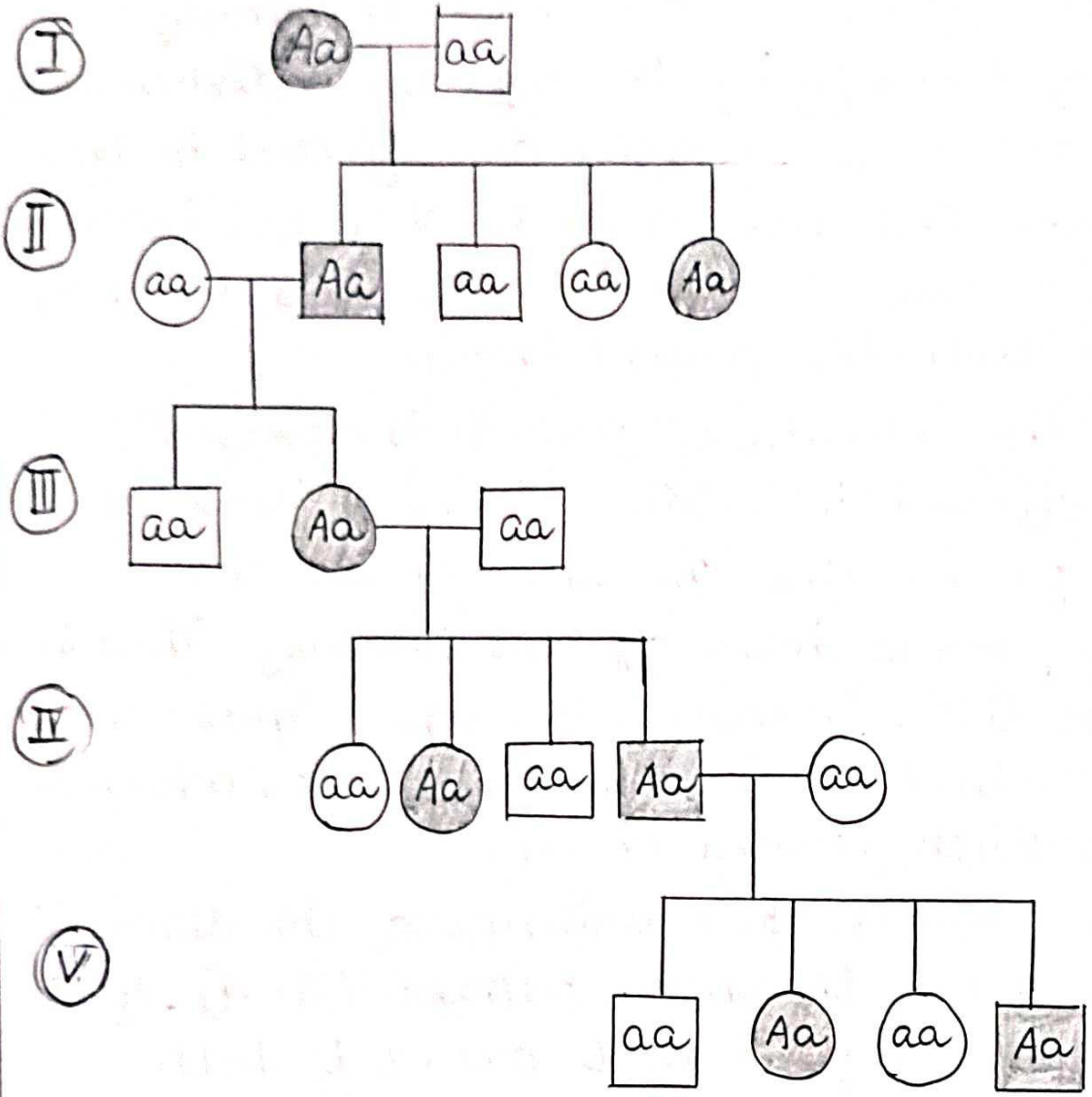


fig. Inheritance of Autosome linked dominant traits

Identifying genotype -

(i) Since it is a dominant disease, heterozygous will be affected. Thus all the normal individuals should be double recessive (say, aa).

(ii) Since every affected individuals has one normal parent (which is a) and at least one normal sibling (which is also aa), therefore in the parent case all affected individuals must have inherited a recessive allele from the normal parent (aa) and a dominant allele from affected parent (A).

Thus all affected individuals in the present chart are heterozygous (Aa).

Conclusion - The analysis of the Pedigree chart reveals the following -

(i) The disease or character is autosomal
(ii) The disease or character is dominant.
(iii) All normal individuals are double recessive.
(iv) All affected individuals are heterozygous.

Example - Polydactylyl, dimple in cheek. of human

2. Autosomal Recessive trait.

Observation -

(i) For the particular character the analysis starts by identifying the affected individuals. If both males and females are affected in the chart, the character can't be Y-linked because this chromosome is absent in females which is the case with the present chart.

(ii) Now the inheritance pattern is observed. Affected individuals may have normal parents so there is absence of vertical transmission so its a recessive disease.

(iii) If the disease is X-linked, the male of the first generation (I-1) could have had the disease because males are hemizygous for all X-linked disease. Thus the pedigree chart refers to neither Y-linked nor X-linked rather autosomal inheritance.

Identifying genotype -

(i) Since it is autosomal recessive disease all affected individuals are double recessive (aa) and all normal individuals have at least one dominant allele.

(ii) Since individuals of the first generation having one 'A' each, have contributed ~~one 'a'~~ each to the offspring, they themselves should be

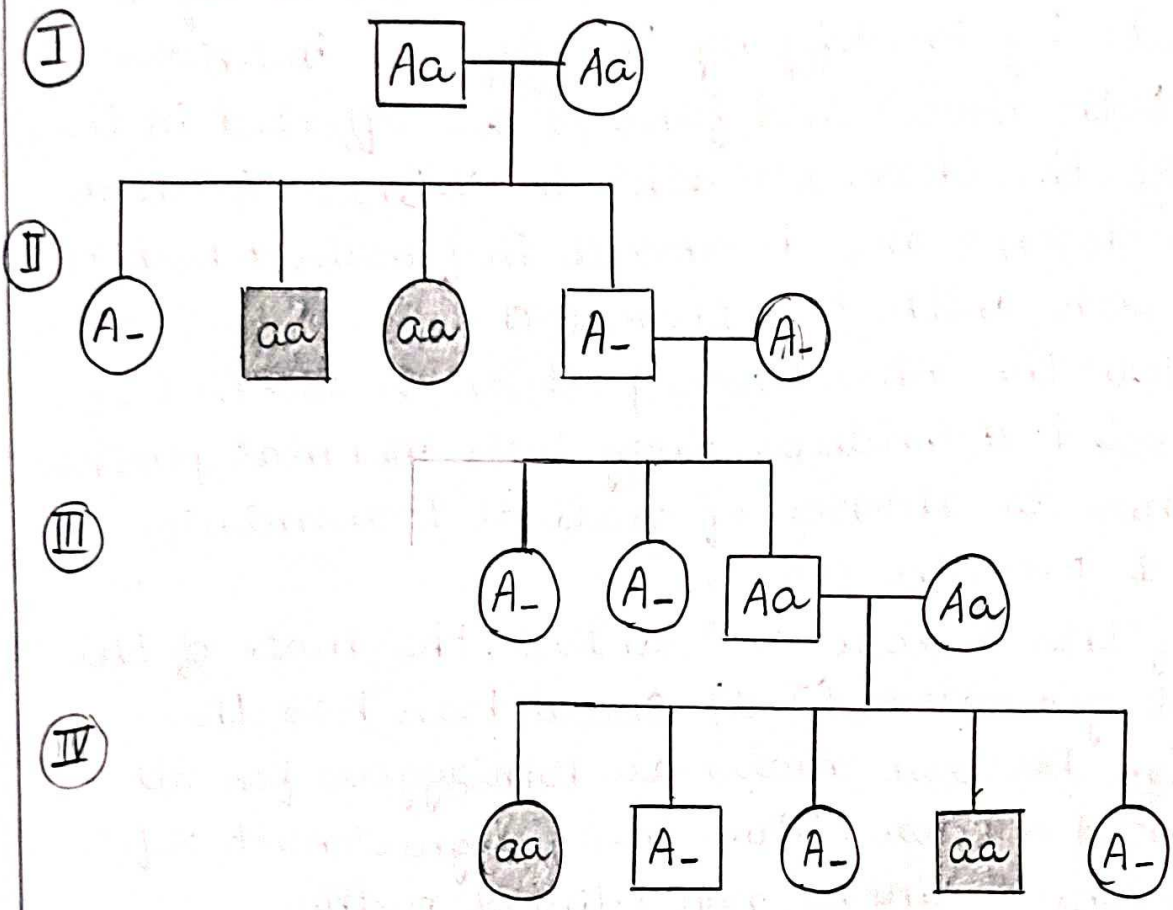


fig. Inheritance of Autosomal linked recessive traits.

having 'a'. Thus with this logic the individuals I-1, I-2, III-3, III-4 are heterozygous.

(iii) There is equal chance that either II-4 or the female with which it mated is heterozygous.

(iv) Genotypes of other individuals can be 'AA' or 'Aa' with equal probability.

Conclusion -

The analysis of the pedigree chart reveals the following —

(i) The disease or character is autosomal

(ii) The disease or character is recessive

(iii) All normal individuals have at least one dominant allele.

(iv) All affected individuals are double recessive or homozygous.

Example - Albinism in humans

3. X-Linked Dominant trait -

Observation -

(i) Since both males and females are affected the disease isn't Y-linked because females don't have this chromosome.

(ii) We observe vertical transmission indicating a dominant disease.

(iii) We find that if male is affected then all his daughters are affected and no son is affected. Again if female is affected both sons and daughters are affected this suggest that the character is X-linked because X-chromosome is the only chromosome which is transmitted from father to daughter but never father to son.

Inference -

The disease demonstrated by this chart is X-chromosome or X-linked dominant.

Identifying genotype -

(i) Since the disease is dominant and X-chromosome every affected individuals has atleast one dominant allele say 'A'.

(ii) Again all normal individuals should have no 'A'.

(iii) Since the disease is X-linked males should have one allele and females will have two allele.

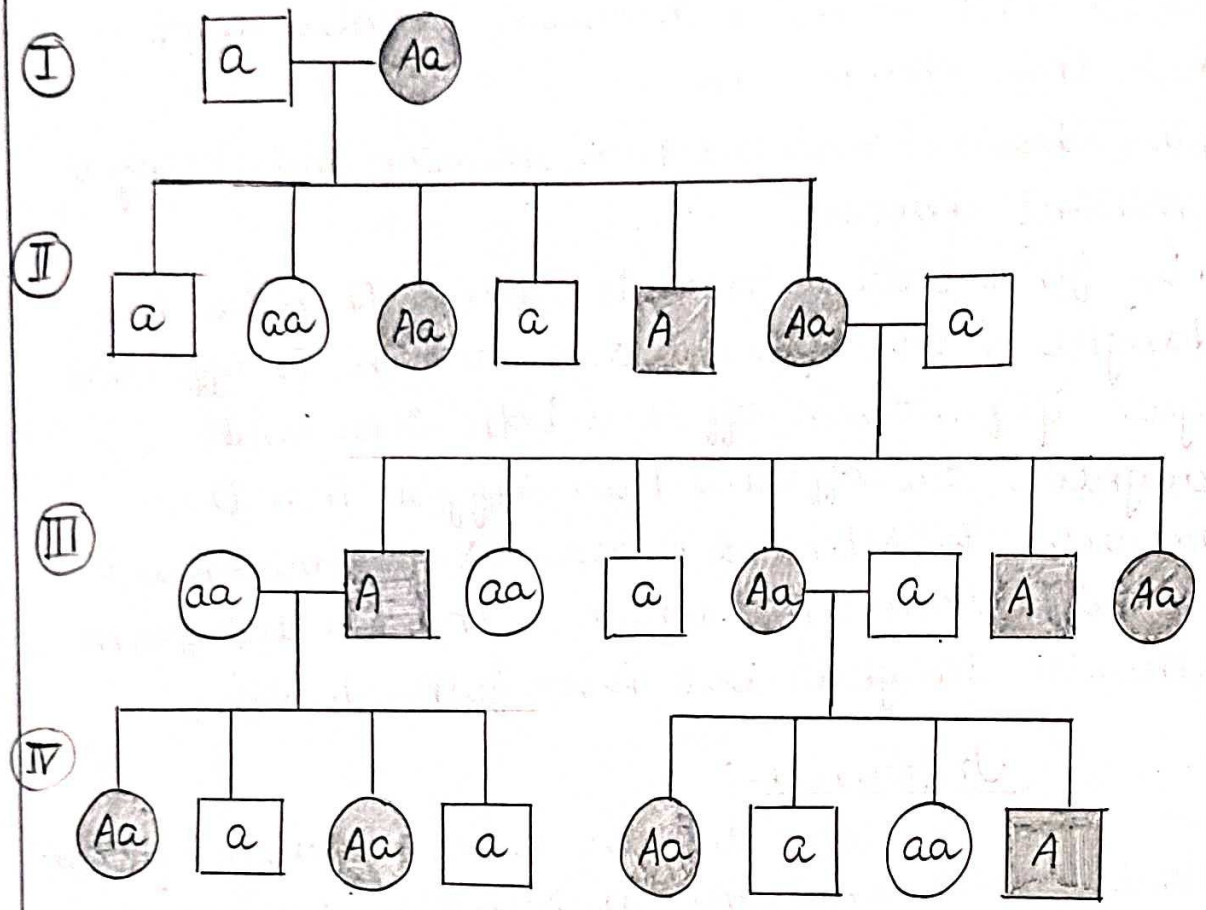


fig. Inheritance pattern of X-linked dominant trait.

(iv) By looking at inheritance pattern, since daughter and sons of affected individuals can be both affected and normal, and since all mating are between one normal and one affected individuals, all the affected females are heterozygous.

Conclusion -

The analysis of the pedigree chart reveals the following —

(i) All normal males are hemizygous
(ii) All normal females are homozygous.
(iii) All affected males are hemizygous.
(iv) All affected females are heterozygous.

Example - Bifid tongue. in human

(4) X-linked Recessive traits -

Observation -

(i) For the particular character the analysis starts by identifying the affected individuals. Both males and females are affected so the disease is either X-linked or autosomal.

(ii) Affected individuals may have normal parents so there is absence of vertical transmission so its a recessive disease.

(iii) Since males are more affected and since males are hemizygous for X-linked genes the pedigree chart indicates X-linked inheritance.

(iv) Out of 6 affected individuals there is only one affected female. Since the disease in recessive this female should be 'aa' thereby inheriting a allele from each parent. In case the disease is X-linked the male parent of the ~~offspring~~ affected female child being hemizygous, should also be affected. This is the case with the present pedigree chart. We therefore conclude that the disease is X-linked.

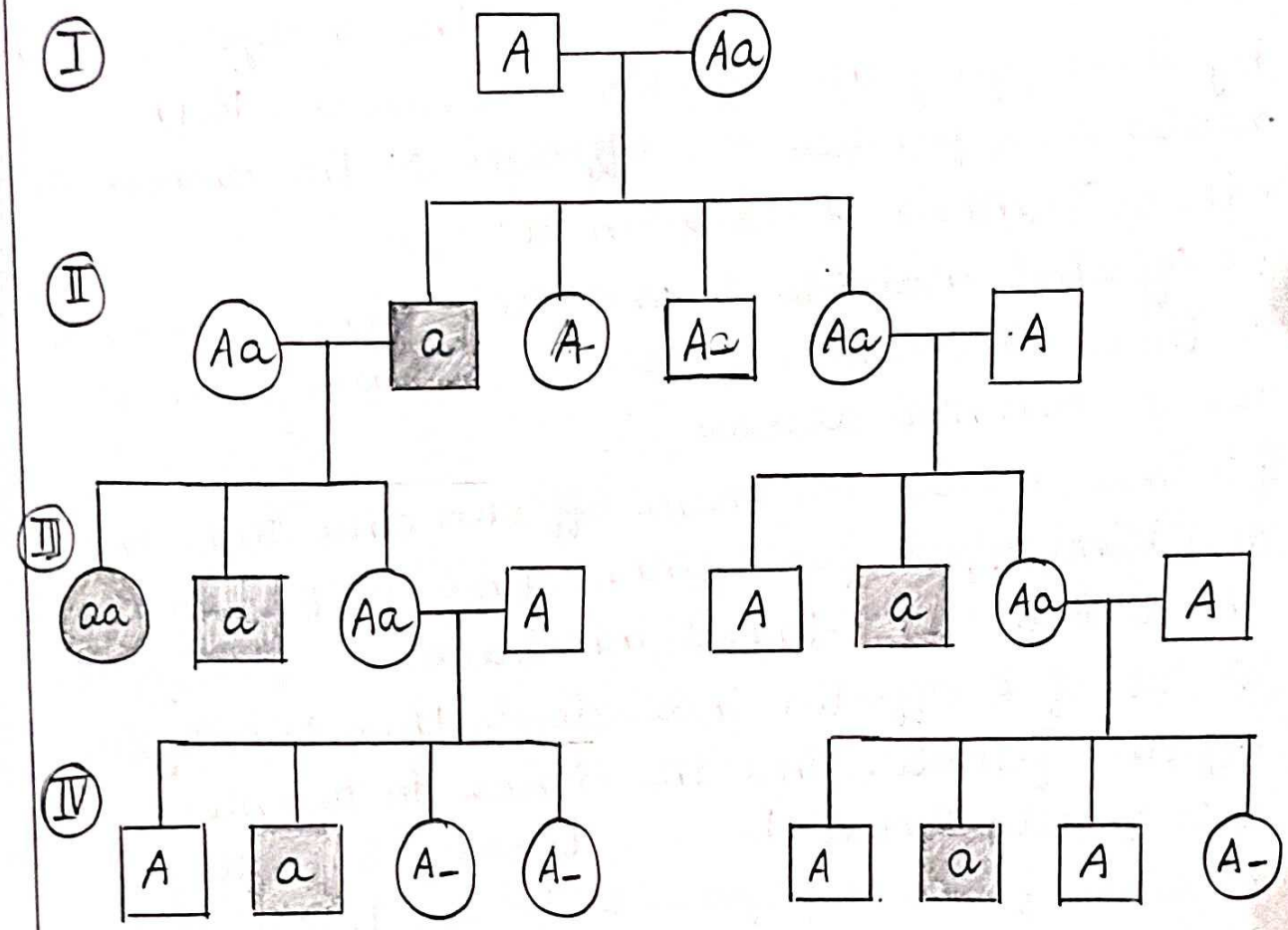


fig. Inheritance pattern of X-linked recessive trait

Identifying genotype -

(i) The disease being autosomal recessive, every normal individuals should have at least one 'A' (normal males will have 'A' while normal females may be double dominant 'AA' or carrier 'Aa').

(ii) All affected individuals are homozygous 'aa' in females and hemizygous 'a' in males.

(iii) Looking at II-1 male which is affected having 'a' being present in X-chromosome must have recessive it from mother I-2. So the first female I-2 must be a carrier (Aa) with this some logic the individuals II-4 and III-3 and III-6 (all females) should be carrier.

(iv) Since the female III-1 is affected having one 'a' from either parent, the female II-1 should be a carrier of 'a'.

(v) The genotype of other females (III-3, IV-3, IV-4, IV-8).

(vi) The genotype of other individuals females can be 'Aa' or 'AA' and we denote them as 'A_'.

Conclusion -

The disease/character identified by the pedigree chart is X-linked recessive.

(i) All affected female individuals are homozygous.
(ii) All affected male individuals are hemizygous. (recessive).
(iii) Every normal males are hemizygous.
(iv) Every normal females are double dominant or carrier.

Example - Hemophilia, colour blindness in humans.

⑤ Y-chromosome linked traits -

Observation -

(i) All affected individuals are male indicating in Y-linked inheritance.

(ii) None of the sons of the affected males are not normal.

(iii) There is no carrier individuals and offsprings of normal individuals are normal therefore the disease is Y-linked.

(iv) Since human males are heterozygous for Y-linked genes, it is always expressed in male individuals and the question of dominants doesn't arrives.

Identifying genotype -

(i) Say Y_1 is the allele cause it the diseased character and Y^2 doesn't produce the character.

(ii) So all affected males will be 'Y^1' and all normal males will be 'Y^2' in hemizygous condition.

(iii) The females being devoid of Y-chromosome have no corresponding gene/allele.

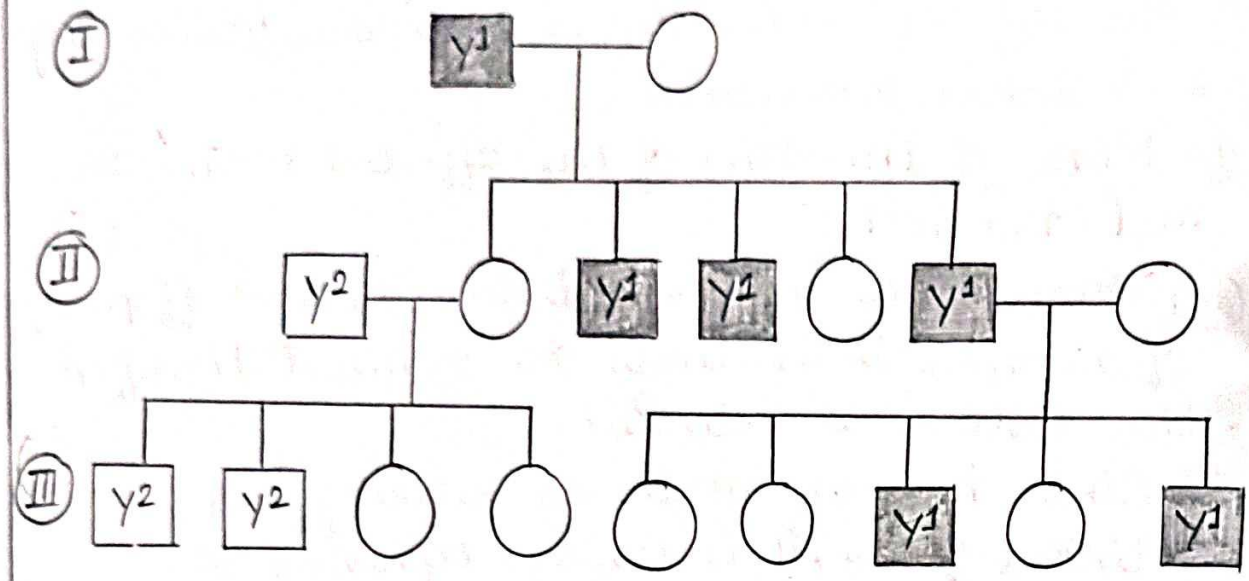


fig. Inheritance pattern of Y-linked traits.

Conclusion -

The disease/character analysing by the pedigree chart is Y-linked.

(i) All affected individuals are male 'Y^2' in hemizygous condition.

(ii) All normal males 'Y^2' in hemizygous condition.

Example - Hypertrichosis (hair in external ear in humans).

good

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