Rules for Inheritance

I. **Autosomal Recessive** → Appears in both sexes with equal frequency trait tend to skip generations.
→ Affected offspring are usually born to unaffected parents.
→ when both parents are heterozygot, approx. 1/4 of the progeny will be affected.
→ Appears more frequently among the children of consanguine marriages.

II. **Autosomal Dominant** → Appears in both sexes with equal frequency.
→ Both sexes transmit the trait to their offspring, doesn't skip generations
→ Affected offspring must have an affected parent unless they posses a new mutation.
→ when one parent is affected (heterozygot) and the other parent is unaffected, approximately 1/2 of the offspring will be affected.
→ Unaffected parents do not transmit the trait.

III. **X-Linked Dominant** → Both males and females are affected, often more females than males are affected, doesn't skip the generation.
→ Affected son must have an affected mother.
→ Affected daughter must have either an affected mother or an affected father.
→ Affected fathers will pass the trait on to all their daughters.
→ Affected mothers if heterozygous will pass the trait on to 1/2 of their sons and 1/2 of their daughters.

IV. **X-linked Recessive** → more males than females are affected.
→ Affected sons are usually born to unaffected mothers thus the trait skip generations.
⇒ Approximately 1/2 of carrier mother's sons are affected.
→ It is never passed from father to son.
→ All daughter of affected fathers are carriers.

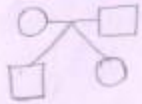
(v) Y-linked Dominant :- → only males are affected. it is passed from father to all sons. It doesn't skip generations.

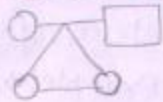
Mitochondrial :- Trait is inherited from mother only. all children of a mother are at risk to be affected or carriers.

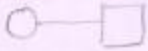
Pedigree Analysis

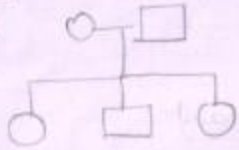
Definition:- A pedigree is a chart of the genetic history of family over several generations.

Construction:- (i) ○ Female (ii) □ Male

(iii) Fraternal twins 

(iv) Identical twins 

(v) Married couple 

(vi) Siblings 

(vii) Affected

(viii) ⊙ X-linked (colour half in)

(ix) Autosomal carries 

(x) ⊘ Deceased.

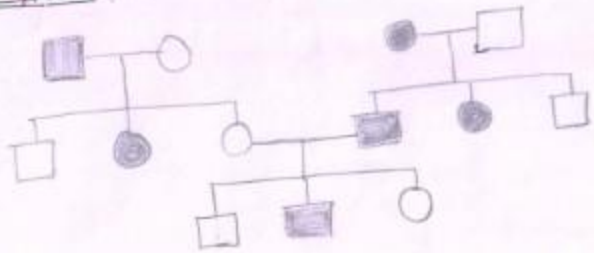
Interpreting a Pedigree chart:-

(I) Determine if the pedigree chart shows an autosomal or X-linked disease??

⇒ If most of the males in the pedigree are affected the disorder is X-linked,

⇒ If it is a 50/50 ratio between men and women the disorder is autosomal.

Example:-



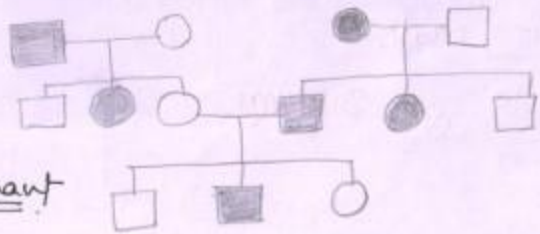
Answer:- Autosomal

(II) Determine whether the disorder is dominant or recessive.

⇒ if the disorder is dominant (one of the parents must have the disorder).

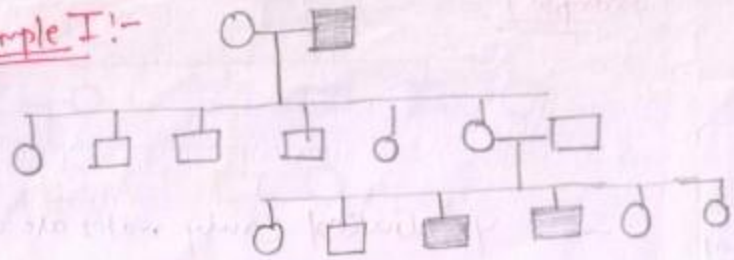
⇒ if the disorder is recessive, neither parent has to have the disorders because they can be heterozygous.

Example!→



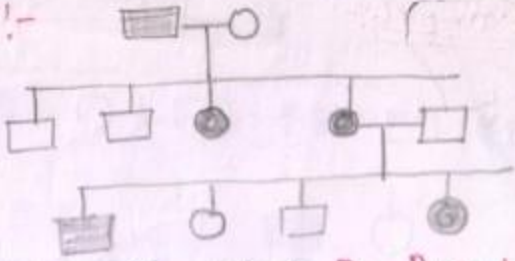
Answer :- Dominant

Example I:-



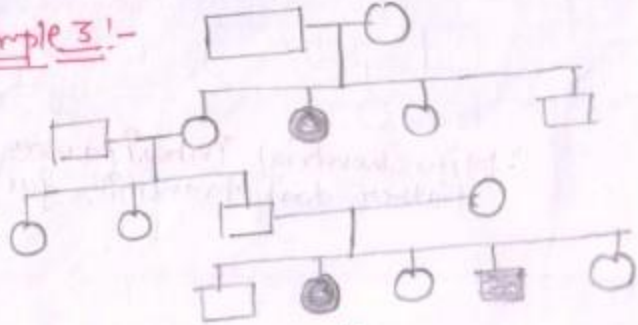
→ only males are affected.
→ Son don't share the phenotype of their father thus X-linked.
→ Expression of Hemophilia skip generation
Recessive.

Example II:-



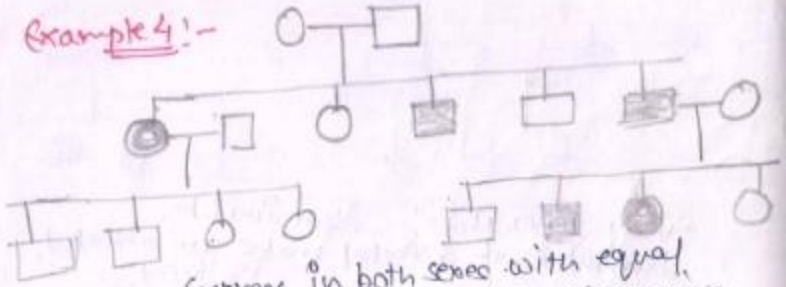
→ Every Generation:- Dominant.
→ X-linked dominant.

Example 3:-



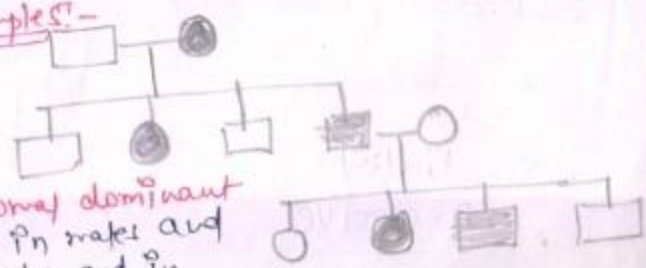
→ Autosomal recessive
(affected children from unaffected parents).

Example 4:-



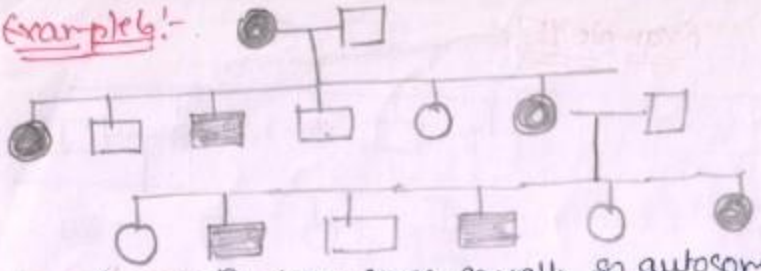
Express in both sexes with equal frequency, autosomal recessive

Example 5:-



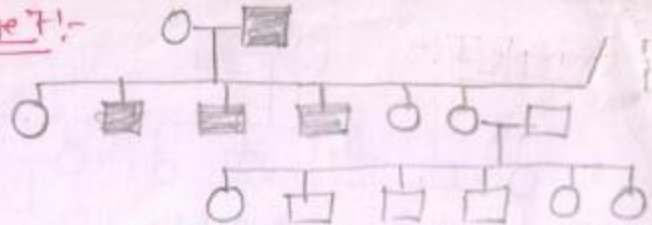
Autosomal dominant
(Equal in males and females, and in every generation).

Example 6:-



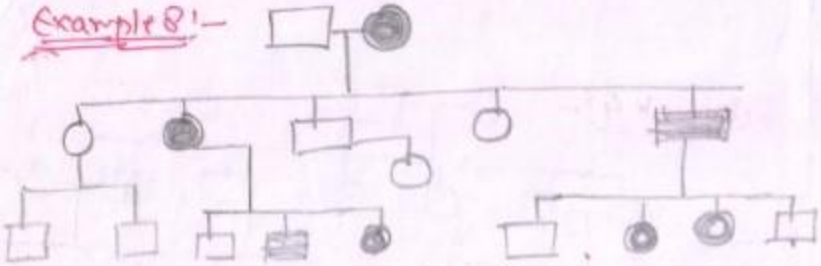
→ Appears in both sexes equally so autosomal
In every generation so dominant.

Example 7:-



Y- linked (only males are affected)

Example 8:-

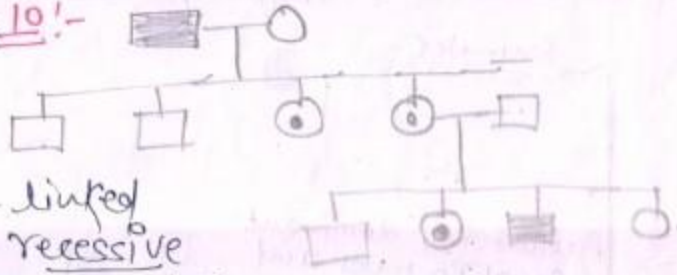


Every generation:- Dominant.
Daughter of affected males are affected,
X- linked.

Example 9:-

Mitochondrial Inheritance
Father don't transmit, just mothers

Example 10:-



X - linked
recessive

Rules of Inheritance

I. Autosomal Recessive

⇒ Appear in both sexes with equal frequency.
⇒ Trait tend to skip generation.
⇒ Affected offsprings are usually born to unaffected parents.
⇒ When both parents are heterozygot, approximately 1/4 of the progeny will be affected.

II. Autosomal Dominant

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⇒ Trait doesn't skip generation.
⇒ Affected offsprings must have an affected parent unless they posses a new mutation.
⇒ When one parent is affected (heterozygot) and the other parent is unaffected approximately 1/2 of the offspring will be affected.

III. X-linked Dominant

⇒ Both males and females are affected, often more females than males.
⇒ doesn't skip the generation.
⇒ Affected Son must have an affected mother.
⇒ Affected daughter must have either an affected mother/father.
⇒ Affected father will pass the trait on to all their daughters.
⇒ Affected mothers if heterozygous will pass the trait on to 1/2 of their sons and 1/2 of their daughters.

IV. X-linked Recessive

⇒ more males than females are affected.
⇒ Trait tend to skip generation.
⇒ Approximately 1/2 of carrier mother's sons are affected.
⇒ It is never passed from father to son.
⇒ All daughters of affected fathers are carries.

V. Y-linked dominant

→ only males are affected, it is passed from fathers to all sons.
⇒ It doesn't skip generation.

VI. Mitochondrial

→ Trait is inherited from mothers only. all children's of a mother are at a risk to be affected / carriers.

Interpreting a Pedigree chart :→

I) Determine if the pedigree chart shows an autosomal or X-linked disease?
⇒ If most of the males in the pedigree are affected the disorder is X-linked.
⇒ If it is a 50/50 ratio b/w men and woman the disorder is autosomal.

II) Determine whether the disorder is dominant or recessive?
⇒ If the disorder is dominant (one of the parents must have the disorder).
⇒ If the disorder is recessive neither parent has to have the disorder because they can be heterozygous.

Crossing over!→ Janssens (1909) was the first person to discover Chiasma formation & related process of crossing over. Morgan (1910) found phenomenon of linkage and Crossing recombination.

⇒ Crossing over is recombination of genes due to exchange of genetic material b/w two synapsed homologous chromosomes. It is mutual exchange of segments of genetic material b/w non-sister chromatids of two homologous chromosomes, so as to produce recombination of new combinations of genes.

⇒ The non-sister chromatids in which exchange of segments has occured are called recombinant or cross over while the other chromatids in which cross-over has not take place are K/a parental chromosome or non cross-over.

Characteristics of Crossing over!→ Crossing over/recombination occurs at two levels!–

1) At gross chromosomal level called chromosomal cross over,

⇒ The crossing over results basically from an exchange of genetic material b/w non sisters chromatids by break and exchange following replication.

2) At DNA level called genetic recombination.

⇒ The frequency of crossing over appears to be closely related to physical distance b/w genes on chromosomes and serves as a tool in constructing genetic maps of chromosomes.

Mechanism of crossing over!– Chromosomes get replicated in S-phase of Interphase. Chromosomes which tend to undergo recombination due to meiotic crossing over necessarily complete two functions!–

1) 99.7% replication of DNA &
2) 75% synthesis of histones, both of which take place prior to onset of Prophase I & attachment of each chromosome by its both ends (telomeres) to the nuclear envelope (i.e to nuclear lamina) via the specialised structure called attachment plaques (plaques) this event occurs during the leptotene stage of prophase I. therefore leptotene chromosomes are double stranded though the two strands are not visible due to presence of nucleoprotein complex in b/w the chromatids.

The process of crossing over comprises 4 steps!–

1) **Synapsis!–** Replicated but apparently single homologous chromosome come to lie side by side with similar gene loci of the two chromosomes exactly opposite. It occurs in the zygotene stage of prophase I. The phenomenon is called synapsis. The synapsed pair of homologous chromosomes are called bivalents. The small amount of unreplicated chromosomes (0.3%) if present also undergoes replication.

⇒ The two homologous chromosomes are held together by a Synaptonemal complex.

2) **Tetrad formation!–** The chromatids of each synapsed chromosome slightly separate & become visible in the pachytene stage of prophase I. A group of 4 homologous chromatids are called a tetrad → the cells proceeds to diplotene stage of prophase I.

3) **Exchange of Chromatids!→** At this time breakage of chromatid segments exchange of non-sister chromatid segments & later their fusion in new places occurs. The Synaptonemal complex begins to dissolve except in the region of crossing over.

⇒ The synaptonemal attachement points b/w the homologous chromosomes are called Chiasmata. The crossing over thus includes the breaking of chromatid segments their transposition & fusion.

4) **Disjunction!→** After the completion of crossing over, the synaptic forces end & the homologous chromosomes move apart by shifting chiasmata to the ends. The phenomenon is called terminalization. Many of them disappear before metaphase I.