Human Genetics

Human genetics studies are different to perform because

- Long Generation time (18-20)
- control and desired crosses are difficult to perform.

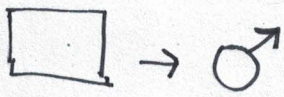
- No. of progency very few
- for many character human show polygenic inheritance.
- No. of chromosomes are very high in comparision to cell size.

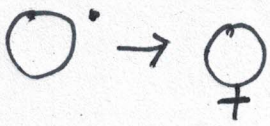
→ To study human genetics following Methods are used:-

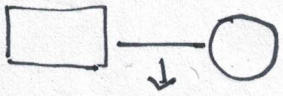
- Biochemical studies (MBBS)
- Chromosomal studies (MBBS)
- Study of Twins (MBBS)
- Cytological studies (MBBS)
- Pedigree Analysis.
- Population Genetics.

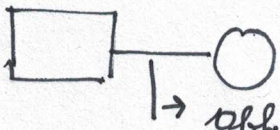
Pedigree Analysis.

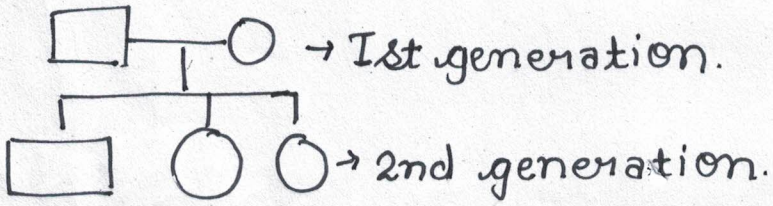
- With the help of Pedigree symbols genetic information is shown in a family chart.

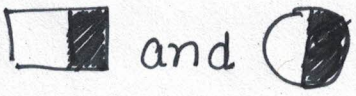
1.) 

2.) 

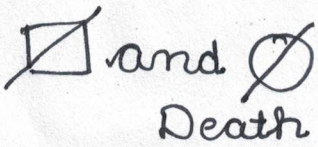
3.) 
Mating line.

4.) 
→ off spring line

5.) 
→ Ist generation.
→ 2nd generation.

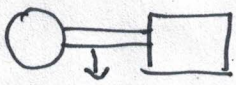
6.) 
and
Heterozygous.

7.) 
and
Disease

8.) 
and
Death

9.) 
carrier (XLR)

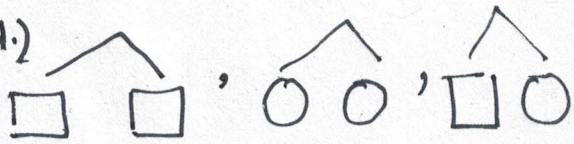
10.) 1 Abortion.

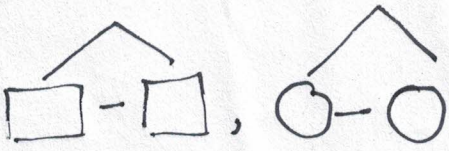
11.) 
Marriage between close relative.

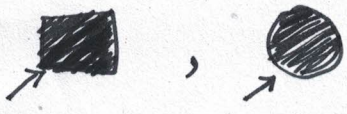
12.) Sex unspecified.

13.) 

· No. of Unaffected time.

14.) 
Dizygotic twins.

15.) 
Monozygotic twins

16.) 
Propositive
Propositive

Autosomal Recessive

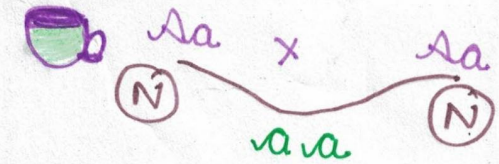
- Generally, generation skip is seen.
- If both parents are normal they can have diseased child
- If both parents are diseased they cannot have normal child.

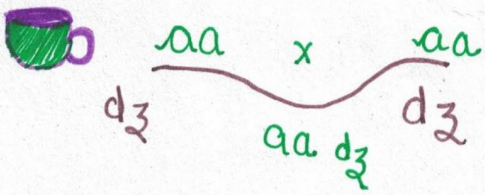
A.R.

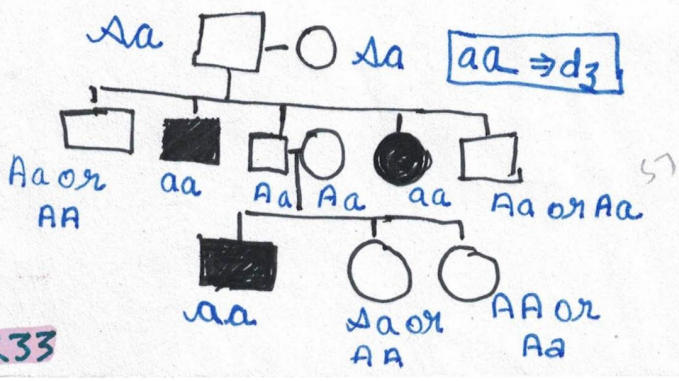
A → Normal a → disease

AA → Normal aa → disease

Aa → carrier / Normal







Autosomal Dominant

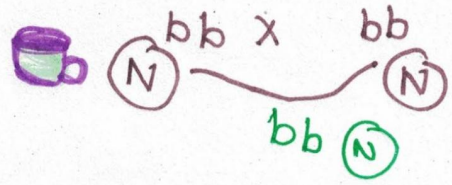
- Generally, generation skip is not seen.
- Normal parents can never have disease child.
- If both parents are diseased they can have normal child.

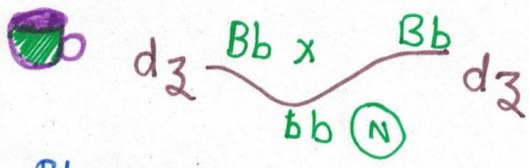
A.D.

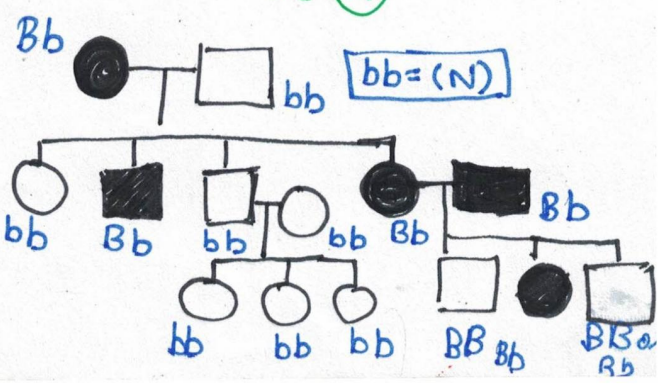
B → disease b → Normal

BB → Disease bb → Normal

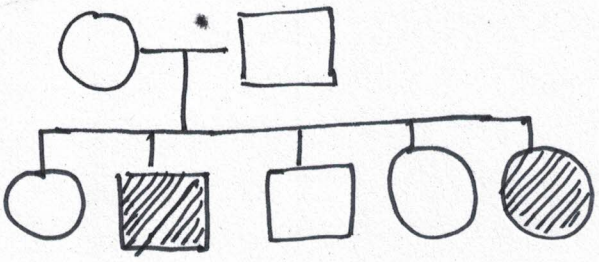
Bb → disease



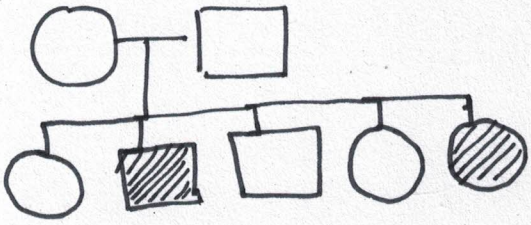


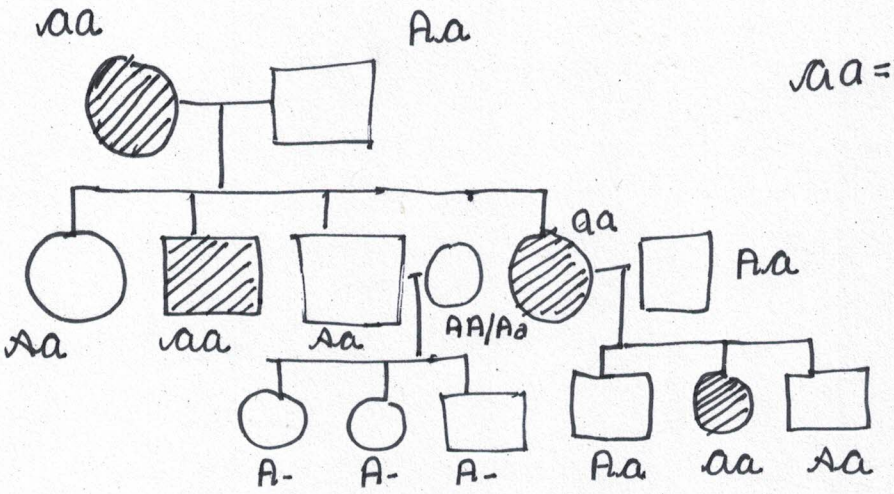


Autosomal Recessive



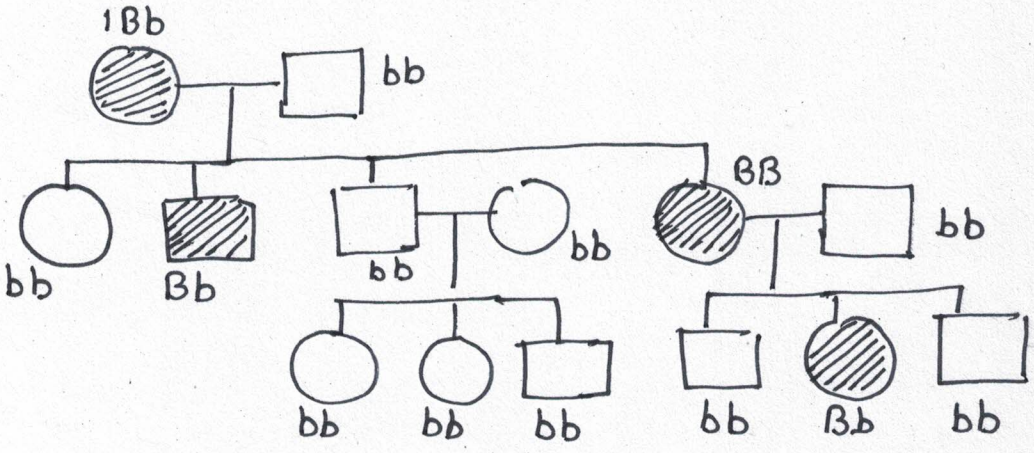
Autosomal Dominant





aa = dz → A.R.

AD



X-Link Recessive Pedigree

Acceptance से ज्यादा rejection पर जोर देंगे।

☞ A Normal father can never have diseased daughter.

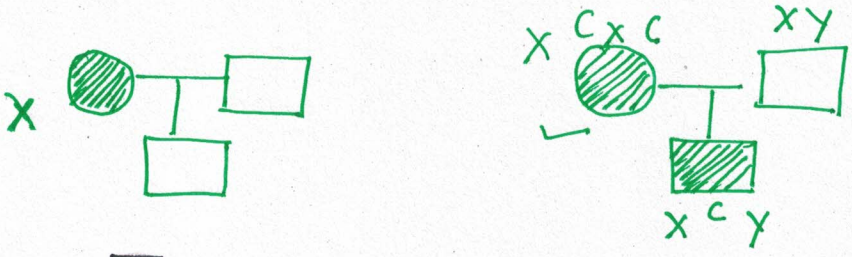
OR

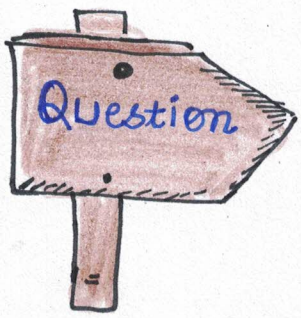
☞ If in a pedigree normal father is having diseased daughter it can never be x-link recessive.

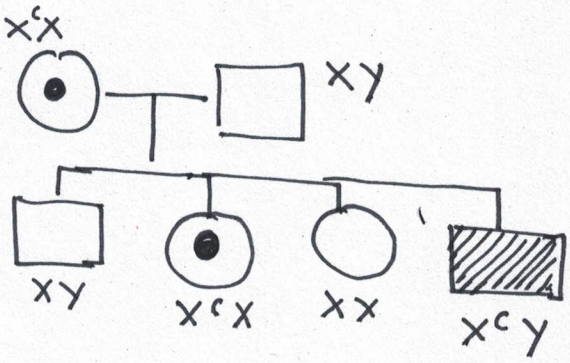
☞ A disease mother can never have normal son.

OR

☞ If in a pedigree mother is diseased than son is normal, this pedigree can never belong to X-link recessive pedigree.

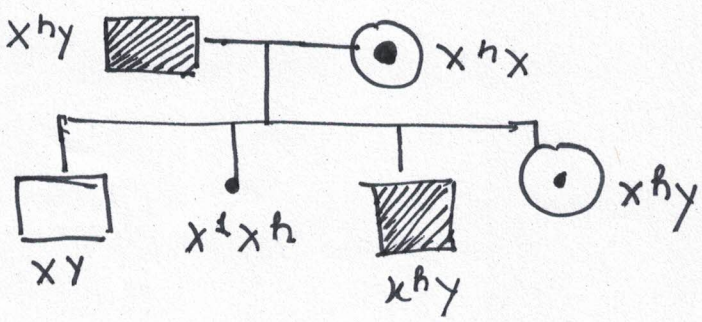






Conclusion:→ X-link Recessive

→ colourblindness, Haemophillia.



conclusion:- X-link Recessive

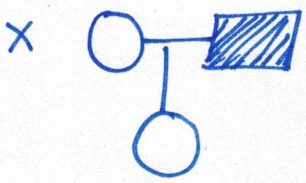
- Haemophilia.

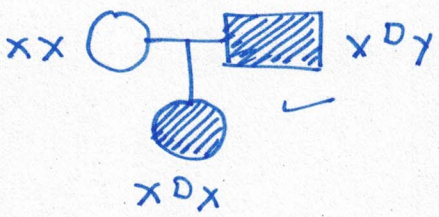
X-Link dominant

- A disease father can never have normal doughter

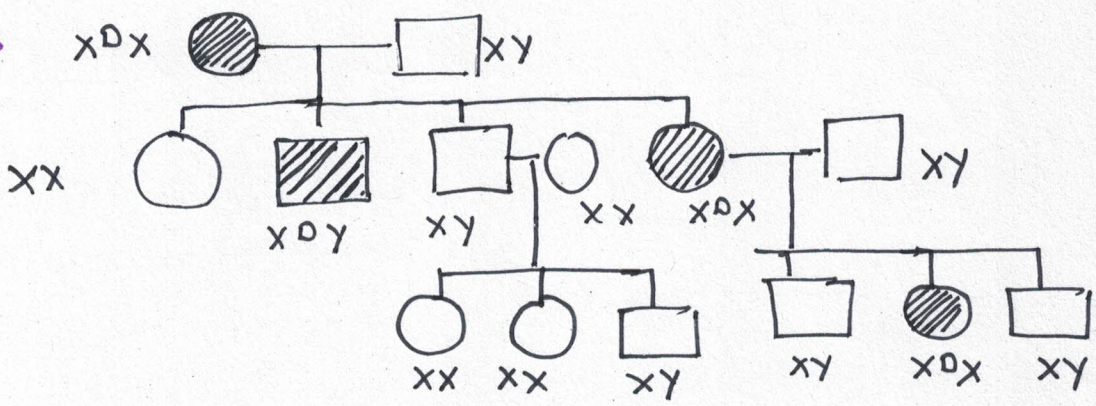
OR

- If in a pedigree father is diseased and doughter is normal then pedigree cannot belong to x-Link Dominant



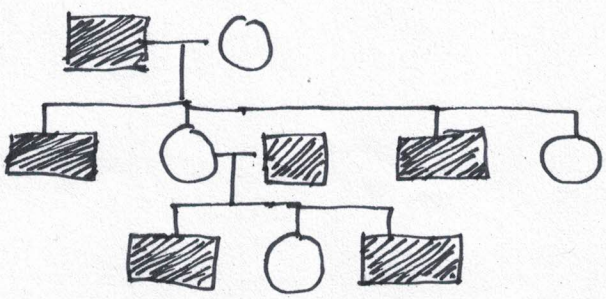


Question



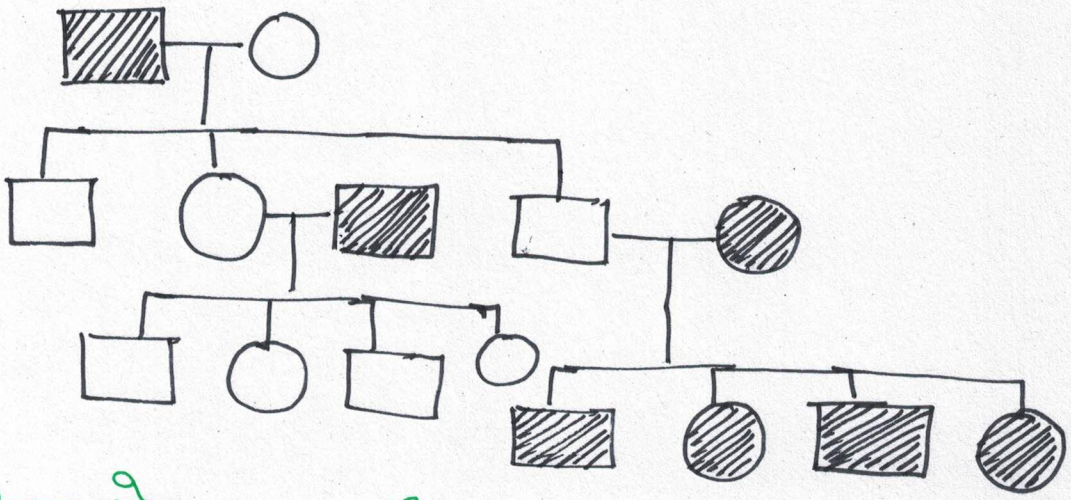
- A.R — ① Albinism / cystic fibrosis
- A.D — ② Hungthingtoris chores / Myothonin Dystrophy
- XLR — ③ C.B. / DHD
- XLD — ④ Pseudorickets

Y-Link Pedigree



All Males will be diseased.

Cytoplasmic Inheritance.



मम्मी बीमार तो खानदान बीमार, पापा बीमार तो बच्चे होशियार।

Order of Preference

- Y-Link
- cytoplasmic
- A-R/A-D
- XLR/XLD

Autosomal Recessive

- Blue eyes
- Straight hairs
- Small Eyes
- Normal cheeks
- fused Ear lobe
- Non tongue roller

Autosomal Dominant

- Black or Brown eyes
- curly hairs
- Large Eyes
- Dimple in cheek.
- free Ear lobe
- Tongue roller

- PTC Non taster
- Left handness
- Rh ⊖
- Albino

- PTC tastes
- Right handedness.
- Rh ⊕
- Nigro.

Two Right handed ~~pepeo~~ people are having left handed son, what is the genotype of parent ?

① RR×RR ② R$_r$×RR ③ rr×rr ④ Rr×Rr

Population Genetics

Hardy Weinberg while studying gene frequency in a population they reached to a conclusion that in case of ideal population gene frequency remain same from one generation to another.

Criteria for Ideal Population:

- Large population.
- No migration.
- No mutation.
- No natural Selection.
- Random Mating.

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Genetic Drift

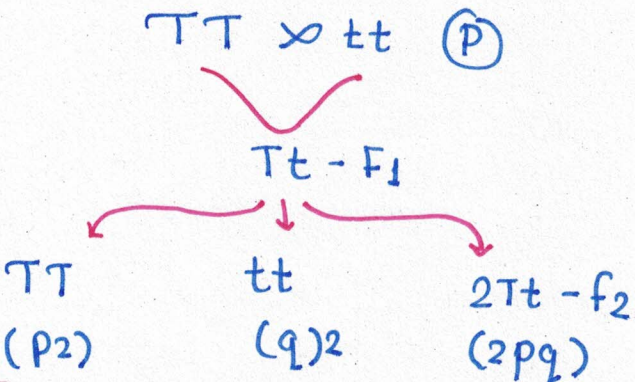
If genetic drift is seen in ideal population then sampling Error must have taken place.

★ Large sample size, small Error

★ According to Hardy Weinberg ★

$p+q=1$ $\qquad$ $p^2+q^2+2pq=1$

- p = frequency of dominant Allele.
- q = frequency of Recessive Allele.
- p^2 = frequency of homozygous dominant phenotype.
- q^2 = frequency of Recessive homozygous phenotype.
- $2pq$ = frequency of dominent heterozygous phenotype.
- p^2+2pq = frequency of dominent phenotype.



Question frequency of Albinism in a population is 25%. what is the frequency of recessive allele.

Albinism → recessive trait → occurs in homozygous state i.e → $q^2 = 25\%$. $\quad q^2 = 25/100 \rightarrow q = 5/10 \Rightarrow q = 0.5$

Question: $q^2 = 81\%$, what is the value of q.

$q^2 = 81/100$, $\Rightarrow q = 9/10 \Rightarrow q = 0.9$

Question: In an Ideal population following Hardy Weinberg Law 3600 people are suffering from albinism out of 10,000 of total population, calculate frequency of recessive Allele.

$\Rightarrow q^2 = \frac{3600}{10000} \Rightarrow q = 6/10 = q = 0.6$

Question 16% people are suffering from Albinism what is the % age of people in carrier state.

$\Rightarrow$ carrier state $\rightarrow 2pq$

Given $q^2 = 16/100 \rightarrow q = 4/10 = q = 0.4$

$p+q = 1 \rightarrow p + 0.4 = 1 \Rightarrow p = 0.6$

$2pq = 2 \times 0.4 \times 0.6$

$= .48$

Question find out the number of Heterozygores free Earlobe person in a population of 3000 of the proportion of fused Ear lobe person is 9%.

$\Rightarrow$ Heterozygous $\rightarrow 2pq$

$q^2 = 9/1000 \rightarrow q = 3/10$, $q = 0.3$

$p+q = 1 \rightarrow p + 0.3 = 1 \rightarrow p = 0.7$

$2pq = 2 \times 0.7 \times 0.3 = .42$

$3000 \times 42/100 = 1260.$

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Question If frequency of recessive phenotype is 9% then find out the no. of homozygous organism in population of 50,000.

$\Rightarrow q^2 = 9/100 \rightarrow q = 3/10 \rightarrow q = 0.3$

$p + q = 1 \Rightarrow p + 0.3 = 1 \Rightarrow p = 0.7$

$p^2 + q^2 = (0.3)^2 + (0.7)^2$

$= 0.09 + 0.49$

$= .58$

$50000 \times 58/100 \rightarrow 29,000.$