Handwritten Notes

On

Pedigree Analysis

There are 6 case in Pedigree Analysis –

Autosomal

Autosomal Recessive disease (AR)

A — Dominat (Normal)

a — Recessive (Disease)

AA → Normal
Aa → Normal but Carrier
aa → Affected

↳ It is used in both ♂ and ♀ due to Autosome.

Eg: Sickle Cell Anaemia
Thalassemia
Phenylketonuria
Alkaptonuria
Albinism

Autosomal Dominant disease (AD)

A — Dominant (Disease)

a — Recessive (Normal)

AA → affected
Aa → affected
aa → Normal

↳ It is also used in both ♂ and ♀

Eg: Huntington's chorea.
**Myotonic dystrophy.
Polydactyly.

X-Linked

X-Linked Recessive (XR)

X^+ — Dominant

X^a — Recessive (Disease)

♀
X^+X^+ = Normal
X^+X^a = Normal but Carrier
X^aX^a = affected

♂
X^+Y = Normal
X^aY = affected

eg: Haemophilia
Colourblindness
G6PD
DMD (Duchenne muscular dystrophy)

X-Linked Dominant (XD)

X^+ — Dominant (Disease)

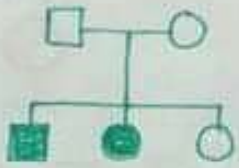
X^a — Recessive

♀
X^+X^+ = affected
X^+X^a = affected
X^aX^a = Normal

♂
X^+Y = affected
X^aY = Normal

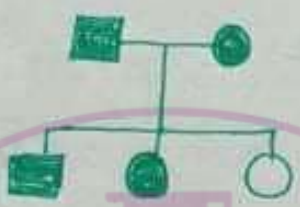
eg: Pseudorickets (Vitamin D resistance rickets)
Defective enamels of teeth

Step 1 :- If both parents are normal and any one child affected than AD and XD do not possible.



~~AD~~
~~XD~~

Step 2 :- If both parents are affected and any one child normal than AR and XR do not possible.



~~AR~~
~~XR~~

** If Step 1 and 2 do not apply than AR and AD possible.

Step 3 :-

XR doesn't possible

- Father normal daughter affected.
- Mother affected son normal.

** If Step 3 does not apply than XR possible.

Step 4 :-

XD doesn't possible

- Father affected daughter normal
- Mother normal son affected

** If Step 4 does not apply than XD possible

● → Affected female

■ → Affected male

◧ ◑ → Heterozygous/Carrier for autosomal reccesive disease

⊙ → Carrier female of sex linked recessive disease

◇ → Sex unspecified

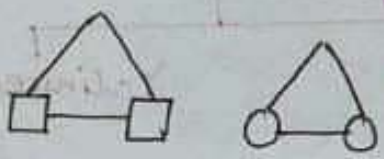
(5) → five unaffected offspring's

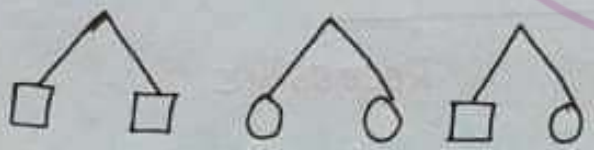
☒ ∅ → Death of individual

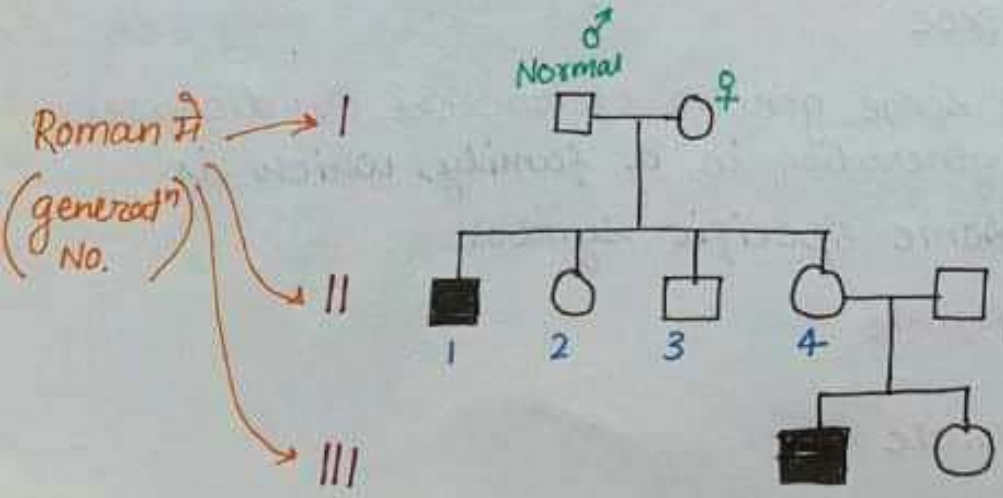
| → abortion or still birth
(death during embryonic stage)

□—○ → Mating (marriage)

□═○ → Consanguineous marriage
(Marriage b/w close relatives)

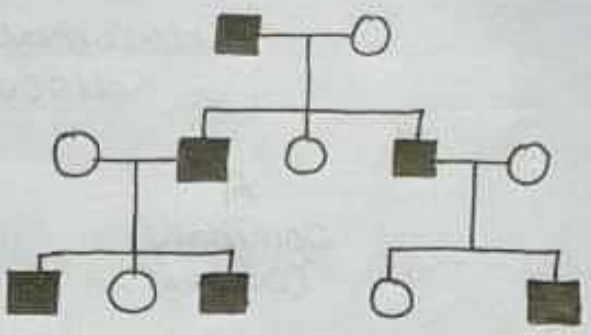
 → Monozygotic twins

 → Dizygotic twins
(fraternal)



*** Y-linked :-

If father affected than all son should be affected.

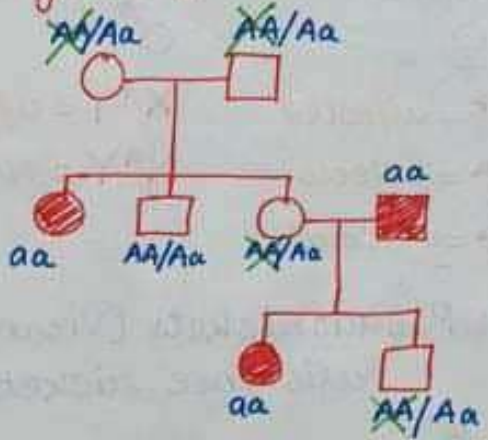


*** Cytoplasmic inheritance/ Maternal inheritance :-

↳ If mother affected than all offspring's should be affected.

↳ If father affected than any offspring's is never affected.

Q. Given pedigree show the inheritance of alkaptonewia. What will be the genotype of all members of given pedigree ?



Alkaptonewia :-

Autosomal recessive disease.

AA :- Normal

Aa :- Normal but Carrier

aa :- affected.