Handwritten Notes

On

Pedigree Analysis
There are 6 cases in Pedigree Analysis:

**Autosomal**

- **Autosomal Recessive disease (AR)**
  - Dominant (Normal): $A$
  - Recessive (Disease): $a$
  - $AA \rightarrow$ Normal
  - $Aa \rightarrow$ Normal but Carrier
  - $aa \rightarrow$ Affected

  - It is used in both $\sigma$ and $\varphi$ due to Autosomal.
  - Eg: Sickle Cell Anaemia
  - Thalassemia
  - Phenylketonuria
  - Alkaptonuria
  - Albinism

- **Autosomal Dominant disease (AD)**
  - Dominant (Disease): $A$
  - Recessive (Normal): $a$
  - $AA \rightarrow$ Affected
  - $Aa \rightarrow$ Affected
  - $aa \rightarrow$ Normal

  - It is also used in both $\sigma$ and $\varphi$.
  - Eg: Huntington's Chorea.
  - Myotonic dystrophy.
  - Polydactyly.

**X-Linked**

- **X-Linked Recessive (XR)**
  - Dominant: $X^+$
  - Recessive (Disease): $X^a$
  - $X^+X^+ = $ Normal
  - $X^+X^a = $ Normal but Carrier
  - $X^aX^a = $ Affected

  - Eg: Haemophilia
  - Colourblindness
  - G6PD
  - DMD (Duchenne muscular dystrophy)

- **X-Linked Dominant (XD)**
  - Dominant (Disease): $X^+$
  - Recessive: $X^a$
  - $X^+Y = $ Affected
  - $X^+X^a = $ Affected
  - $X^aX^a = $ 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.

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

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

Step 3:—
- Father normal daughter affected.
- XR doesn’t possible

** If step 3 does not apply than XR possible—

Step 4:—
- Father affected daughter normal
- XD doesn’t possible

** If step 4 does not apply than XD possible
- Affected female
- Affected male
- Heterozygous/Carrier for autosomal recessive disease
- Carrier female of sex linked recessive disease
- Sex unspecified
- 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)

Roman m (generations)
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.

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

Alkaptonuria:
Autosomal recessive disease.

<table>
<thead>
<tr>
<th>Genotype</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>AA</td>
<td>Normal</td>
</tr>
<tr>
<td>Aa</td>
<td>Normal but carrier</td>
</tr>
<tr>
<td>aa</td>
<td>Affected</td>
</tr>
</tbody>
</table>