Basic Tool in human genetics:

① Family study – Upto 3/4 generation, character is studied

Technically we make a pedigree chart and then analyse the pattern of inheritance by observing on traits

Family study
↓
"Pedigree Chart"
↓
"Analysis of inherited traits"
↓
Pattern of inheritance.

Pedigree chart: A pedigree chart is a chart depicting biological relationships of the members of the families ~~through generation~~ and the inheritance of the selected traits through generations.

Through pedigree we can know –
- mutual relationship b/w family members.
- Pattern of inheritance of particular trait.

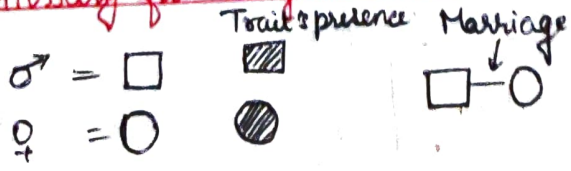
How to prepare a pedigree –

① Identify a family (through a "proband" for inherited disorders)
↳ first person who have been presented to us having this trait.
↓
generally used in contst of disorders

② Collect the information from the family regarding relationship of members and ~~traits~~ presence of traits.

③ Make the pedigree chart and in consultation with family ensures that it is correct in information.

Glossary for Pedigree Chart –



Generations I

II

III

Proband (first patient encountered to clinic for the disease diagnosis)

Roman numeral gives generation (I, II, III)

Arabic numeral gives offspring sequence (1, 2, 3, 4, 5)

- Death

- Sex not specified — diamond shape (if any foetus is aborted) without knowing it's sex

- Consultant — Unaffected and he/she brought the proband to clinic

Proband

- → Carrier for autosomal recessive (This is required for analysis only but not for making the pedigree)

- → Carrier for sex linked recessive

- → Consanguineous marriage.

- Adopted i.e. given out to other family in adoption.

- Adopted in family from outside not their own child

- monozygotic Twins

- dizygotic Twins.

- Spontaneous abortion (without any disease)

- Termination of pregnancy

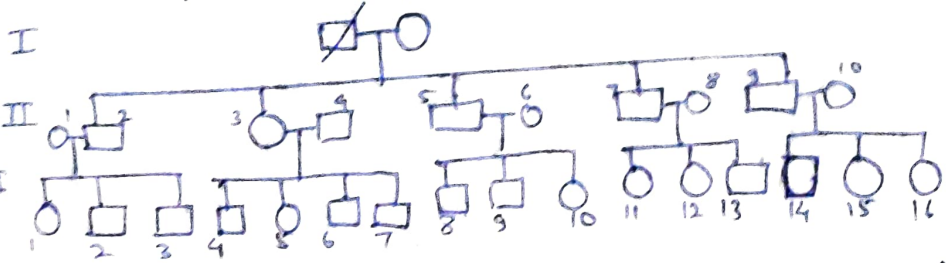
- Spontaneous abortion (if family has any disease)

- induced abortion

- Pregnancy (sex unknown of foetus)

- Pedigree with multiple disease.

→ Total confidentiality of a family has to be maintained.



Amphitreatre pedigree — → semicircle / circle

Gene → anything which produces transcription product

Patterns of Inheritance :-

Broadly divided into 2 types.

1) Autosomal inheritance
2) Sex-linked inheritance.

In this type of inheritance, there is no linkage of with sex-chrom., some but having difference in expression with reference to sex of individuals. These are of 2 types = 1. sex-limited
2. sex-influenced.

3) Mitochondrial inheritance.

If mutation has occured in such an allele that even its single copy is sufficient for ~~no~~ expressing this mutation's effect then it is called as dominant mutation. If mutation has occured in an allele & expresses itself only in homozygous mutant cond. then it is c/a recessive mutation.

NOTE: Gain of function - of new function
Restoration of function.

Generally, in mutation, wild type fn is either reduced or lost.

Basic 4 patterns -

	Dominant	Recessive
Autosomal	① Autosomal dominant	② Autosomal Recessive
Sex-linked or X-linked	③ Sex-linked dominant	④ Sex-linked Recessive

⑤ - Y-linked
⑥ Mit. inheritance.

A majority part of Y-chromosome is heterochromatic. There are only a few genes ~~@~~ on it & are functional.

NOTE:

Disease - Very well defined phenotype.
Disorder - One or more gene involved. Several effects phenotypically but no relation b/w them could be established.
Syndrome - One or more gene involved. Several unrelated phenotypes.

All 6 above given, follow MENDELIAN PATTERN OF INHERITANCE

① If we are able to find dominance & recessiveness.
② If it shows independent assortment in pedigree.

Then we can say that such a trait is following the Mendelian Pattern of Inheritance.

eg Hutington Disease (autosomal dominant) & Haemophilia & Thallesemia.