# Chapter 5: Human Genetics

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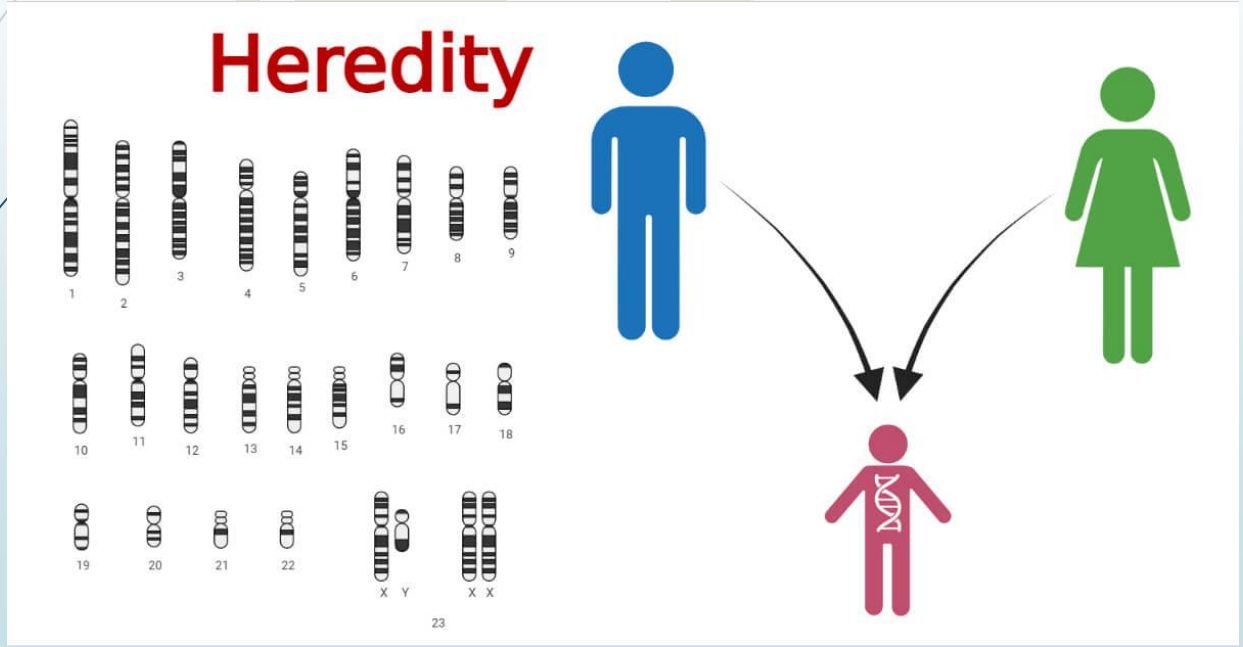
- Document 3: Sex-linked Diseases
- Document 4: Chromosomal Abnormalities
- Document 5: Prenatal Diagnosis

# Document 1

## Inheritance of Genetic Traits



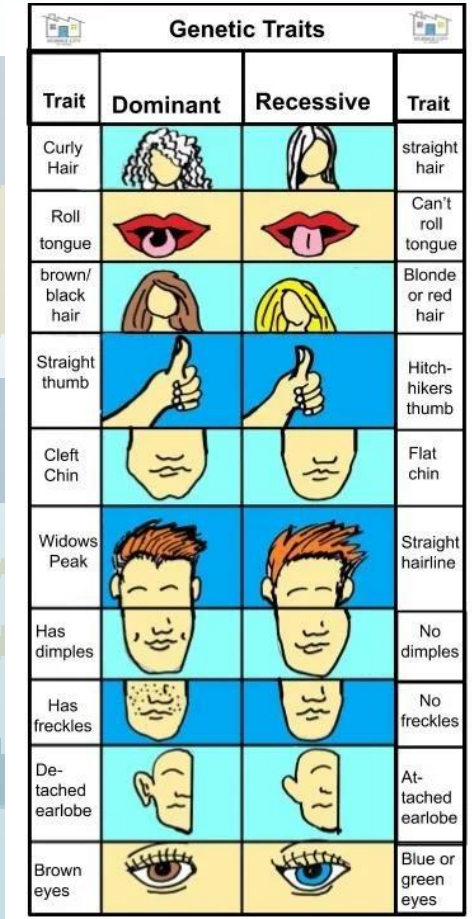
# I. Normal and Pathological Hereditary traits

- **In humans, traits could be**:

- **Morphological:** ex: height, hair color..
- **Physiological:** ex: synthesis of structural or functional proteins (hemoglobin, insulin, muscle protein dystrophin..)
- **Pathological:** ex: synthesis of abnormal proteins or non-functional proteins

- For each trait, there exist many different phenotypes:

→ example:



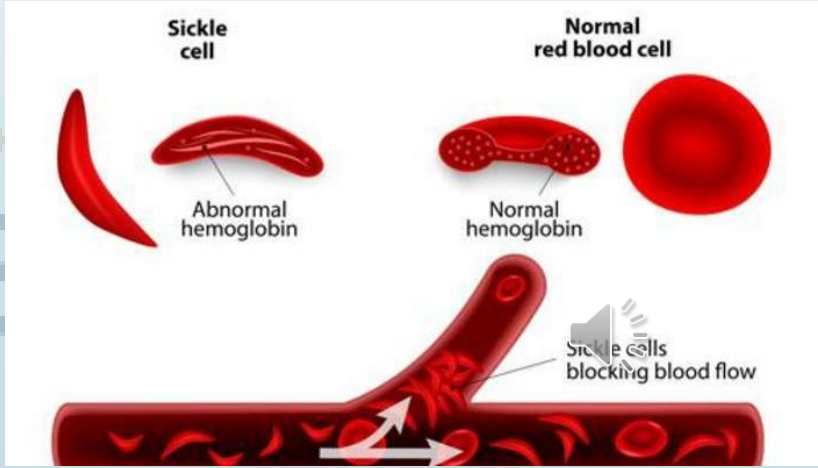
| Genetic Traits | | | |
|---|---|---|---|
| Trait | Dominant | Recessive | Trait |
| Curly Hair | | | straight hair |
| Roll tongue | | | Can't roll tongue |
| brown/ black hair | | | Blonde or red hair |
| Straight thumb | | | Hitch-hikers thumb |
| Cleft Chin | | | Flat chin |
| Widows Peak | | | Straight hairline |
| Has dimples | | | No dimples |
| Has freckles | | | No freckles |
| De-tached earlobe | | | At-tached earlobe |
| Brown eyes | | | Blue or green eyes |

Therefore, many different phenotypes exist for each trait. For example:

–The skin is more or less pigmented in different individuals; albinos lack pigmentation.

–A healthy individual has a normal hemoglobin, whereas individuals having thalassemia or sickle cell anemia have an abnormal hemoglobin.

– A healthy individual has dystrophin which is a protein normally bound to the muscle membrane, whereas it is absent in individuals suffering from Duchenne muscular dystrophy.



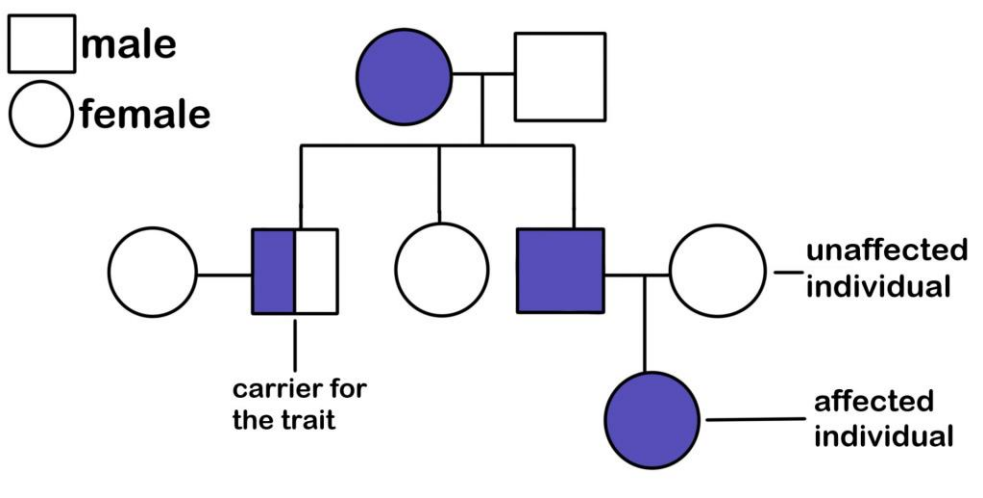
# II. Constructing a pedigree

- Pedigree: Is a chart of the genetic history of family over generations.
- **The study of a pedigree allows to determine**:

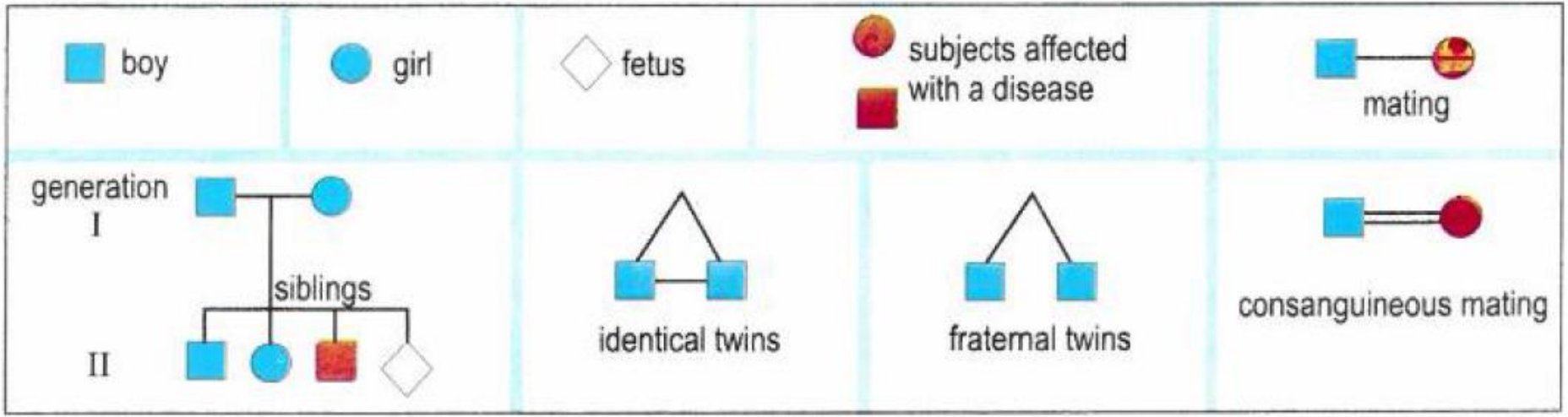
- The mode of transmission of a disease or trait (dominant, recessive..)

- The possible genotypes of the family members.

- The risk of a couple to have an affected child.




Be Smart ACADEMY




***Doc.e*** *Symbols used to construct a pedigree. I, II... are used for generations and 1, 2, 3... for the individuals of the same generation.*