

Patterns of Inheritance and Variance

Some scientists and their contributions:-

1. Reginald C. Punnett

-Punnett square.

2. Sutton and Boveri

-Chromosomal Theory of Inheritance (CTI).

-from parallel behavior.

3. Henking

-Discovered X-bodies.

4. Thomas Hunt Morgan

-Experimentally verified CTI.

-subject of experimentation: fruit fly- *Drosophila melanogaster*.

5. deVries, Correns and vonTschermak

-independently rediscovered Mendel's work.

6. Sturtevant

-mapped chromosomes in 1911.

-used recombination frequency.

Mendelian Diseases:-

a) Recessive Allele:

1. Haemophilia (or Bleeder's Disease)

- gene on X-chromosome.
- Queen Victoria.
- single protein affected which initiates a cascade.
- much increased clotting time.

2. Sickle Cell Anemia

- allele commonly called HbS.
- gene on chromosome 11.
- valine instead of glutamic acid at the 6th position of the beta globin chain of the haemoglobin molecule.
- undergoes polymerization under low oxygen conditions.

3. Phenylketonuria

- gene on chromosome 12.
- “inborn error of metabolism.”
- patient lacks enzyme to convert phenylalanine to tryosine.
- accumulates and gets partly converted to phenylpyruvic acid.
- causes mental retardation.
- name comes from passing out in urine due to poor absorption by kidney.

4. Thalassemia

- Beta on chromosome 11.
- Alpha on chromosome 16.
- abnormal haemoglobins form, resulting in anemia.

5. Alkaptonuria

- gene on chromosome 3.
- inability to metabolise tryosine.
- alkapton() accumulates and is passed down urine.
- urine turns black when exposed to air.

6. *Color blindness*

-on X chromosome.

7. *Gaucher's Disease*

-gene on chromosome 1.

-defect in lipid storage and metabolism.

8. *Cystic Fibrosis*

-gene on chromosome 7.

-lung infections due to excess mucus secretion.

b) Dominant Allele:

1. *Huntington's Chorea*

-gene on chromosome 4.

-abnormal involuntary jerky writhing movements.

-late onset disease. (35-44 years usually.)

2. *Myotonic Dystrophy*

-gene on chromosome 19.

-gradual degeneration of muscles.