TOPIC 3.4: MODES OF INHERITANCE

Principles of Inheritance

Gregor Mendel established the principles of inheritance via experimentation (he crossed large numbers of pea plants)

His findings pioneered current scientific understanding:

- Organisms have heritable factors (genes)
- Parents contribute equally to inheritance by supplying one version of the gene each (alleles)
- Gametes contain only one allele of each gene (haploid)
- Fusion of gametes results in zygotes with two alleles of each gene (diploid)

It is now known that the separation of the two alleles of each gene into separate haploid gametes occurs via meiosis

Genotype versus Phenotype

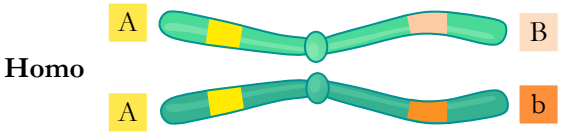
A **genotype** is the allele combination for a specific trait

There are three possible types of allele combinations:

- *Homozygous* – Both alleles are the same (e.g. AA)
- *Heterozygous* – Alleles are different (e.g. Aa)
- *Hemizygous* – Only one allele (e.g. X/Y genes in males)

A **phenotype** is the physical expression of a specific trait

- It is determined by genotype *and* environmental factors



Modes of Inheritance

Complete Dominance

One allele is expressed over another

- Dominant allele is expressed in heterozygote (capital letter)
- Recessive allele is masked in heterozygote (lower case letter)

A recessive phenotype can only be expressed in homozygotes

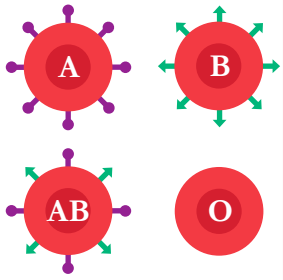
- Heterozygotes will display the dominant phenotype

Phenotype	Black	Black	Brown
Genotype	BB	Bb	bb

Codominance

Both alleles are equally expressed in the phenotype

- Heterozygotes have a distinct phenotype (superscript letter)
- An example of codominance is the ABO blood system

Blood Type	Genotype	Phenotypes
A	$I^A I^A$ *or* $I^A i$	
B	$I^B I^B$ *or* $I^B i$	
AB	$I^A I^B$	
O	i i	

Genetic Diseases

Genetic diseases can be due to recessive, dominant or codominant alleles

- Recessive conditions are most common, as heterozygotes are carriers

Autosomal Recessive

- Cystic fibrosis is caused by a mutated CFTR gene (chromosome 7)
- Produces thick mucus that clogs airways and causes respiratory issues

Autosomal Dominant

- Huntington's disease is caused by a mutated HTT gene (chromosome 4)
- An amplification of CAG repeats (>40) leads to neurodegeneration

Autosomal Codominant

- Sickle cell anemia is caused by a mutated HBB gene (chromosome 11)
- Sickling of blood cells leads to anemia and other complications

Radiation Exposure

Radiation and mutagenic chemicals increase mutation rates and can cause genetic diseases

- Most genetic diseases in humans are rare

Two examples of radiation exposure are:

- Nuclear bombing of Hiroshima (1945)
- Accident / meltdown in Chernobyl (1986)

Some long-term consequences included:

- An increased incidence of cancer
- Reduced immunity (⬇ T cell count)
- Congenital abnormalities (Chernobyl only)
- A variety of organ-specific health effects (e.g. liver cirrhosis, cataract induction, etc)