

INTRODUCTION TO GENETICS

- ◆ Genetics is the study of heredity, that is, how characteristics such as eye colour are inherited from parents to offspring.
- ◆ Genes are the chemicals in the nuclei of cells that determine the characteristics that are inherited. Each human cell has thousands of genes in the nucleus. Genes are made of DNA (deoxyribonucleic acid).
- ◆ Chromosomes are fine thread-like structures in the nucleus of all body cells. Genes are situated on chromosomes.
- ◆ Numbers of Human Chromosomes – Human body cells (somatic cells) have 23 pairs or 46 chromosomes. Human gametes (sperm and egg) have only 23 chromosomes.
- ◆ Homologous Chromosomes – Homologous chromosomes is the name given to the pairs of chromosomes in body cells.
- ◆ Human Sex Chromosomes – Of the 46 chromosomes in human body cells, 1 pair or 2 sex chromosomes only determine whether a person is male or female. Human females have 2 X-shaped sex chromosomes (XX). Human males have 1 X-shaped and 1 Y-shaped chromosome (XY).
- ◆ Autosomes are the other 22 pairs or 44 chromosomes in human body cells.

ALLELES

- ◆ Genes are made of DNA. Genes help us to make proteins (e.g. muscle protein, hormones, enzymes, antibodies, skin collagen, hair keratin) that will determine certain characteristics we inherit.
- ◆ For a specific characteristic (e.g. eye colour), there may be one or more types (e.g. blue, brown) that may be determined by slightly different variations of the DNA in the gene. These are called alleles. For example, in eye colour, there may be brown eye colour determined by a gene or allele B, or for blue eye colour, it is determined by a slightly different gene or allele b.

MULTIPLE ALLELES

- ◆ For a particular characteristic, there may be more than 2 alleles. For example, in Blood Type, there are 3 alleles – A, B and O.)

GENOTYPES AND PHENOTYPES

- ◆ Each characteristic (e.g. eye colour) is determined by a pair of genes/alleles. One of those genes came from the father via sperm, and the other came from the mother via egg.
- ◆ Genotype is the pair of genes/alleles written in letter form (e.g. BB).
- ◆ Phenotype is the characteristic that appears as a result of that genotype (e.g. brown eyes).
- ◆ Example 1 – Eye Colour in Humans

GENOTYPE	PHENOTYPE
BB	Brown-eyed person
Bb	Brown-eyed person
bb	Blue-eyed person

- ◆ Example 2 – Height in Pea Plants

GENOTYPE	PHENOTYPE
TT	Tall pea plant
Tt	Tall pea plant
tt	Short pea plant

- ◆ Homozygous (or Purebred) – The genotype for the characteristic has the same genes/alleles (e.g. BB, bb).
- ◆ Heterozygous (or Hybrid) – The genotype for the characteristic has different genes/alleles (e.g. Bb).
- ◆ Wild-type - The most common phenotypes in a population are called the wild-type. These may be dominant (e.g. brown eye colour) or recessive (e.g. green pea colour).

MENDEL'S LAWS

- ◆ Gregor Mendel (1822-1884) was an Austrian monk whose work with pea plants laid the foundation for genetics.
- ◆ Principle of Segregation states that the alleles of a gene separate when gametes (sperm or ova) are formed. Each gamete receives one or the other allele.
- ◆ Principle of Independent Assortment states that the segregation of alleles of one gene is independent of the segregation of alleles of another gene during gamete formation.

PEDIGREES (OR FAMILY TREES)

- ◆ Pedigrees are drawn to examine a characteristic being studied (e.g. eye colour, haemophilia).
- ◆ Symbols used are:

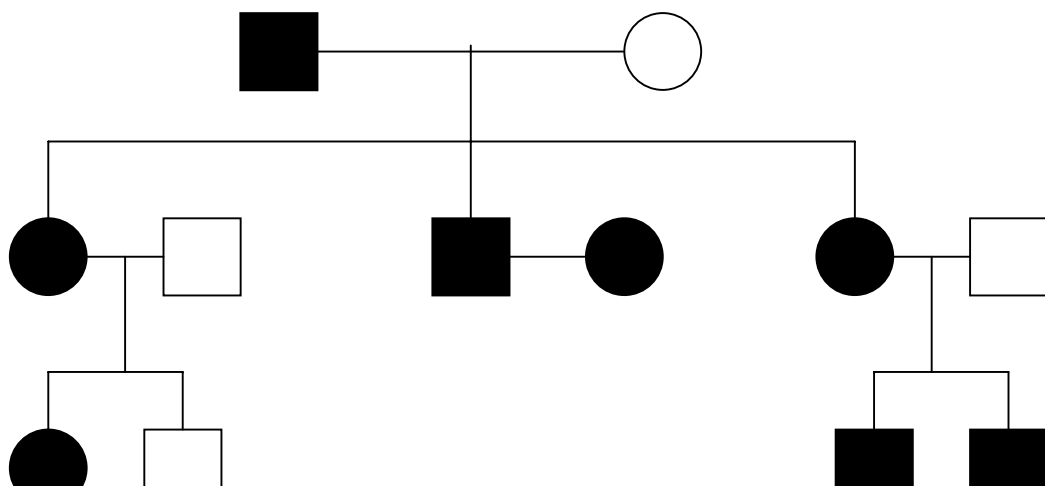
□ Male without characteristic being studied

○ Female without characteristic being studied

■ Male with characteristic being studied

● Female with characteristic being studied

- ◆ Example of Pedigree



SEX DETERMINATION

- ◆ Because the sperm or egg will contain only $\frac{1}{2}$ of the genes and chromosomes of a person, then each sperm could contain either an X or a Y chromosome. Each egg contains either an X or the other X chromosome.
- ◆ To have a daughter, the sperm from the father and the egg from the mother must both contain X chromosomes.
- ◆ To have a son, the father's sperm must have a Y chromosome to join with the mother's egg that has an X chromosome.
- ◆ It is the father who determines the sex of a child.

