# CIE Biology A-level

## Topic 16: Inherited change

Notes

**Meiosis** is a form of cell division that gives rise to **genetic variation**. The main role of meiosis is **production of haploid gametes** as cells produced by meiosis have half the number of chromosomes.

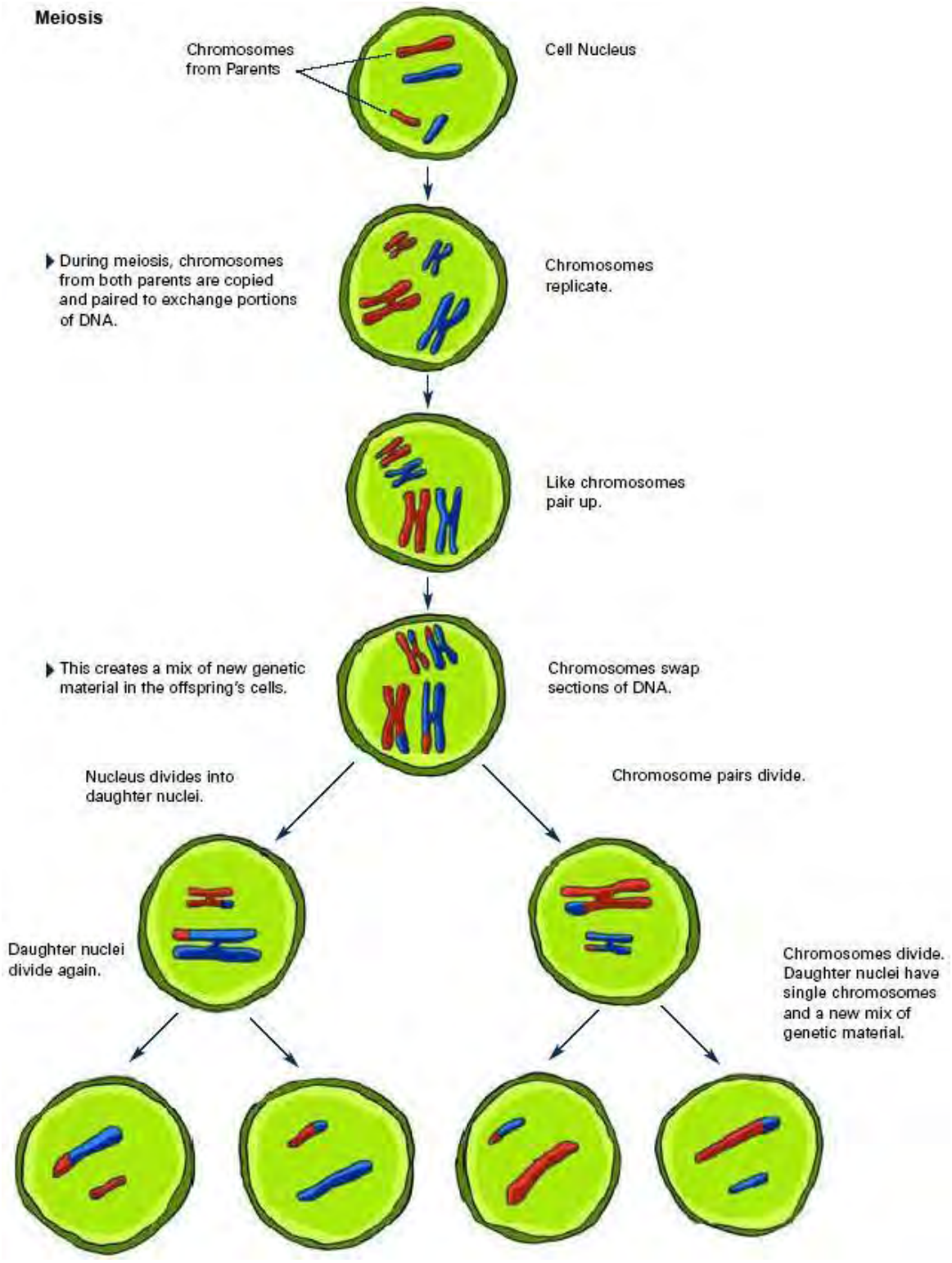


*Figure SEQ Figure * ARABIC 1 Biologypost - Meiosis*

**Meiosis produces genetically different cells; genetic variation is achieved through:**

- **Crossing over of chromatids** where pairs of chromosomes line up and exchange some of their genetic material
- **Independent assortment of chromosomes** – there are various combinations of chromosome arrangement

**Keywords:**

**Allele** – one of a number of alternative forms of a gene

**Locus** – the specific position of a gene on a chromosome, the two alleles of a gene are found at the same loci on the chromosome pairs

**Haploid** – haploid cells carry a single set of chromosomes in the nucleus. These include sperm and egg cells. Produced through meiosis

**Diploid** –diploid cells carry two sets of chromosomes in the nucleus. Produced through mitosis

**Phenotype** – observable characteristics of an organism which are as a result of genotype and environment

**Genotype** – the alleles present within cells of an organism, for a particular trait or characteristic

**Dominant** – only a single allele is required for the characteristic to be expressed, that is the allele is always expressed in the phenotype

**Recessive** – the characteristic is only expressed if there is no dominant allele present

**Homozygous** – two identical alleles

**Heterozygous** – two different alleles

**Homologous chromosomes** – a pair of chromosomes, one from the father and one from the mother. Each has the same genes in the same position, although the alleles of these genes may be different.

**Codominance** – both alleles contribute to the phenotype

**Linkage** is the phenomenon where genes for different characteristics, located at different loci on the same chromosome are linked.

**Monogenic inheritance** – when a phenotype or trait is controlled by a single gene. For instance, cystic fibrosis where the individuals with doubly recessive phenotype are affected.

**Dihybrid cross** – inheritance of two genes

**Sex linkage** – expression of an allele dependent on the gender of the individual as the gene is located on a sex chromosome, for instance, males are more likely to inherit an X-chromosome linked condition because they only have a single copy of the X chromosome. An example of sex linkage is haemophilia which is a recessive condition (hh).

**Autosomal linkage** – genes which are located on the same chromosome and tend to be expressed together in the offspring

**Codominance** – when both alleles are expressed in a heterozygote, that is, both alleles contribute towards the phenotype. Examples include blood type.

**Epistasis** – the interaction of different loci on the gene, one gene locus affects the other gene locus. One gene loci can either mask or suppress the expression of another gene locus.

**Recessive epistasis** occurs when the presence of a recessive allele prevents the expression of another allele at a second locus. Recessive epistasis gives the ratio of **9:3:4.**

**Dominant epistasis** is when a dominant allele at one locus completely masks the alleles at a second locus. Dominant epistasis gives a ratio of **12:3:1.**

**Monohybrid and Dihybrid Crosses:**

- Used to predict the ratios of inherited characteristics in a population
- **Monohybrid crosses** are used to determine the outcome of **one gene**. It will have **four outcomes** and have a ratio of **3:1**, where the dominant trait is more common. An example of this would be crossing heterozygous green and yellow pea plants:

  G= green (dominant allele)
  g= yellow (recessive allele)

  (Gg x Gg)

| | **G** | **g** |
|---|---|---|
| **G** | GG | Gg |
| **g** | Gg | gg |

The outcomes are GG, Gg, Gg and gg. As G is dominant, there is a 75% chance that the offspring will display this allele in the phenotype and be green. There is a 25% chance that the offspring will be yellow.

- **Dihybrid crosses** are used to determine the outcome of **two genes,** each of which have **two alleles**. This results in **16 outcomes.**
  E.g. Pea plants can be tall or short, and have purple or white flowers.

  P= purple
  p= white
  T= tall
  t= short

  Crossing two heterozygous parents (PpTt x PpTt):

  The gametes are PT, Pt, pT, and pt

| | PT | Pt | pT | pt |
|---|---|---|---|---|
| PT | PPTT | PPTt | PpTT | PpTt |
| Pt | PPTt | PPtt | PpTt | Pptt |
| pT | PpTT | PpTt | ppTT | ppTt |
| pt | PpTt | Pptt | ppTt | pptt |

  This results in a **9:3:3:1** ratio.

# Chi-squared test

$$X^2 = \sum \frac{(\text{observed} - \text{expected})^2}{\text{expected}}$$

The **chi squared test** is a **statistical** test which can be used to establish whether the difference between **observed and expected results** is small enough to occur purely due to chance.

- It can be used if the **sample size** is sufficiently large, that is over 20. It can only be used for **discontinuous variation** data in the form of raw counts.

- The chi squared test can be used to determine whether the **null hypothesis** is correct or not. The null hypothesis is the assumption that there is no difference between observed and expected results.

- The value obtained is compared to the **critical value**, and in a case where the value obtained is less than the critical value, the null hypothesis is accepted as the difference due to chance is not significant

- Whereas in a case where the $x^2$ value is greater than critical value, the null hypothesis is rejected meaning that the difference between observed and expected results is not due to chance, as is significant.

# Mutations

**Mutations** are changes in the sequence of nucleotides in DNA molecules. Types of mutations include:

- **Insertion/deletion mutations** where one or more nucleotide pairs are inserted or deleted from the sequence. This type of mutation alters the sequence of nucleotides after the insertion/deletion point known as a frameshift.
- **Point mutation/substitution** occurs where one base pair is replaced by another (this may have no effect).
- A **nonsense mutation** is one where a translation is stopped early thus giving rise to a truncated polypeptide due to premature introduction of a stop codon.
- A **missense mutation** is a codon change which results in the production of a different amino acid, thus resulting in altered tertiary structure of the protein. The extent of the effect of this is determined by which amino acid is replaced. For example, if it is an amino acid which makes up an active site, it may make an enzyme inactive.
- A **silent mutation** is a codon change which does not affect the amino acid sequence produced.

Mutations can either have **neutral effects** where the mutation causes no change to the organism, for example in a case where the mutation occurs in **a non-coding region** of DNA or is a **silent mutation**, as described above. A mutation can also be neutral when a change in tertiary structure of the protein has no effect on the organism.

Some mutations are beneficial, for instance, humans developed **trichromatic vision** through a mutation. Harmful mutations include a mutation in the **CFTR protein** which causes **cystic fibrosis.**

**Conditions caused by gene mutations:**

| Human condition | Effect of mutation on phenotype |
|---|---|
| Albinism | Albinism is a result of a mutation in a gene that causes the production of melanin. This causes **little or no melanin to be produced**, resulting in light hair and skin colour, and vision impairment. |
| Sickle cell anaemia | Caused by a **missense mutation** in the beta-haemoglobin gene. This causes **red blood cells to become sickle shaped**. Sickle cells carry less oxygen and can block blood vessels. |
| Haemophilia | Caused by a mutation in the FVII or FIX genes located on the **X chromosome**, which code for proteins that are important in **blood clotting**. The result is that blood cannot clot correctly. |
| Huntington's disease | Huntington's is caused by a mutation to the HTT gene, which makes a protein called huntingtin. It leads to the **degeneration of nerve cells** in the brain, causing cognitive and movement problems. |

# Controlling gene expression

**Gene expression** can be controlled at the transcriptional, post-transcriptional, translational and post-translational levels.

An example of transcriptional control is **the lac operon**, which is a length of DNA composed of structural genes and control sites which controls the expression of **beta-galactosidase** responsible for hydrolysis of lactose in E.coli. The operon consists of a **promoter region** which is the binding site for RNA polymerase to initiate transcription, **operator region** where the inhibitor binds and **structural genes** which give rise to 3 products, beta galactosidase, lactose permease and another enzyme. The **inhibitor** is coded for by a regulator gene, located outside the operon which binds to the operator region.

In a case where the concentration of glucose is high and the concentration of lactose is low, the transcription of the structural genes is inhibited due to binding of the **repressor** to the operator region. However, in a case where the concentration of glucose is low and concentration of lactose is **high, lactose binds the repressor** thus causing the shape of its

active site to change, therefore making it ineffective. This means that it can no longer bind to the operator region and transcription of the structural genes takes place.

Gene expression can also be controlled by **transcription factors** which have the ability to switch genes on and off. They do so through interaction with the promoter sequence of DNA to either initiate or inhibit transcription.

Gene expression is controlled at post-transcriptional level by **editing of the primary mRNA transcript**, during which the non-coding regions called **introns are** removed, thus creating a mature transcript consisting only of protein-producing regions known as **exons.**

Gene expression can be controlled at the **post-translational level.** For example, proteins such as adrenaline can be activated with the help of **cyclic AMP**. This occurs when adrenaline binds to a complementary receptor, which activates the enzyme **adenylate cyclase** which converts ATP to cyclic AMP which starts a **cascade of enzyme reactions** within the cell, thus activating the protein.

**Gibberellin** is a plant hormone that controls plant growth and seed germination by controlling the production of amylase. It does this by breaking down **DELLA**, a **repressor protein** which inhibits the binding of a transcription factor to the gene promoter region. As a result, the transcription factor can bind and transcription of the gene can occur which increases the synthesis of amylase.