

# CAIE Biology A-level

## Topic 16: Inherited change

### Notes

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**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. **Reduction division** takes place where the chromosome number halves from diploid to **haploid**.

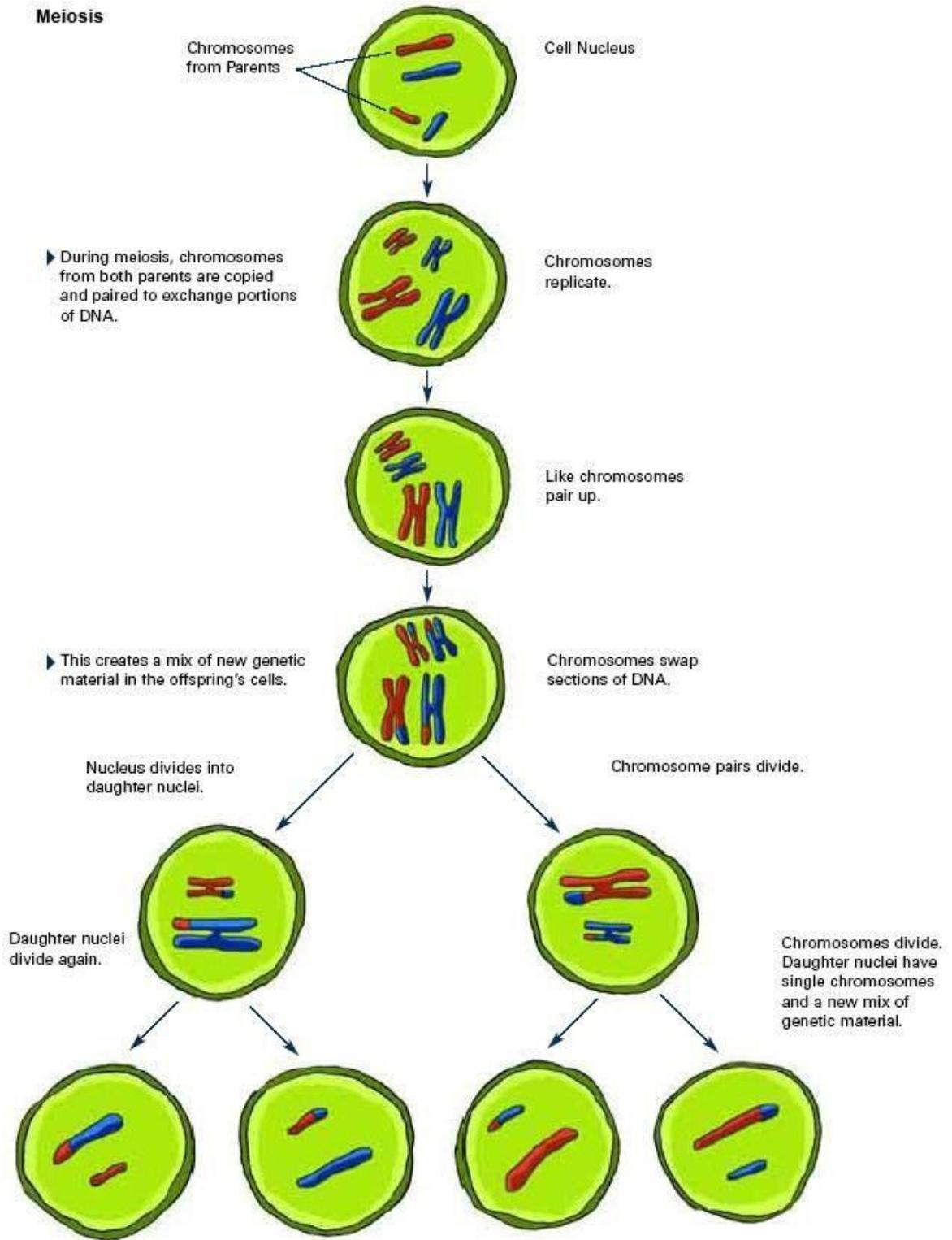


Figure SEQ Figure \\* ARABIC 1 Biologypost - Meiosis



**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

During **fertilisation** the **random fusion** of gametes also increases genetic variation in offspring.

**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)

	<b>G</b>	<b>g</b>
<b>G</b>	GG	Gg
<b>g</b>	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	Pp tt
pT	PpTT	PpTt	ppTT	ppTt
pt	PpTt	Pp tt	ppTt	pp tt

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

### Albinism

Albinism is a condition that affects a person's melanin production in the skin and leads to them having white hair, light eyes (may affect their eyesight) and pale skin.

The **TYR gene** is responsible for the production of **tyrosinase**. This is the enzyme that controls melanin production. If a **mutation** in the TYR gene occurs, tyrosinase production is hindered resulting in the person developing albinism.

### Sickle cell anaemia

A condition where a person is deficient in the amount of healthy red blood cells it has. If a person has sickle cell anaemia they have **crescent-shaped** red blood cells. This means they have abnormal hemoglobin which can't carry sufficient oxygen. The **HBB gene** is responsible for producing a protein which is a subunit of haemoglobin. If a mutation occurs in the HBB gene then the red blood cell's **haemoglobin** will be altered leading to sickle cell anemia.

### Hemophilia

A **sex-linked recessive disorder** which only affects **males**. It is carried on the **X chromosome** and females can be **carriers** of the disorder. It is when a person's blood doesn't clot normally due to the lack of the blood-clotting factor. If this isn't treated it can lead to prolonged bleeding which can be fatal. A mutation in the **FB gene** leads to a **factor VIII** deficiency causing haemophilia.

### Huntington's disease

A **neurodegenerative disease** which affects the brain and the spinal cord (central nervous system). The **HTT gene** codes for a protein called **huntingtin** which plays a role in normal functioning of neurones. A mutation in the HTT gene causes Huntington's disease.

### 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 <b>little or no melanin to be produced</b> , resulting in light hair and skin colour, and vision impairment.
Sickle cell anaemia	Caused by a <b>missense mutation</b> in the beta-haemoglobin gene. This causes <b>red blood cells to become sickle shaped</b> . Sickle cells carry less oxygen and can block blood vessels.
Haemophilia	Caused by a mutation in the FVII or FIX genes located on the <b>X chromosome</b> , which code for proteins that are important in <b>blood clotting</b> . 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 <b>degeneration of nerve cells</b> 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. The height of plants is controlled by whether the active form of gibberellin is present, which is determined by the plants genes: if the **dominant allele (Le)** is present, gibberellin is active and the plant grows tall, if the **recessive allele (le)** is homologously present, a **non-functioning** gibberellin enzyme is coded for, thus the plant remains short.

