CAIE Biology A-level

Topic 16: Inherited Change

Flashcards

Explain the terms ‘haploid’ and ‘diploid’.

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- Haploid (n) - having half of a set of homologous chromosomes e.g. gametes
- Diploid (2n) - having the full set of homologous chromosomes e.g. somatic cells

Explain the term ‘homologous chromosomes’.

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In diploid cells, the chromosomes are in pairs - they are homologous chromosomes. Each chromosome of a pair carries the same genes, but may have different alleles. Therefore, there are two loci for each gene.

Humans have 23 pairs of chromosomes, or 46 chromosomes in total.

One chromosome of each pair is from the mother (maternal) and the other is from the father (paternal).

Why must the number of chromosomes be halved before fertilisation?

Why must the number of chromosomes be halved before fertilisation?

Each of the gametes contains half the number of chromosomes (23 in humans) so that when they fuse during fertilisation the diploid number is restored (46 in humans). If this did not occur, the number of chromosomes would double with each generation.

Name the process of cell division that gives rise to gametes.

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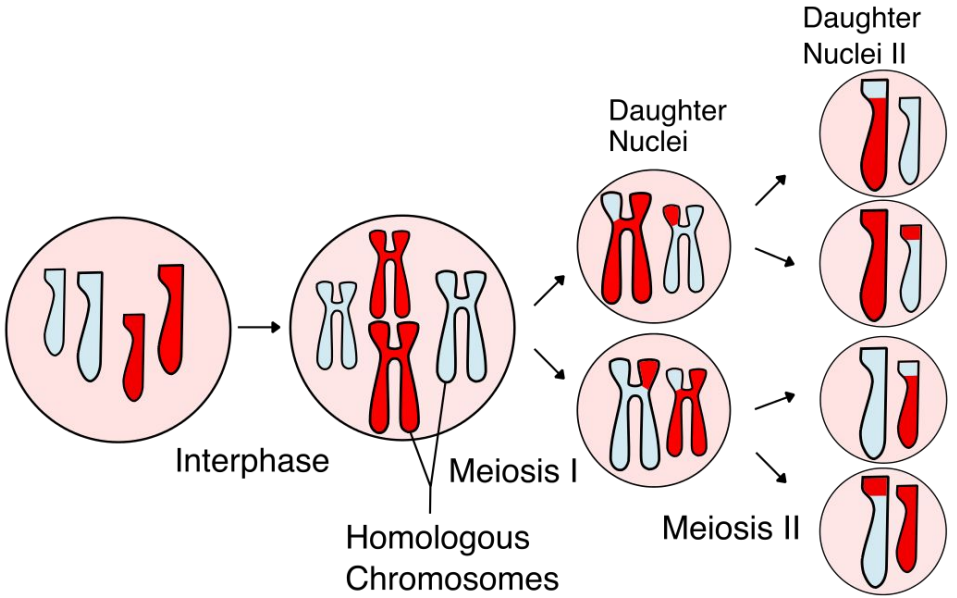
Meiosis



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Describe the process of spermatogenesis.

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- A diploid (2n) spermatogonium divides by mitosis to produce a diploid primary spermatocyte (2n)
- The primary spermatocyte undergoes meiosis I, resulting in two secondary spermatocytes (n)
- The two secondary spermatocytes enter meiosis II to produce four spermatids (n)
- The spermatids mature into motile sperm, capable of reaching and fertilising an egg

Describe the process of oogenesis.

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- Before birth, oogonia (2n) divide by mitosis to form primary oocytes (2n)
- The primary oocytes begin meiosis I but pause at prophase I until puberty
- During and after puberty, the primary oocyte continues meiosis I, producing a secondary oocyte (n) and a polar body. The secondary oocyte begins to undergo meiosis II, but stops at metaphase II
- If the secondary oocyte is fertilised by a sperm cell, meiosis II finishes and the fertilised egg now has the diploid number of chromosomes

Describe the events of meiosis I.

Describe the events of meiosis I.

- **Prophase I** - centrosomes move to opposite poles of the cell; homologous chromosomes pair up. Crossing over occur here.
- **Metaphase I** - homologous chromosomes line up along the equator of the cell
- **Anaphase I** - homologous chromosomes are separated and pulled to opposite poles of the cell by spindle fibres
- **Telophase I** - the nuclear envelope forms around the genetic material at each pole and the cell divides (cytokinesis)

Describe the events of meiosis II.

Describe the events of meiosis II.

- **Prophase II** - centrosomes move to opposite poles of the cell
- **Metaphase II** - the chromosomes align along the equator of the cell
- **Anaphase II** - spindles pull the sister chromatids apart to the opposite poles of the cell
- **Telophase II** - the nuclear envelopes reform. Four genetically different daughter cells are produced, each with the haploid number of chromosomes.

Explain how crossing over in meiosis increases genetic variation.

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During prophase I, the chromatids within a homologous pair of chromosomes twist around each other. Parts of the chromatid break off and may be rejoined with the chromatid of the other homologous chromosome. The same loci are exchanged. This creates **new combinations of alleles** and therefore increases genetic variation.

Explain how random assortment in meiosis increases genetic variation.

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During metaphase I, the homologous chromosomes align randomly along the equator. This means that in the daughter cells of meiosis I, there will be a mix of maternal and paternal chromosomes, increasing genetic variation.

Why is the random fusion of gametes important?

Why is the random fusion of gametes important?

It increases genetic variation due to the random combination of chromosomes generated.

Define the term ‘gene’.

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A gene is a sequence of DNA that codes for a polypeptide (protein) or RNA.

What is meant by the term ‘locus’?

What is meant by the term ‘locus’?

A locus is the specific position of a gene on the chromosomes.

What is an allele?

What is an allele?

An allele is one of the different versions of a gene. With homologous chromosomes, there are two loci for each gene; these may carry different, or the same alleles.

What is meant by homozygous and heterozygous?

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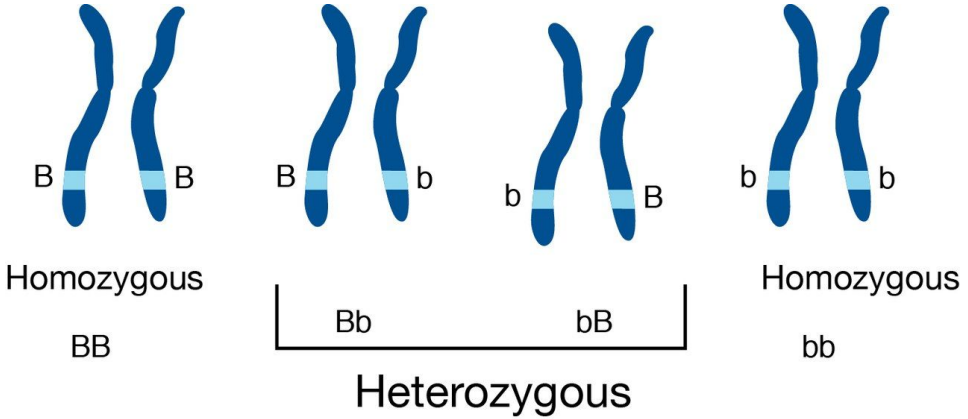
Homozygous - the alleles on each of the homologous chromosomes in a pair are the same.

Heterozygous - the alleles in the homologous pair are different.



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State what is meant by dominant and recessive alleles.

State what is meant by dominant and recessive alleles.

A **dominant allele** is a form of a gene that will be expressed even if only one is present (heterozygous) - it can override recessive alleles.

Recessive alleles will only be expressed if they are homozygous.

Define the term ‘codominant’.

Define the term ‘codominant’.

Both alleles (which are different) are fully expressed and contribute to the phenotype.

Explain what is meant by ‘linkage’.

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Two genes are said to be linked if they are close to each other on the same chromosome. They are unlikely to be separated during crossing over of meiosis, and so are inherited together.

What is a test cross?

What is a test cross?

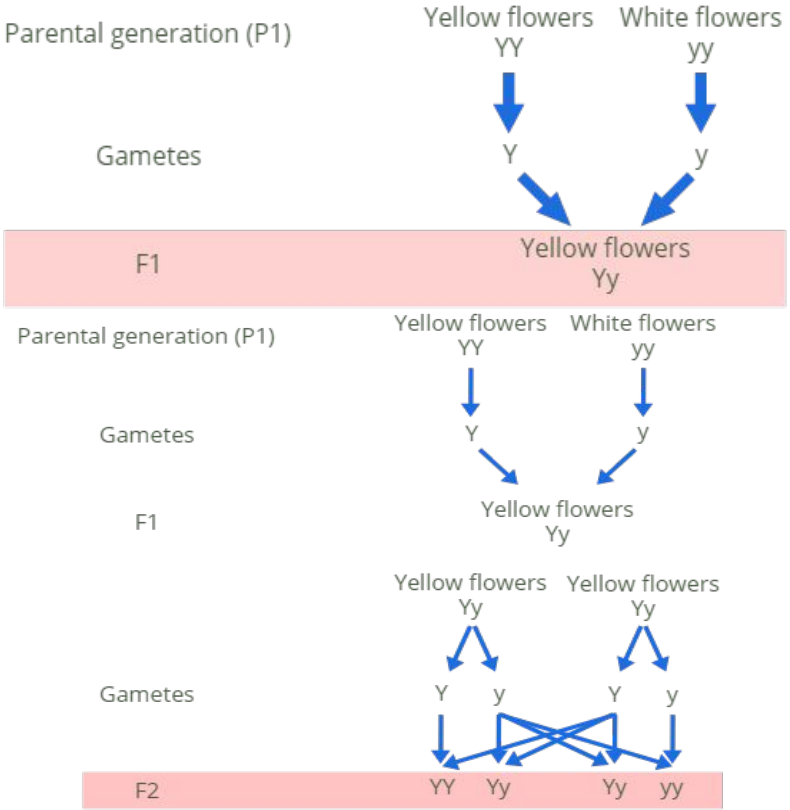
A genetic cross to determine the genotypes of the parents and offspring. An organism with a recessive genotype is crossed with an organism whose genotype is unknown, but their phenotype shows a dominant allele is present. The phenotypes of the offspring will determine whether the parent organism is heterozygous or homozygous.

What is meant by F1 and F2?

What is meant by F1 and F2?

F1 is the **first generation**, produced from one homozygous dominant parent and one homozygous recessive parent. The F1 generation is therefore heterozygous.

F2 is the **second generation**, produced from inbreeding of the F1 generation.



Define ‘phenotype’.

Define ‘phenotype’.

The observable features of an organism. The phenotype is a result of the interaction between the genotype and environment.

Define ‘genotype’.

Define ‘genotype’.

All of the alleles that an organism possesses.

What does a monohybrid cross show?

What does a monohybrid cross show?

The possible genotypes of the offspring for one gene only.

What is autosomal linkage?

What is autosomal linkage?

When two genes are positioned close together on the same autosome (any chromosome except the sex chromosomes) they are likely to be inherited together and so are autosomally linked.

Explain what is meant by ‘sex linkage’.

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Sex-linked genes are any genes found on the X or Y chromosomes.

Why are males at greater risk of sex-linked disorders caused by recessive alleles?

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Females = XX

Males = XY

The X chromosome is much larger than the Y chromosome, so for many of the genes on the X chromosome there is no homologue on the Y chromosome. Therefore, recessive alleles on the X chromosome will appear more frequently in the phenotype in males, because they only have one copy of that gene.

Why is the chi-squared test used?

Why is the chi-squared test used?

To test whether the difference between the observed and expected frequencies is significant, or whether the difference is due to chance.

Describe the types of gene mutations that can occur.

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- **Substitution** - one nucleotide base is swapped for another
- **Deletion** - one or more bases are removed from the DNA sequence
- **Insertion** - one or more bases are added into the DNA sequence

Explain how a gene mutation can affect the gene product.

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- **Silent mutation** - a change in one base does not affect the amino acid the codon codes for. This is due to the degenerate nature of the genetic code
- **Nonsense mutation** - the new base creates a stop codon, which means the protein is not fully produced
- **Missense mutation** - the new codon codes for a different amino acid. It may change the shape of the protein produced
- **Frameshift** - deletions or insertions shift the sequence, so every codon downstream is read differently, resulting in different amino acids and potentially a completely different protein.

Describe how albinism can arise as a result of a mutation.

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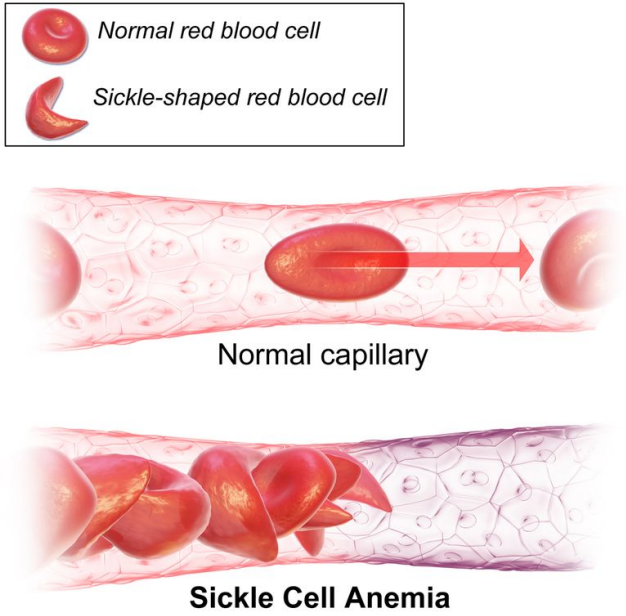
One form of albinism is caused by mutations of the tyrosinase (TRY) gene. Such mutations alter the tyrosinase enzyme which is responsible for the production of melanin; an inactive or absent enzyme results in albinism.

Outline how a mutation can cause sickle cell anaemia.

Outline how a mutation can cause sickle cell anaemia.

There is a glutamine-to-valine substitution in the HBB gene for β-globin. This changes how the haemoglobin molecules interact; they form strands and produce sickle-shaped red blood cells.

Sickle-shaped cells are less efficient at transporting oxygen, and may get stuck in the capillaries.



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Describe how a mutation can lead to haemophilia.

Describe how a mutation can lead to haemophilia.

The F8 gene, which codes for factor VIII (involved in clotting) is mutated in haemophilia. This gene is found on the X chromosome, so this is a sex-linked disorder caused by a recessive allele.

Explain how Huntington's disease can arise from a mutation.

Explain how Huntington's disease can arise from a mutation.

In people with Huntington's disease, the HTT gene, which codes for the protein huntingtin, contains a large number of CAG repeats within the DNA sequence - a 'stutter'.

Describe the difference between a regulatory gene and a structural gene.

Describe the difference between a regulatory gene and a structural gene.

A **regulatory gene** controls the expression of other genes.

A **structural gene** is one that codes for a protein or RNA that does not regulate the expression of other genes.

What is the difference between a repressible enzyme and an inducible enzyme?

What is the difference between a repressible enzyme and an inducible enzyme?

The synthesis of a **repressible enzyme** is only stopped when a repressor protein is activated.

The synthesis of an **inducible enzyme** is only started when a substrate is added.

What type of operon is the *lac* operon?

What type of operon is a *lac* operon?

An inducible operon.

How is the genetic control of protein production achieved using the *lac* operon?

How is the genetic control of protein production achieved using the *lac* operon?

- The expression of the genes in the *lac* operon depends on whether lactose is present. The *lac* repressor senses lactose.
- When **lactose** is **not present**, the repressor binds to the operator and prevents transcription of the genes.
- When **lactose** is **present**, it binds to the repressor and changes its shape. It can no longer block the transcription of the *lacZ, lacY* and *lacA* genes, which code for enzymes involved in lactose metabolism.

Name the molecules that regulate gene expression in eukaryotes.

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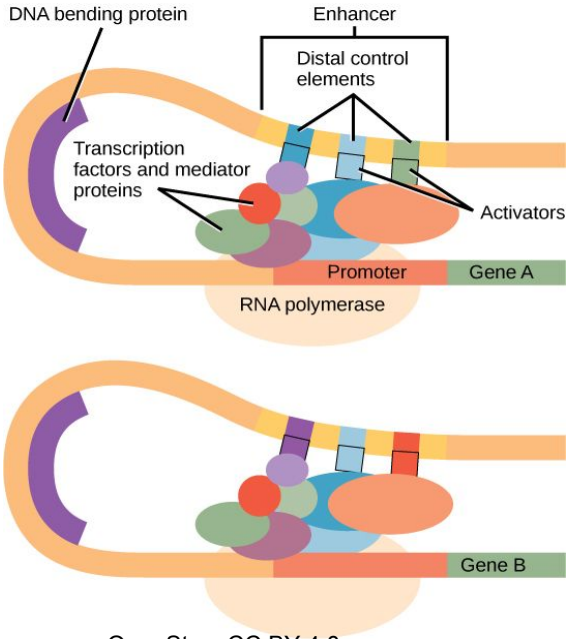
Transcription factors



Explain how transcription factors control gene expression.

Explain how transcription factors control gene expression.

Transcription factors bind to the promoter regions of genes. They may activate or repress the expression of a gene by changing how easy it is for RNA polymerase to access the DNA for transcription.

Explain how gibberellin can activate the expression of genes.

Explain how gibberellin can activate the expression of genes.

Gibberellin causes the DELLA protein repressors, which normally inhibit transcription factors, to break down. This means the gene can be transcribed (such as the gene for amylase).