WJEC (England) Biology A-level

Topic 2.5: Inheritance

Notes

Allele – alternative form 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

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

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). Other examples include Duchenne muscular dystrophy.

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.**

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 the 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 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.
- **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
- **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.**

An example of chromosomal mutation is Down's syndrome where a third copy of chromosome 21 is present.

Controlling gene expression

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