WJEC (Wales) Biology A-level

Unit 4.3 - Inheritance

Flashcards

What is a gene?

What is a gene?

A length of DNA on a chromosome that codes for the production of one or more polypeptide chains and functional RNA.

Define genotype.

Define genotype.

The genetic constitution of an organism.

Define phenotype.

Define phenotype.

The expression of an organism's genetic constitution, combined with its interaction with the environment.

What is a locus?

What is a locus?

The fixed position on a DNA molecule occupied by a gene.

What is an allele?

What is an allele?

Different forms of a particular gene, found at the same locus on a chromosome. A single gene could have many alleles.

Define homozygous

Define homozygous

Having two identical alleles of a gene

e.g. FF or ff

Define heterozygous

Define heterozygous

Having two different alleles of a gene

e.g. Ff

What is a dominant allele?

What is a dominant allele?

- An allele that is always expressed
- Represented with a capital letter e.g. F

What is a recessive allele?

What is a recessive allele?

- An allele that is only expressed if 2 copies are present
- Represented with a small letter e.g. f

What is meant by codominant alleles?

What is meant by codominant alleles?

Two dominant alleles that both contribute to the phenotype, either by showing a blend of both characteristics, or the characteristics appearing together.

What is an autosome?

What is an autosome?

A chromosome that is not an X or Y chromosome.

What is a sex chromosome?

What is a sex chromosome?

A chromosome that determines the sex of an organism, e.g. X and Y chromosomes in humans and other mammals.

What is the F_1 generation?

What is the F_1 generation?

The first generation of offspring resulting from the cross of two individuals in the parental generation.

What is the F_2 generation?

What is the F_2 generation?

The second generation of offspring resulting from the cross of two individuals in the F_1 generation.

Define monohybrid inheritance.

Define monohybrid inheritance.

Where one phenotypic characteristic is controlled by a single gene.

What is Mendel's first law of inheritance?

What is Mendel's first law of inheritance?

- **Law of segregation**
- Alleles separate randomly into gametes
- Each parent passes one allele to their offspring

What is Mendel's second law of inheritance?

What is Mendel's second law of inheritance?

- **Law of independent assortment**
- The alleles of genes assort independently of other genes during gamete formation

Draw an example of a genetic cross diagram.

Draw an example of a genetic cross diagram.

Parental phenotypes	Brown eyes Blue eyes
Parental genotypes	Bb bb
Gametes	B b b b
*Offspring genotypes (draw a **Punnett square**)*	Bb, Bb, bb, bb
Offspring phenotypes	2:2 brown eyes:blue eyes

A female who is homozygous recessive for cystic fibrosis (ff) has a child with a heterozygous male (Ff). Draw a punnett square to illustrate this monohybrid inheritance.

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		Female genotype	
		f	f
Male genotype	F	Ff	Ff
	f	ff	ff

PKU is a recessive condition. Two heterozygous parents have offspring. Predict the proportion of offspring that will have PKU.

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75% chance of normal phenotype

25% chance of PKU phenotype

		Female genotype	
		P	p
Male genotype	P	PP	Pp
	p	Pp	pp

What is the purpose of a test cross?

What is the purpose of a test cross?

Used to determine whether an individual with a dominant trait is heterozygous or homozygous dominant.

Describe how a test cross is carried out.

Describe how a test cross is carried out.

- An individual of **unknown genotype** is bred with a **homozygous recessive** individual
- Offspring phenotypes are observed to determine the genotype of the unknown individual

Define dihybrid inheritance.

Define dihybrid inheritance.

The determination of a trait by the inheritance of two genes.

What does Mendelian inheritance assume?

What does Mendelian inheritance assume?

It assumes that the genes involved are not linked.

What is meant by sex-linkage?

What is meant by sex-linkage?

- An allele is located on one of the sex chromosomes
- Its expression depends on the sex of the individual

Give an example of a phenotype which shows sex-linkage.

Give an example of a phenotype which shows sex-linkage.

- Haemophilia
- Duchenne muscular dystrophy

Describe haemophilia.

Describe haemophilia.

- X-linked recessive condition
- Results in excessive bleeding and blood that is slow to clot

Describe Duchenne muscular dystrophy.

Describe Duchenne muscular dystrophy.

- X-linked recessive condition
- Characterised by muscle degeneration and weakness

Why are haemophilia and Duchenne muscular dystrophy more common in males?

Why are haemophilia and Duchenne muscular dystrophy more common in males?

They are X-linked recessive conditions. Males only inherit one X chromosome so are more likely to express the gene in their phenotype.

What is linkage?

What is linkage?

- Two or more genes positioned on the same autosome
- Unlikely to be separated by crossing over during meiosis so often inherited together

What is the chi-squared test?

What is the chi-squared test?

A statistical test used to determine whether the difference between observed and expected data is due to chance or a real effect. Can be used to compare expected phenotypic ratios with observed ratios.

How is a chi-squared test performed?

How is a chi-squared test performed?

- Make a **null hypothesis**
- Used Mendelian ratios to calculate the expected numbers
- Calculate **chi-squared value** using chi-squared equation
- Calculate the **degrees of freedom**
- Select an appropriate significant level (normally 0.05)
- Find the **critical value**
- **Compare** the chi-squared value with the critical value
- Accept or reject the null hypothesis

How are degrees of freedom calculated?

How are degrees of freedom calculated?

The number of categories minus 1.

If the chi-squared value is greater than or equal to the critical value, is the null hypothesis accepted or rejected?

If the chi-squared value is greater than or equal to the critical value, is the null hypothesis accepted or rejected?

Rejected

What does it mean if the null hypothesis is rejected?

What does it mean if the null hypothesis is rejected?

There is a significant difference between the observed and expected results.

If the chi-squared value is less than the critical value, is the null hypothesis accepted or rejected?

If the chi-squared value is less than the critical value, is the null hypothesis accepted or rejected?

Accepted

What does it mean if the null hypothesis is accepted?

What does it mean if the null hypothesis is accepted?

The difference between the observed and expected results is not significant.

The results occured due to chance.

What is a mutation?

What is a mutation?

- An alteration to the volume, arrangement or structure of DNA
- May affect a single gene or a whole chromosome

When do most mutations occur?

When do most mutations occur?

- Crossing over in prophase I
- Non-disjunction in anaphase I and II

What is a gene mutation?

What is a gene mutation?

- A change to at least one nucleotide base in DNA or the arrangement of bases
- May occur spontaneously during DNA replication and can be beneficial, damaging, or neutral

Give an example of condition caused by a gene mutation.

Give an example of condition caused by a gene mutation.

Sickle cell anaemia

Describe sickle cell anaemia.

Describe sickle cell anaemia.

- Recessive genetic disorder caused by a substitution mutation on chromosome 11
- Results in abnormal haemoglobin which distorts red blood cells

What is a chromosome mutation?

What is a chromosome mutation?

A change in the structure or number of chromosomes, affecting many genes.

Give an example of condition caused by a chromosome mutation.

Give an example of condition caused by a chromosome mutation.

Down's syndrome

Describe Down's syndrome.

Describe Down's syndrome.

- Genetic disorder characterised by delayed development and learning disabilities
- Due to non-disjunction, an affected individual possesses **three copies** of **chromosome 21**

What is chromosome nondisjunction?

What is chromosome nondisjunction?

Failure of homologous chromosomes to separate in meiosis I or sister chromatids to separate in meiosis II. Results in gametes with one extra or less chromosome than normal.

What is a mutagen?

What is a mutagen?

A chemical, biological or physical agent that increases the rate of gene mutations above normal level.

Give some examples of mutagens.

Give some examples of mutagens.

- Ionising radiation (gamma radiation, UV, X-rays)
- Chemicals (e.g. polycyclic hydrocarbons)

What is a carcinogen?

What is a carcinogen?

A type of mutagen that causes cancer.

What is an oncogene?

What is an oncogene?

Mutations of proto-oncogenes that are activated continuously, resulting in uncontrolled cell division.

Define epigenetics.

Define epigenetics.

The study of changes in gene expression that are not due to alterations in the nucleotide base sequence of DNA.

How can histone modification affect gene expression?

How can histone modification affect gene expression?

- Alteration of histones by the addition of methyl, acetyl or phosphate groups
- Increases or decreases gene expression by causing the histone to coil more tightly or loosely

How can DNA methylation affect gene expression?

How can DNA methylation affect gene expression?

- Addition of a methyl (CH_3) group to cytosine bases
- Prevents recognition of the bases, reducing gene expression