OCR (B) Biology A-level

5.1.1 - Patterns of inheritance

Flashcards

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.

What is meant by a dominant allele?

What is meant by a dominant allele?

An allele whose characteristic will always appear in the phenotype, whether one or two are present.

What is meant by a recessive allele?

What is meant by a recessive allele?

An allele whose characteristic only appears in the phenotype if no dominant allele is present, meaning two must be present.

What is meant by homozygous and heterozygous?

What is meant by homozygous and heterozygous?

Homozygous= both alleles are dominant, or both alleles are recessive.

Heterozygous= one allele is dominant, the other is recessive.

Define monogenic inheritance.

Define monogenic inheritance.

Where one phenotypic characteristic is controlled by a single gene.

What is a mutation?

What is a mutation?

An alteration to the DNA base sequence. Often arise spontaneously during DNA replication. Can cause extreme changes to the protein coded for.

Give examples of conditions caused by a mutation.

Give examples of conditions caused by a mutation.

- Cystic fibrosis
- Sickle cell anaemia
- Phenylketonuria
- Huntington's disease

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 meant by multiple alleles?

What is meant by multiple alleles?

A gene with more than two alleles.

Give examples of phenotypes which show both codominance and multiple alleles.

Give examples of phenotypes which show both codominance and multiple alleles.

- Blood groups; three alleles (A, B, O). A and B display codominance (i.e. blood group AB).
- HLA antigens; many types and isoforms, which also display codominance.

What is meant by sex-linkage?

What is meant by sex-linkage?

Where an allele is located on one of the sex chromosomes, meaning its expression depends on the sex of the individual.

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

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Haemophilia. Gene present on the X chromosome and is recessive, meaning males are more likely to express the gene in their phenotype.

What is meant by autosomal linkage?

What is meant by autosomal linkage?

Where two or more genes are located on the same (non-sex) chromosome. In this case, only one homologous pair is needed for all four alleles to be present. For genes that aren't linked, two homologous pairs are needed.

Give examples of phenotypes which show autosomal linkage.

Give examples of phenotypes which show autosomal linkage.

- Blood group
- Nail patella syndrome

How can model organisms be used to investigate inheritance?

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Drosophila used to investigate dihybrid inheritance. Wing length and body colour appears in the 9:3:3:1 ratio expected of dihybrid characteristics.

What is the chi-squared test?

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A statistical test used to find out 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?

The formula results in a number, which is then compared to a critical value (for the corresponding degrees of freedom). If the number is greater than or equal to the critical value, we conclude there is no significant difference and the results occured due to chance.

What is chromosome nondisjunction?

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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.

Briefly describe syndromes that may be caused by chromosome non-disjunction.

Briefly describe syndromes that may be caused by chromosome non-disjunction.

- Down's syndrome = one extra chromosome 21. Causes facial deformations and slow development.
- Turner's syndrome = lacking one of the X chromosomes (XO), only occurs in females. Causes learning disabilities, obesity and short stature.
- Klinefelter's syndrome = one extra X chromosome (XXY), only occurs in males. Causes fertility problems and reduced muscle strength.

How can genetic diseases be tested for?

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- Pedigree analysis; studying the pattern of people in the family with the disease.
- Genetic testing; screening for the gene within the DNA of an individual.

Give ethical issues around testing for genetic diseases.

Give ethical issues around testing for genetic diseases.

Distressing for the family. May face a choice between terminating a pregnancy or raising a child with low quality of life. Genetic counselling helps them to make a decision.