Edexcel IAL Biology A-level

2.15-2.18 - Gene Expression

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.

What is meant by the term genotype?

What is meant by the term genotype?

The genotype is the genetic makeup of an organism which includes the different alleles which the organism possesses.

Define the term phenotype

Define the term phenotype

An organism's observable characteristics. It is determined by the interactions between the genotype and the environment.

What are alleles?

What are alleles?

Different versions of the same gene

What is a dominant allele?

What is a dominant allele?

A version of a gene where only one copy is needed for it to be expressed

What is a recessive allele?

What is a recessive allele?

A version of a gene where two copies are needed for it to be expressed

What is meant when an organism is homozygous?

What is meant when an organism is homozygous?

When an organism has two copies of the same allele (two recessive or two dominant)

What is meant when an organism is heterozygous?

What is meant when an organism is heterozygous?

When an organism has two different versions of the same gene (one dominant and one recessive)

What is codominance?

What is codominance?

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

Define monohybrid inheritance

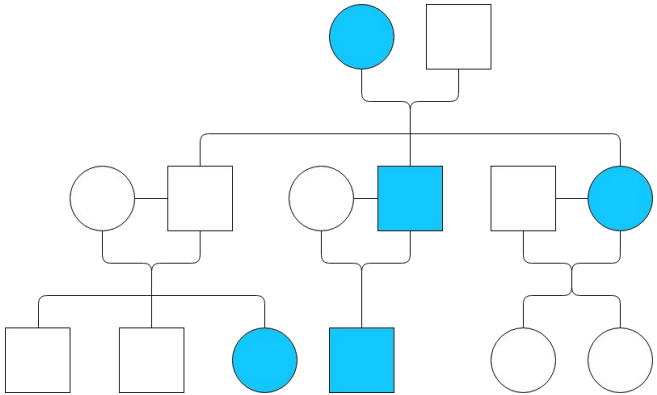
Define monohybrid inheritance

Where one phenotypic characteristic is controlled by a single gene

What do family pedigrees show?

What do family pedigrees show?

The inheritance of an allele over multiple generations



What is a sex-linked characteristic?

What is a sex-linked characteristic?

- A characteristic where the gene responsible for its located on a sex chromosome
- This makes it more common in one sex than another

Give one example of a sex-linked trait

Give one example of a sex-linked trait

Red/Green colourblindness is a sex-linked trait as it is inherited on the X chromosome and so much more common in males

What is red/green colour blindness?

What is red/green colour blindness?

An X-linked genetic condition which results in an inability to distinguish between red and green due to a lack of red or green photoreceptors

Why is red/green colour blindness significantly more common in males?

Why is red/green colour blindness significantly more common in males?

- The faulty gene which causes red/green colour blindness is located on the X chromosome
- Males only inherit one copy of the X chromosome and so they cannot inherit another non-faulty copy of the gene

What is cystic fibrosis?

What is cystic fibrosis?

An inherited **autosomal recessive** genetic condition which impairs the functioning of the gaseous exchange, digestive and reproductive systems by producing a faulty copy of the protein involved in the production of mucous, digestive juices and other fluids.

What is the genotype of individuals with cystic fibrosis?

What is the genotype of individuals with cystic fibrosis?

Homozygous recessive (ff)

What is the genotype of carriers of cystic fibrosis?

What is the genotype of carriers of cystic fibrosis?

Heterozygous (Ff)

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 inheritance

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 inheritance

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

Draw a Punnett square for a cross between two heterozygous cystic fibrosis carriers (Ff)

Draw a Punnett square for a cross between two heterozygous cystic fibrosis carriers (Ff)

	F	f
F	FF	Ff
f	Ff	ff

25% healthy	FF
50% carriers	Ff
25% have CF	ff

What is meant by genetic screening?

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Determining if an individual's DNA contains a certain allele, usually one that may result in a genetic disorder. This can allow prenatal diagnosis, and for treatment to be started earlier

What is pre-implantation genetic diagnosis (PGD)?

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The determination of genetic diseases during in-vitro fertilisation (IVF) before implantation of the embryo into the uterus

Describe the process of chorionic villus sampling

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A sample of embryonic tissue is taken from the placenta at around 8 to 12 weeks of pregnancy. It is screened for various disorders and results are available quickly

Describe the process of amniocentesis

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A sample of amniotic fluid is taken at around 14 to 16 weeks of pregnancy. The foetal cells have to be grown for 2-3 weeks before screening can take place, meaning results are slower than CVS

Give some social and ethical issues surrounding prenatal genetic screening

Give some social and ethical issues surrounding prenatal genetic screening

- Procedures carry risk of harming foetus
- May result in abortion, which many people object to
- High cost of bringing up a baby with a genetic disorder
- Emotional and mental stress on parents

What is genetic screening?

What is genetic screening?

A method of testing an individual's genome for faulty alleles that may make them more susceptible to certain diseases and disorders

Outline how genetic testing can be used to improve healthcare (3)

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- Enables awareness of potential risks and the introduction of lifestyle changes to reduce these associated risks
- Enables early treatment plans to begin
- Prediction of a patient's reaction to certain drugs - 'personalised medicine'

Outline the drawbacks of using genetic testing in healthcare (2)

Outline the drawbacks of using genetic testing in healthcare (2)

- Discrimination by employers, insurance firms etc. if a person is likely to develop a disease
- Person may develop anxiety, depression, etc