WJEC (Eduqas) Biology A-level

Topic 2.7 - Application of reproduction and genetics

Flashcards

What is the Human Genome Project?

What is the Human Genome Project?

An international research project involving thousands of scientists which used Sanger sequencing to successfully map the entire human genome.

What is DNA sequencing?

What is DNA sequencing?

Identifying the base sequence of a DNA fragment.

What is Sanger sequencing?

What is Sanger sequencing?

A method of DNA sequencing that only sequences relatively short sections of DNA at a time. It takes a long period of time.

Outline the potential benefits of the Human Genome Project.

Outline the potential benefits of the Human Genome Project.

- Allows for the development of targeted, **personalised medical treatments** and greater **accuracy** of diagnosis
- Increased opportunities for screening genetic conditions and **early detection** of disease
- Enables the study of **incidences of mutation** in different genes

What is the 100K Genome Project?

What is the 100K Genome Project?

A UK Government project that aims to study variation in the human genome amongst 100,000 UK citizens. It uses Next Generation Sequencers (NGS).

Describe Next Generation Sequencing (NGS).

Describe Next Generation Sequencing (NGS).

A faster, cheaper and more accessible method of sequencing that can sequence an entire genome in a few hours.

Describe genetic counselling.

Describe genetic counselling.

- Service that provides information and advice to people affected by or at risk of genetic diseases
- Helps individuals and families to make informed decisions

What is genetic screening?

What is genetic screening?

- Testing individuals for certain faulty alleles
- Used to detect disorders such as cystic fibrosis, Huntington's disease and thalassemia

Outline the potential disadvantages of genetic screening.

Outline the potential disadvantages of genetic screening.

- Screening for conditions such as cancer and Alzheimer's disease only indicates an increased risk - may cause unnecessary stress and anxiety
- What happens to the test result data? Discrimination from employers or insurance companies? Misuse of information?
- Risk of false positives or false negatives
- Who should be screened? Limited funds and time
- Screening embryos could lead to 'designer babies'

Give some examples of organisms other than humans whose genomes have been sequenced.

Give some examples of organisms other than humans whose genomes have been sequenced.

- Chimpanzees and other primates
- *Anopheles gambiae*, the mosquito
- *Plasmodium* parasite

How has sequencing the genome of the mosquito, *Anopheles gambiae*, been useful to humans?

How has sequencing the genome of the mosquito, *Anopheles gambiae*, been useful to humans?

- *Anopheles* has developed insecticide resistance
- Sequencing has enabled the development of chemicals that make *Anopheles* susceptible to insecticides

Outline the advantage of sequencing the genome of the *Plasmodium sp.* to humans.

Outline the advantage of sequencing the genome of the *Plasmodium sp.* to humans.

- *Plasmodium sp.* has developed multi-drug resistance
- Enables the development of more effective drugs

What is genetic fingerprinting?

What is genetic fingerprinting? State some applications of genetic fingerprinting.

- A technique used to genetically identify an organism
- Applications in forensics, screening for hereditary diseases, paternity testing, selection for clinical trials

What are exons?

What are exons?

Region of DNA that code for an amino acid sequence.

What are introns?

What are introns?

Non-coding sequences of DNA.

What are STRs?

What are STRs?

- Short Tandem Repeats
- Sections of repeated nucleotides within introns that produce variation in individuals

What techniques can be used to produce a genetic fingerprint?

What techniques can be used to produce a genetic fingerprint?

PCR and gel electrophoresis

What is PCR?

What is PCR?

- Polymerase Chain Reaction
- An *in vitro* technique used to rapidly amplify fragments of DNA

Describe the reaction mixture in the first stage of PCR.

Describe the reaction mixture in the first stage of PCR.

Contains the DNA fragment to be amplified, primers that are complementary to the start of the fragment, free nucleotides to match up to exposed bases, and Taq DNA polymerase to create the new DNA.

What is Taq DNA polymerase?

What is Taq DNA polymerase?

A thermally stable enzyme that synthesises a double-stranded molecule of DNA from a single template strand using complementary nucleotides.

Summarise the process of amplifying DNA fragments using PCR.

Summarise the process of amplifying DNA fragments using PCR.

1. Heated (90-95°C) to break hydrogen bonds between DNA strands
2. Cooled (55-60°C) to allow primers to bind - **annealing**
3. Heated (70°C) to activate Taq DNA polymerase and allow free nucleotides to join
4. New DNA acts as a template for next cycle

What is gel electrophoresis?

What is gel electrophoresis?

A technique that separates nucleic acid fragments or proteins by size using electric current.

How does gel electrophoresis work?

How does gel electrophoresis work?

- DNA fragments of varying lengths are placed at one end of a slab of agarose gel
- Electric current applied. DNA fragments move towards the positive end of the gel
- Shorter fragments travel further. The pattern of bands created is unique to every individual

What is genetic engineering?

What is genetic engineering?

The modification of the genome of an organism by the insertion of a desired gene from another organism. This enables the formation of organisms with beneficial characteristics.

What is recombinant DNA?

What is recombinant DNA?

A combination of DNA from two different organisms.

Summarise the process of using restriction enzymes to produce DNA fragments.

Summarise the process of using restriction enzymes to produce DNA fragments.

- Gene identified using **gene probe**
- **Restriction endonucleases** cut DNA at specific palindromic sequences producing **sticky ends**

Summarise the process of using reverse transcriptase to produce DNA fragments.

Summarise the process of using reverse transcriptase to produce DNA fragments.

- **mRNA** complementary to the target gene used as a template
- **Reverse transcriptase** synthesises **cDNA** (complementary)
- Mixed with free nucleotides which match up to their base pairs. **DNA polymerase** joins nucleotides forming second strand

Outline the advantages of using reverse transcriptase to produce cDNA.

Outline the advantages of using reverse transcriptase to produce cDNA.

- Don’t have to locate the gene
- Gene not cut into non-functional fragments by restriction enzymes
- Introns not present in cDNA
- Doesn’t require post-transcriptional processing to produce functional mRNA

Summarise the process of inserting a DNA fragment into a vector.

Summarise the process of inserting a DNA fragment into a vector.

- Plasmid (circular DNA from bacteria) used as the vector
- Plasmid cut using the **same restriction enzymes** as the DNA, so that the sticky ends are **complementary**
- **DNA ligase** joins the fragment and plasmid together

Describe how antibiotic-resistance genes are used in the identification of recombinant bacteria.

Describe how antibiotic-resistance genes are used in the identification of recombinant bacteria.

Antibiotic resistance genes can be inserted into plasmids at the same time as DNA fragments. The transformed cells are then placed on a plate with antibiotics. Only the cells that successfully took up the vector will grow.

Give an application for genetic modification of bacterial cells.

Give an application for genetic modification of bacterial cells.

Human gene for insulin production can be inserted into a vector, so that the bacterial cell will produce insulin. Useful in medicine e.g. treatment of diabetes.

Outline the disadvantages of using recombinant DNA to make human products.

Outline the disadvantages of using recombinant DNA to make human products.

- Identifying the required gene may be difficult
- Some eukaryotic genes can’t be expressed in prokaryotes
- Antibiotic-resistance genes could be transferred to pathogenic bacteria
- Expensive

What are GM organisms?

What are GM organisms?

Organisms that have had their genome altered.

Outline the benefits of GM crop production.

Outline the benefits of GM crop production.

- Improves nutritional value of foods
- Longer shelf-life of products
- Greater crop yields & reduces crop losses
- Reduces need for pesticides
- Reduces need for land clearing
- Can produce human medicine and vaccines

Outline the risks of GM crop production.

Outline the risks of GM crop production.

- Reduction in biodiversity
- Unknown effects on health
- Cross-pollination could result in herbicide-resistant weeds
- May increases costs for farmers
- Insect eating predators may be damaged by toxins in the plant
- Transferred gene may spread to wild population and cause problems

What is gene therapy?

What is gene therapy?

A therapeutic technique in which a faulty allele is replaced with a functional allele in order to treat or prevent disease.

Name the two types of gene therapy.

Name the two types of gene therapy.

- Somatic cell therapy
- Germ line therapy

Differentiate between somatic cell therapy and germ line therapy.

Differentiate between somatic cell therapy and germ line therapy.

- **Somatic** - allele introduced to target cells only, short-term, must be repeated
- **Germ line** - allele introduced to embryonic cells so it is present in all resultant cells, permanent, passed onto offspring

What is a vector?

What is a vector?

A carrier used to transfer a gene from one organism to another, e.g. plasmid or virus.

What is Duchenne Muscular Dystrophy (DMD)?

What is Duchenne Muscular Dystrophy (DMD)?

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

What is the cause of DMD?

What is the cause of DMD?

It is caused by one or more mutations in the dystrophin gene that prevent the production of dystrophin.

Outline how DMD can be treated using gene therapy.

Outline how DMD can be treated using gene therapy.

1. Healthy gene inserted into vector (e.g. virus)
2. Vector inserted into muscle tissue
3. Virus delivers gene to muscle cells
4. New gene incorporated into DNA of cell
5. Transcription and translation of gene produces normal dystrophin protein
6. Symptoms of DMD alleviated

What is drisapersen?

What is drisapersen?

An experimental drug that aims to treat DMD by exon skipping.

Explain how drisapersen works.

Explain how drisapersen works.

It introduces a ‘molecular patch’ over the mutated exon, enabling the gene to be read. A shorter, more functional type of dystrophin is synthesised.

Discuss the ethical issues surrounding the use of gene therapy.

Discuss the ethical issues surrounding the use of gene therapy.

- Health implications - may produce an immune response, activation of oncogenes etc.
- Is it right to alter the genotype of an unborn child?
- What conditions should be treated using gene therapy?
- Could lead to healthcare inequalities
- Expensive - money could be better spent elsewhere

What are stem cells?

What are stem cells?

Cells that are unspecialised and retain the ability to differentiate into a range of cell types.

What is tissue engineering?

What is tissue engineering?

An extension of gene therapy that aims to replace, repair or improve biological function by replacing organs and tissues.

What is the main advantage of using stem cells?

What is the main advantage of using stem cells?

Rapid production of genetically identical cells and organisms.

Outline the disadvantages of using stem cells.

Outline the disadvantages of using stem cells.

- Expensive and unreliable in mammals
- In plants, disease and pathogens can cause issues
- Inadvertent selection of disadvantageous alleles, unknown long-term effects

What are the ethical issues related to the use of stem cells from embryos?

What are the ethical issues related to the use of stem cells from embryos?

- Embryos used to provide stem cells are destroyed which is seen as unethical and a waste of potential human life
- Could lead to the ‘farming’ of embryos for stem cells
- May lead to the reproductive cloning of humans