Study Notes: Module 6

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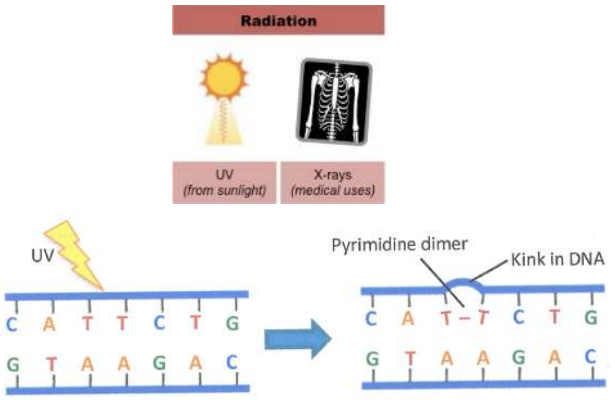
Inquiry Question 1:

How does mutation introduce new alleles into a population?

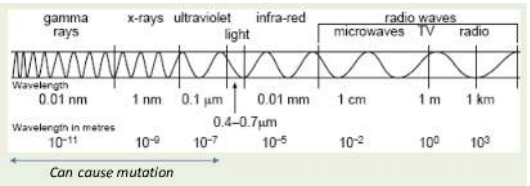
DOT POINTS:

- *explain how a range of mutagens operate, including but not limited to:*
  - *electromagnetic radiation sources*
  - *chemicals*
  - *naturally occurring mutagens*

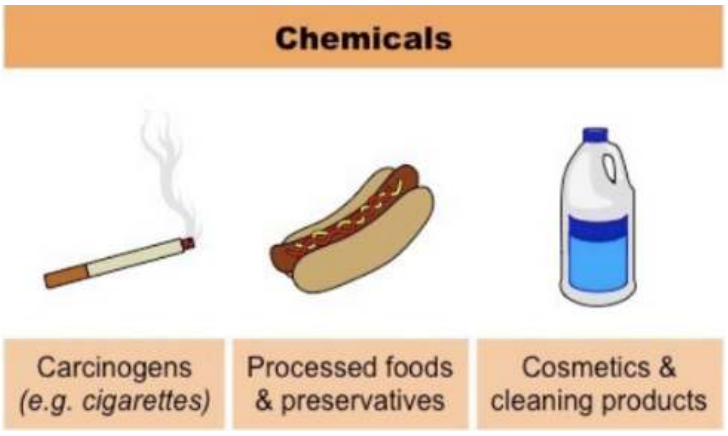
Electromagnetic Radiation sources



- Electromagnetic spectrum is a continuum of electromagnetic waves, which are arranged in order of frequency and wavelength.
- It's ability to penetrate cells is inversely proportional to its wavelength
  - **Shorter** the **wavelength** the **stronger** it is
- High energy radiation can ionise atoms (remove electrons from atoms)
  - Is called Ionising radiation
  - If it enters the DNA, it creates ions in the cell contents and can alter the way the two strands of DNA interact, shift sections of DNA in chromosomes and cause covalent bonds to form between adjacent bases,

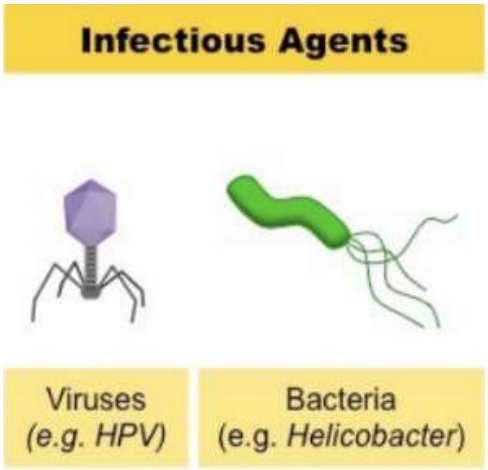


Chemicals



- Chemical mutagens interact in different ways, it can:
  - produce mutagenic compounds,
  - Cause incorrect pairings during DNA replication
  - Some chemicals insert themselves into the DNA resulting in the molecule changing shape
  - Change the properties of the bases
- Examples:
  - Alcohol, tar in tobacco smoke,
  - Charred and fatty foods and food additives and preservatives (such as nitrites) eg. Sausages
  - Organic solvents (for example, benzene), cleaning products, asbestos, coal tars, heavy metals, pesticides and some hair dyes.

Naturally Occurring Mutagens

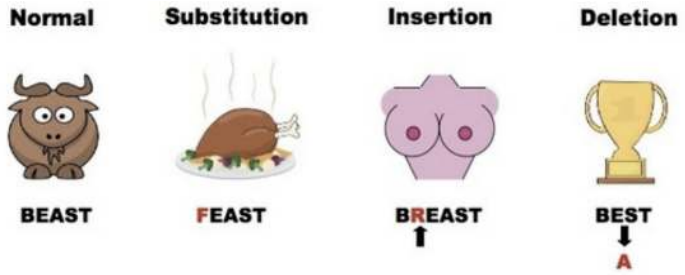


- Mutagenic agents that are present at normal levels within natural environments and may cause mutation.
- Can cause mutations and cancers by interfering with the functions of oncogenes and tumour-suppressor genes.
    - A transposon is a section of DNA that spontaneously fragment and relocate or multiply within the genome.
        - When they insert, they disrupt DNA functioning
- Microbes are naturally occurring biological mutagens.
    - viruses (such as hepatitis B virus, HIV, Epstein-Barr virus, Rubella virus)
    - bacteria (such as Helicobater)

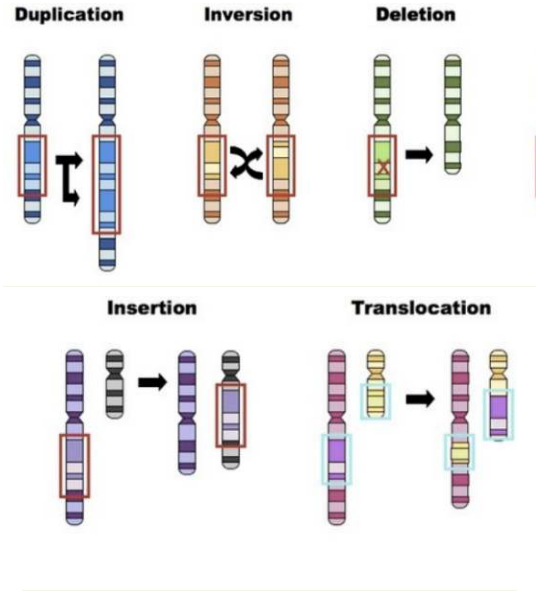
DOT POINTS:

- *compare the causes, processes and effects of different types of mutation, including but not limited to:*
    - *point mutation*
    - *chromosomal mutation*

Point Mutation

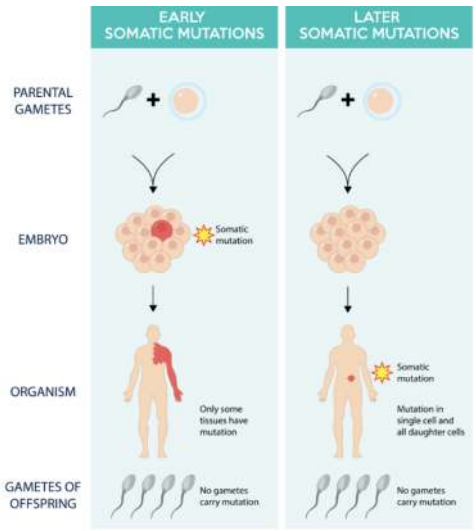
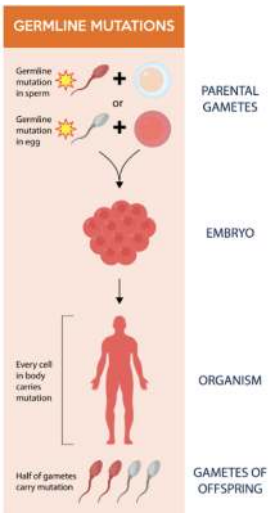


- Changes to a single base pair of DNA and affect only a single gene (gene mutation)
- **Substitution** (where ATG become ACG)**:**
    - **Silent** mutation: Results in a new codon that still codes for the same amino acid.
    - **Missense** mutation: Results in an amino acid being replaced. It will still produce a protein but may not function properly.
    - **Nonsense** mutation: Results in an amino acid being changed to a stop codon. Result in incomplete and usually non-functional proteins.
- **Insertion** (e.g. ATG becomes ATCG)
- **Deletion** (e.g. ATG becomes AG)
- Both insertion and deletion cause a massive impact as they result in a **frameshift** where every codon from that point on is affected, which means that incorrect amino acids are being created, resulting in non-functional proteins

Chromosomal Mutation: Block Mutations 	• Whole groups of genes are affected. There is a change in the structure of a chromosome involving large pieces being rearranged • **Duplications**: a part of the chromosome is copied, resulting induplicate segments • **Inversions**: a segment of a chromosome is removed and then replaced in reverse order • **Deletions**: a portion of a chromosome is lost • **Insertions**: a part of one chromosome is removed and added to another chromosome • **Translocations**: segments of two chromosomes are exchanged

Chromosomal Mutation: Changes in Chromosome Number	• **Aneuploidy:** Loss or gain of **whole** (singular) chromosomes.
	  ○ E.g. In humans there could be 45 or 47 chromosomes instead of the normal 46 chromosomes in the nucleus.
	  ○ Down syndrome, caused by an extra copy of chromosome 21.
	• **Polyploidy:** Loss or gain of complete **sets** (two) of chromosomes.

DOT POINTS: • *distinguish between somatic mutations and germ-line mutations and their effect on an organism*	

Somatic Mutations  	• Occur in somatic cells (non-reproductive body cells) after conception. • Affect the **individual** and **cannot be inherited** by offspring nor passed onto the next generation. • Affect only patches of tissue derived from the mutated cell • Are more severe if they occur early in development.
Germ-line Mutations  	• Occur in the germ cells (cells that form the gametes, and the gametes themselves). • Are **hereditary** in nature (**can be passed** down) ○ they occur in the gametes which participate in fertilization • Are present at conception • The zygote formed will be heterozygous for that mutation in all cells. • Every cell in the body carries the mutation
DOT POINTS: • *assess the significance of 'coding' and 'non-coding' DNA segments in the process of mutation*	
Coding DNA	• Is a sequence of DNA that codes for protein. • makes up about 1.5% of the human genome. • Mutations can affect the type or sequence of amino acids used to produce a protein. • The type of mutation will determine how significant the effect on the protein produced will be which then transfers to how significant the effect on the cell will be.

<table>
<tr><td>Non-coding DNA</td><td>

Does not provide instructions for making proteins instead, it contains sequences that act as regulatory elements, determining when and where genes are turned on and off
Makes up about 98.5% of the human genome.
A mutation in noncoding DNA can cause a protein to be expressed in the wrong place or at the wrong time or can reduce or eliminate the expression of an important protein when it is needed.

</td></tr>
<tr><td colspan="2">DOT POINTS:
● investigate the causes of genetic variation relating to the processes of fertilisation, meiosis and mutation</td></tr>
<tr><td>Summary of Fertilisation, Meiosis and Mutation</td><td>
<table>
<tr><td>Fertilisation</td><td>A unique set of chromosomes are made as there is one set of chromosomes from each parent</td></tr>
<tr><td>Meiosis</td><td>Occurs due to the random segregation of the chromosomes that cross over during prophase I</td></tr>
<tr><td>Mutation</td><td>It’s a permanent change in DNA</td></tr>
</table>
</td></tr>
<tr><td colspan="2">DOT POINTS:
● evaluate the effect of mutation, gene flow and genetic drift on the gene pool of populations</td></tr>
<tr><td>Mutation</td><td>

Mutations lead to the formation of new alleles, due to changes in DNA that arise during meiosis.
Advantageous and detrimental mutations are passed on.

Very few mutations are advantageous and selected to increase in frequency.
Detrimental mutations are eliminated,
Neutral mutations remain

Introduction of new genes into the gene pool

Can cause variations in species

If successful, can cause the creation of new species

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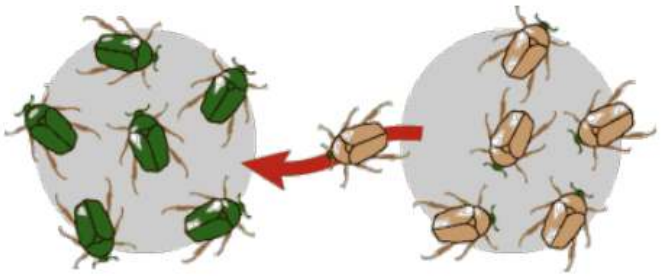
Gene Flow



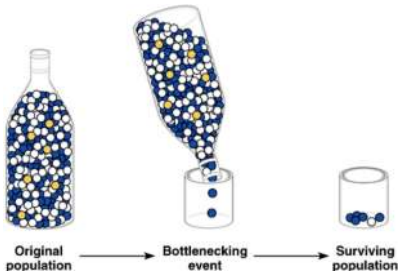
- Occurs when new alleles are added or removed from the gene pool, due to the movement of fertile individuals or gametes.
- Essentially an organism from one group enters (immigration) or exits (emigration) another group which in turn introduces new alleles
- can be a very important source of genetic variation and makes possible new combinations of traits.
- Changes the allele frequency (%) due to the mixing of new individuals
- Factors that affect gene flow:
  - Geographic isolation (mountain)
  - Behavioural barriers (courtship rituals)

Genetic Drift

- An unpredictable chance event which results in changes in allele frequencies from one generation to the next.
- Occurs as a random event and **NOT** natural selection
- Causes individuals to be different
- Doesn't necessarily lead to adaptations to environmental conditions

- **Founder Effect:**
  - Occurs when a few individuals become isolated from a larger population, creating a new population that differs from the original population

- **Bottleneck Effect:**
  - Occurs when a large population reduces suddenly in size, e.g. by a natural disaster or human activities.
    - The surviving population has a much smaller gene pool than the original population.



Inquiry Question 2:

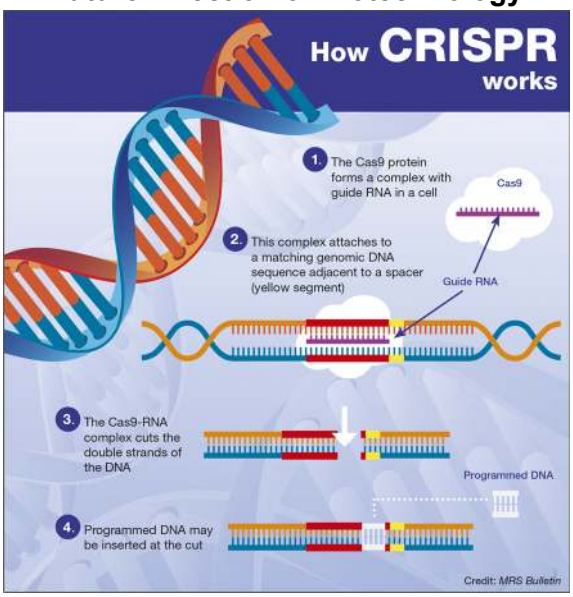
How do genetic techniques affect Earth's biodiversity?

DOT POINTS:

- *investigate the uses and applications of biotechnology (past, present and future), including:*
  - *analysing the social implications and ethical uses of biotechnology, including plant and animal examples*
  - *researching future directions of the use of biotechnology*
  - *evaluating the potential benefits for society of research using genetic technologies*
  - *evaluating the changes to the Earth's biodiversity due to genetic techniques*

Ancient Biotechnologies	• Selective breeding - animals and crops and Fermentation **Impact on Society:** • Plants: Increases the yield and the quality of the harvest. Includes an abundance of food, new type of products • Animals: Food animals (pigs) are bigger, more tender and grow faster. Working animals (horses, dogs) could be bred for specific tasks. **Changes in biodiversity:** • Short term - more biodiversity • Long term - reduced biodiversity • Selective breeding uses one characteristic, so others may be lost • With the same traits, they all may die to one illness they are all susceptible to
Present Biotechnologies	• DNA Manipulation - Recombinant DNA **Impacts on Society** • Allows for the mass production of a certain gene **Changes in biodiversity:** • Short term - more biodiversity • Long term - reduced biodiversity

Social implications and ethical uses	• Referring to issues of non-harm, ethical decisions, individual rights and autonomy, equity and justice, privacy
	• Dolly the Sheep - Cloned via SCNT
	  ○ Raised questions about the cloning of humans and poor health that arose from the first attempts at cloning mammals
	  ■ Dolly Lived only half of her lifespan
	• BT Cotton
	  ○ Uses the Bt bacterium toxin gene to provide immunity to pests, decreases biodiversity, meaning a whole crop could be wiped out by environmental change/insects more likely to develop immunity

Future Direction of Biotechnology
 
 	
	• Biotechnology research has the potential to make revolutionary advances in the fields of medicine, agriculture and the environment.
	• Examples include:
	  ○ Improved diagnosis of diseases
	  ○ predisposition to medical conditions
	  ○ 'Designer drugs' → individualised to a patient's specific genetic profile
	  ○ New Vaccines
	  ○ Treatment for chronic diseases
	• CRISPR
	  ○ Gene-editing tool
	  Process
	  ■ Cas9 protein forms a complex guide RNA in a cell
	  ■ attaches to a matching genomic DNA sequence
	  ■ Cas9-RNA cuts the double strands of the DNA
	  ■ Programmed DNA can be inserted into the cut

Benefits of Biotechnology for Society and Research	• The fundamental aim of biotechnology is to meet human needs or demands in order to improve our quality of life. • Biotechnologies can help society as they can prevent fatalities in some cases • They also allow scientists to complete further research into certain bacteria and other genes to help solve a problem the world is currently facing • They allow for advancements to create higher-quality and more quantity in less time. • Gene therapy → treat human disease, • Genetically modified food → alleviate hunger, • Plant banks/animal cryopreservation—maintain biodiversity and conservation, • Plant/algae based resources → biofuels

Changes to Earth's Biodiversity due to Biotechnology	• Selective breeding requires the decision-making of which traits are better and more productive than others.
	  ○ The increase of these traits initially increases the biodiversity of the Earth but as these traits become more commonly used due to their productivity, the biodiversity will decrease due to the same traits being used.
	  ○ Short term → New traits introduced (increased biodiversity)
	  ○ Long term → As selective breeding occurs, traits are lost while others are seen as more common (decreased biodiversity)

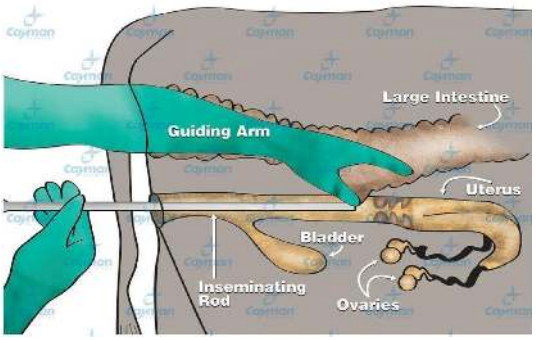
Inquiry Question 3:

Does artificial manipulation of DNA have the potential to change populations forever?

DOT POINTS:

- *investigate the uses and advantages of current genetic technologies that induce genetic change*
- *compare the processes and outcomes of reproductive technologies, including but not limited to:*
  - *artificial insemination*
  - *artificial pollination*

Artificial Insemination

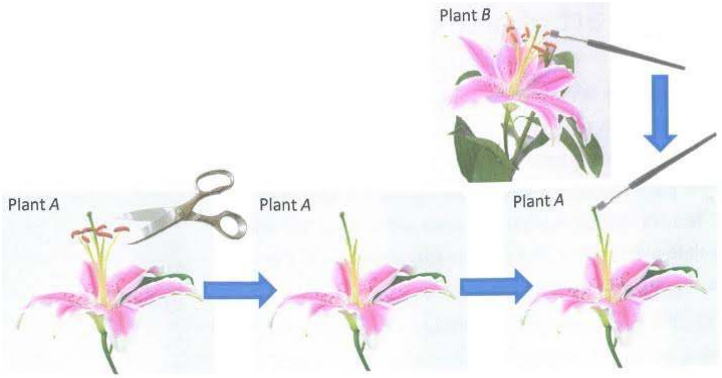


- A reproductive technology that involves the injection of male semen into the vagina or cervix of a female without sexual intercourse
- In agriculture, semen is collected from a male specimen from an artificial vagina or mechanical stimulation
- The semen is then kept cool in liquid nitrogen to keep the sperm cells from drying out
- In the case of agriculture, the sperm is put into the cervix/uterus using an inseminating rod/gun

Advantages:

- Desirable characteristics/traits
- Conservation
- Cheap

Artificial Pollination



- Involves humans taking the pollen from the stamen (male part) of one plant that has certain desired features and placed on the stigma (female part) of another flower of the same species
- Plants can be artificially pollinated in two main ways:
  - **Hand Pollination**
    - Involves the gardener or farmer using a small brush to transfer the pollen from one plant to the stigma of the other plant
  - **Mechanical Pollination**
    - Involves the mass-dusting of pollen onto plants from small aircraft or large blowers

Comparison of Artifical Pollination and Artifical Insemination

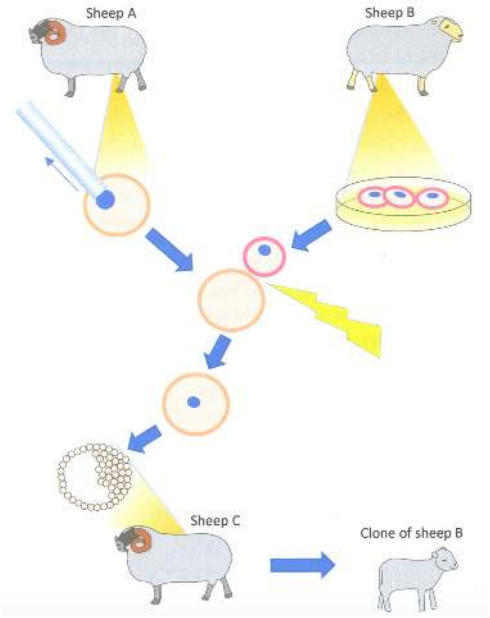
	Artificial Pollination	**Artificial Insemination**
Organisms bred with this reproductive technology	Wheat, flowers, plants etc	Cows, horses, other livestock
Method of collecting male gametes	Collecting pollen from plants with a brush or mechanical tool	Through an artificial vagina or mechanical stimulation
Method of transferring male gametes to female	Apply pollen on the stigma by the brush or mechanical tool	Inserting an inseminating rod/gun into the animal
General purpose of this reproductive technology	Better traits in plants	Better traits in animals

DOT POINTS:

- *investigate and assess the effectiveness of cloning, including but not limited to:*
  - *whole organism cloning*
  - *gene cloning*

Whole Organism Cloning in Plants	• People have been cloning plants, using cuttings and grafting techniques since the beginning of the agricultural revolution which began about 10 000 years ago • Some plants can grow new roots if they have part of their stem cut and placed in soil or water. • In **grafting**, sections of two plants are joined together, a stem cutting (scion) is grafted to the roots of another plant (stock) • In **Plant tissue culturing**, tissue scrapping is taken, is then chemically treated and grown and initially will grow as undifferentiated cells. After time plantlets will grow, which can then be separated and moved to grow elsewhere.

Whole Organism Cloning in Animals



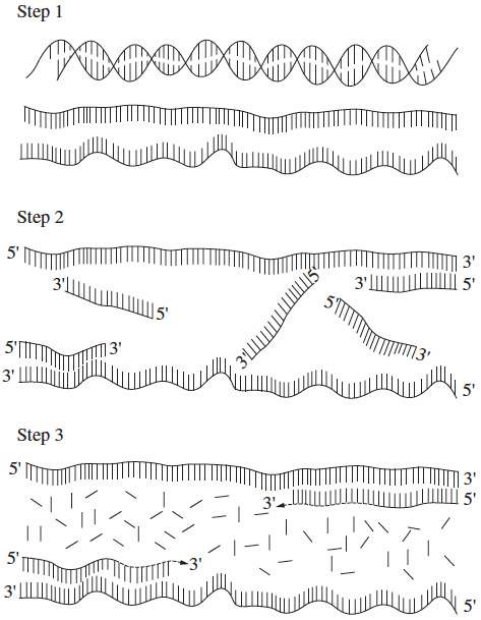
- Creating another organism that has the exact same genetic material as another organism.

Steps of Somatic Cell Nuclear Transfer

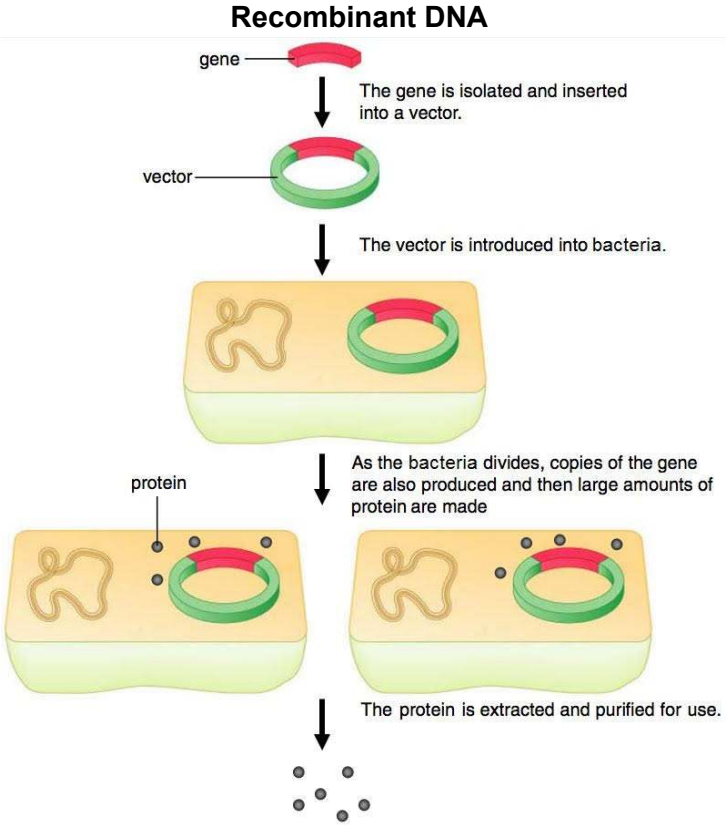
1. A somatic cell is taken from an individual to be cloned
    - The nucleus contains all of the genetic material
2. The nucleus is transferred to a donor egg which has had its nucleus removed (enucleated egg)
3. Electricity is used to fuse together the enucleated egg and somatic cell
4. Once they are fused, specific chemicals stimulate mitosis
5. Once mitosis has occurred, it is implanted into a surrogate mother
    - The surrogate will be given hormones to help the pregnancy
6. The surrogate will give birth to an identical clone

- It took many tries for this to work in a mammal without the baby dying after birth. The first mammal cloned was a sheep in 1996 called Dolly.

Polymerisation chain reaction (PCR)



- A technique that is used to make millions of copies of a gene (amplification)
- PCR mimics what happens when DNA replication occurs before cell division, however, it happens in a laboratory instead.
- Takes 2-3 hours to get a billion copies
- One cycle results in two double-stranded sequences of target DNA, each containing one newly made strand and one original strand.
- **Ingredients in PCR:**
    - DNA segment of interest
    - Primers
    - DNA Nucleotides
    - DNA polymerase enzymes
    - Buffer
- **Step 1:** Denaturation is where the temperature rises to 95ºC so the DNA helix breaks
- **Step 2:** Annealing is where primers bind to the target DNA and initiate polymerisation, the temperature is at 50ºC here
- **Step 3:** Extension is where new strands of DNA are made using the original as a template, by joining the free nucleotides



- The DNA that results from moving a section of DNA from its normal location and inserting it into another site
- Used to clone genes in bacteria
- Large quantities of the desired gene can be obtained if the recombinant DNA is introduced into a host and allowed to replicate

Process

- A DNA fragment is isolated and grown in a lab
- An appropriate plasmid vector is isolated from a bacterial cell
- The Human DNA and plasmid are treated with the same restriction enzyme
    - The enzyme cuts the DNA molecule at a recognition site
    - The cuts produce a fragment that has two 'sticky ends' where nucleotide bases are exposed
    - The DNA is cut so that it can be joined with fragments that match the sticky ends
- The complimentary sticky ends attach to each other through base-pairings and DNA ligase is added to bond them (annealing)
- Plasmid is introduced to a bacterial cell
- Gene cloning occurs bacterium with the plasmid reproduces

DOT POINTS:

- *describe techniques and applications used in recombinant DNA technology, for example:*
    - *the development of transgenic organisms in agricultural and medical applications*

Agricultural Uses of Transgenic Species

- A transgenic species is a species that has genes injected from another species into its genetic code
- In agriculture, transgenic crops increase crop productivity, provide resistance to insect predation and prevent disease.

BT Cotton

- Developed in the mid-1990s, created with strains from the bacterium Bacillus thuringiensis to make it immune to larvae of insects that eat the plant, acting as a pesticide
- India and USA, the two largest producers of cotton, use over 95% Bt cotton.

Golden Rice

- Modified by inserting a gene from maize and a gene from bacteria found in soil → biosynthesise beta-carotene
    - Used by humans to create vitamin A which is good for eyesight

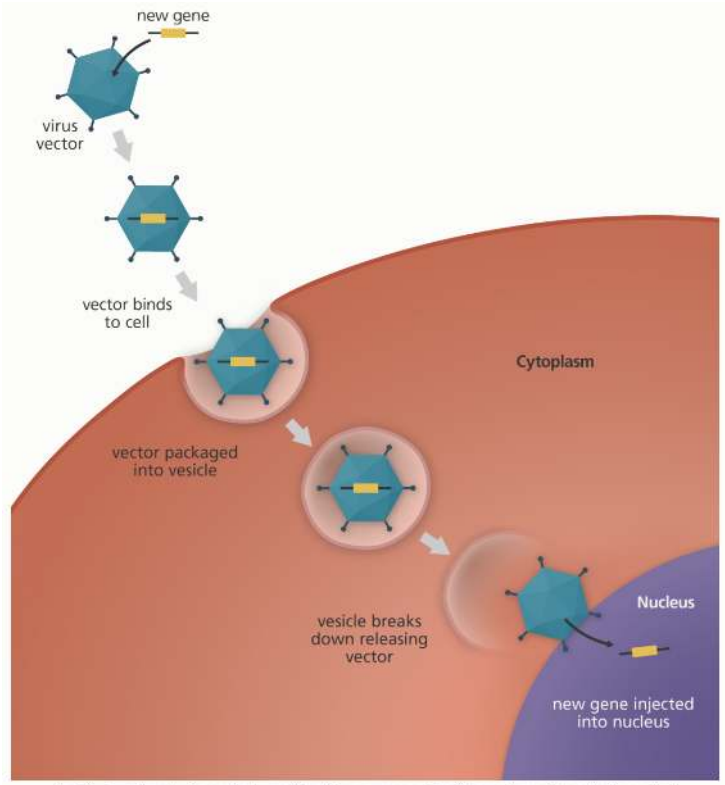
Advantages:

- Supplies nutrition
- Reduces illness → economic burden
- Reduced pesticide usage

Disadvantages:

- Over-regulation → higher prices
- Not enough nutrients produced
- Money to research could've used to supply education to these countries

Medical Uses of Transgenic Species: Gene Therapy



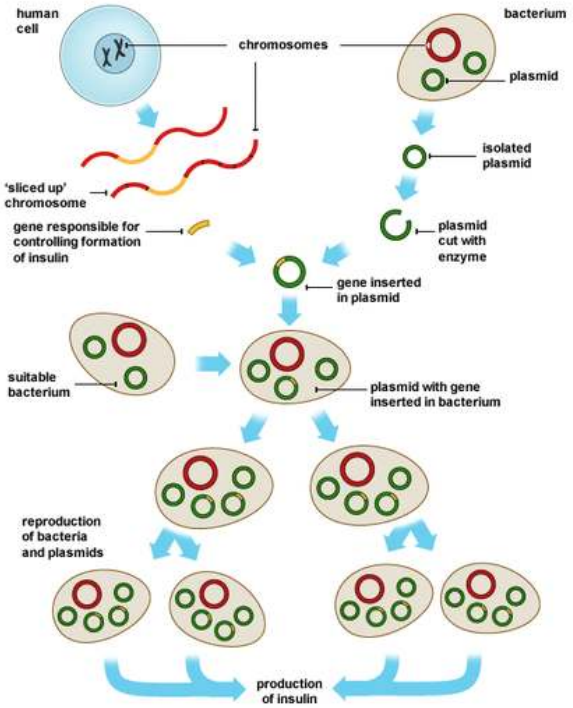
An illustration to show the transfer of a new gene into the nucleus of a cell via a viral vector. Image credit: Genome Research Limited

- An experimental technique that uses genes to treat or prevent disease
- Can Involve:
  - Replacing a mutated gene that causes disease with a healthy copy of the gene.
  - Inactivating, or "knocking out," a mutated gene that is functioning improperly.
  - Introducing a new gene into the body to help fight a disease.
- There are two different types of gene therapy which are:
  - Somatic gene therapy: transfer of a section of DNA to any cell of the body that doesn't produce sperm or eggs.
  - Germline gene therapy: transfer of a section of DNA to cells that produce eggs or sperm → passed onto children

Applications of Gene Therapy:

- X-SCID:
  - X-linked severe combined immune deficiency
  - Have a faulty gene meaning they have no working immune system and therefore have to live in a sanitary bubble
  - With gene therapy, bone marrow from the boy is removed to 'harvest' stem cells
  - The stem cells are infected with a working copy of the X-SCID gene and are put back into the boy
  - 7 out of 10 infants have restored immune function
  - Two of the children initially formed leukaemia → transferred cell cause activation of a cancer-causing gene

Medical Uses of Transgenic Species: Recombinant DNA



- Used to mass produce medical products such as insulin, human growth hormones, blood clotting factors, vaccines and many other drugs

Process:

- Human cells with a gene for healthy insulin are selected.
- Chromosome is removed from the cell
- Insulin gene is cut from the chromosome using restriction endonuclease enzymes
- A bacterial cell is selected → Some of the DNA is in the form of circular plasmids
- Plasmids are removed from bacterial cell
- Plasmids are cut open using the same restriction enzyme
- Insulin gene is inserted into the plasmids using the ligase enzyme
- Plasmids are returned to the bacterial cell
- Bacterial cell is allowed to reproduce in a fermenter
    - All cells contain the human insulin gene

DOT POINTS:

- *evaluate the benefits of using genetic technologies in agricultural, medical and industrial applications*
- *interpret a range of secondary sources to assess the influence of social, economic and cultural contexts on a range of biotechnologies*

Benefits of GMO's (Agricultural)

Social Advantages:

- Addresses matters of global inequality → poverty and food security
- Can increase dialogue between scientists and communities → improve scientific literacy
- Reduced environmental footprint
- Require less tillage → fewer greenhouse gas emissions

Economic Advantages:

- Stimulates agricultural economy
- Provides farmers in 3rd-world countries the ability to grow crops easily and quickly
- Poor-nutrient soil can still grow nutrient-rich foods
- May help desertified ecosystems

Cultural Advantages:

- Essential part of cultural practices

	• Agricultural practices can be preserved in changing climates • Can preserve important foods and maintaining industries
Benefits of Genetic Technologies (Medical)	**Social Advantages:** • Allows a better life for individual affected (somatic) • Can allow for future generations to not be affected with illness (Germline) **Economic Advantages:** • Products such as human insulin can be made on a large commercial scale, reducing costs **Cultural Advantages:** • These processes can be available for people with religious beliefs that wouldn't be allowed to use products from animals to help them survive
Benefits of Spider Silk (Industrial)	**Social Advantages:** • Strong material **Economic Advantages:** • High Yield • Cost Effective • Quality control • Versatility **Cultural Advantages:** •
DOT POINTS: • *evaluate the effect on biodiversity of using biotechnology in agriculture*	
Effect on Biodiversity in Agriculture Biotechnology	• Genetically engineered organisms have the ability to increase the genetic diversity of species • Introduction of genes from different populations such as animals into plants • Salmon anti-freeze is used in strawberries → they can grow in climates that are colder and unsuitable that would cause extreme frost damage to them • Recombinant DNA technologies can also decrease diversity • Transgenic tomatoes that look and taste better will want to be used more than regular tomatoes • Pest resistant crops (such as BT Cotton) will produce higher yields and will be more popular.