Genetic Change Study Notes

TOPIC 1 - Mutation:

Inquiry Question: How does mutation introduce new alleles into a population?

1.1 Mutation Operations

- *Explain how a range of mutagens operate, including:*
    - *Electromagnetic radiation sources*
    - *Chemical*
    - *Naturally occurring mutagens*

<u>Mutagens:</u>

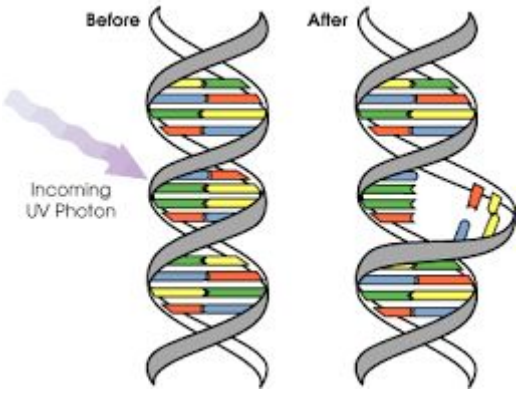
- Definition → an agent which causes a genetic mutation
- DNA:
    - DNA has to replicate exactly and has to be maintained in order for cells to function correctly
    - DNA is usually able to detect changes to the DNA and fix the errors (becomes a problem when the repair system is compromised)
    - Mutagens are any agent that causes a mutation to the DNA these are often carcinogenic (cancer-causing)
    - Mutation → a change to the DNA sequence
- Types of mutagens:
    - Chemical
    - Naturally occurring mutagens (biological and non-biological)
    - Electromagnetic radiation (any form of radiation)

1.1.1 Electromagnetic Radiation

- EMR = short wavelength and high energy
- This causes atoms in the cell to become charged meaning that this weakens the hydrogen bonds and the nitrogen bases break chemical bonds. The cell could either die or live on with mutated DNA
- Radiation is highly penetrative because it is composed of high energy waves. It is therefore able to enter cells from external sources and interfere with DNA molecules in the nucleus
- Interference by electromagnetic radiation can cause bonds within the DNA structure to break. When this occurs there may be a change to the chemical composition of the DNA molecule. Parts may be deleted or rearranged or the shape of the entire molecule may change. This may lead to a mutation if the DNA repair system is unable to repair the change or repairs it incorrectly
- Radiation may also damage DNA directly by ionising other molecules in the cell to produce free radicals. These free radicals are highly reactive (unpaired number of electrons) meaning they want to take electrons from other molecules. These may react with DNA to damage it.
- Radiation is energy being transmitted through space from a source e.g. the sun transmits electromagnetic radiation (EMR)
    - UV light is a form of radiation from the sun
    - It isn't always harmful but damage can arise in high oses

- Excitation of adjacent pyrimidines (T and C) by UV induces the formation of covalent linkages

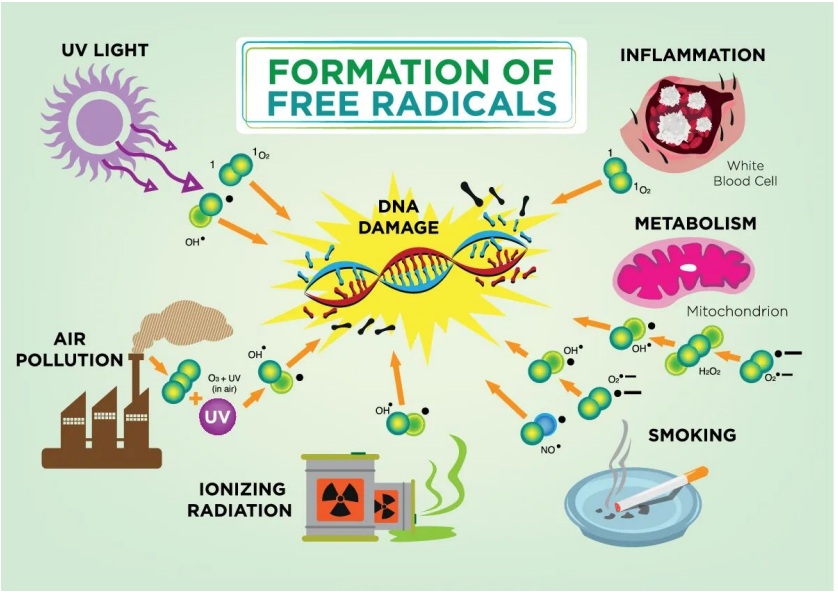
- Some forms of radiation are more harmful than others
- Prolonged or overexposure can be very dangerous
- EMR is all around us - ranging from low energy radio waves up to the very high-frequency gamma rays
- Higher energy EMR including UV, X-ray and gamma rays are able to damage the atoms in our DNA
- UV from the sun can displace (move) the electrons in atoms, ionizing the bases in DNA (giving them a charge). This changes the shape of the DNA → leads to skin cancer cells



- X-rays and gamma rays are often used in medical imaging or sadly used in warfare
- Direction mutation → High energy radiation can break the sugar-phosphate backbone of DNA. this can distort or displace a whole section of DNA
- Indirect mutation → When the radiation causes other molecules to lose an electron. These reactive molecules now interact and interfere with the DNA causing mutations. DNA is very specific and needs all its aspects to function properly otherwise it becomes unstable.

1.1.2 Chemical Mutagens

- If you are exposed to a particular chemical every single day, it will build up in your system and could cause mutations (high frequency and for long periods of time)
- Chemical mutagens come from free radicals which are highly reactive and try to become more stable by taking electrons from other molecules like DNA which damages it
- Free radicals are elements that have become positively charged and they want to take electrons from other molecules like DNA to become stable



- Free radicals are found in alcohol, tobacco smoke, heavy metals, chemical warfare, industrial solvents, UV light and pollution
- Example → Mustard gas
- How do chemicals mutate:
    1. Chemicals can accidentally be incorporated into DNA, instead of proper nucleotides. 5-BDU is a chemical that tricks DNA polymerase into thinking it is thymine.
    2. Chemicals can actually insert itself into DNA and it creates a bulge in DNA which prevents DNA replication from happening
    3. Chemicals make gaps in DNA

1.1.3 Naturally Occurring Mutagens

- Mycotoxins - fungi toxin can insert itself into DNA and change shape/structure
- Biological mutagens - bacteria, fungi, viruses can make free radical toxins that damage DNA
- Certain viruses e.g. HPV can alter the host DNA, damaging the replication cycle and cause cancer - HPV-16 and HPV-32 have both been linked to cervical cancer and a vaccine has been developed for the prevention
- Viruses replicate by inserting their DNA into host cells. This creates a disruption in normal cell function and may lead to lasting mutational changes
- Bacterial infections can induce inflammation which may reduce the efficiency of DNA repair systems and therefore increasing the rate of mutation i.e. *Heliobacter pylori* has been linked to the development of stomach cancer
- Non-biological mutagens include heavy metals such as mercury, lead, chromium and cadmium
- These metals can cause mistakes in the accuracy of DNA repair and so mistakes are easily made when there is prolonged exposure to these metals

1.2 Processes and Effects of Mutations

- *Compare the causes, processes and effects of different types of mutation, including but not limited to:*
    - *Point mutation*
    - *Chromosomal mutation*

1.2.1 Causes of Mutation

- Mutations → 'mistakes' in a cell's DNA that lead to abnormal protein production
- Mutations can result in a number of different processes
- Mutagens are not the only sources of mutation, the process of DNA replication can also cause mutation
- Errors in the replication of the genetic code are common and usually, the DNA is able to repair itself, sometimes when the rate of mutation rises above the average, the efficacy of repair can be diminished.

1.2.2 Types of Mutations

1. Point mutations
2. Chromosomal mutations

MUTATION	POINT	CHROMOSOMAL
DEFINITION	Only one nucleotide is changed	Change in structure of chromosome
CAUSES	Error in DNA replication	Error in cell division
PROCESSES	Frameshift (insertion & deletion) or substitution (mis-sense, nonsense & silent)	Deletion, inversion, translocation, duplication & non-disjunctions
EFFECTS	Frameshift - all codons after mutation are affected (= different amino acids) Mis-sense - different amino acid Non-sense - unfinished protein Silent - no effect	All - breakage in middle of gene destroys the gene Deletion, inversion & translocation - genes in another place Duplication - changes in amount Non-disjunction - varied effect
EXAMPLE	Sickle cell anaemia - point mutation in haemoglobin gene (GAG → GTG)	Trisomy 21 - non-disjunction during gamete formation (3 #21)

Point mutations:

- When one DNA base is replaced with another
- Only alters, adds or removes one nucleotide from a sequence of DNA or RNA
- Also called SNPs = single nucleotide polymorphism
- This change in a base in DNA will change the corresponding RNA and therefore the protein too
- This mutation is classified by its effect on DNA and not for how it affects the resulting protein
- Example → Sickle Cell Anemia (substitution)
- Typically only affect one gene
- Includes substitution, insertion and deletion
- Substitution → when one nucleotide is switched out for a different one (e.g. an A is swapped for a C)
- Insertion → when nucleotides are added into a sequence (frameshift)
- Deletion → when nucleotides are deleted from a sequence (frameshift)

Frame-shift mutations:

- When a base is added to a DNA sequence
- Can be a deletion or insertion of one or more bases, this results in a shift in the frame. The reading becomes wrong, codons are going to code for a different amino acid
- This change leads to an additional base being in the resulting messenger RNA sequence
- The wrong proteins are being made and this means that the entire cell might not be able to function at all
- This mutation changes the reading frame of the RNA
- All codons will be slightly different as an extra codon will cause a change in the sequence
- Usually have more significant effects than point mutations
- This mutation is classified by its effect on DNA and not for how it affects the resulting protein

Non-sense mutations (a type of point):

- Any genetic mutation that leads to the RNA sequence becoming a stop codon instead
- Effect the resulting protein more than missense mutations do as the new stop codon could take away a huge section of the protein instead of changing one amino acid to another
- This means it could stop too early or too late in the sequence

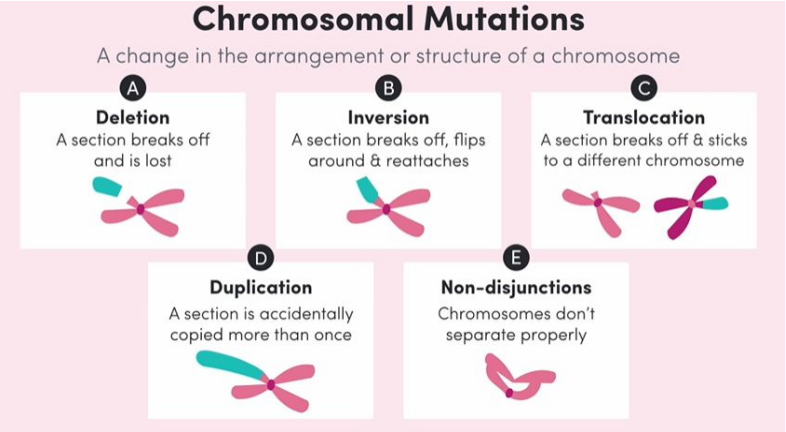
- Occurs in translation and tells it to stop the polypeptide chain being produced → can have detrimental effects

Missense mutations (a type of point):

- Any genetic mutation that changes an amino acid from one to another
- Silent mutations:
  - Doesn't actually affect the protein at all
  - This is because many different RNA codons can code for the same amino acid, it's possible that the mutation might not affect the protein at all
- Conservative mutations:
  - When the new amino acid is of the same type as the original
- Non-conservative mutations:
  - When a new amino acid is of a different type from the original

Chromosomal mutations:

- Large sections of chromosomes are altered or changed
- A chromosomal mutation (e.g translocation or non-dysfunction) is when there is a change to the structure of the actual chromosome itself eg a whole chromosome added or missing- altering the number of chromosomes in total- this can happen both germline and somatic cells for different reasons

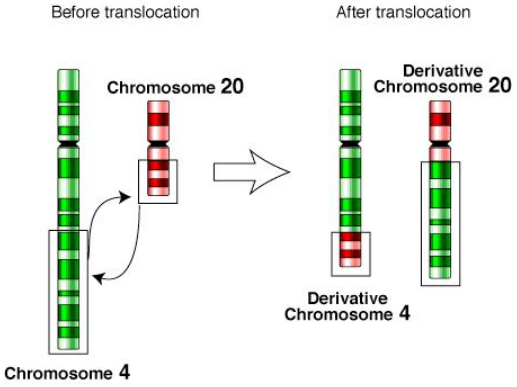


- Can involve changing the structure of a chromosome, the loss or gain of part of a chromosome
- Have serious implications on growth, development and survival of individuals (due to large amounts of chromosomal DNA being affected)
- **Aneuploidy** is a chromosomal mutation where there is one or more extra chromosomes, or one or more fewer chromosomes. **Polyploidy** is a chromosomal mutation in which a cell has entire extra sets of chromosomes.
- Types: deletion, inversion, translocation, nondisjunction and duplication

1. Deletion:
   - Due to breakage
   - A piece of a chromosome is **lost/removed**
   - A chromosome deletion is possible, where an entire section of a chromosome is deleted. Diseases that can be caused by deletion mutation can include 22q11. 2 deletion syndrome, cystic fibrosis, Turner syndrome, and Williams syndrome.

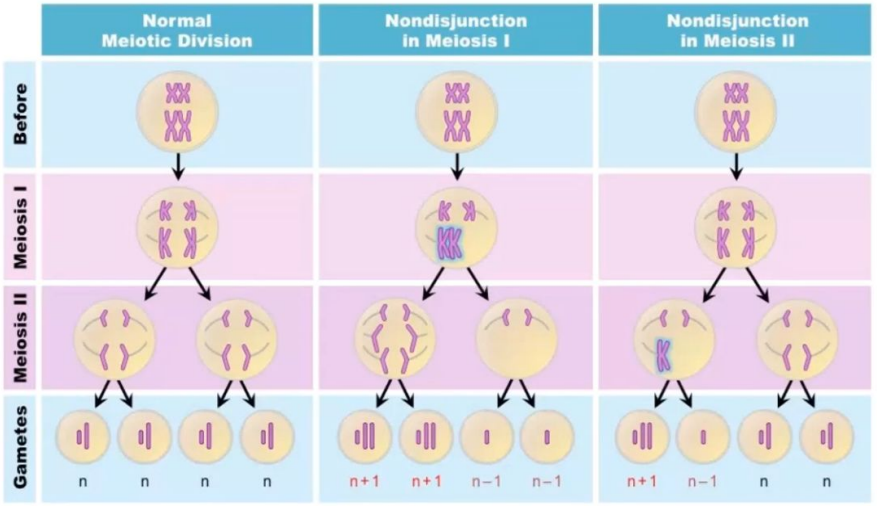
2. Inversion:
   - Chromosome segment breaks off
   - Segment **flips** around backwards
   - Segment reattaches

3. Translocation:
   - Involves two chromosomes that aren't homologous
   - Part of one chromosome is **transferred** to another chromosome



4. Duplication:
   - Occurs when a gene sequence is **repeated**
   - The mutated chromosome has an extra copy of genes

5. Nondisjunction:
   - Failure of chromosomes to separate during meiosis
   - Causes gametes to have too many or too few chromosomes



EXAMPLE - Down Syndrome:

- Most common type = trisomy 21 (all cells have 3 copies of chromosome #21 instead of 2)
- This is an example of a nondisjunction that occurs during the formation of gametes
- Down's syndrome causes a distinct facial appearance, intellectual disability and developmental delays. It may be associated with thyroid or heart disease.
- Other symptoms include:
  - Developmentally → delayed development, learning disability, short stature, or speech delay in a child
  - Eyes → lazy eye or spots
  - Also common → difficulty thinking and understanding, brachycephaly, upslanting palpebral fissures, atlantoaxial instability, bent little finger, congenital heart disease, displacement of the tongue, excess skin on the back of the neck, flaccid muscles, hearing loss, immune deficiency, low-set ears, mouth breathing, obesity, obstructive sleep apnea, polycythemia, seborrheic dermatitis, single line on palm, thickening of the skin of the palms and soles, thyroid disease, or vision disorder

1.3 Somatic and Germ-line Mutations

- Distinguish *between somatic mutations and germline mutations and their effect on an organism*

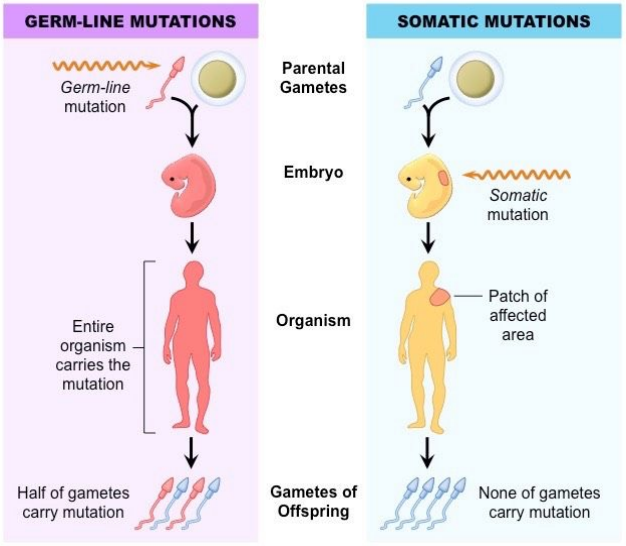
Somatic mutations:

- Mutations that occur in body cells
- Often occur early in the cell cycle but should be repaired before mitosis occurs
- If the repair fails, then the resulting daughter cells from mitosis will carry the mutation
- These mutations usually only affect the individual and are rarely passed on to the next generation
- Some examples of somatic mutations are thalassemia, cystic fibrosis and Tay-Sachs disease
- Cancers and tumours are also often a result of somatic mutations
- Skin cancer - skin cells are altered by UV rays and this can mean uncontrolled cell growth
- This can result in a change in the organism's phenotype in affected areas such as body, arms, legs
- Can lead to uncontrollable growth of unspecialised cells
- Can also result in a change in the organism's physiological characteristics such as rendering the organism unable to produce a protein leading to the death of brain cells known as Tay Sachs disease

Germline mutations:

- Germline mutations occur in the reproductive cells and can be passed onto the offspring - sometimes called gametic mutations
- A germline mutation is any mutation that occurs within a germ cell (egg or sperm) so it could be a dna base point mutation, gene mutation or a chromosomal mutation
- Carried in the sperm or ovum

- If a gamete carrying the mutation is fertilised, then the resulting zygote and all of its cells will also carry this mutation
- Example → BRCA1 and BRCA2, sickle cell anemia, colour-blindness, cystic fibrosis
- Any inherited or predisposed to inheriting cancer is considered to be a germline mutation
- gametes/germline cells are the basis of all other cells in the body, when fertilisation occurs, a paternal and maternal gamete combine to form a zygote
- Germline mutations are caused by both internal and external factors



MUTATION	GERM-LINE	SOMATIC
DEFINITION	Change in DNA of germ cell (AKA a cell which forms sex cells)	Change in DNA of somatic cell (AKA a body cell)
EFFECT: ON ORGANISM	None	All daughter cells from the original, mutated cell will have the mutation
EFFECT: ON OFFSPRING	All cells will have the mutation	None
EXAMPLE	Down syndrome	Lung cancer

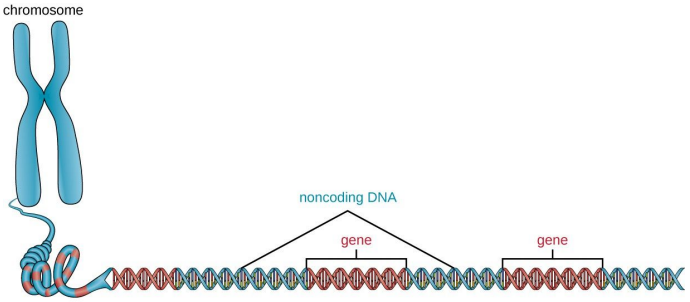
1.4 Coding and Non-Coding DNA

- *Assess the significance of 'coding' and 'non-coding' DNA segments in the process of mutation*

- The human genome is composed of over 3 billion base pairs but not all of this information actually codes for a protein in the cell
- There are roughly 20,000 protein-coding genes, comprising only about 1.5% of the entire genome → expressed through transcription and translation
- The rest of the human genome is 'non-coding' DNA and most of it doesn't have an identified purpose in the cell
- Some portions of non-coding DNA have been identified as serving important functional purposes

Non-coding (introns):

- Codes in DNA that are not used in the final process of translation - no actual protein produced
- 95% of DNA is non-coding



- Although it doesn't code for protein it still important
- It makes functional RNA such as tRNA or rRNA → important for protein synthesis
- It codes for the regulation of the gene (what makes the gene switch on and off) we well as when and where the protein is made
- Regulatory sequences act to increase or decrease the amounts of protein being produced:
  - Promoters = regions of DNA before a gene which help RNA polymerase to bind to the coding DNA
  - Operators = regions of DNA where repressors combine to spot protein synthesis from occurring
- Therefore introns play a role in controlling gene expression and if mutated will affect the gene expression
- Meaning, any particular change in non-coding DNAs will result in a protein to be expressed in the wrong time or place
- Repetitive sequences:
  - Regions of DNA which are repeated continuously
  - Most repetitive sequences are introduced by viruses
  - Some is non-functional in that they are inactive copies of genes
- Mutations in the non-coding region also affect the ability of introns to be removed prior to translation which can lead to uncontrolled cell growth (cancer)
- Many regions of noncoding DNA play a role in the control of gene activity, determining when and where certain genes are turned on or off. These mutations include changes in single DNA building blocks (point mutations), insertions, deletions, duplications, and translocations.

Coding DNA (exons):

- Coding DNA segments play a vital role in the production of a protein that controls the function of many biological processes in our system
- A single change in coding segments will cause a mishap in the processes
- If a mutation occurs in the coding sequence it will alter the polypeptide chain, the structure or function of the protein produced and therefore directly affecting the phenotype of the organism
- These mutations are often associated with a genetic disease

- Example → haemophilia, cystic fibrosis
- Mutations in coding segments can stop a protein from functioning normally
- Some mutations play a role in increasing adaptability of an organism in changing environment conditions - this is good for evolution

MUTATION	CODING DNA	NON-CODING DNA
DEFINITION	DNA which codes for the amino acid sequence of a protein	DNA which doesn't code for proteins
EFFECT OF MUTATION	Vary depending on type of mutation and what cell it's in	Makes functional RNA: change in efficiency of protein synthesis Regulatory sequences: change in amount of protein produced Repetitive sequences: none
EXAMPLE	Sickle cell anaemia	Lung cancer

1.5 Causes of Genetic Variation

- *Investigate the causes of genetic variation relating to the processes of fertilisation, meiosis and mutation*

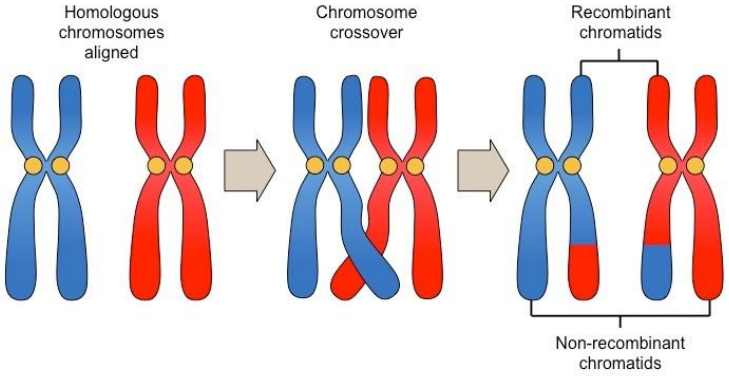
Genetic Variation:

- Variation is essential for survival and is the driving force of evolution → allows populations to adapt to the environment and ensuring survival in the face of selection pressures
- Mutations lead to variation within a species as they affect the composition of genes and proteins → this could either be positive (leading to a desirable trait) or negative (causing the organism to die)
- The processes of meiosis and fertilisation also allow for increased variation in the population. The human genome is designed so that we have a 'back-up' for most genes in the form of multiple alleles
- During sexual reproduction, genes separate, rearrange and are combined in new ways, so that what we inherit is always slightly different to our parents

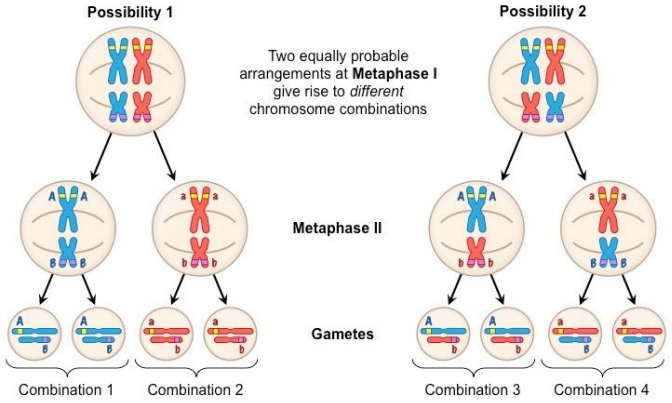
Summary of genetic variation		
	Process	Variation
Meiosis	- The production of gametes (sperm and ovum) - One parent cell becomes four daughter cells, each with half the number of required chromosomes (one chromatid of each chromosome)	- Mutation during DNA replication - Crossing over - Random segregation - Independent assortment
Fertilisation	- Two gametes (one sperm and one egg) come together to form a zygote - The full set of chromosomes is restored (23 pairs, 46 in total)	- Random selection of gametes - Interaction of dominant and recessive genes

1.5.1 Mistakes during meiosis

- Crossing over and independent assortment are two points during meiosis where mutations and mistakes can arise
- Mutations lead to variation which is necessary for evolution
- Crossing over:
  - Involves the exchange of chromosome segments between paired homologous chromosomes that occurs during prophase I
  - This means that some of the genes originating on the maternal chromosome get mixed in with genes on the paternal chromosome
  - Sometimes crossing over isn't equal and one chromosome gets a longer piece than the other
  - This can result in gene duplication in the chromosome that got more DNA
  - This can then give rise to new genes because the extra gene can have mutations while the duplicate gene continues to carry out its normal function



- Independent assortment:
  - When meiosis is complete the resulting eggs or sperm have a mixture of maternal and paternal chromosomes
  - The homologous chromosomes are randomly intermixed during meiosis so that each egg or sperm cell has a unique combination of maternal and paternal alleles



- Random segregation:
  - Random segregation is the random separation of chromatids from every homologous pair into gametes during anaphase, which increases variation by ensuring that inherited homologous chromosomes come from both paternal and maternal genetic information as opposed to all chromatids from the parent's father or mother.
- How much variation is produced?
  - The number of combinations of 23 maternal and paternal homologues that can result from independent assortment is 223, about 8 million
  - This doesn't account for variation caused by mutations or crossing over
  - This is why is it practically impossible to have two siblings identical unless they are twins

1.5.2 Fertilisation Variation

- Random fertilisation is a further mechanism that produces genetic variation in the process of sexual reproduction
- Fertilisation randomly brings together two gametes produced in two different individuals
- For any man and woman, the number of unique combinations of genes that could occur in the offspring is 8 million times 8 million - not counting variation caused by crossing over and mutation
- Fertilisation increases variation because it requires two gametes from two different parent organisms to combine in order to restore a full set of chromosomes
- This is a random process meaning that potential combinations occur with equal statistical probability
- Each individual inherits one set of genes from each parent meaning at least two alleles for each trait, the dominant and recessive interaction of these alleles during expression will increase population variability

1.6 Population Genetics

- *Evaluate the effect of mutation, gene flow and genetic drift on the gene pool of populations*

<u>Gene pool:</u>

- Total collection of alleles for a population
- The gene pool retains all of the genetic information of the population, including the number of genes, number of alleles and the allele frequency
- Gene pools are dynamic = they constantly change. They change due to:
  - How often different alleles come up in the population
  - New alleles arising

<u>Population Genetics:</u>

- Definition → the study of genetic differences within and between populations
- Largely involves the tracking of genes and their alleles across space and time

- Understanding population genetics is important to evolutionary biology because it allows us to comprehend how traits have become prevalent in populations and therefore how populations have changed and evolved into new species
- Theories in population genetics rely on the fundamental principles of the theory of evolution

KEY CONCEPTS

FACTOR	WHAT IS IT	CHANGE IN ALLELES IS DUE TO ...	EFFECT ON THE NEXT GENERATION
Selective pressure	The main selective pressure is natural selection (Darwin)	variations that are passed on because they make individuals more likely to survive (and more virile)	Alleles that make individuals 'fitter' – more likely to survive and live to reproductive age – become most frequent
Sexual selection	Certain individuals are more attractive to mates and therefore more likely to breed	non-random mating (mating is not random; some individuals mate more than others)	Alleles of individuals who are most successful at mating are more common in the gene pool
Mutation	New genes arise due to 'errors' in DNA replication during meiosis (gametogenesis); they may be beneficial, neutral or harmful	new alleles arising during gametogenesis being introduced into a population	New alleles that are beneficial become more frequent in the population
Genetic drift (more obvious in smaller populations)	Random events (e.g. a tornado) lead to a change in gene frequency because some individuals are wiped out	random chance (non-selective; does not depend on genetic make-up)	Causes individuals within a population to be different (not necessarily more successful) due to random chance
Gene flow (more obvious in smaller populations)	Individuals with different genes come into a population and spread their alleles	mixing with new genetically different individuals (e.g. immigration, emigration)	Allele frequency in the population changes

<u>Evolution</u>:

- A process that is responsible for bringing about diversity in individuals within a population
- The change in allele frequency within a gene pool in a population over time is known as microevolution
- The four main mechanisms that have played a fundamental role in driving microevolution forward include: mutation, genetic drift, gene flow and natural selection

<u>Selection Pressures:</u>

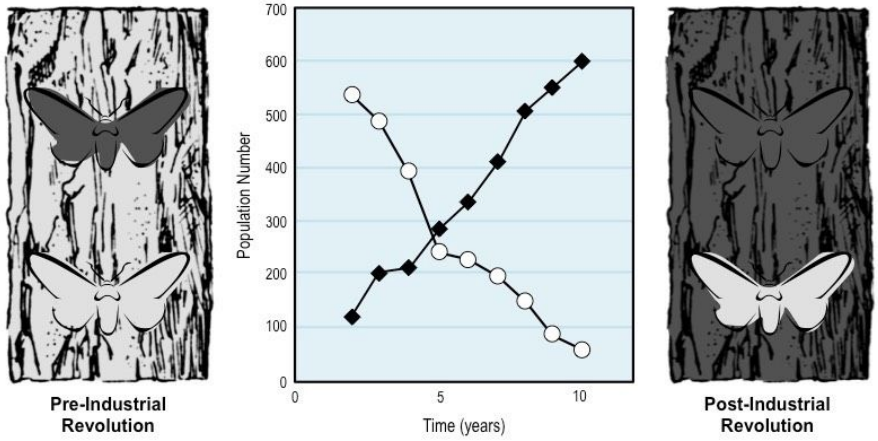
- Refers to any external factor which affects an organism's ability to survive in its environment
- Includes: weather, temperature, predators, food availability and mate choice
- Selection pressures change the allele frequency in a population as organisms with advantageous alleles are more likely to survive
- A positive selection pressure works to increase the frequency of an allele in a gene pool
- A negative selection pressure works to decrease the frequency of an allele in a gene pool

<u>Mutations:</u>

- Any change to DNA caused by a mutagen, DNA replication process or during meiosis

Natural selection:

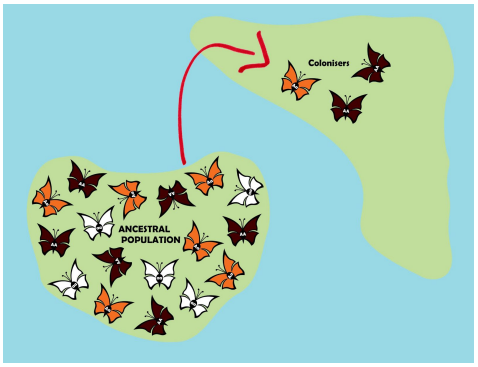
- The theory put forward by Charles Darwin
- A process through which a particular allele of a physical characteristic becomes more or less common in a population over a few generations
- The physical characteristic that provides an adaptive advantage are selected and become more common in the population over generations
- Example → Peppered moth:
  - Before the Industrial Revolution, the dark peppered moth was quite rare in the United Kingdom
  - During the Industrial Revolution, the trees on which these moths rested became covered with soot
  - This resulted in a better chance of survival of these moths to reproduce efficiently, resulting in an increase in the population of these moths significantly.



Genetic Drift:

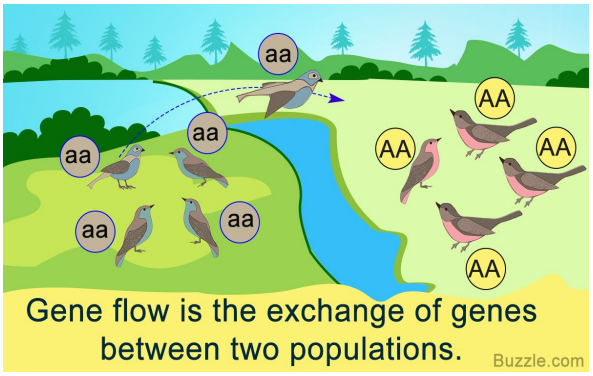
- Depends on luck and chance
- Changes allele frequencies due to chance, not exactly more fit. Randomly sampling or chance events
- Has the most change in relation to allele frequency for smaller population
- Occurs 2 ways:
  1. **Bottleneck effect**: Requires a bottleneck 'event' and it's just bad luck because the event was at that location. (e.g. fires and koalas, already endangered, roughly 40-50% of koalas died in fires, this means the gene pool becomes much more limited and the variation is less). A drastic event that may cause a sudden change in the allele frequency due to reduced population size. This will lead to over-representation of certain alleles and underrepresentation of others in the population. This population size will reduce further as interbreeding between the survivors continues and may increase the chances of an allele becoming homozygous.

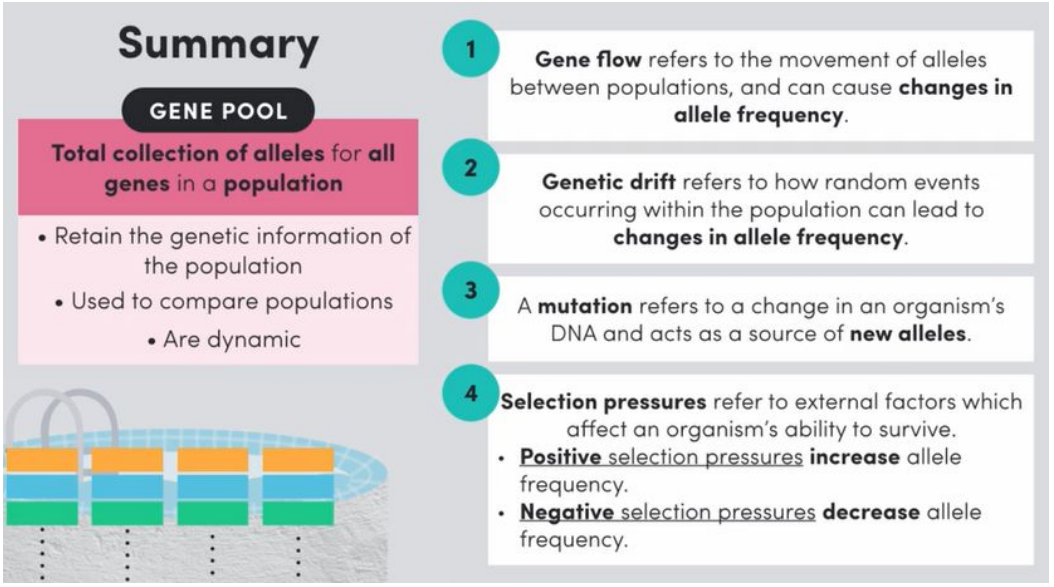
2. **Founder**: A random sample of a population that migrates to a new area and 'found' a new population. When individuals of one population migrate to a new isolated geographical area (that is not inhabited by any other population) to constitute the gene pool of that area. These new individuals make up the gene pool and are a representation of the original population in the new habitat but in a smaller proportion.



Gene Flow:

- It is a change in the allele frequency brought about by the transfer of alleles or gametes from one population to another
- This occurs when individuals migrate from one population to another
- New alleles are introduced into the gene pool of that population, leading to a change in the allele frequency of that population.



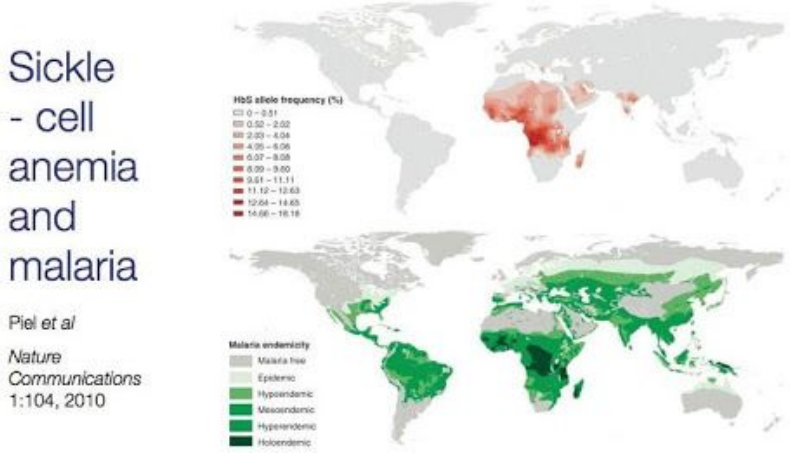


EXAMPLE - Sickle Cell Disease:

- In the US between 70,000 -125,000 people have sickle cell disease (doesn't take into account migrants)
- In African Amercians the incidence is 1/500
- High frequencies of people carrying the trait near the coast and the lake and low frequencies in the mountains.
- Where there was a really high level of sickle cell there was also a very high level of malaria
- Sickle cell is known to be genetic but there is a link between high population of malaria in that area
- The children carrying the character for sickle cell disease had a lower parasite count as if they had it partially (heterozygous). Much lower than people with sickle cell
- Study of 5,000 individuals → reliable because he used a lot of subjects and the same frequencies were found in different areas

	N	n
N	NN	Nn
n	Nn	nn

- NN and nn are both at a disadvantage and have high levels of malaria. Only protected from malaria if you are heterozygous. NN means you are very vulnerable to malaria whilst nn means you have sickle cell disease.
- Natural selection is what enabled the sickle cell gene to move along with the malaria disease → the movement of that allele can be contributed to gene flow



The sickle cell gene is found in high frequency where malaria is endemic - it evolved through natural selection processes and if carrying the gene (heterozygous) you are innately protected from malaria. With the advent of travel and migration this allele has mixed into other populations where there is no advantage as malaria is not found there e.g. North America and Australia.

TOPIC 2 - Biotechnology:

Inquiry Question: How do genetic techniques affect Earth's biodiversity?

- *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*

2.1 Applications of Biotechnology

Biotechnology:

- Definition → any technology that exploits biological systems or living organisms for use of medicine, agriculture, industry or food production. (Example: farming, fermentation, antibiotics, genetic engineering and GMOs)

2.1.1 Past, Present and Future Applications

Past technologies:

- The history of biotechnology began over 10,000 years ago with the agricultural revolution
- Farming began at this time in the Middle East but was invented independently in other parts of the world too
- This is arguably the most important invention in human history
- 40,000 years ago Aboriginal Australians were using aquaculture by incorporating sophisticated fish traps engineered using knowledge of fish migration and river systems. The Abroiginal fish traps in the Brewarrina region of NSW still exist today.

Present:

- **Antibiotics** → a medicine (such as penicillin or its derivatives) that inhibits the growth of or destroys microorganisms. Antibiotics have no effect on viral infections and only work properly on bacteria. Antibiotics are produced by microorganisms. Antibiotics are considered a biotechnology as they are naturally produced by organisms.
- **Artificial Insemination (AI)** → involves taking sperm from a male and artificially inserting it into one or several females

Advantages	Disadvantages
Makes breeding easy and cost effective	Disturbs species and their natural habitats
Allows for genetic preservation	Not effective on all species
More resources (e.g. milk and meat)	Ethically not seen as a natural process

High profits	Inbreeding leads to diseases
Makes remote mating possible - transport of sperm to other communities to increase genetic variation of the species	Decreased genetic variation as only certain traits are selected and therefore extinction is likely due to decreased gene pool

- **GMOs / Transgenic species →** A transgenic species is an organism that has had part of another species' genome transferred into its own through the techniques of genetic engineering. Transgenic crop examples include BT cotton and golden rice. Some animals that have been transgenically modified include sheep, goats, pigs, cows, rabbits, rats, mice, fish, insects, parasites and even humans.
- **Cloning →** Cloning is the process of producing genetically identical individuals of an organism either naturally or artificially. In nature, many organisms produce clones through asexual reproduction. Cloning in biotechnology refers to the process of creating clones of organisms or copies of cells or DNA fragments.
- **Gene therapy →** A medical treatment where a healthy copy of a gene is inserted into a non-germline tissue in a developed organism. It can be transported directly into a cell or via an adeno-associated virus vector (a small virus that causes no diseases and causes a very mild immune response). Has the potential to replace conventional medical treatments for diseases and disorders.

Future:

- **Gene editing (CRISPR)** → CRISPR stands for 'clusters of regularly interspaced short palindromic repeats.' It is a specialized region of DNA with two distinct characteristics: the presence of nucleotide repeats and spacers. Repeated sequences of nucleotides — the building blocks of DNA — are distributed throughout a CRISPR region

Selective Breeding:

- Also known as artificial selection: have been used to select certain traits on a single species
- Mainly used for domesticated plants and animals to increase the characteristics most favourable for human use, consumption and enjoyment.
- Examples → wheat, dogs, cattle (meat, milk)
- The process by which humans use animals and plant breeding to selectively develop particular characteristics (phenotypic traits) in a population of individuals by choosing which 2 specific animals or plants should sexually reproduce
- The outcome isn't always achieved as you are not controlling which alleles are passed onto the offspring
- Artificial pollination and artificial insemination are examples of selective breeding
- Artificial pollination:
  - Artificially controlling plant reproduction is artificial pollination
  - In angiosperms (flowering plants), the stamen of a flower are removed and the pollen is dusted onto the stigma of a flower from the same (self-pollination) or another (cross-pollination) plant.
  - Example → Domestication of wheat (10,000 years ago). The key traits of the crop were rapidly modified. One trait that features in domestic wheat over wild wheat is

that it has non-shattering spikes, an adaptation that allows the plant to better retain its seeds and to be harvested more easily.

- Fermentation → a metabolic process that produces chemical changes in organic substrates through the action of enzymes. In biochemistry, it is narrowly defined as the extraction of energy from carbohydrates in the absence of respiration. Chemical breakdown of bacteria, yeast and other microorganisms.
- Pasteurisation → is a process in which water and certain packaged and non-packaged foods are treated with mild heat, usually to less than 100 °C, to eliminate pathogens and extend shelf life.
- Impact of selective breeding:

Issue	Impact
Health of the animal or plant	Crops usually appear unaffected by a transgene but animals may experience adverse effects that affect growth rates
Uncontrollable pest plant species	Only a concern if the transgene promote rapid growth and there was a selective advantage to having the genes in a wild environment
Loss of biodiversity	Reduction of genetic variation may be harmful with environmental changes. However, GM technology may accelerate the trend of used to re-introduce characteristics into the population specifically with seed banks
Reduced genetic variation	Reduces genetic variation, increasing the likelihood of detrimental homozygous recessive traits in offspring

2.1.2 Social and Ethical Implications

- Biotechnologies have become significantly prevalent within society due to new technologies
- Since this area of science is fast developing and there is a lot of unknown aspects, there are ethical issues involved

<u>Positive Uses:</u>

- The fundamental aim of biotechnology is to improve people's quality of life by meeting the growing needs of society
- Using biological tools means solutions to issues such as disease can be created/found
- Genetic diversity:
    - Many biotechnologies involve editing genetic material within and across species
    - This has the potential to create new arrangements of genes, increasing the diversity of traits we see around us
    - This can be positive for evolution as we continue to re-combine traits in order to best survive and thrive in our environments

Concerns:

- Ownership of information:
  - Information is an important commodity and there is controversy surrounding data mining and distributing information in the technology industry
  - Personal and private information is valuable to large companies
  - Biotechnologies and gene technologies allow us to understand more about humans on a fundamental level and the value of this information has the potential to be exploited
- Consumer rights and choices:
  - With biotechnologies such as GM foods, it can often be difficult to identify products which have been edited and those which haven't
  - This may infringe on the consumer's rights to choose the food they would prefer to eat based on their own morals
  - This is why initiatives such as clear and explanatory labelling of GMOs are important
- Regulation:
  - In terms of globalisations, governments need to legislate biotechnology so that they can safely control its development without stifling innovation
  - In addition to considering local security, we should think about how we regulate biotechnology on a global scale
  - This is because biotechnologies and their use will not only affect citizens in certain countries but the whole world
- Biohacking:
  - Due to the emergence of open-access information, availability of molecular biology resources and overall improved scientific literacy in the community, there has been a rise in biohacking
  - This democratisation of research has the potential to advance social good but also poses an interesting dilemma for regulation when individuals are not operating in large and structured institutions

Effect on Biodiversity:

- The use of biotechnology has the potential to be dangerous and can damage species or their environment
- Editing life at its fundamental level may have detrimental effects so it's important to realise that it is the responsibility of humans to create these tools and use them in an appropriate manner
- In editing ecosystems, we have created new species by influencing the emergence traits which are considered desirable
- Biotechnologies have the ability to decrease biodiversity and even wipe a species out or make them endangered

TOPIC 3 - Genetic Technologies:

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

3.1 Current Technologies

- *Investigate the uses and advantages of current genetic technologies that induce genetic change*

Ethics → The ethics surrounding this particular field of science is interesting to consider. Are we working towards the greater good and using this to benefit society and medicine or could this technology be taken and used for more sinister reasons.

<u>Biotechnology and its implications for society:</u>

- Biotechnology → use of biological materials as tools
- Biotechnology can be used for:
  - Medicine
  - Agriculture
  - Industry
  - Environment
- Social implications
  - Refers to the effects on society or part of society or an event or action
  - There can be harm to the environment, bioterrorism, production safety, ethical issues
  - Bioethics involves using an ethical approach to make decisions about biological issues
- Ethical frameworks:
  - Rights and responsibilities: the rights of one imply the responsibilities of another to ensure those rights
  - Consequentialism: weighing the benefits and harms resulting from our actions
  - Autonomy: should we have the right to choose for themselves or is it a unified decision
  - Virtue ethics: a virtue is something that a community accepts as being 'good' such as honesty, kindness and patience. Virtue ethics emphasise decisions that are in line with these characteristics.

<u>Biotechnology - Past, Present and Future:</u>

Period	*Biotechnology*	*Social implications*	*Ethical implications*
Past	- Called 'classical technology' - Incl. yeast to make bread and beer/wine fermentation - Introduced by Mendel who introduced the	Social implications of past technologies would've been concerned with the production of wine/beer	No real ethical issues as animals weren't being manipulated

	idea of genetic information being transferred from one generation to the next		
Present	- Introduction of DNA meant that biotechnology became much more advanced - The applications have varied over time with uses in agriculture, medicine and within the environment - Originally started with animal and plant breeding - Growing crops meant genetically modified crops was introduced	More than 13.3 million farmers around the world use agricultural biotechnology to increase yields, prevent damage from insects and pests and reduce farming's impact on the environment. Growing biotech crops can also help lower the cost of production, cutting back on expenses like fuel, water and herbicides.	Ethical issues usually involve medicine and the unknown impacts they would have in the future for humans. There are also issues with modifying plants and animals as it can sometimes be harmful for the organisms and have negative effects. These ethical issues raise questions into whether biotechnology is okay.
Future	- Involves future treatment of disease, cancer and other genetic disorders. - Cellular metabolic pathways are being manipulated to enable a range of outcomes including edible vaccines, manufacturing cells and organs and the transplantation of organs from animals to humans	Like all technologies, biotechnology offers the potential of enormous benefit but also potential risks. Biotechnology could help address many global problems, such as climate change, an aging society, food security, energy security and infectious diseases, to name just a few	Ethical issues that arise from modern biotechnologies include the availability and use of privileged information, potential for ecological harm, access to new drugs and treatments, and the idea of interfering with nature. Applications include agriculture and health care. Because future technologies aren't being made yet, uncertainty can raise issues.

3.2 Reproductive Technologies

- *Compare the processes and outcomes of reproductive technologies:*
  - *Artificial insemination*
  - *Artificial pollination*

<u>Artificial Pollination:</u>

- Take the pollen from one plant with certain favorable characteristics and placing the pollen on the stigma of another flower also with favourable characteristics
- Used in agriculture, farming, crop growing
- Used by Mendel in his pea plant experiments
- Advantages:
  - Produce crops with favourable characteristics (e.g. larger fruit, increased yield)

    - Can change the genetic composition of a population very quickly to suit the breeders needs
- Disadvantages:
    - Overuse leads to a decrease in genetic diversity and these varieties with less favourable characteristics may contain useful genes which could be used for other purposes
    - When less favourable traits e.g. being vulnerable to attack by pests or disease are inherited along with the favourable traits e.g. high yields of fruit

Artificial Insemination:

- Involves taking the sperm from one animal and artificially inserting it into the female to produce offspring
- Common in agriculture e.g. cattle, horses, sheep, pigs
- Reason → select the best breeding characteristics from the male to inseminate a female with similar or beneficial characteristics
- Advantages:
    - Sperm can be frozen and transported anywhere in the world
    - One male can be used to inseminate many more females than in normal mating
    - Many more offspring with favourable characteristics can be produced than by normal mating
- Disadvantages:
    - Can be quite expensive as the specialised equipment and high quality sperm can be costly
    - Although we may be breeding for desirable traits, they may also pass on less favourable characteristics such as aggressive animals at the same time
    - Overuse of the sperm from one particular breeding line can reduce genetic diversity, causing problems if recessive characteristics and genetic diseases show up in the phenotype

Impact on genetic biodiversity:

- Both techniques can be used to increase the number of offspring that can be generated by one parent and therefore can result in decreased genetic diversity in the population
- Other individuals in the population do not contribute to the next generation thereby limiting the gene pool
- Example → semen from the same bull can be used to impregnate hundreds of cows or pollen from one male flower is more likely to be transferred to several female flowers
- Both reproductive techniques can overcome geographical barriers and therefore allows the genes to be spread more widely across the world
- These techniques could increase genetic diversity by allowing interbreeding between geographically separated organisms and generating new hybrids
- Banks of sperm and pollen can be created to preserve any endangered genes and allow them to be more prevalent in subsequent generations. This helps to prevent loss of genetic diversity

IVF:

- The process by which an egg is fertilised by sperm outside the body (in vitro0
- Evolved as a method for treating infertility
- Differs from artificial insemination in that an egg is fertilized by a sperm outside the mother's body
- IVF is often carried out in conjunction with MOET to maximise the high genetic merit of female cattle. MOET allows cows (who usually only reproduce once a year) to become stud breeders when surrogate mothers are used.
- Steps involved:
  - Ovaries are stimulated with hormones to stimulate egg production
  - Eggs are harvested and sperm introduced for fertilisation
  - Time is required for cells to divide into blastocyst stage
  - Viable embryos are either stored in liquid nitrogen or transferred immediately for implantation
  - If implantation is successful a pregnancy should result

3.3 Cloning

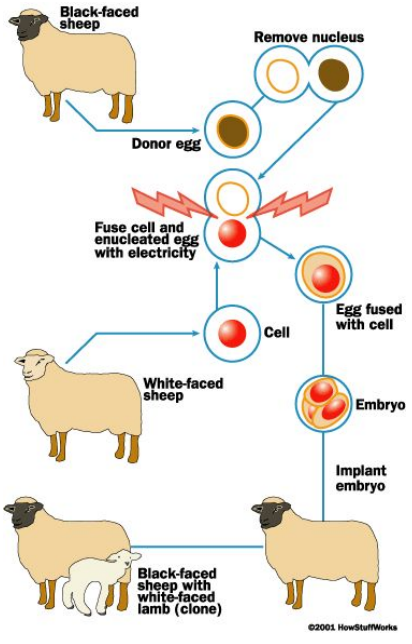
- *Investigate and assess the effectiveness of cloning, including:*
  - *Whole organism cloning*
  - *Gene cloning*

Issues within cloning include:

- Concern for animal welfare
- Moral and religious concerns
- Unforeseen health risks
- Unequal access due to expense

3.3.1 Somatic Cell Nuclear Transfer (Whole Organism Cloning)

- Method of producing genetically identical organisms
- Genes, cells or whole organisms (plants and animals) can be cloned
- Offspring of asexual reproduction are clones
- Techniques include nuclear transfer technology, embryo splitting and therapeutic cloning
- Cloning sheep:
  1. Egg is removed from black-faced sheep. DNA is removed from the cell.
  2. Somatic cells are taken from white-faced sheep (has to be somatic because it needs a diploid set of chromosomes).
  3. Somatic cell and empty donor egg are fused together using electricity ← gives jolt to start dividing
  4. Embryo begins to divide and it put into the original black sheep
  5. The sheep that is produced is genetically identical to the white faced sheep



Cloning Whole Plants:

- Cuttings:
  - Very easy to do
  - A section is removed from the parent plant and placed in soil or water. This cutting then develops its own roots, stems and leaves and it eventually grows into a full-sized version of original plant
- Grafting:
  - More effort than cuttings
  - A cutting from the stem of the plant is bound to the cut stem of another plant with already developed roots. The two stems fuse and grafting grows as if it's part of the whole plant
- Tissue cultures:
  - A section of the parent plant is pulverised releasing individual plant cells. Cells are then grown on a nutrient and hormone containing medium and they then grow to form small sprouts. Sprouts are moved onto another medium to grow further or are re-introduced to natural growing environment

Applications of Whole Organism Cloning:

1. Scientific research:
   - Animal testing plays a key role in the development of drugs and medical devices
   - Cloned animals are useful in drug response testing because their reactions to the drugs are theoretically uniform → means they are reliable
2. Agriculture:
   - Cloning enables desirable traits to be passed onto offspring → this is faster and more reliable than natural breeding
3. Wildlife conservation:
   - Cloning can be used to prevent the extinction of critically endangered species
   - Cloning may be used to bring back species from extinction

Advantages	Disadvantages
- Scientific research → cloning can produce genetically uniform animals that produce reliable drug responses. Plus, cloning can be used to help save human lives by enabling the production of "grown-to-order" tissues (embryonic stem cell technology). - When it comes to agriculture, the characteristics of an organism can be precisely controlled. That is, cloning can be used to produce crops and livestock with certain "desirable" characteristics. Cloned organisms can also be produced in a short time period, compared to normal breeding techniques. Genetic uniformity in crops results in more consistent requirements, growth rates and harvesting times. - For wildlife conservation, cloning can be used to	- Very expensive - Ethical questions surrounding cloning. For example, the consumption of cloned foods could have effects on humans that we don't know about yet, and cloned animals tend to suffer more from adverse health issues and have higher mortality rates. - Cloning can reduce genetic diversity. This means that the population is less likely to survive sudden environmental changes and

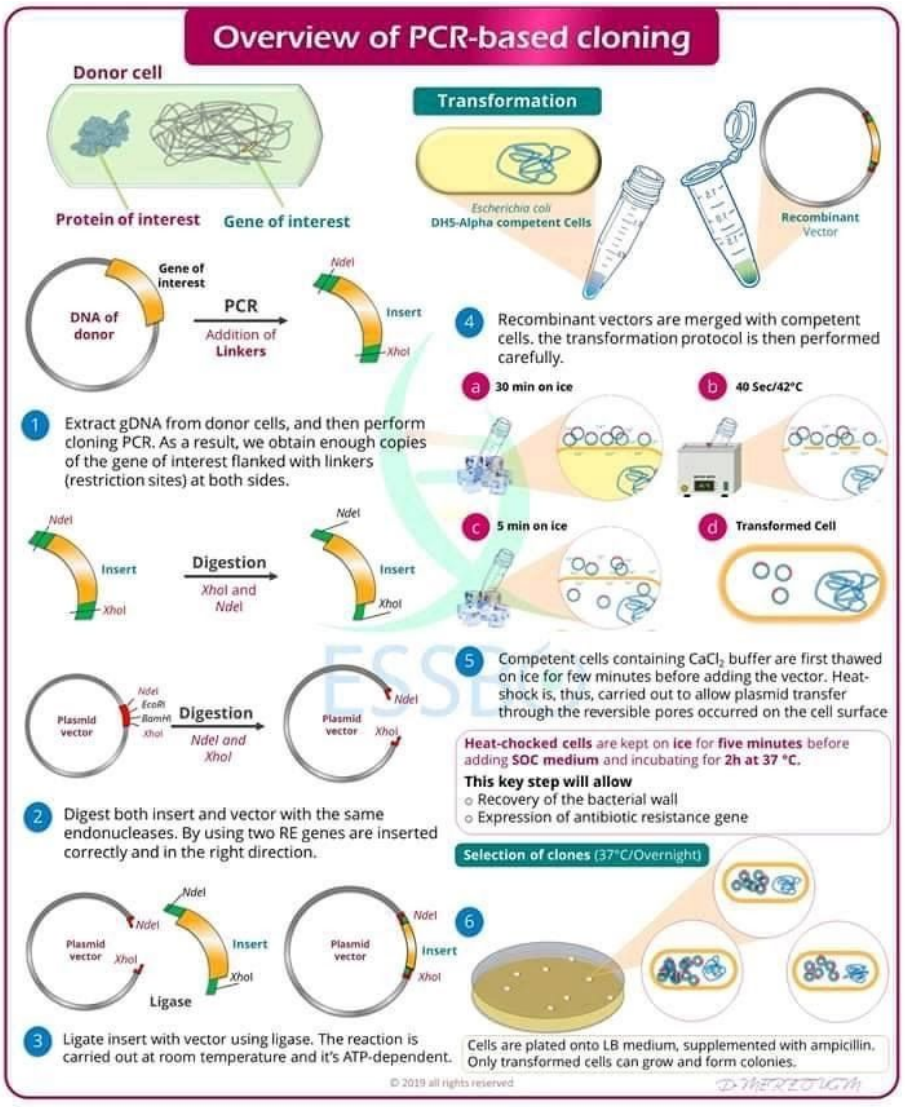
save critically endangered species. As of yet, this hasn't been applied for animals, but has been used to save rare plants (like the Wollemi pine) from extinction.	is more likely to suffer from interbreeding.

3.3.2 Gene Cloning

- Purpose → the process of gene cloning is using to locate and copy a specific gene from the entire genome
- Definition → the process of making an exact copy of a particular gene (usually of interest due to the protein it codes for)
- Steps include:
  1. The DNA is first extracted from the organism and put in a test tube
  2. Restriction enzymes are added to the test tubes
  3. In the second test tube, bacterial plasmids are used and they are also cut with the same restriction enzyme
  4. The test tubes are mixed together, this will allow the gene of interest to recombine with the bacterial plasmid → **this is how recombinant DNA is made**
  5. Bacteria are added to the test tubes and treated with electrical impulses, this creates holes in the cell wall of the bacteria and allows the recombinant plasmids into the bacteria → this bacteria is now a transgenic species (it has recombinant DNA)
  6. Plasmids contain an antibiotic resistance gene, this means they can grow in the presence of antibiotics (they're resistant), if they have been transformed
  7. The bacteria are now grown in a culture containing the antibiotic, they will only grow if they have been transformed
  8. Each colony that grows will represent a different gene, scientists need to find the correct colony with the gene of interest and then grow a pure culture of it

<u>PCR:</u>

- PCR = polymerase chain reaction
- Definition → a technique used to make lots of copies of a specific region of DNA
- In vitro = in a test tube
- Cycling the reaction mixture through a process of:
  - Denaturation (95 degrees) → DNA separates to form single strands
  - Annealing (55 degrees) → primers bind to DNA strands
  - Extension (72 degrees) → DNA polymerase synthesises new DNA (using the sample of DNA as a template)



<u>Recombinant DNA Technology:</u>

- In vivo = in a living organism such as a bacterium to make lots of copies of a target gene for us
- The target gene is inserted into the bacterium
- Involves cutting and pasting the target gene into a plasmid using special enzymes. This plasmid is then used to transport the foreign gene into bacteria. The bacteria is now able to express the gene. As the bacteria grows and divides it also makes copies of the target gene

Advantages	Disadvantages
- Crucial to ensure there is a large enough sample for analysis. This is important, since we may only have a small DNA sample available. DNA can be amplified to larger amounts by gene cloning so that we can identify the individual responsible for the	- Very expensive - Gene cloning raises ethical questions. The use of gene cloning for DNA sequencing and profiling doesn't bring up too many ethical issues generally,

crime! - Necessary to produce the genetic material which is inserted into another organism when forming a transgenic organism. For example, gene cloning is necessary to produce copies of the human insulin gene, which can then be inserted into E. coli and expressed so that insulin can be produced in the large amounts needed to treat people with diabetes.	so long as the DNA is collected with a person's consent and privacy is upheld. However, the ethical issues surrounding transgenic organisms are fairly extensive. For example, genetically modified foods may have detrimental effects on human health.

3.4 Applications of Genetic Technology

- *Describe techniques and applications used in recombinant DNA technology:*
  - *The development of transgenic organisms in agricultural medical application*

3.4.1 Recombinant DNA Technology

- Definition → methods to join together DNA from two different species, in order to produce new genetic combinations
- These technologies are very widely developed because they allow us to get the best out of the world around us and to exploit all manner of biological phenomena for our own advantage
- By combining various genes in new ways across organisms, we are able to create better biological machines
- Plasmids (small, circular piece of DNA) are the most commonly used scaffold DNA
- Steps involved:
  1. Isolation:
     - DNA fragments are extracted from their natural sources
     - The target gene (gene needed because of its protein) and the scaffold DNA are needed for this process
  2. Digestion:
     - Involves using the same restriction enzyme to cut the DNA fragments (both the target gene and the scaffold DNA)
     - Restriction enzymes only cut DNA at a specific base sequence and the same enzyme needs to be used so that they are complementary to each other
  3. Insertion:
     - The ends bind so that the target gene is inserted into the scaffold DNA
  4. Ligation:
     - Even though the DNA bases have bonded together, the backbone isn't connected properly
     - DNA ligase glues the backbone of the DNA together
- PCR and DNA sequencing allow the 'right' plasmids to be chosen
- Bacterial transformation can be used to make lots and lots of copies of the recombinant DNA (amplified)
  - Bacterial transformation is the process of introducing foreign DNA into a bacterial cell

3.4.2 Transgenesis

- Definition → introduction of exogenous genetic material (DNA from an external source/different organism) into a living organism. This is performed so that the organism exhibits a new trait and transfers this trait to its offspring
- All living organisms are made of the same fundamental building blocks: nucleic acids → meaning that a defined sequence will encode the same protein in all organisms
- This consistency within the biosphere allows us to exploit the individual tools which organisms have by inserting them into other species

<u>ATryn Goats:</u>

- Purpose → ATryn is indicated for the prevention of peri-operative and peripartum thromboembolic events in patients with hereditary antithrombin deficiency. HD is a genetic disorder leading to excessive clotting because of a lack of normal antithrombin activity.
- The AT-III protein is what is deficient in the human and the goats are transgenically modified to obtain this protein
- Advantages:
  - Means less human blood donations are required
  - An effective alternative for people that don't have AT-III
  - Goats typically produce high volumes of milk meaning efficiency
- Disadvantages:
  - Can be expensive
  - Ethical issues associated with using animals for human benefit.

<u>BT Cotton:</u>

- Purpose → BT cotton is an insect-resistant transgenic crop designed to combat the bollworm.
- Contains a gene from soil bacterium = Bacillus thuringiensis. This gene codes for a toxin disrupting the digestive system of the cotton-eating caterpillar
- Bt cotton was created by genetically altering the cotton genome to express a microbial protein from the bacterium Bacillus thuringiensis.
- Bt cotton was developed using the common soil bacterium Bacillus thuringiensis (Bt). Bt cotton produces proteins that are toxic to the specific Helicoverpa spp.
- Advantages:
  - Increases yield of cotton due to effective control of three types of bollworms, viz. American, Spotted and Pink bollworms.
  - Reduction in insecticide use
  - Reduction in predators
  - No health hazards due to rare use of insecticides
- Disadvantages:
  - High cost of seeds
  - Potential negative long term effects

3.4.3 Gene Sequencing

- Definition → techniques used to determine the sequence of nucleotides in a section of DNA
- Gene sequencing is widely used of different purposes and it's progression allowed for things such as the Human Genome Project
- The Human Genome Project (HGP) was an international scientific research project with the goal of determining the base pairs that make up human DNA, and of identifying and mapping all of the genes of the human genome from both a physical and a functional standpoint.
- Gene sequencing involves isolating DNA and identifying the sequential order of the nucleotides present in a section of genetic material. From this information, computational programs allow us to transcribe and translate genes in silico

3.4.4 Gene Therapy

- Definition → the correction of genetic disorders by introducing normal, functional genes into cells
- Gene therapy is an important area of research for the medical field because it has the potential to make lasting changes to an individual's health rather than just treating symptoms
- Gene therapy involves the insertion of a corrected, functioning gene into a cell in which there is a defect
- Introducing this healthy genetic material means that the offspring of the cell will inherit this healthy gene and therefore repair the genetic disorder
- There are a number of different techniques used to insert new genetic material into cells, including gene guns, inorganic nanoparticles and viruses
- Scientists have exploited the ability of viruses to evade the immune system and insert themselves and their genetic material into human cells
- By swapping out viral DNA for human genes, and then allowing these non-infectious vectors to enter host cells, we are able to transfect cells with the desired information
- Gene therapy can be administered in two was:
    1. In vivo (within the living)
    2. Ex vivo (out of the living)
- Human gene therapy has been attempted on somatic (body) cells for diseases such as cystic fibrosis, adenosine deaminase deficiency, familial hypercholesterolemia, cancer, and severe combined immunodeficiency (SCID) syndrome.

3.4.5 CRISPR-Cas9

- Uses an RNA guide and it takes the protein complex to the gene it wants to edit, it matches and the CRISPR-Cas9 complex cuts out the DNA and adds in a new base.
- Definition → CRISPR-Cas9 is a unique technology that enables geneticists and medical researchers to edit parts of the genome by removing, adding or altering sections of the DNA sequence.
- It is currently the simplest, most versatile and precise method of genetic manipulation and is therefore causing a lot of talk about CRISPR-Cas9
- Steps:
    1. Identity section of the genome causing health problems

2. Create a specific strand of guide RNA to recognise this specific sequence
3. The guide RNA attaches to the Cas-9 (enzyme)
4. The complex is introduced into the target cells
5. Locates the sequence and cuts the DNA
6. Sequences are then edited, added or removed

- Benefits:
  - Has the potential to correct the genetic errors that cause disease – may one day be able to cure the disease forever
  - It can eliminate microbes that cause disease
  - May resurrect extinct species
  - E.g. bring back the woolly mammoth to help with global warming
  - Could create new healthier food, may help with bigger crop yields – genes that determine size, architecture and shape of a plant for a greater harvest
  - Could eradicate dangerous pests – mosquitos
- Limitations:
  - Potential for it to trigger an immune response, resulting in toxicity
  - Ethics associated – unborn child can't give consent
  - May alter the function and stability of a gene
  - May not be able to recognise plasmid DNA in the cells
- Uses in humans:
  - Functionally inactive genes in human cell lines and cells
  - Target underlying causes of a disease and possibly cure it by modifying the patient's genome
  - Remove allergies, diseases such as HIV, cystic fibrosis, eliminate cancers
- Uses in plants and animals:
  - Allergy-free goods
  - Eliminate mosquitos
  - Greener fuel
  - De-extinction

3.5 Benefits of Genetic Technologies

- *Evaluate the benefits of using genetic technologies in agricultural, medical and industrial applications*

<u>ATryn Goats → MEDICAL:</u>

- This is a transgenic example
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  - Reduction in insecticide use
  - Reduction in predators
  - No health hazards due to rare use of insecticides
- Disadvantages:
  - High cost of seeds
  - Potential negative long term effects

Spider Goats→ INDUSTRIAL:

- This is a transgenic example
- Spider genes - genes that produce the silk in spiders are inserted into the goat
- Purpose → Due to its strength and elasticity, spider silk fiber could have several industrial uses, such as for making bulletproof vests and improved car airbags
- Advantages:
  - Can be used in cosmetic and medical procedures
  - Easier to milk goats than spiders
  - More cost effective than milking spiders
- Disadvantages:
  - Long-lasting effects are unknown (could be harmful to humans)

Application	Techniques	Benefits
Agriculture	- Selective breeding - Artificial pollination - Transgenesis	- Creation of crop and livestock species which exhibit favourable traits (higher yields, higher nutritional value) - Creation of organisms which don't require use of insecticides or herbicides, decreasing the impact on the environment - Increased food security as a solution to global poverty and food shortages
Medicine	- Therapeutic cloning - Gene cloning	- Personalised medicine leading to preemptive diagnosis of disorders for better treatment

	- Gene sequencing - Gene therapy - CRISPR	- Treatment for genetic diseases leading to potential cures, rather than merely addressing symptoms - Improved diagnostic tools - Cheaper, faster tools which can be used in remote locations, increasing access to healthcare - Improved creation of important biological molecules for treatment of disease (e.g. insulin)
Industry	- Gene cloning - Transgenesis	- Increased speed of chemical reactions leading to more efficient industrial processes - Creation of organisms which produce industrially significant products (e.g. biofuels, energy)

3.5 Evaluation of Genetic Technology

- *Evaluate the effect on biodiversity of using biotechnology in agriculture*

<u>Biodiversity:</u>

- Biodiversity → The variety of plant and animal life on Earth, involving genetic diversity, species diversity and ecosystem diversity.
- All species are interconnected, they depend on one another. With less biodiversity, these connections weaken and sometimes break, harming all the species in the ecosystem
- Biodiversity is important to people in many ways, as biodiversity decreases these systems break down
- Biodiversity is critical in maintaining healthy ecosystems and thereby sustaining plant and animal life on Earth (including human life
- Biologists and breeders in agriculture realise they need to conserve diversity in living organisms for the long-term survival of species and to feed the growing human population
- Modern biotechnology gives the humans the potential to alter the path of evolution by artificially combining the qualities of organisms that were once separate species
- This could increase biodiversity in the short term, however in the long term, biodiversity will be reduced if these organisms with desirable characteristics are reproduced and bred, using reproductive technologies

<u>Genetic Engineering:</u>

- Genetic engineering increasing biodiversity: Genetic engineering has the potential to increase genetic variation as it allows gene to ‘break the species barrier’
- Selective breeding can reduce a gene pool to desirable traits encoded by alleles already present in the species. Recombinant DNA technology allows scientists to take genes from desirable traits from one species and splice them into another - creating potentially limitless biodiversity.
- Example → Genetically modified crop varieties that are created to be economically advantageous encourages farmers to adopt new GM varieties, severely reducing the number of non-GMO varieties being grown. Much of the genetic variety in these traditional varieties may be lost.

- Benefits:
  - Gene therapy to treat human disease
  - Genetically modified food to alleviate hunger
  - Plant banks and animal cryopreservation to maintain biodiversity and for wildlife conservation
  - Plant and algae-based resources to develop next-generation biofuels
  - Potential to eliminate certain genetic disorders through gene editing tools.

Advantages and Disadvantages of Biotechnologies:

Advantages	Disadvantages
- Can be applied to save species that are on the brink of extinction. - Improve biodiversity farming practices - Alleviate hunger in resource-poor areas of the world - Puts into place biodiversity conservation measures to ensure future resilience in these species - Insect and herbicide resistance (not required to use harsh chemicals) - Proliferation of knowledge-based agriculture - Potential to deliberately and effectively increase genetic diversity in crops through transgenics	- Have the potential to reduce genetic diversity in the long term by selectively breeding desired gene combinations and therefore increase the risk of populations being wiped out in response to disease or sudden environmental change. - Human survival may also be compromised if we influence the path of evolution to the extent that a species becomes extinct - Ability to out-compete un-modified crops - Horizontal gene transfer into native ecosystems

3.7 Secondary Sources

- *Interpret a range of secondary sources to assess the influence of social, economic and cultural contexts on a range of biotechnologies*

3.7.1 Golden Rice

- A stain of rice which was developed through genetic engineering techniques
- The variety has been designed to produce beta-carotene, a precursor to vitamin A
- The fortified rice is intended to be grown in areas whose populations have a general shortage of vitamin A
- Golden rice was created by inserting two genes for beta-carotene biosynthesis into the plant's genome:
  - psy (phytoene synthase), derived from daffodil
  - crtl (carotene desaturase), derived from a soil bacterium
- These genes were inserted into the genome under the control of the endosperm promoter so that the would only be expressed in the edible part of the plant

Advantages	Disadvantages
- Public health benefits (aiding with vitamin A deficiency, usually responsible for nearly 2 million deaths) - Introduction of new alleles into the gene pool, increasing variation across rice species - Development of this technology paves the way for future research into nutritionally augmented foods - The Golden Rice Project sets an example of how biotechnologies can be implemented for humanitarian use rather than pure capital raising	- Potential loss of biodiversity in the surrounding area due to unsustainable monoculturing practices - Existing issues with agriculture are exacerbated by corporate control of the product - Unforeseeable risks of introducing new molecules into diet - Fears that widespread use of golden rice will divert attention away from continuing structural inequalities that are the cause of vitamin A deficiency

3.7.2 Genetically Modified Foods

- Organisms whose genomes have been altered by genetic engineering techniques
- The aim of genetically modified foods is to introduce new traits (often derived from different organisms) which confer a benefit such as resistance to insects or herbicides or increased nutritional value
- Examples include:
  - Bt cotton
  - Golden rice
  - Virus resistant papaya

Advantages	Disadvantages
Social impacts: - Addresses matters of global inequality such as poverty and food security - May increase the dialogue between communities and scientists and improve scientific literacy - Reduction in environmental footprint is beneficial to the global community as there is less leaching of chemicals into ecosystems - GM crops require less tillage, meaning there are fewer greenhouse gas emissions and the production of drought-resistant crops enables water conservation Economic impacts: - Stimulates agricultural economy - May provide farmers in third world countries with tools to grow crops easily and quickly - Enables farmers with nutrient-poor soil or	Social impacts: - May increase socioeconomic disparity if implemented incorrectly (rich get richer) - Lack of consistent regulation internationally may restrict the ability for Gm foods to be improved effectively, which may impact negatively on farmers whose only choice given their environment and its challenges is to use Gm crops Economic impacts: - Potential for monopolisation by large biotechnology companies - Exploitation of patents on GM crop strains to increase profits - May cut small-scale and 3rd world farmers out of the market - Development of 'terminator seed' technologies may create dependence on companies and continual re-purchasing of products may lead to uncertainty for farmers in terms of practice consistency

poor access to water to continue growing nutrient rich foods - May help improve desertified ecosystems Cultural impacts: - Food has been an essential part of cultural practices for centuries - Agricultural practices which are often central to global cultures, can be preserved in the face of changing climates - May provide a tool for preserving important foods and maintaining significant industries in certain areas	Cultural impacts: - Traditional, region-specific farming practices may be eradicated in favour of large-scale agricultural methods - Lack of sufficient scientific communication with regards to GM foods has led to widespread mistrust amongst communities and a rise in anti-science beliefs - Backlash from religious groups on ethical grounds may lead to debate

Feature	Atryn Goat	BT cotton	Spider-Goat
Name of organism	Atryn Goat	Bt cotton	Spider-goat
Gene inserted	AT-III protein	Bt cotton was developed using the common soil bacterium Bacillus thuringiensis (Bt). Bt cotton produces proteins that are toxic to the specific Helicoverpa spp.	Spider genes - genes that produce the silk in spiders are inserted into the goat
Advantages	- Takes pressure off blood donations - Good alternative for people that don't have this protein - Provides high volume of milk meaning efficient	- Increases yield of cotton due to effective control of three types of bollworms, viz. American, Spotted and Pink bollworms. - Reduction in insecticide use - Reduction in es and predators - No health hazards due to rare use of insecticides	- Can be used in cosmetic and medical procedures - Easier to milk goats than spiders - More cost effective than milking spiders
Disadvantages	- Can be expensive - Ethical issues associated with	- High cost of seeds - Negative long term effects	- Long-lasting effects are unknown (could be harmful to

	using goats		humans)
Use (Medical/Industrial /Agriculture)	Used for humans who don't produce AT-III and this then ensures that their blood clots. This is important for their health and safety.	Bt cotton is an insect-resistant transgenic crop designed to combat the bollworm. Bt cotton was created by genetically altering the cotton genome to express a microbial protein from the bacterium Bacillus thuringiensis.	For instance, due to its strength and elasticity, spider silk fiber could have several medical uses, such as for making artificial ligaments and tendons, for eye sutures, and for jaw repair. The silk could also have applications in bulletproof vests and improved car airbags