Module 6: Genetic Change

Outcomes

A student:

- solves scientific problems using primary and secondary data, critical thinking skills and scientific processes BIO11/12-6
- communicates scientific understanding using suitable language and terminology for a specific audience or purpose BIO11/12-7
- explains natural genetic change and the use of genetic technologies to induce genetic change BIO12-13

Content Focus

Students learn about natural and human-induced causes and effects of genetic change, including mutations, environmental pressure and uses of biotechnology. Students investigate how the processes of inheritance and evolution are applied.

The work of scientists in various fields of work, including agriculture, industry and medicine, can be explored within the context of biotechnology. The impact of biotechnology on biological diversity is also explored in this module.

Working Scientifically

In this module, students focus on analysing trends and patterns and solving problems using evidence from data and information. Students also focus on communicating ideas about genetic change for a specific purpose. Students should be provided with opportunities to engage with all Working Scientifically skills throughout the course.

Content

Mutation

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

Students:

- explain how a range of mutagens operate, including but not limited to:
  - electromagnetic radiation sources
  - chemicals
  - naturally occurring mutagens
- compare the causes, processes and effects of different types of mutation, including but not limited to:
  - point mutation
  - chromosomal mutation
- distinguish between somatic mutations and germ-line mutations and their effect on an organism (ACSBL082, ACSBL083)
- assess the significance of 'coding' and 'non-coding' DNA segments in the process of mutation (ACSBL078)
- investigate the causes of genetic variation relating to the processes of fertilisation, meiosis and mutation (ACSBL078)

- evaluate the effect of mutation, gene flow and genetic drift on the gene pool of populations (ACSBL091, ACSBL092)

Biotechnology

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

Students:

- investigate the uses and applications of biotechnology (past, present and future), including: (ACSBL087)
  - 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

Genetic Technologies

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

Students:

- 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
- investigate and assess the effectiveness of cloning, including but not limited to:
  - whole organism cloning
  - gene cloning
- describe techniques and applications used in recombinant DNA technology, for example:
  - the development of transgenic organisms in agricultural and medical applications (ACSBL087)
- evaluate the benefits of using genetic technologies in agricultural, medical and industrial applications (ACSBL086)
- evaluate the effect on biodiversity of using biotechnology in agriculture
- interpret a range of secondary sources to assess the influence of social, economic and cultural contexts on a range of biotechnologies

MODULE 6: GENETIC CHANGE

Mutation: change in DNA
Mutagen: factors that introduce mutations

Mutation Operations:

Electromagnetic radiation sources

- highly penetrative waves that can alter the chemical composition of DNA — break structure, delete or rearrange

Includes Heat + Ionising radiation (X rays, UV + gamma rays)
↳ Physical Mutagens

Physical mutagens may operate by:
- breaking bonds in sugar-phosphate backbone of DNA
- breaking in multiple sites to rearrange/delete nucleotides or inverted order.

Chemical mutagens:

- chemicals that cause mutations if cells are exposed to them
  - alter DNA and protein function

Includes:

Inter-calculating Agents:
- insert themselves to change shape of DNA → errors in DNA replication

Base Analog:
- chemicals similar to nitrogenous bases that incorporate into DNA instead of regular bases → not functional DNA

Reactive Chemicals:
- species that reacts directly with DNA to alter structure
  eg reactive oxygen.

Eg: Nitrous Acid – swap bases out
Benzo pyrene – inserts itself between bases

Naturally Occuring Mutagens:

- present at normal levels in the environment
  - ↳ living or non-living

- Cycasin – poison in Cycad
  - ↳ changes DNA sequence to effect protein synthesis
    damaging DNA
- Chromium – metal in sedimentary rocks
  - ↳ modifies chemical nature of guanine to pair with adenine not cytosine

Types of Mutations:

Point Mutations:

- affect only one (or few) nucleotides within a gene
- may include:
  - Substitution → swapping one base for another
  - Insertion → base added to sequence
  - Deletion → removing base from sequence.

  } frameshift (shift other bases)
- Point Mutations can be:
  - silent → no effect on codon / amino acid
  - Missense → affects one codon, new amino acid
  - Nonsence → introduces stop codon → dysfunctional protein

Chromosomal Mutations:

- affect large section of chromosome
- Includes:
  - Deletion → section is removed
  - Insertion → section is added
  - Inversion → section is inverted + re-inserted.
  - Translocation → section moved to non-homologous chromosome
  - Duplication → section is doubled

Comparing Point to Chromosomal Mutations:

Similarities
- both can lead to dysfunctional protein/DNA change
- Both are mutations

Differences
- Some point mutations can be good while chromosomal are always bad
- Chromosomal → germline

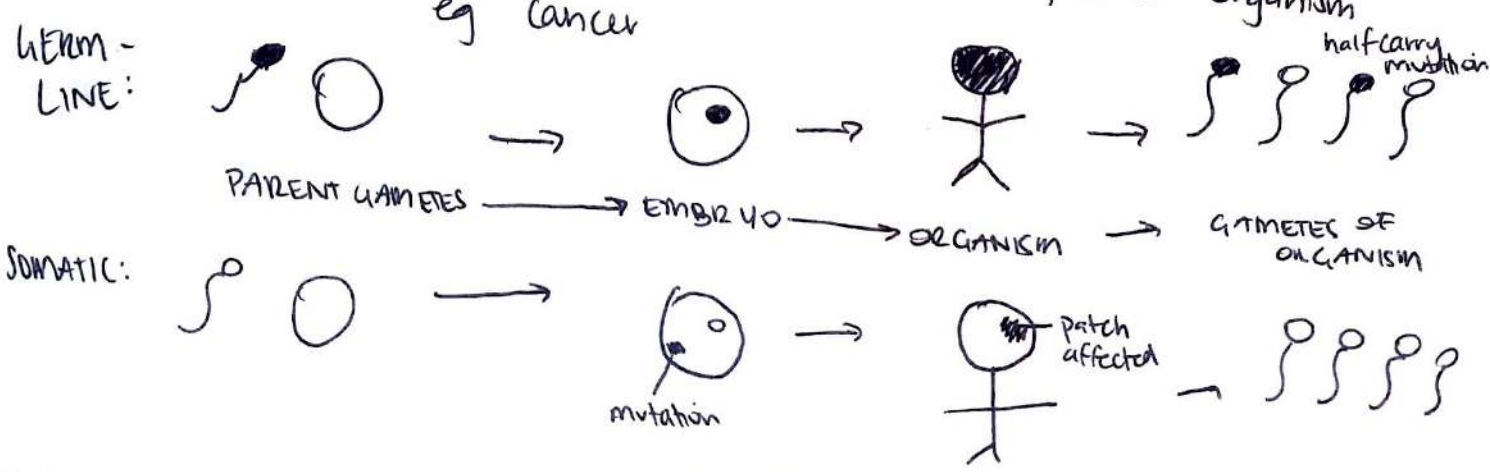
Somatic vs Germ-line + effect on organism

Germ-line Mutations: (in sperm/egg)

- mistake in meiosis – effects every cell in the body
- passed onto offspring which inhent, effecting all cells of the organism
- cause of Diseases eg sickle cell anaemia, cystic fibrosis + colour blindness

Somatic Mutations: (non-reproductive cells)

- mistake in mitosis – localised to one part of organism, does not effect whole organism
- Can be passed onto daughter cells in mitosis
  ↳ not inherited by offspring
- Can be caused by environmental/external factors
- Localised effect, may lead to tumor in one part of organism eg cancer



Coding + Non-coding DNA in Mutations

- Coding DNA (exons): – DNA in genome that produces amino acids / polypeptides
  - ↳ Proteins have structural, functional + regulatory importance in the cell.
  - translated + transcribed
  - ◦ Mutation may effect protein produced, and have serious effects on cellular function

Non-coding DNA: – essential to gene activity / cell function but not protein synthesis

- ◦ Includes: – junk DNA → mutations have little effect
  - enhancers/silencers → mutation may result in over or under expression of genes
  - Promoters → mutation may inhibit ability to transcribe gene
  - Introns → mutation may effect gene splicing
  - Terminators → mutation may result in incorrect elongated mRNA
  - non-coding RNA → mutation may result in alterations of molecules such as tRNA

- Non-coding DNA mutations may have impact on processes of transcription + translation

CAUSES OF GENETIC VARIATION

Fertilisation variation

- Two gametes from two different parents
- Randomised pairing of sperm with ovum
- Two each of genes means two alleles - increased genetic variation

Meiosis variation

- Independent Assortment — alleles for different traits are unrelatedly sorted in gametes
  ↳ increases variation
- Crossing Over — homologous chromosomes line up + exchange segments of DNA to produce new gene combination (unique) within sister chromatids
- Random Segregation — ↳ greater variation in gametes
  alleles separate randomly from one another during gamete formation
  ↳ gametes are all unique

Mutations on variation:

- increases genetic variation of a species / gene pool
- introduces new alleles / gene combinations + traits

Effect of Mutation/s on Populations

- Mutations introduce new alleles in the population which enter the gene pool, hence increases the gene pool.
- Genetic Drift - allele frequency of one allele increases, the frequence of other alleles decreases, hence decreases the gene pool.
- Gene flow - recombines DNA between two populations, hence increase gene pool
- Bottlenecking → abrupt reduction in population, causes loss of diversity in gene pool
- Founder Effect → new population of small number organisms (separate from larger) has genetic loss leading to new speciation events + evolutionary pathways

Applications of Biotechnology

Past Uses:

In agriculture: (mainly)

Selective Breeding – Plants + Animals
eg. Corn + Mules

Cross Breeding – Mule for transport

Fermentation – breakdown of sugars anaerobically
eg. Beer, Yogurt, wine

Present Uses:

- Sperm transfer / selective breeding through artificial pollination / insemination
- mRNA vaccines
- Biofuel (ethanol) from sugarcane.

Future Uses: Cloning

Social Issues / Ethical Issues

Positives:

- improve quality of life / meet societal needs
- Genetic modification — increase diversity + evolution
  ↳ best survival

Negatives / Concerns:

- Ownership of individual's DNA – discrimination
- Religious views on intervening with nature through Biotechnology
- harmful to living organism
- may reduce variation by selectively certain traits.

Future Directions:

- Uses in Medicine – improved treatment + disease detection
  - + better health, less suffering, ↑ LE
  - – equity, insurance, overpopulation
- Uses in Agriculture – synthetic meat production
  - + world hunger, vegetarians, environmentally better
  - – impact farmers / agricultural industry.
- Cloning – whole organism minimally successful atm
  - – + genetic variation
- Genetic Engineering –

Potential Benefits for society

- allows for faster growth rate
  - eg AquaAdvantage salmon
- Create extended life
  - resistance to common forms of Death, reduce health risks
- Develop specific traits:
  - Desirable traits for better consumption / use (animals)
- New products created:
  - eg alter energy content of plants/animals
- Greater yield produced
- Reduce risk to local water supply

Changes to Earth's Biodiversity

- Can result in decrease of biodiversity

Positives: – Creation of new species by influencing emergence of desirable traits
– Implement change faster

Negatives: – can crops favored, fewer crop varieties → more susceptible to disease or evolution
– can animals breeding w/ natural population – unforeseen consequences

Genetic Technologies – DNA manipulation

Uses + Advantages of current technologies on genetic change

	Technology:	Uses:	Advantages:
Reproductive Technologies	◦ Artificial Insemination	◦ Livestock (agriculture) ◦ Fertility treatments (humans)	◦ efficient, bypass infertility + synchronise pregnancy
	– IVF	– Fertility (humans)	– Freeze embryos – genetic screening
	◦ Artificial Pollination	◦ Pollinate crops ◦ genetic experiments	◦ control inheritance of desirable traits
Cloning Techniques	– Whole organism cloning	– Livestock industry	– Definite inheritance of desirable traits.
	◦ Therapeutic cloning	◦ Medicine (stem cell technologies)	◦ Stem cell able differentiate into any cell.
	– Gene Cloning	– Medicine + industry	– Production of biologically relevant proteins eg insulin for industry
Recombinant DNA techniques.	◦ Transgenesis	◦ Agriculture (crops) ↳ Bt corn ◦ Environmental biotechnology	◦ Create organisms w/ many functions, favorable traits, ◦ reduce pesticide use
	– Gene sequencing	– Medicine – Genetic research ↳ identify new genes	– identify genetic factors/risk factors – understand evolution – forensic biotechnology
	◦ Gene Therapy	◦ Medicine	◦ Treatment for disease eg Cystic Fibrosis
	– CRISPR	– Molecular Biology ↳ gene editing	– cost-effective for gene therapy + transgenics

Reproductive Technologies:

Artificial Insemination – Uses in Agriculture + Medical Industry

Process:
- Involves collecting + banking sperm from male + inserting into vagina of female
- Semen can be cryogenically stored indefinitely + used to inseminate females

Outcomes:
- Transporting sperm overcomes transporting whole animals
  - ↳ cost effective + reduces risk of injury
- Many females can be inseminated by same male.

– may reduce biodiversity, costly, but could save endangered species.

Artificial Pollination – used in Agriculture

Process:
- Involves brushing pollen from male of one flower onto female stigma of another flower
- Pollinated flower is covered to prevent pollination from other flowers.

Outcomes:
- controlled inheritance of favorable traits
- Hybridisation able – eg corn is hybrid for ↑ germination rate, greater yield + uniformity.

– total control of breeding, overuse lead to monoculture

IVF – Used in Medicine (humans)

Process:
- Stimulate egg production to collect from ovaries
- Fertilise egg, incubate to produce embryos
- Embryos are implanted into the uterus or frozen (future use)

Outcomes:
- Favorable genes passed to offspring
- Allows for genetic screening to avoid disease
- Overcome infertility

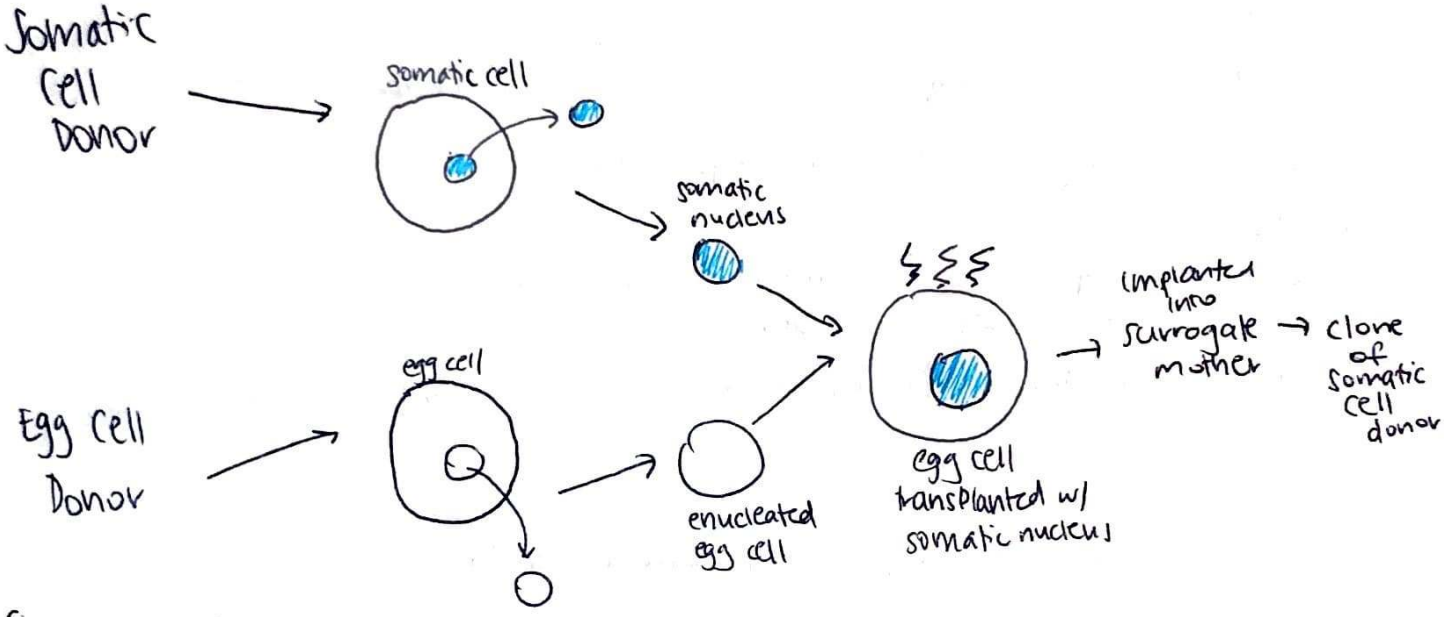
– Expensive, not 100% successful.

Cloning Effectiveness

Whole Organism Cloning:

- creating new genetically identical organism. Currently limited to cattle, chickens, sheep + dogs.
- Disadvantages: – high cost
  – high mortality rate
  – increased health problems

Process through Somatic Cell Nuclear Transfer:



Gene Cloning

- producing multiple copies of specific DNA sequence

Process:

1 – gene is isolated + cut from cell via restriction enzymes.

2 – gene fragments have matching sticky ends, Plasmid cut with same enzymes

3 – Gene pasted into plasmid to form recombinant DNA

4 – Transformation: Plasmid inserted back into host cell

5 – Host cell replicates.

- Allows genes lacking to be amplified at fast + efficient rate.

Therapeutic Cloning:

- Cloning techniques for therapy for disease
  - Stem cells genetically identical to donor used to treat disease
- Involves the process/use of somatic cell nucleus transfer
- Stem cells treat the disease to replace dysfunctional cells
  - ↳ no immunological rejection

Recombinant DNA technology

- When DNA from 2 different sources is joined

Techniques / Applications:

Transgenic Organisms:

- Introducing genetic material from one species into the DNA of another species

Used in Agriculture

- Bt Cotton → gene from soil bacteria inserted into cotton plant to countermeasure parasitic caterpillar (resistant to pesticides)

Used in Medicine

- Plasmid w/ insulin-coding genes makes cell produce large amounts of insulin which can be used to treat diabetes.

Agriculture - Aqua Advantage Salmon → grows bigger + faster with growth gene (hormone) from eel + other salmon species.

Benefits of Genetic Technologies

Application:	Techniques:	Benefits:
Agriculture	• selective breeding • Artificial pollination • Transgenesis	• Creation of crops + livestock species w/ favorable traits (high yield, nutritional value, pest resistant, temperament) • Creation of crops without need for herb/pesticide → decrease environmental impact • Increased food supply / security
Medicine	• Therapeutic cloning • Gene cloning • Gene sequencing • Gene therapy • ELISA • CRISPR	• Personalised medicine for better treatment • Treatment of genetic disease lead to potential cures • Improved diagnostic tools • Cheaper / faster tools → increased healthcare access • Improved creation of biological molecules for treatment eg Insulin for Diabetes.
Industry	• Gene Cloning • Transgenesis	• Increased speed of chemical reactions for efficient industrial processes • Creation of organisms to produce industrially significant products (eg biofuels, energy)

Effect of biotechnology on Biodiversity

SHORT TERM: broaden the gene pool
LONG TERM: higher risk of extinction due to reduced variability

Advantages:

- Increase genetic diversity in crops, if natural varieties maintained
- decrease risk issues of biodiversity in environmentally impacted - GMOs suited to environment
- Create organism not needing insecticides/herbicides → environmentally better

Disadvantages:

- Selective breeding can impact species' reaction to abiotic/biotic stresses over time
- If profit prioritised, loss of natural species to GMOs, gene pool compromised.
- Could cause rapid evolution of resistance to pesticides

Influence of context on Biotechnology

Social Impacts:

Advantages:

- reduction in environmental footprint → less chemicals in ecosystem
- GM crops require less tillage → less emissions + water (drought resistant crops)

Disadvantages:

- Increase socioeconomic disparity → rich get richer

Economic Impacts:

Advantages:

- Provides disadvantaged farmers w/ tools to grow crops cheap/quick
- Enable farmers w/ nutrient-poor soil to continue growing
  - ↳ increase amount of food provides

Disadvantages:

- Monopolisation by large biotechnology companies

Cultural Impacts:

Advantages:

- Food is essential for many cultures
- Aid in preserving crop cultivation / farming practices

Disadvantages:

- Backlash from religious groups on ethical grounds lead to divise debeats.