SCQF level 6
Unit Specification

Biology: DNA and the Genome

SCQF: level 6 (6 SCQF credit points)

Unit code: J4A6 76

Unit outline

The general aim of this Unit is to develop skills of scientific inquiry, investigation and analytical thinking, along with knowledge and understanding of DNA and the genome. Learners will apply these skills when considering the applications of DNA and the genome on our lives. This can be done by using a variety of approaches, including investigation and problem solving.

The Unit covers the key areas of structure of DNA; replication of DNA; gene expression; cellular differentiation; the structure of the genome; mutations; evolution; and genomic sequencing.

Learners will research issues, apply scientific skills and communicate information related to their findings, which will develop skills of scientific literacy.

Learners who complete this Unit will be able to:

1 Apply skills of scientific inquiry and draw on knowledge and understanding of the key areas of this Unit to carry out an experiment/practical investigation
2 Draw on knowledge and understanding of the key areas of this Unit and apply scientific skills

This Unit is a free-standing Unit. The *Unit Support Notes* in the Appendix provide advice and guidance on delivery, assessment approaches and development of skills for learning, skills for life and skills for work. Exemplification of the standards in this Unit is given in *Unit Assessment Support*.

Recommended entry

Entry to this Unit is at the discretion of the centre. However, learners would normally be expected to have attained the skills, knowledge and understanding required by one or more of the following or equivalent qualifications and/or experience:

- National 5 Biology Course
- free-standing SCQF level 5 Biology Units

Equality and inclusion

This Unit Specification has been designed to ensure that there are no unnecessary barriers to learning or assessment. The individual needs of learners should be taken into account when planning learning experiences, selecting assessment methods or considering alternative evidence. For further information, please refer to the Appendix: *Unit Support Notes.*

Standards

Outcomes and Assessment Standards

Outcome 1

The learner will:

1 Apply skills of scientific inquiry and draw on knowledge and understanding of the key areas of this Unit to carry out an experiment/practical investigation by:

1.1 Planning an experiment/practical investigation
1.2 Following procedures safely
1.3 Making and recording observations/measurements correctly
1.4 Presenting results in an appropriate format
1.5 Drawing valid conclusions
1.6 Evaluating experimental procedures

Outcome 2

The learner will:

2 Draw on knowledge and understanding of the key areas of this Unit and apply scientific skills by:

2.1 Making accurate statements
2.2 Solving problems

Evidence Requirements for the Unit

Assessors should use their professional judgement, subject knowledge and experience, and understanding of their learners, to determine the most appropriate ways to generate evidence and the conditions and contexts in which they are used.

The key areas covered in this Unit are structure of DNA; replication of DNA; gene expression; cellular differentiation; the structure of the genome; mutations; evolution; and genomic sequencing.

The following table describes the evidence for the Assessment Standards. Exemplification of assessment is provided in *Unit Assessment Support*.

Assessment Standard	**Evidence required**
Planning an experiment	The plan must include: ♦ a clear statement of the aim ♦ a hypothesis ♦ a dependent and independent variable ♦ variables to be kept constant ♦ measurements/observations to be made ♦ the equipment/materials ♦ a clear and detailed description of how the experiment/practical investigation should be carried out, including safety considerations
Following procedures safely	The learner must be seen to follow procedures safely.
Making and recording observations/measurements correctly	The raw data must be collated in a relevant format, for example a table.
Presenting results in an appropriate format	One format from: bar graph or line graph.
Drawing a valid conclusion	Must include reference to the aim and be supported by the results.
Evaluating experimental procedures	Provide one evaluative statement about the procedures used **and** suggest one improvement for the experiment. **or** Provide two evaluative statements about the procedures used. **or** Suggest two improvements for the experiment. Appropriate justification must also be provided whichever option is chosen.

Assessment Standard	Evidence required
Making accurate statements and solving problems	Achieve at least 50% of the total marks available in a holistic assessment. A holistic assessment must include: ♦ an appropriate number of opportunities to make accurate statements for each key area of the Unit ♦ at least one opportunity to demonstrate each of the following problem-solving skills: — make generalisations/predictions — select information — process information, including calculations, as appropriate — analyse information

Assessment Standard thresholds

Outcome 1

Learners are not required to show full mastery of the Assessment Standards to achieve Outcome 1. Instead, five out of the six Assessment Standards for Outcome 1 must be met to achieve a pass. Learners must be given the opportunity to meet all Assessment Standards.

Outcome 2

Learners are assessed using a holistic assessment that assesses Assessment Standards 2.1 and 2.2. To gain a pass for Outcome 2, learners must achieve 50% or more of the total marks available in the assessment.

Transfer of evidence

Evidence for the achievement of Outcome 1 for this Unit can be used as evidence for the achievement of Outcome 1 in the SCQF level 6 Units: Biology: Metabolism and Survival (J4A7 76) and Biology: Sustainability and Interdependence (J4A8 76).

Evidence for the achievement of Outcome 2 for this Unit is **not** transferable between the SCQF level 6 Units: Biology: Metabolism and Survival (J4A7 76) and Biology: Sustainability and Interdependence (J4A8 76).

Re-assessment

SQA's guidance on re-assessment is that there should only be one or, in exceptional circumstances, two re-assessment opportunities. Re-assessment must be carried out under the same conditions as the original assessment.

Outcome 1

Learners can re-draft their original Outcome 1 report or carry out a new experiment/practical investigation.

Outcome 2

Learners must have a full re-assessment opportunity, ie a holistic assessment. To achieve Outcome 2, learners must achieve 50% of the total marks available in the re-assessment.

Development of skills for learning, skills for life and skills for work

It is expected that learners will develop broad, generic skills through this Unit. The skills that learners will be expected to improve on and develop through the Unit are based on SQA's *Skills Framework: Skills for Learning, Skills for Life and Skills for Work* and drawn from the main skills areas listed below. These must be built into the Unit where there are appropriate opportunities.

1 Literacy

1.2 Writing

2 Numeracy

2.1 Number processes
2.2 Money, time and measurement
2.3 Information handling

5 Thinking skills

5.3 Applying
5.4 Analysing and evaluating
5.5 Creating

Amplification of these is given in SQA's *Skills Framework: Skills for Learning, Skills for Life and Skills for Work.* The level of these skills should be at the same SCQF level of the Unit and be consistent with the SCQF level descriptor. Further information on building in skills for learning, skills for life and skills for work is given in the Appendix: *Unit Support Notes.*

Appendix: Unit Support Notes

Introduction

These support notes are not mandatory. They provide advice and guidance on approaches to delivering and assessing this Unit. They are intended for teachers and lecturers who are delivering this Unit. They should be read in conjunction with:

- *Unit Assessment Support*

Developing skills, knowledge and understanding

Teachers and lecturers are free to select the skills, knowledge, understanding and contexts that are most appropriate for delivery in their centres.

Approaches to learning and teaching

DNA and the genome		
Key areas	**Depth of knowledge required**	**Suggested learning activities**
1 The structure of DNA (a) Structure of DNA — nucleotides (deoxyribose sugar, phosphate and base), sugar–phosphate backbone, base pairing (adenine–thymine and guanine–cytosine) by hydrogen bonds and double-stranded antiparallel structure, with deoxyribose and phosphate at 3′ and 5′ ends of each strand respectively, forming a double helix.	The base sequence of DNA forms the genetic code.	Examine research that led to an understanding of the structure of DNA. Studies could include Chargaff's base ratios, X-ray crystallography of Wilkins and Franklin, and Watson and Crick's development of the double helix model. Compare DNA extraction from peas and kiwi fruit (possible false positive result in latter as DNA is obscured by pectin).
(b) Organisation of DNA — prokaryotes have a single, circular chromosome and smaller circular plasmids. Eukaryotes all have linear chromosomes, in the nucleus, which are tightly coiled and packaged with associated proteins. They also contain circular chromosomes in their mitochondria and chloroplasts. Yeast is a special example of a eukaryote as it also has plasmids.	The associated proteins are called histones.	

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Key areas	**Depth of knowledge required**	**Suggested learning activities**
2 Replication of DNA (a) Replication of DNA by DNA polymerase and primers	Prior to cell division, DNA is replicated by a DNA polymerase. DNA polymerase needs primers to start replication. A primer is a short strand of nucleotides that binds to the 3′ end of the template DNA strand allowing polymerase to add DNA nucleotides.	Carry out digital or physical modelling of DNA replication. Examine Meselson and Stahl's experiments on DNA replication.
DNA polymerase adds DNA nucleotides, using complementary base pairing, to the deoxyribose (3′) end of the new DNA strand that is forming. Fragments of DNA are joined together by ligase.	DNA is unwound and hydrogen bonds between bases are broken to form two template strands. DNA polymerase can only add DNA nucleotides in one direction resulting in the leading strand being replicated continuously and the lagging strand replicated in fragments.	
(b) Polymerase chain reaction (PCR) amplifies DNA using complementary primers for specific target sequences. Repeated cycles of heating and cooling amplify the target region of DNA.	In PCR, primers are short strands of nucleotides that are complementary to specific target sequences at the two ends of the region of DNA to be amplified. DNA is heated to between 92 and 98°C to separate the strands. It is then cooled to between 50 and 65°C to allow primers to	Carry out PCR using a thermal cycler or water baths.

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Key areas	**Depth of knowledge required**	**Suggested learning activities**
	bind to target sequences. It is then heated to between 70 and 80°C for heat-tolerant DNA polymerase to replicate the region of DNA.	
Practical applications of PCR	PCR can amplify DNA to help solve crimes, settle paternity suits, and diagnose genetic disorders.	Use gel electrophoresis to analyse DNA samples (from kits) to determine criminality or paternity.
3 Gene expression (a) Gene expression involves the transcription and translation of DNA sequences.	Only a fraction of the genes in a cell are expressed.	
Transcription and translation involve three types of RNA (mRNA, tRNA and rRNA).	RNA is single-stranded and is composed of nucleotides containing ribose sugar, phosphate and one of four bases: cytosine, guanine, adenine and uracil.	Carry out digital or physical modelling of transcription and translation.
Messenger RNA (mRNA) carries a copy of the DNA code from the nucleus to the ribosome.	mRNA is transcribed from DNA in the nucleus and translated into proteins by ribosomes in the cytoplasm. Each triplet of bases on the mRNA molecule is called a codon and codes for a specific amino acid.	
Transfer RNA (tRNA) folds due to	A tRNA molecule has an anticodon (an	

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Key areas	**Depth of knowledge required**	**Suggested learning activities**
complementary base pairing. Each tRNA molecule carries its specific amino acid to the ribosome. Ribosomal RNA (rRNA) and proteins form the ribosome.	exposed triplet of bases) at one end and an attachment site for a specific amino acid at the other end.	
(b) The role of RNA polymerase in transcription of DNA into primary mRNA transcripts RNA splicing forms a mature mRNA transcript. The introns of the primary transcript are non-coding regions and are removed. The exons are coding regions and are joined together to form the mature transcript.	RNA polymerase moves along DNA, unwinding the double helix and breaking the hydrogen bonds between the bases. RNA polymerase synthesises a primary transcript of mRNA from RNA nucleotides by complementary base pairing. Uracil in RNA is complementary to adenine. The order of the exons is unchanged during splicing.	
(c) tRNA is involved in the translation of mRNA into a polypeptide at a ribosome. Translation begins at a start codon and		

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Key areas	**Depth of knowledge required**	**Suggested learning activities**
ends at a stop codon. Anticodons bond to codons by complementary base pairing, translating the genetic code into a sequence of amino acids. Peptide bonds join the amino acids together. Each tRNA then leaves the ribosome as the polypeptide is formed.		
(d) Different proteins can be expressed from one gene, as a result of alternative RNA splicing. Different mature mRNA transcripts are produced from the same primary transcript depending on which exons are retained.		
(e) Amino acids are linked by peptide bonds to form polypeptides. Polypeptide chains fold to form the three-dimensional shape of a protein, held together by hydrogen bonds and other interactions between individual amino acids. Proteins have a large variety of shapes, which determine their functions. Phenotype is determined by the proteins produced as the result of gene expression.	Details of other interactions and levels of protein structure are not required. Environmental factors also influence phenotype.	Use digital resources to examine the shape and structure of proteins. Carry out experiments to separate and identify fish proteins by agarose gel electrophoresis. Carry out experiments to separate and identify amino acids, using paper chromatography.

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Key areas	**Depth of knowledge required**	**Suggested learning activities**
4 Cellular differentiation (a) Cellular differentiation is the process by which a cell expresses certain genes to produce proteins characteristic for that type of cell. This allows a cell to carry out specialised functions. Differentiation into specialised cells from meristems in plants; embryonic and tissue (adult) stem cells in animals	Meristems are regions of unspecialised cells in plants that can divide (self-renew) and/or differentiate. Stem cells are unspecialised cells in animals that can divide (self-renew) and/or differentiate. There is no requirement to learn examples of differentiated animal and plant cells.	
(b) Embryonic and tissue stem cells Cells in the very early embryo can differentiate into all the cell types that make up the organism and so are pluripotent. Tissue stem cells are involved in the growth, repair and renewal of the cells	All the genes in embryonic stem cells can be switched on, so these cells can differentiate into any type of cell. Tissue stem cells are multipotent as they can differentiate into all of the types of cell	

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Key areas	**Depth of knowledge required**	**Suggested learning activities**
found in that tissue. They are multipotent.	found in a particular tissue type. For example, blood stem cells located in bone marrow can give rise to all types of blood cell.	View digital resources on the origin of blood cells and their functions.
Therapeutic and research uses of stem cells Therapeutic uses involve the repair of damaged or diseased organs or tissues.	The therapeutic uses of stem cells should be exemplified by how they are used in corneal repair and the regeneration of damaged skin.	Study potential therapeutic uses of stem cells.
Research uses involve stem cells being used as model cells to study how diseases develop or being used for drug testing.	Stem cells from the embryo can self-renew, under the right conditions, in the lab.	
The ethical issues of using embryonic stem cells	Stem cell research provides information on how cell processes, such as cell growth, differentiation and gene regulation work. Use of embryonic stem cells can offer effective treatments for disease and injury; however, it involves destruction of embryos.	Debate the ethics surrounding stem cell research and the sources of stem cells.

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Key areas	**Depth of knowledge required**	**Suggested learning activities**
5 The structure of the genome The genome of an organism is its entire hereditary information encoded in DNA. A genome is made up of genes and other DNA sequences that do not code for proteins. DNA sequences that code for protein are defined as genes. Other sequences regulate transcription and others are transcribed but never translated.	Most of the eukaryotic genome consists of non-coding sequences. Details of regulation of transcription (for example Jacob–Monod hypothesis) not required. tRNA and rRNA are non-translated forms of RNA.	
6 Mutations (a) Mutations are changes in the DNA that can result in no protein or an altered protein being synthesised.		Carry out experiments to investigate the effects of UV radiation on UV-sensitive yeast.
(b) Single gene mutations involve the alteration of a DNA nucleotide sequence as a result of the substitution, insertion or deletion of nucleotides.		

DNA and the genome		
Key areas	**Depth of knowledge required**	**Suggested learning activities**
Nucleotide substitutions — missense, nonsense and splice-site mutations	Missense mutations result in one amino acid being changed for another. This may result in a non-functional protein or have little effect on the protein. Nonsense mutations result in a premature stop codon being produced, which results in a shorter protein. Splice-site mutations result in some introns being retained and/or some exons not being included in the mature transcript.	Study human conditions caused by single gene mutations. Examples could include sickle-cell disease (missense), PKU (missense), Duchenne muscular dystrophy (nonsense) and beta thalassemia (splice-site mutation).
Nucleotide insertions or deletions result in frame-shift mutations.	Frame-shift mutations cause all of the codons and all of the amino acids after the mutation to be changed. This has a major effect on the structure of the protein produced.	Study human conditions caused by frame-shift mutations. Examples could include Tay-Sachs disease (frame-shift insertion) and cystic fibrosis (frame-shift deletion).
(c) Chromosome structure mutations — duplication, deletion, inversion and translocation	Duplication is where a section of a chromosome is added from its homologous partner. Deletion is where a section of a chromosome is removed.	Study human conditions caused by chromosome structure mutations. For example: ♦ Cri-du-chat syndrome — caused by deletion of part of the short arm of chromosome 5.

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Key areas	**Depth of knowledge required**	**Suggested learning activities**
	Inversion is where a section of chromosome is reversed. Translocation is where a section of a chromosome is added to a chromosome, not its homologous partner. The substantial changes in chromosome mutations often make them lethal.	♦ Haemophilia A — one cause is an inversion within the gene that produces a clotting factor (factor VIII). ♦ Chronic myeloid leukaemia — caused by a reciprocal translocation of sections of chromosome 22 and chromosome 9.
(d) Importance of mutations and gene duplication in evolution	Duplication allows potential beneficial mutations to occur in a duplicated gene while the original gene can still be expressed to produce its protein.	
7 Evolution (a) Evolution — the changes in organisms over generations as a result of genomic variations		
(b) Selection Natural selection is the non-random increase in frequency of DNA sequences that increase survival and the non-random reduction in the frequency of deleterious sequences.		

DNA and the genome		
Key areas	**Depth of knowledge required**	**Suggested learning activities**
The changes in phenotype frequency as a result of stabilising, directional and disruptive selection Natural selection is more rapid in prokaryotes. Prokaryotes can exchange genetic material horizontally, resulting in faster evolutionary change than in organisms that only use vertical transfer.	In stabilising selection, an average phenotype is selected for and extremes of the phenotype range are selected against. In directional selection, one extreme of the phenotype range is selected for. In disruptive selection, two or more phenotypes are selected for. Horizontal gene transfer is where genes are transferred between individuals in the same generation. Methods of horizontal transfer are not required. Vertical gene transfer is where genes are transferred from parent to offspring as a result of sexual or asexual reproduction.	
(c) Speciation Speciation is the generation of new biological species by evolution as a result of isolation, mutation and selection.	A species is a group of organisms capable of interbreeding and producing fertile offspring, and which does not normally breed with other groups.	Research the London Underground mosquito.

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Key areas	**Depth of knowledge required**	**Suggested learning activities**
The importance of isolation barriers in preventing gene flow between sub-populations during speciation Geographical barriers lead to allopatric speciation and behavioural or ecological barriers lead to sympatric speciation.		
8 Genomic sequencing (a) In genomic sequencing the sequence of nucleotide bases can be determined for individual genes and entire genomes. Comparison of genomes from different species Comparison of genomes reveals that many genes are highly conserved across different organisms.	Computer programs can be used to identify base sequences by looking for sequences similar to known genes. To compare sequence data, computer and statistical analyses (bioinformatics) are required. Many genomes have been sequenced, particularly of disease-causing organisms, pest species and species that are important model organisms for research.	Research how sequencing technologies use techniques, such as fluorescent tagging of nucleotides, to identify the base sequence. Study potential uses of bioinformatics.

DNA and the genome		
Key areas	**Depth of knowledge required**	**Suggested learning activities**
(b) Evidence from phylogenetics and molecular clocks to determine the main sequence of events in evolution	Phylogenetics is the study of evolutionary history and relationships.	Study the evolution of bears and primates using Geneious software.
The sequence of events can be determined using sequence data and fossil evidence.	Use of sequence data to study the evolutionary relatedness among groups of organisms. Sequence divergence is used to estimate time since lineages diverged.	Compare number and proportion of shared genes between organisms, such as *C. elegans*, Drosophila and humans.
Comparison of sequences provides evidence of the three domains of life — bacteria, archaea and eukaryotes.	Use of sequence data and fossil evidence to determine the main sequence of events in evolution of life: cells, last universal ancestor, prokaryotes, photosynthetic organisms, eukaryotes, multicellularity, animals, vertebrates, land plants.	Research the importance of the Fugu genome as an example of a very small vertebrate genome with a high rate of chromosome deletion.
	Molecular clocks are used to show when species diverged during evolution. They assume a constant mutation rate and show differences in DNA sequences or amino acid sequences. Therefore, differences in sequence data between species indicate the time of divergence from a common ancestor.	Compare human and chimp genomes to show the rapid change in genes for immune system and regulation of neural development over the last six million years.

DNA and the genome		
Key areas	**Depth of knowledge required**	**Suggested learning activities**
(c) An individual's genome can be analysed to predict the likelihood of developing certain diseases. Pharmacogenetics and personalised medicine	Pharmacogenetics is the use of genome information in the choice of drugs. An individual's personal genome sequence can be used to select the most effective drugs and dosage to treat their disease (personalised medicine).	

Administrative information

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Superclass: RH

History of changes to National Unit Specification

Version	Description of change	Authorised by	Date