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## Biology: DNA and the Genome

**SCQF:** level 6 (6 SCQF credit points)

**Unit code:** H4KD 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; control of gene expression; cellular differentiation; the structure of the genome; mutations; evolution; 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 available as 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 or relevant component 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; control of gene expression; cellular differentiation;  
the structure of the genome; mutations; evolution; genomic sequencing.

Evidence can be drawn from a variety of sources and presented in a variety of formats.

The following table describes the evidence for the assessment standards which require exemplification. Evidence may be presented for individual outcomes, or gathered for the unit. If the latter approach is used, it must be clear how the evidence covers each outcome.

<b>Assessment Standard</b>	<b>Evidence required</b>
Planning an experiment	The plan should 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
Presenting results in an appropriate format	One format from: table, line graph, chart, key, diagram, flow chart, summary, extended text or other appropriate format
Drawing a valid conclusion	Include reference to the aim
Evaluating experimental procedures	Suggest two improvements with justification
Making accurate statements	At least half of the statements should be correct across the key areas of this Unit.
Solving problems	One of each: make generalisations/predictions select information process information, including calculations, as appropriate analyse information

Exemplification of assessment is provided in Unit assessment support packs.  
Advice and guidance on possible approaches to assessment is provided in the Appendix: *Unit Support Notes*.

## Assessment Standard Thresholds

### Outcome 1

Candidates 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. Candidates must be given the opportunity to meet all assessment standards. The threshold has been put in place to reduce the volume of re-assessment where that is required.

#### Transfer of evidence

Evidence of Outcome 1 in a unit is transferrable between the other units at SCQF level 6.

#### Re-assessment

Candidates can be given the opportunity to re-draft their original Outcome 1 report or to carry out a new experiment/practical investigation.

### Outcome 2

There is no requirement to pass assessment standard 2.1 (making accurate statements) and assessment standard 2.2 (solving problems) independently. Candidates can be assessed using a single test that contains marks and a cut-off score.

A suitable unit assessment will cover all of the key areas (assessment standard 2.1) **and** assess each of the problem-solving skills (assessment standard 2.2).

Where a candidate achieves 50% or more of the total marks available in a single unit assessment, they will pass Outcome 2 for that unit. Existing unit assessment support packs (UASPs) can be used, or centres can replace the questions with suitable alternatives of a similar standard

Unit assessment support pack 1 contains questions on all of the key areas (AS 2.1) and questions covering each of the problem solving skills (AS 2.2), and may be adapted for use as a single assessment. The number of marks available for each question should be combined to give the total number of marks available. A cut-off score of 50% should be applied to the unit assessments.

Outcome 2: assessment activity 2 – tests contain questions covering assessment standards 2.1 and 2.2 in a single assessment. These do not require to be adapted.

**Important note:** Centres can continue to assess AS 2.1 and 2.2 separately using the existing UASPs. If this option is chosen, 50% or more of the KU statements (AS 2.1) made by candidates must be correct in the unit assessment and at least one correct response for each problem solving skill (AS 2.2) is required to pass outcome 2. However, if a candidate is given more than one opportunity in a unit assessment to provide a response for a problem solving skill, then they must answer 50% or more correctly.

## **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 should be carried out under the same conditions as the original assessment. It is at the teacher or lecturer's discretion how they re-assess their candidates. Candidates may be given a full re-assessment opportunity, or be re-assessed on individual key areas and/or problem-solving skills. As there is no requirement to pass assessment standard 2.1 (making accurate statements) and assessment standard 2.2 (solving problems) independently, candidates must achieve 50% of the 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:

- ◆ the *Unit Assessment Support packs*

## Developing skills, knowledge and understanding

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



## Approaches to learning and teaching

key areas	Suggested learning activities	Exemplification of key areas
<p><b>1 The structure of DNA</b></p> <p>(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.</p>	<p>Case study examining the experimental evidence of the bacterial transformation experiments of Griffiths and identification of DNA as the transforming principle by Avery et al., phage experiments of Hershey and Chase, Chargaff's base ratios and the X-ray crystallography of Wilkins and Franklin.</p> <p>Watson and Crick's double-helix model as an evidence-based conclusion.</p> <p>Case study on Meselson and Stahl experiments on DNA replication. DNA gel electrophoresis.</p> <p>Comparison of DNA extraction from peas and kiwi fruit (false positive result in latter as DNA is obscured by pectin).</p>	<p>All cells store their genetic information in the base sequence of DNA. The genotype is determined by the sequence of bases.</p>
<p>(b) Organisation of DNA — circular chromosomal DNA and plasmids in prokaryotes. Circular plasmids in yeast. Circular chromosome in mitochondria and chloroplasts of eukaryotes.</p>		

key areas	Suggested learning activities	Exemplification of key areas
<p>DNA in the linear chromosomes in the nucleus of eukaryotes is tightly coiled and packaged with associated proteins.</p>		
<p><b>2 Replication of DNA</b>            (a) Replication of DNA by DNA polymerase and primer. Directionality of replication on both template strands. DNA polymerase adds complementary nucleotides to the deoxyribose (3') end of a DNA strand. Fragments of DNA are joined together by ligase.</p>	<p>Virtual or physical modelling of DNA replication.</p>	<p>Prior to cell division, DNA is replicated by a DNA polymerase. DNA polymerase needs a primer to start replication.</p> <p>DNA is unwound and unzipped to form two template strands. This process occurs at several locations on a DNA molecule. DNA polymerase can only add DNA nucleotides in one direction resulting in one strand being replicated continuously and the other strand replicated in fragments.</p>
<p>(b) Polymerase chain reaction (PCR) amplification of DNA using complementary primers for specific target sequences.</p> <p>DNA heated to separate strands, then cooled for primer binding. Heat-tolerant DNA polymerase replicates the region of DNA. Repeated cycles of heating and cooling amplify this region of DNA. Positive and negative controls. Practical applications of PCR.</p>	<p>Case study on the use of PCR, including practical using thermal cycler or water baths.</p> <p>Emphasise the 'needle in a haystack' accuracy of primers and the amplification of 'a haystack from the needle' by PCR.</p> <p>Investigating plant evolution using chloroplast DNA and PCR.</p>	<p>The polymerase chain reaction (PCR) is a technique for the amplification of DNA in vitro.</p> <p>In PCR, primers are complementary to specific target sequences at the two ends of the region to be amplified.</p> <p>Cooling allows primers to bind to target sequences.</p>

key areas	Suggested learning activities	Exemplification of key areas
<p><b>3 Control of gene expression</b></p> <p>(a) The phenotype is determined by the proteins produced as the result of gene expression, influenced by intra- and extra-cellular environmental factors. Only a fraction of the genes in a cell are expressed.</p> <p>Gene expression is controlled by the regulation of transcription and translation.</p>	<p>Separation and identification of fish proteins by agarose gel electrophoresis.</p> <p>Investigation of the shape and structure of fibrous and globular proteins using RasMol or protein explorer software.</p> <p>Separation and identification of amino acids using paper chromatography.</p>	<p>The genetic code used in transcription and translation is found in all forms of life.</p> <p>mRNA is transcribed from DNA in the nucleus and translated into proteins by ribosomes in the cytoplasm.</p> <p>Proteins have a large variety of structures and shapes resulting in a wide range of functions.</p> <p>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</p>
<p>(b) Structure and functions of mRNA. Single strand, replacement of thymine with uracil and deoxyribose with ribose compared to DNA. mRNA (messenger) carries a copy of the DNA code from the nucleus to the ribosome. rRNA (ribosomal) and proteins form the ribosome. Each tRNA (transfer) carries a specific amino acid.</p>		

key areas	Suggested learning activities	Exemplification of key areas
<p>(c) Transcription of DNA into primary and mature RNA transcripts to include the role of RNA polymerase and complementary base pairing. The introns of the primary transcript of mRNA are non-coding and are removed in RNA splicing. The exons are coding regions and are joined together to form mature transcript. This process is called RNA splicing.</p>	<p>Modelling transcription and translation using virtual and physical resources.</p>	<p>RNA polymerase moves along DNA unwinding and unzipping the double helix and synthesising a primary transcript of RNA from RNA nucleotides by complementary base pairing.</p>
<p>(d) Translation of mRNA into a polypeptide by tRNA at the ribosome. tRNA folds due to base pairing to form a triplet anticodon site and an attachment site for a specific amino acid. Triplet codons on mRNA and anticodons translate the genetic code into a sequence of amino acids. Start and stop codons exist. Codon recognition of incoming tRNA, peptide bond formation and exit of tRNA from the ribosome as polypeptide is formed.</p>		
<p>(e) Different proteins can be expressed from one gene, as a result of alternative RNA splicing and post-translational modification. Different mRNA molecules are produced from the same primary transcript depending on which exons are included in the mature RNA transcript. Post translation protein structure modification by cutting and combining polypeptide chains or by adding</p>		

key areas	Suggested learning activities	Exemplification of key areas
phosphate or carbohydrate groups to the protein.		
(f) Proteins are held in a three-dimensional shape — peptide bonds, folded polypeptide chains, hydrogen bonds, interactions between individual amino acids		
<p><b>4 Cellular differentiation</b></p> <p>(a) Cellular differentiation is the process by which a cell develops more specialised functions by expressing the genes characteristic for that type of cell.</p> <p>Differentiation into specialised cells from meristems in plants; embryonic and tissue (adult) stem cells in animals.</p> <p>(b) Embryonic and tissue (adult) stem cells. Research and therapeutic uses of stem cells by reference to the repair of damaged or diseased organs or tissues. Stem cell research provides information on how cell processes such as cell growth, differentiation and gene regulation work. Stem cells can be used as model cells to study how diseases develop or for drug testing. The ethical issues of stem cell use and the regulation of their use.</p>	<p>Tissue culture of plant material.</p> <p>Case study on use of stem cells in repair of diseased or damaged organs (eg skin grafts, bone marrow transplantation and cornea repair).</p> <p>Case study on ethics of stem cell research and sources of stem cells. For example, embryos must not be allowed to develop beyond 14 days, around the time a blastocyst would be implanted in a uterus.</p> <p>Sources of stem (adult) cells include embryonic stem cells, tissue stem cells and attempts to reprogram specialised cells to an embryonic state (induced pluripotent stem cells). Ethical issues could include: regulations on the use of embryo stem cells, the use of induced pluripotent stem cells and the use of nuclear transfer techniques.</p>	<p>Meristems are regions of unspecialised cells in plants that are capable of cell division.</p> <p>Stem cells are unspecialised somatic cells in animals that can divide to make copies of themselves (self-renew) and/or differentiate into specialised cells.</p> <p>Cells in the very early embryo can differentiate into all the cell types that make up the organism (are pluripotent). These cells don't self-renew in vivo, but can under the right conditions in the lab. It is then they are termed embryonic stem cells and are used as a source of stem cells in research.</p> <p>Tissue (adult) stem cells are needed for growth, repair and renewal of tissues. They replenish differentiated cells that need to be replaced and give rise to a</p>

key areas	Suggested learning activities	Exemplification of key areas
		<p>more limited range of cell types (are multipotent), eg blood stem cells found in the bone marrow produces the various blood cell types.</p> <p>Once a cell becomes differentiated it only expresses the genes that produce the proteins characteristic for that type of cell.</p> <p>The therapeutic uses of stem cells should be exemplified by reference to the repair of damaged or diseased organs, eg corneal transplants, and skin grafts for burns.</p>
<p><b>5 The structure of the genome</b></p> <p>The genome of an organism is its entire hereditary information encoded in DNA. DNA sequences that code for protein are defined as genes.</p> <p>The structure of the genome — coding and non-coding sequences.</p> <p>A genome is made up of genes and other DNA sequences that do not code for proteins.</p> <p>Non-coding sequences include those that regulate transcription and those that are transcribed to RNA but are never translated. Some non-coding sequences have no known function.</p>	<p>Non translated forms of RNA include tRNA, rRNA and RNA fragments.</p>	<p>Most of the eukaryotic genome consists of these non-coding sequences. Non-translated forms of RNA include tRNA, rRNA and RNA fragments.</p>

key areas	Suggested learning activities	Exemplification of key areas
<b>6 Mutations</b> (a) Mutations are changes in the genome that can result in no protein or an altered protein being expressed.	Investigate mutant yeast or germination rates of irradiated seeds.	
(b) Single gene mutations involve the alteration of a DNA nucleotide sequence as a result of the substitution, insertion or deletion of nucleotides. Single-nucleotide substitutions include: missense, nonsense and splice-site mutations. Nucleotide insertions or deletions result in frame-shift mutations or an expansion of a nucleotide sequence repeat.	Investigate how point mutations can be silent, neutral, missense, nonsense or frame-shift. Research reasons for geographical variation in incidence of post-weaning lactose tolerance or sickle-cell trait in humans as examples of point mutation.	Regulatory sequence mutations can alter gene expression. Splice site mutations can alter post-transcriptional processing.
(c) Chromosome structure mutations — duplication, deletion, inversion and translocation.	Analyse evidence for formation of human chromosome 2 by fusion of two ancestral chromosomes. Gene duplication and alpha and beta globins in haemoglobin.	Alterations to the structure of one or more chromosomes.
(d) Importance of mutations and gene duplication in evolution.		
(e) Polyploidy — errors during the separation of chromosomes during cell division (nondisjunction) can result in cells with whole genome duplications. Importance of polyploidy in evolution and human food crops	Research polyploidy in plants and importance in origin of crop plants. Research rarity of polyploidy in animals.	Polyploidy examples include banana (triploid) and potato (tetraploid), as well as swede, oil seed rape, wheat and strawberry.

key areas	Suggested learning activities	Exemplification of key areas
<p><b>7 Evolution</b></p> <p>(a) Evolution — the changes in organisms over generations as a result of genomic variations.</p>		
<p>(b) Gene transfer. Vertical (inheritance) - from parent to offspring as a result of sexual or asexual reproduction. Prokaryotes can exchange genetic material horizontally, resulting in rapid evolutionary change. Prokaryotes and viruses can transfer sequences horizontally into the genomes of eukaryotes.</p>		
<p>(c) Selection. Natural selection is the non-random increase in frequency of DNA sequences that increase survival and the non-random reduction in deleterious sequences. Sexual selection is the non-random increase in frequency of DNA sequences that increase reproduction. The differences in outcome as a result of stabilising, directional and disruptive selection.</p>	<p>Gather data on sexual selection in brine shrimp.</p>	
<p>(d) Genetic drift. The random increase and decrease in frequency of sequences, particularly in small populations, as a result of neutral mutations and founder effects.</p>		
<p>(e) Speciation is the generation of new biological species by evolution as a result of isolation,</p>	<p>Research different definitions of the term species (eg biological species concept,</p>	<p>A species is a group of organisms capable of interbreeding and producing</p>



key areas	Suggested learning activities	Exemplification of key areas
<p>mutation and selection. The importance of geographical barriers in allopatric speciation. The importance of behavioural or ecological barriers in sympatric speciation. Hybrid zones.</p>	<p>phylogenetic species concept) and the difficulty of applying species definition to asexually reproducing organisms.</p> <p>Research the London Underground mosquito.</p> <p>Collaborative data gathering of hooded crow and carrion crow hybrid zone in Scotland.</p>	<p>fertile offspring, and which does not normally breed with other groups.</p> <p>The formation of hybrid zones in regions where the ranges of closely related species meet.</p>
<p><b>8 Genomic sequencing</b> (a) Genomic sequencing — the sequence of nucleotide bases can be determined for individual genes and entire genomes. To compare sequence data, computer and statistical analyses (bioinformatics) are required.</p>	<p>Research how sequencing technologies use techniques such as fluorescent tagging of nucleotides to identify the base sequence.</p>	
<p>(b) Evidence from phylogenetics and molecular clocks to determine the main sequence of events in evolution: last universal ancestor, prokaryotes, photosynthesis, eukaryotes, multicellular organisms. The sequence of events can be determined using sequence data and fossil evidence. Comparison of sequences provides evidence of the three domains (bacteria, archaea and eukaryotes)</p>	<p>Case study on the evolution of bears and primates using Geneious software.</p> <p>Highly conserved DNA sequences are used for comparisons of distantly related genomes.</p> <p>Compare number and proportion of shared genes between organisms such as <i>C. elegans</i>, <i>Drosophila</i> and humans.</p>	<p>The use of sequence data to study the evolutionary relatedness among groups of organisms. Sequence divergence is used to estimate time since lineages diverged.</p> <p>The 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,</p>

key areas	Suggested learning activities	Exemplification of key areas
		vertebrates, land plants.
<p>(c) Comparison of genomes from different species.</p> <p>Comparison of genomes reveals that many genes are highly conserved across different organisms.</p>	<p>Research the importance of the Fugu genome as an example of a very small vertebrate genome with a high rate of chromosome deletion.</p> <p>Comparison of human and chimp genomes reveals rapid change in genes for immune system and regulation of neural development over last 6 million years.</p>	<p>Many genomes have been sequenced, particularly of disease-causing organisms, pest species and species that are important model organisms for research.</p>
<p>(d) Personal genomics and health.</p> <p>Pharmacogenetics</p> <p>Analysis of an individual's genome may lead to personalised medicine through knowledge of the genetic component of risk of disease and likelihood of success of a particular treatment.</p> <p>Difficulties with personalised medicine.</p>	<p>Comparison of individual's genomes focuses on point mutations, repetitive sequence errors and blocks of duplication and deletion.</p>	<p>The difficulties in distinguishing between neutral and harmful mutations in both genes and regulatory sequences, and in understanding the complex nature of many diseases.</p>

# Administrative information

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**Superclass:** RH

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## History of changes to National Unit Specification

Version	Description of change	Authorised by	Date
2.0	Page 1 – the description of key areas under ‘Unit outline’ has been revised to give more information  Page 4 – in Outcome 1.3, the word ‘accurately’ has been replaced by ‘correctly’.  Pages 4 - 5 – the Evidence requirements have been rewritten to better explain what is required  Page 5 – information has been added on Transfer of Evidence	Qualifications Development Manager	April 2014
3.0	Assessment Standards 2.2 & 2.3 removed	Qualifications Development Manager	June 2014
4.0	Level changed from Higher to SCQF level 6. Unit support notes added. Assessment standard threshold added.	Qualifications Manager	September 2018

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