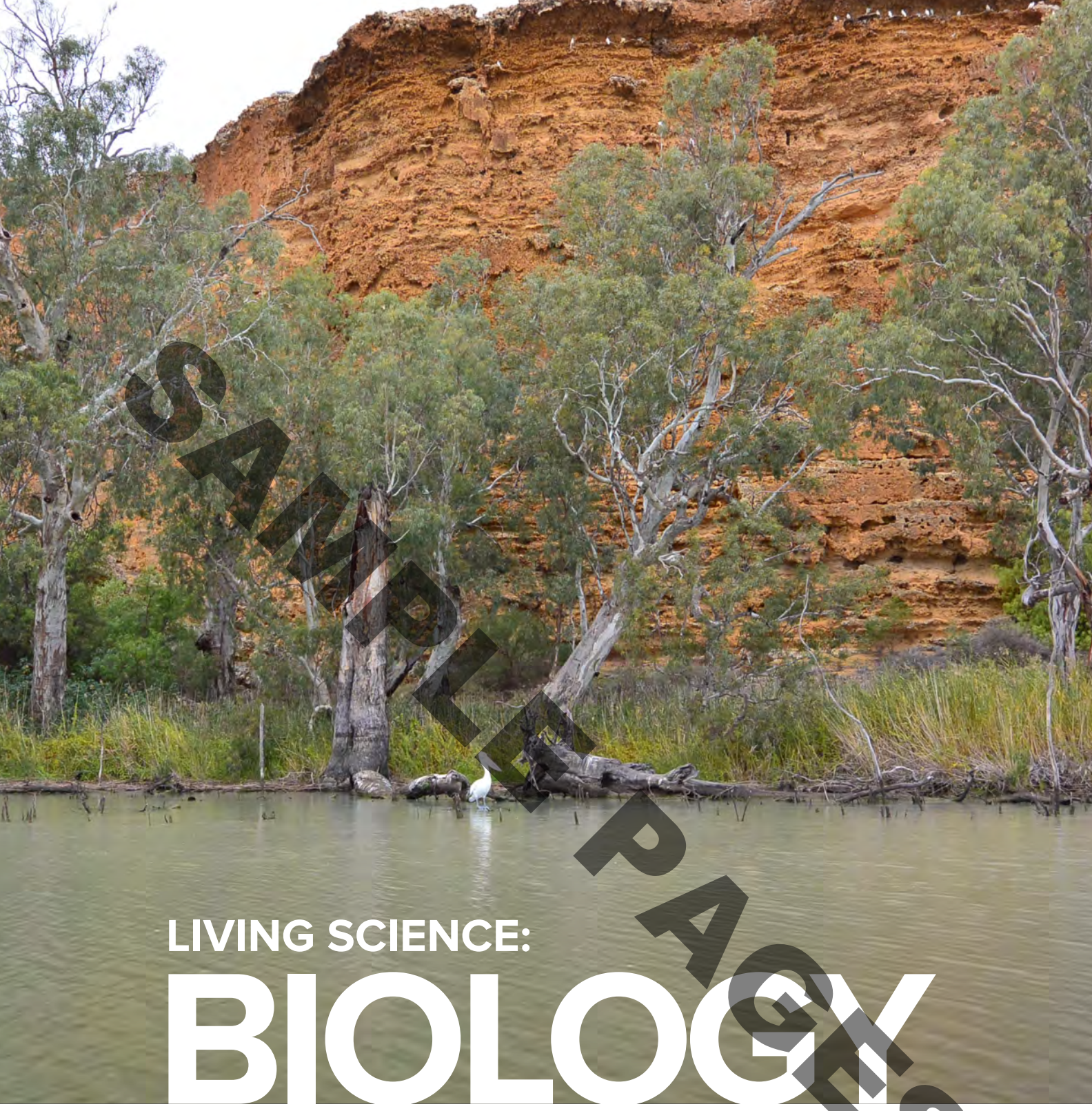

LIVING SCIENCE:
BIOLOGY

FOR SACE STAGE 2

SASTA
South Australian Science Teachers Association

Supporting Teachers of Science
Advancing Science Education

AUSTRALIAN CURRICULUM EDITION

DR KATHY ADAMS

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LIVING SCIENCE: BIOLOGY FOR SACE STAGE 2 Australian Curriculum Edition

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Cover photograph taken by Michael Adams ©
The Australian Pelican (*Pelecanus conspicillatus*) is often seen swimming along the River Murray. This photo taken on the River Murray at Wongulla, South Australia shows the pelican relaxing by the bank on a fallen tree with the characteristic red cliffs that flank the river in the background with white cockatoo (*Cacatua galerita)* lining the highest edges.

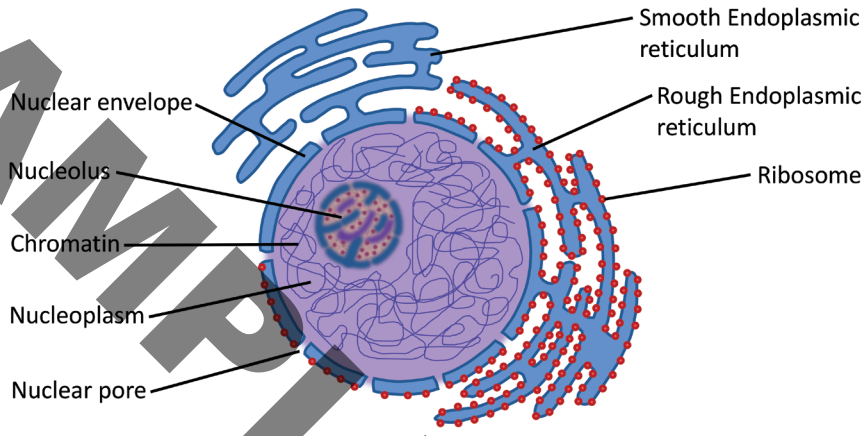
CHAPTER 1 DNA AND PROTEINS

DNA: Structure of Chromosomes in Prokaryotes and Eukaryotes

DNA in Eukaryotes is Linear

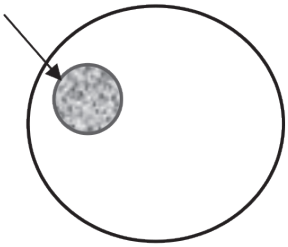
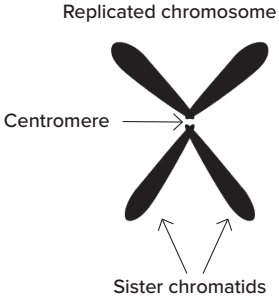
Eukaryotic cells have a membrane bound nucleus (Figure 1.6). The DNA is surrounded by a nuclear membrane, which has nuclear pores, to enable molecules to enter and exit the nucleus.

Figure 1.6: Nucleus.



The DNA in the nucleus, looks like multiples pieces of string that have been randomly squeezed inside the membrane. The reality is that the DNA is in a diffuse state and is called chromatin (Table 2). However, if it was possible to get close enough to view it, these "pieces of string" are actually multiple independent linear chromosomes, where the DNA molecules are bound to and coiled around proteins (called histones). Therefore, the packaging of DNA inside the nucleus is highly ordered.

Table 2: Different forms of DNA.

CHROMATIN	CHROMOSOME	CHROMATID
		
The form of DNA found in the nucleus of eukaryotic cells. The DNA is bound and coiled around proteins called histones.	Chromosomes are formed when chromatin condenses during cell division. Made up of two sister chromatids connected by a centromere. Sister chromatids are 2 copies of the same genetic information.	Sister chromatids separate during cell division. A single molecule of DNA that represents a copy of a chromosome in new cells. Single chromatids are not visible, becomes chromatin.

Later in *this book*, more detail about the structure and function of two organelles present in eukaryotic cells: mitochondria and chloroplasts will be provided. It has been hypothesised that these two organelles evolved from ancestral prokaryotes that had the ability to undergo aerobic respiration and photosynthesis and were engulfed by a larger prokaryotic cell, where they formed an endosymbiotic relationship. These organelles each contain their own circular DNA, resembling the prokaryote chromosome. This genetic information contains all the information needed for the mitochondria and chloroplasts to undertake their respective functions and for their own division, by binary fission. The presence of these circular DNA molecules in these organelles is hypothesised to be due to their prokaryotic origins. This theory known as the endosymbiotic theory will be described in detail in *Chapter 4*.

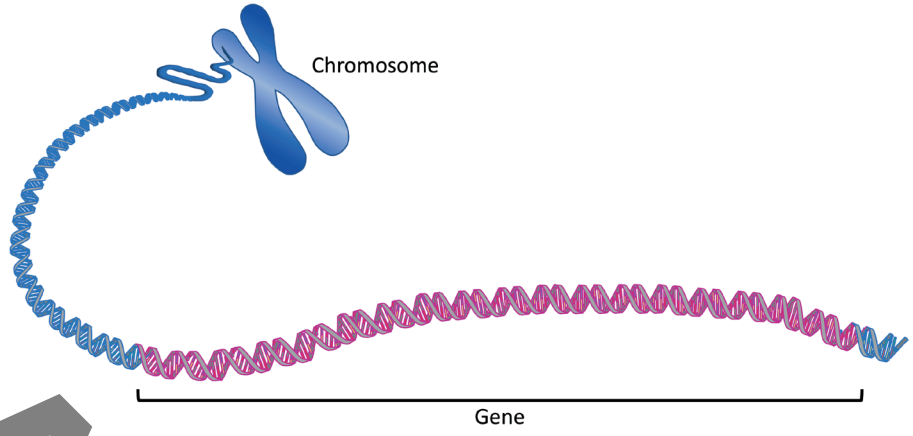
Table 3 compares the features and characteristics of the chromosomes and associated processes in prokaryotes and eukaryotes.

Table 3: A comparison of chromosomes in Prokaryotes and Eukaryotes

PROKARYOTE CHROMOSOME	EUKARYOTE CHROMOSOMES
Simple genome Most contain a single circular chromosome*	Complex genome Contain multiple linear chromosomes
Are condensed into the nucleoid region of the cell, usually due to the ability of the circular DNA molecule to coil onto itself^.	Are condensed into a membrane bound nucleus due to the presence of histone proteins.
Contain one copy of each gene No introns	Contain 2 copies of a gene Genes contain exons and introns
No homologous pairs	Chromosomes are organised into homologous pairs and observed as a karyotype.
DNA replication, transcription and translation occur in the cytoplasm.	DNA replication and transcription (first part of protein synthesis) occur in the nucleus. Translation (second part of protein synthesis) occurs in the cytoplasm on ribosomes.
Small quantity of non-coding DNA and repetitive sequences.	Contain large quantities of non-coding DNA and repetitive sequences.
Contain extrachromosomal DNA called plasmids.	Rare to find extrachromosomal DNA.

DID YOU KNOW? There are generally always exceptions to the rule in Biology! Here are some rare exceptions to the rule that prokaryotes have a singular circular chromosome. The bacterial species *Borrellia spirochetes* (which causes Lyme disease) contains a linear chromosome^ rather than a circular one and *Vibrio cholerae* (which causes the disease cholera), contains two overlapping circular chromosomes*. Eubacteria do not have histone proteins, however, recent studies have shown that the DNA may associate with other cell proteins to assist with its packaging into the cytoplasm, and in another twist, archaebacterial species do have histones and package their DNA like eukaryotes!

Figure 1.10: The relationship between a chromosome, gene and DNA.



An individual's genotype, the DNA sequences of each gene inherited cannot be altered (except by mutation in a particular cell(s)). However, there are other mechanisms of DNA modification such as methylation that may alter the way genes are expressed and hence affect the phenotype of an individual. A phenotype is the actual physical expression of the characteristics an individual possesses and displays e.g. eye or skin colour.

Exons and Introns

The basic function of genes are the same regardless of the organism, they are the unit of heredity but the structure of genes are different in eukaryotes compared to prokaryotes.

In eukaryotes, the gene is composed of two regions called exons and introns. Exons have coding sequences that will enable the product of the gene to be expressed while the intron regions of a gene are considered to be non-coding and interrupt the sequence for the gene product, and therefore must be removed from the RNA template before a gene product can be made.

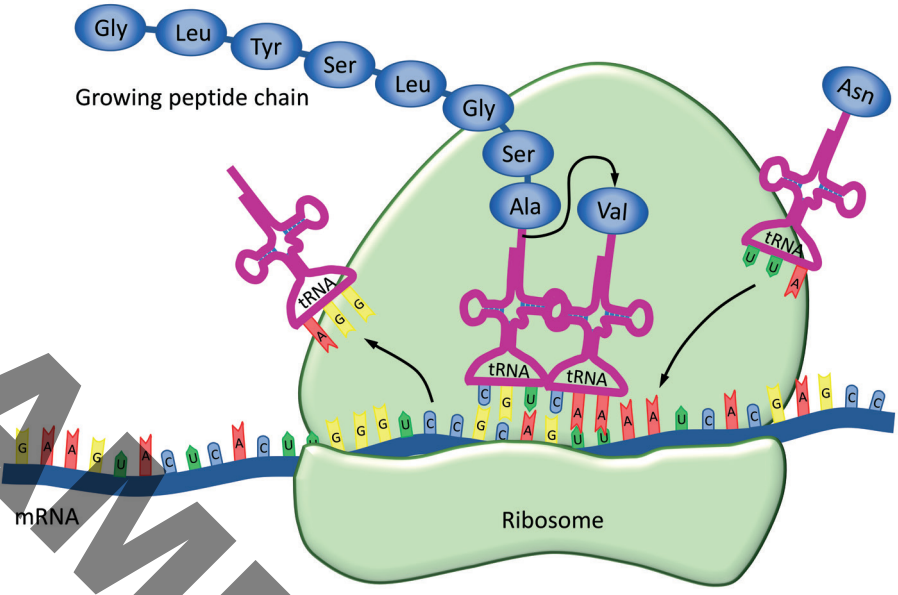
DID YOU KNOW? An easy way to remember the role of exons and introns is to remember that the exons are **ex**pressed, and share the prefix '**ex**'. Introns, **in**terrupt the coding regions and share the same prefix '**in**'.

Introns are removed after transcription

To remove the introns requires specific processing, known as RNA splicing after the first process of protein synthesis, called transcription has occurred (see later in *this chapter* for the details of protein synthesis including transcription and translation).

DID YOU KNOW? An enzyme known as RNA-splicing endonuclease is responsible for the removal of the introns from mRNA that will form other RNA molecules such as transfer RNA (tRNA), and a RNA-protein complex known as a spliceosome removes introns from mRNA destined to be functional protein.

Figure 1.15: Translation



DID YOU KNOW? The ribosomes in prokaryotes and eukaryotes differ. The prokaryotic ribosome is smaller, containing less rRNA and fewer proteins. The differences are shown below.

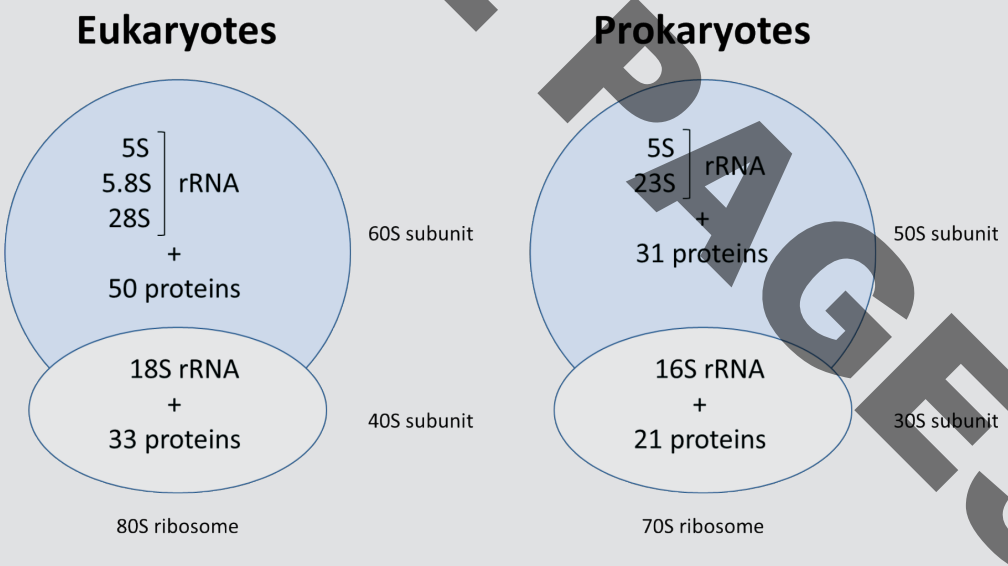


Table 7: The role of DNA, mRNA, tRNA and rRNA in Protein Synthesis.

COMPONENT	ROLE IN PROTEIN SYNTHESIS
DNA	The double helix unwinds and the two strands separate. The coding strand (which contains the genetic information of genes and therefore the correct order of bases to produce its specific product) is not used as a template in protein synthesis. The other strand, the non-coding strand, or template strand is used in transcription to synthesise mRNA.
mRNA	Synthesised during transcription, mRNA is a working copy of the gene. It enables the protein encoded by the gene to be made. It has the same sequence as the coding strand of DNA, except uracil (U) will be found instead of thymine (T). mRNA carries codons which are triplets of bases, that are complementary to an anti-codon on specific tRNA molecules which corresponds to a particular amino acid. This is how mRNA directs the order of amino acids that are joined together to form the polypeptide. mRNA is synthesised in the nucleus of eukaryotic cells and in the cytoplasm of prokaryotic cells. mRNA then travels to a ribosome.
tRNA	Carry amino acids to the ribosome to enable the synthesis of the polypeptide. Each tRNA has a specific amino acid associated with it, according to the anti-codon sequence on the molecule. The tRNA will specifically interact with the codons present on mRNA, again utilising the complementary base pair rule. Adenine will pair specifically with uracil and cytosine and guanine will specifically base pair.
rRNA	Ribosomes are made up of rRNA and proteins. The rRNA is the catalytic component of a ribosome. It is this rRNA component of the ribosome that enables the polypeptide to be synthesised.

Table 8: The relationship between DNA and RNA codons, anti-codons and amino acids.

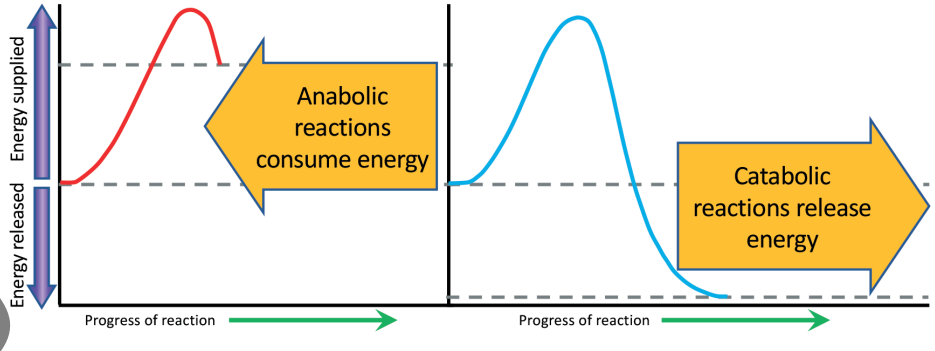
COMPONENT	RELATIONSHIP TO OTHER COMPONENTS
DNA	Coding strand carries the genetic information required to produce a gene product from a gene. The template strand is used to produce a copy of the coding strand in the form of messenger RNA (mRNA).
RNA	mRNA is translated into a polypeptide chain, through its interaction with a ribosome, where the codons (triplet bases) are aligned with complementary bases (anti-codons) on tRNA molecules. The codons have the same sequence as the coding strand of DNA, except where a thymine base (T) is present in DNA, it will be a uracil (U) base in RNA.
Anti-codons	Found on tRNA molecules. They will specifically bind (due to complementary base pairing) to a codon on the mRNA in the presence of a ribosome. The anti-codon is specific for the amino acid bound to the tRNA molecule. The anti-codons have the same sequence as the template strand of DNA, except where a thymine base (T) is present in DNA, it will be a uracil (U) base in RNA.
Amino acids	There are 20 amino acids that can be used to produce a polypeptide. The order of amino acids is determined by the gene sequence. Amino acids can only bind to a particular tRNA with the anti-codon that corresponds to that amino acid.

Review Questions: Check your understanding.

14. Outline the steps involved in semi-conservative DNA replication. Use a diagram to aid your explanations if you wish.
15. Describe the role of complementary base pairing in DNA replication.
16. Suggest why it is necessary to have an exact copy of DNA replicated.
17. What is a gene? Use your own words to provide a definition.
18. Describe the difference between exons and introns.
19. Why are introns removed from the mRNA after transcription?
20. Describe the steps involved in transcription.
21. Describe the steps involved in translation.
22. Describe the relationship between codons, anti-codons and amino acids.
23. Why is the DNA base sequence found in a gene critical to the product produced by protein synthesis?
24. State the functions of the different forms of RNA.
25. Suggest a reason why codons are three nucleotide bases long?
26. Use one of the codon usage tables provided, and state the following:
    i) the amino acid with the most synonymous codons.
    ii) the amino acid with the least synonymous codons.
    iii) the three stop codons.
    iv) at least one codon that represents the amino acids:
        - Arginine.
        - Serine.
        - Glutamic acid.

    v) the effect if the codon
        - UAC was changed to UAU.
        - UGC was changed to UGA.
        - GUU was changed to GCU.

Figure 1.21: Graphs of Anabolic and Catabolic Reactions.



Anabolic reactions synthesise larger substances required by the cell from smaller substrates and requires an input of energy (which is usually in the form of adenosine tri-phosphate, ATP, *see Chapter 2*).

Enzymes reduce the activation energy needed for these types of reactions, as they bring the substrates into close proximity in the active site, and orientate them such that the reaction can proceed at a lower energy level. Anabolic reactions can be identified in energy graphs as the products will have more energy than the substrate.

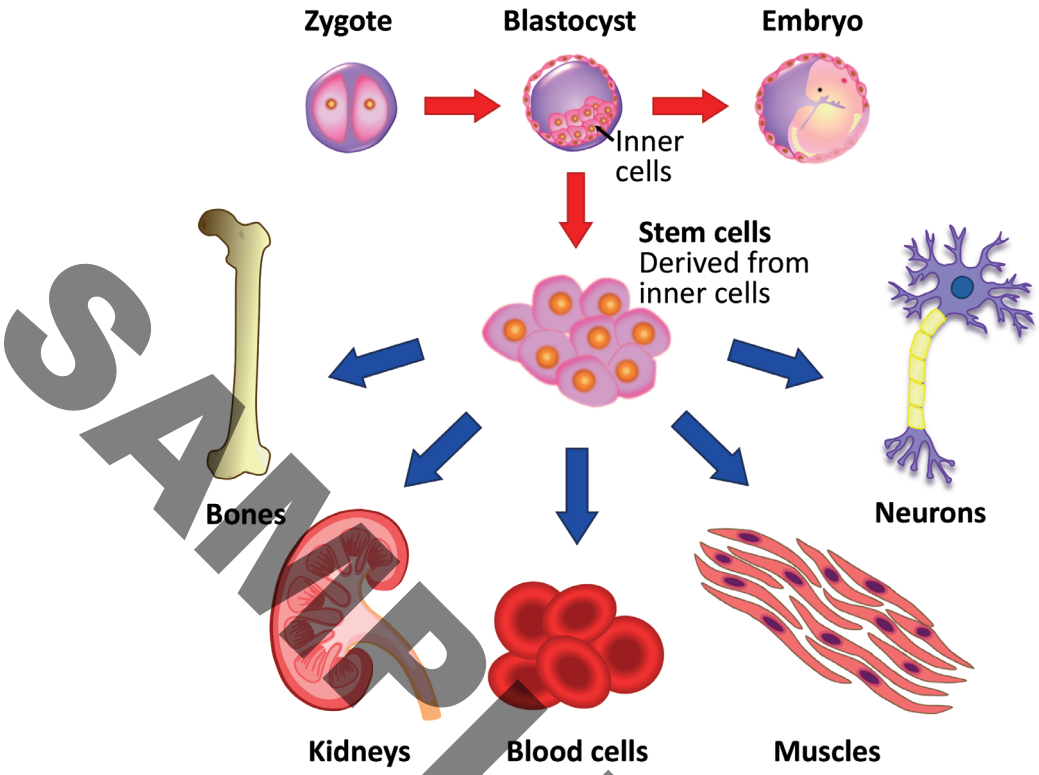
DID YOU KNOW? The synthesis of all the complex molecules found in a cell, such as carbohydrates, lipids, proteins and nucleic acids, are generated by anabolic reactions. Important cell processes like photosynthesis, protein synthesis, and DNA replication also rely on anabolic reactions.

Catabolic reactions occur when larger molecules are broken down into smaller products, which releases a net gain of energy, which can be utlised to undertake energy requiring functions in the cell. Catabolic reactions occur during processes such as cellular respiration and the metabolism of glucose.

Enzymes reduce the activation energy needed for these types of reactions, as they orientate the substrate in the active site in a particular manner that causes stress on the bonds which leads to the bonds being broken, and the reaction can then proceed at a lower energy level. Catabolic reactions can be identified in energy graphs as the products will have less energy than the substrate.

Enzymes are highly specific for their substrate(s). Substrates will bind to an enzyme if there is a complementary shape to the active site on the enzyme. The interaction between the substrate and the enzyme can be described by the **induced fit model**. This model describes how the initial complementary binding results in slight changes in the shape of the enzymes active site that causes the substrate to better fit into this space (this moulding is called induced fit binding,

Figure 1.29: Cell differentiation: Zygote to Differentiated Cells in Humans.



DID YOU KNOW? In the human body alone, there are over 200 different types of cells. In contrast, plants have typically three cell types, parenchymal, collenchymal and sclerenchymal. Cells on their own in a multicellular organism would not function effectively and so the cells of similar structure and function are arranged into tissues. Tissues of different types are then packaged together to perform a common function as an organ. These organs will then work with other organs and tissues to work together in an organ system.

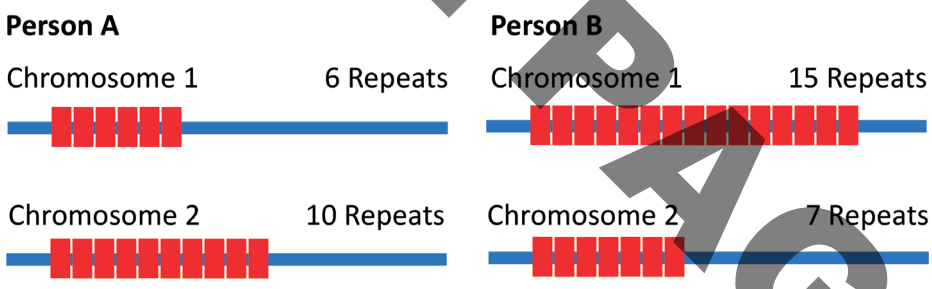
DID YOU KNOW? There are two types of stem cells: Embryonic stem cells and adult stem cells. Embryonic stem cells are called pluripotent as they can become any cell type of the body and are present in the developing embryo, and after birth in umbilical cord blood. The second type of stem cell, adult stem cells in theory could also divide into any cell type, however, they are limited in their ability to differentiate due to their tissue of origin. Scientists are currently investigating the differences between these cell types, to determine if adult stem cells can be used to potentially treat human diseases instead of using embryonic stem cells, which are ethically more complex to use. Adult stem cells could be taken directly from the patient, reducing the need to locate genetic matches and reduce the chance of rejection by the host's immune system.

More modern techniques of DNA profiling use sections of the DNA called variable number tandem repeats (VNTRs) and microsatellites, also referred to as short tandem repeats (STRs). These sections of DNA can be specifically amplified using PCR or obtained by digesting DNA with particular restriction enzymes and purifying the required DNA fragments.

VNTRs are generally 6 to 100 base pairs in length (e.g. ACAGGGTGTGGGG) and are found in numerous locations throughout human chromosomes. VNTRs are not used as often in modern DNA profiling as they can be at times difficult to interpret when comparing profile patterns. STRs are more commonly used today as the method of analysis is also more cost effective and automated. STR loci are spread throughout the genome including all the autosomal chromosomes and the X and Y sex chromosomes. They have a repeat pattern of generally between two and eight base pairs in length (e.g. AGCT) and the repeats typically range from 50 to 300 base pairs.

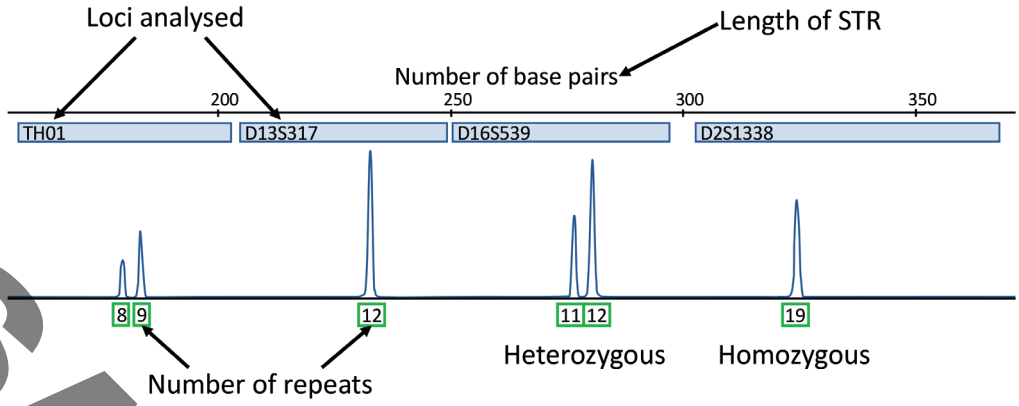
Figure 1.41 demonstrates two DNA profiles using STRs for two individuals for one loci. In person A, they have for the loci shown an STR pattern of 5, 10. While person B has a profile of 15, 7. What does this mean? Remember that you inherit a copy of your mother's DNA and a copy of your father's. Each provide to you one copy of each gene needed to encode for the characteristics that you need to function. Therefore each loci has 2 genes. It means that Person A inherited 6 repeats for this loci from one parent and 10 from the other. Person B has a different repeat profile, and therefore different DNA profile as they inherited 15 repeats from one parent and 7 from the other.

Figure 1.41: STR profile of Person A and Person B for one loci.



Through electrophoresis, these STRs can be represented on a different type of electropherogram. Figure 1.42 represents an electrophoregram of multiple STRs in one individual. These profiles are generated so that scientists can determine the number of repeats in each allele at a loci, the total length of the STR, and if an individual is homozygous or heterozygous for the STR. These pieces of information enable scientists to determine with relative accuracy the profile of the DNA and therefore match it to an individual.

Figure 1.42: An electropherogram showing an STR profile of 4 loci in one individual.



The information in the electropherogram above (Figure 1.42) can also be presented as a table of data (Table 15).

Table 15: Data from an electropherogram can be presented in a tabular format.

LOCUS	LOCATED ON CHROMOSOME	STR REPEAT SEQUENCE	ALLELE VALUES (Number of Repeats)
TH01	11	AATG	8, 9
D13S317	13	TATC	12, 12
D16S539	16	GATA	11, 12
D2S1338	2	TGCC	19, 19

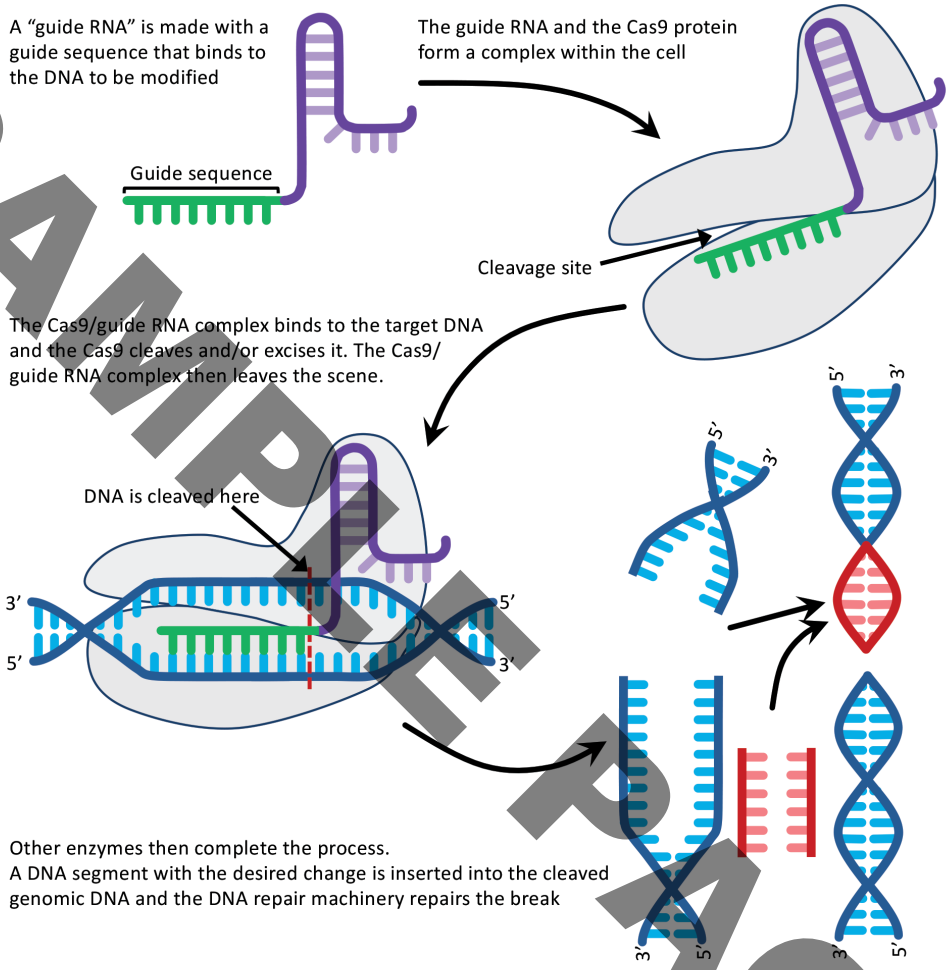
DID YOU KNOW? Another type of variation increasingly used in DNA profiling are Single Nucleotide Polymorphisms (SNPs). These types of changes occur due to mutation from one generation to the next and account for many of the differences seen in DNA sequences between individuals. It is estimated that there is a mutation every 1200 base pairs, so each individual shares 99.9% of their DNA with another individual. SNPs which are found throughout the genome are identified by looking at each allele, and determining if there is a sequence change between the two genes. That is, the maternal gene may carry a G, while the paternal gene has a T. This is a SNP. As there are thousands, multiple SNPs provide a reliable method to determine a DNA profile of any individual. While an individual may share one SNP, it is extremely unlikely that two individuals will have 100 the same (except for identical twins).

Useful resources: Nikolay's Genetic Lessons: How to read STR electropherogram, 2018. https://www.youtube.com/watch?v=W1tiMcZ8aSc

Bob Trenwith, 2018. Introduction to Forensic Science - 4.4 Short Tandem Repeats STR https://www.youtube.com/watch?v=HdzBh6sMLA0

DID YOU KNOW? CRISPR/Cas9 is not the only gene editing mechanism used by scientists. Others include Zinc finger nucleases and the TALEN system.

Figure 1.50: CRISPR



SHE Alert: Since the discovery of CRISPR-Cas9 technology multitudes of applications have been published in hundreds of scientific journals and newspapers around the world for scientists and society to be informed (*Communication and Collaboration*). The hope for CRISPR is that it is going to cure cancer, give us personalised medicines and solve all the issues facing the agricultural industry as they grapple with increasing human demand and climate change. All it takes to see the impact this technology is having is to look at the headlines: "CRISPR editing of plants and animals gets green light in Australia. Now what? Anna Salleh, ABC Science, https://www.abc.net.au/news/science/2019-04-30/crispr-gene-editing-in-the-food-chain/11053622, Eric Nilner, National Geographic, https://www.nationalgeographic.com/environment/future-of-food/food-technology-gene-editing/, John Ueland, MIT Technology Review, https://www.technologyreview.com/s/612997/the-crispr-twins-had-their-brains-altered/