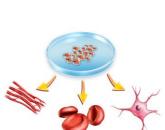
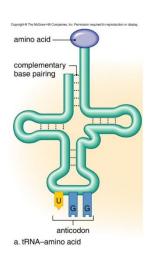
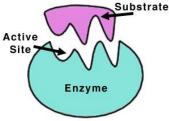
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UNIT 1 - NOTES

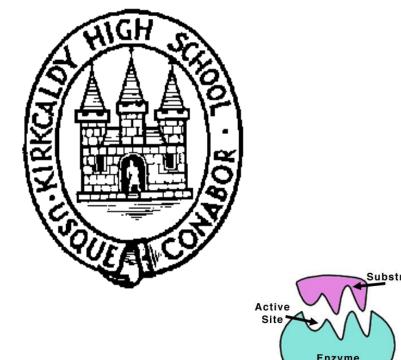


Higher Human BIOLOGY





< A



Introduction

This course allows candidates to acquire a deeper understanding of cellular processes, physiological mechanisms and their impact on health, aspects of the nervous system, and defence mechanisms as they apply to the human species.

The course aims to:

- develop and apply knowledge and understanding of human biology
- develop an understanding of human biology's role in scientific issues and relevant applications of human biology, including the impact these could make in society
- develop scientific inquiry and investigative skills
- develop scientific analytical thinking skills, including scientific evaluation, in a human biology context
- develop the skills to use technology, equipment and materials safely in practical scientific activities
- develop planning skills
- develop problem-solving skills in a human biology context
- use and understand scientific literacy to communicate ideas and issues and to make scientifically informed choices
- develop the knowledge and skills for more advanced learning in human biology
- develop skills of independent working

There are three units for Higher Human Biology:

- Unit 1 Human Cells
- Unit 2 Physiology and Health
- Unit 3 Neurobiology and Immunology

<u>Assessment</u>

Assessment of Higher Human Biology has three components:

Question paper 1 25 Multiple choice questions

Question paper 2 *95 mark written paper*

Assessed in SQA exams in May

Assignment: Produced in school under exam conditions

Apparatus and techniques

In addition to the key areas, candidates must have knowledge of the following pieces of apparatus and become familiar with the following techniques. You should check these off as we progress through the course:

<u>Apparatus</u>

- 🗆 beaker
- balance
- measuring cylinder
- □ dropper/pipette
- □ test tube/boiling tube
- □ thermometer
- funnel
- syringe
- timer/stopwatch
- 🗆 Petri dish
- water bath
- □ colorimeter
- pulsometer
- □ sphygmomanometer

<u>Techniques</u>

- using gel electrophoresis to separate macromolecule, for example DNA fragments
- using substrate concentration or inhibitor concentration to alter reaction rates
- neasuring metabolic rate using oxygen, carbon dioxide and temperature probes
- □ using a respirometer
- measuring pulse rate and blood pressure
- measuring body mass index

The small print: Key Area 1 Division and differentiation in human cells (a) Division of somatic and germline cells. • Somatic stem cells divide by mitosis to form more somatic cells. • Germline stem cells divide by mitosis and by meiosis. • Division by mitosis produces more germline stem cells. • Division by mitosis produces haploid gametes.
 (b) Cellular differentiation 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. Embryonic and tissue stem cells. Cells in the very early embryo can differentiate into all the cell types that make up the individual and so are pluripotent. Tissue stem cells are involved in the growth, repair and renewal of the cells found in that tissue. They are multipotent.
 (c) Therapeutic and research uses of stem cells. Therapeutic uses involve the repair of damaged or diseased organs or tissues
 (d) Cancer cells Cancer cells divide excessively because they do not respond to regulatory signals This results in a mass of abnormal cells called a tumour Cells within the tumour may fail to attach to each other, spreading through the body where they may form secondary tumours

<u>Ur</u> Unit 1: Prior knowledge

- □ Tissues, organs and systems
- Cell division
- □ Cell ultrastructure and function
- □ Cell division and chromosomes
- Base sequence and base pairing of DNA
- □ Function of proteins
- Enzymes
- □ Summary equation for respiration
- ATP and energy

To ensure success, it is **Vital** that you are secure in your previous Biology knowledge.

Sure success, it is **Vital** that be secure in your previous by knowledge. First **homework exercise** will bu an opportunity to check have a good basis to move on at r level. Your first homework exercise will give you an opportunity to check you have a good basis to move on at Higher level.

(a) <u>Division of Somatic and Germline cells</u>



Cardiac muscle cells





Nerve cells

The human boay is made up of many ______ cells that have specific structural, ______ and biochemical properties. Specialised cells arise from the differentiation of unspecialised cells during ______ development.

Specialised cells with similar functions are grouped into _____. Similar tissues are grouped into _____.

During differentiation, certain genes that express ______ important for the function of a specific cell are '______'. This allows it to develop a more specialised structure to carry out a ______ function. Once a cell becomes differentiated it only expresses the ______ that produce the proteins characteristic for that type of cell.

Somatic Stem cells

Stem cells are	the ability	cells	What is a stem cell?
that have		to	A single cell that can
cells or specialised cells	_ to make more of one or more	into	differentiate into man cell types.

Image prepared by Catherine Terraney for the Votiand Academies, Understanding Stem Cells: An Overview of the Science and Issues from the Vational Academies. Mar. / Away national analysis on the Vational Academies. Mar. / Away national analysis on the Vational Academies. Mar. / Away national analysis on the Vational Academies. Mar. / Away national analysis of the Vational Academies. Mar. / Away national academics on the Vational Academies. Mar. / Away national academics on the Vational Academics of the Va

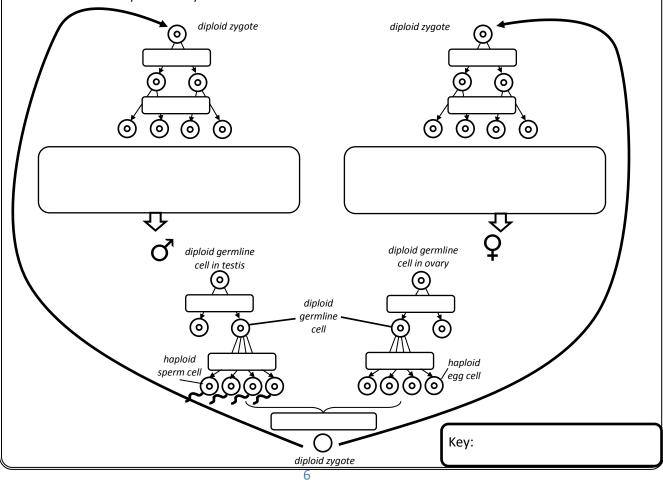
FACT CHECKER: Somatic stem cells:

Write a note above on <u>somatic</u> stem cells.

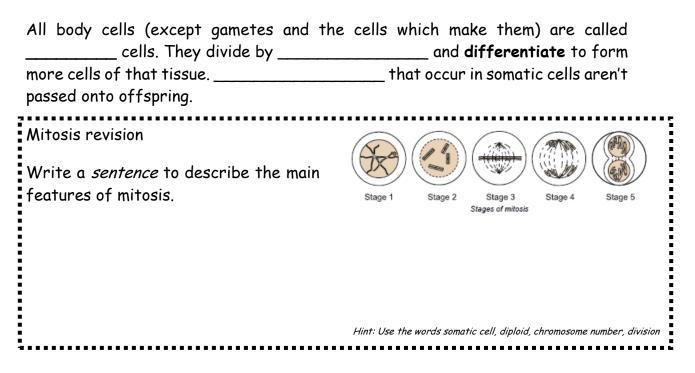
Germline Stem cells

Germline cells are gametes and the cells of the body that eventually form sex cells (______). Like somatic cells, germline cells are ______. This means that the nucleus contains two sets of chromosomes and are therefore able to undergo mitosis to make more germline cells.

FACT CHECKER: Germline cells:
Write a note above on <u>germline</u> cells.
Use the Torrance textbook (page 7) to complete the diagram below. It shows how the human life cycle
alternates between haploid and diploid cells.



Division in somatic cells



Body organs are formed from a variety of tissues made from somatic cells.

Somatic cells make copies of themselves by mitosis.

Division in germline cells

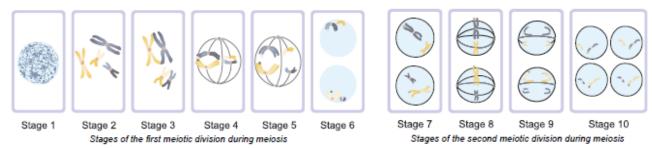
Gametes and the cells that produce gametes are called germline cells. Germline cells can divide in two ways:

- By _____ to produce more diploid germline cells
- By _____ to produce haploid gametes

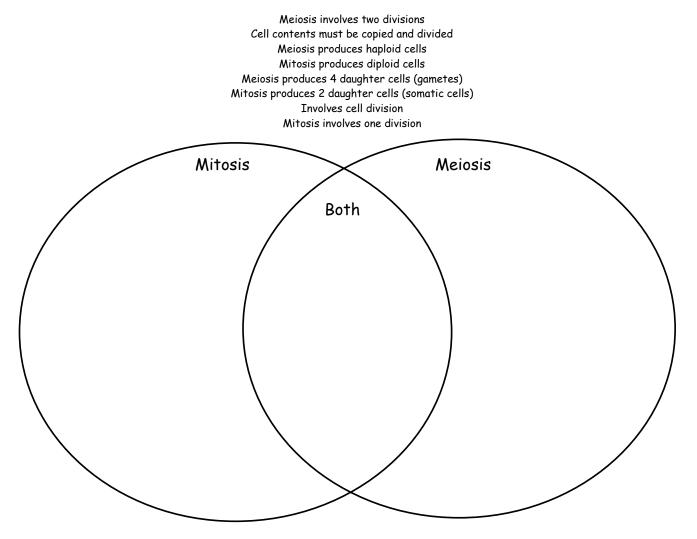
The nucleus of a germline stem cell can divide by mitosis to maintain the diploid chromosome number. Diploid cells have 23 pairs of homologous chromosomes.

The nucleus of a germline stem cell can divide by meiosis. It undergoes two divisions, firstly separating homologous chromosomes and secondly separating chromatids. Haploid gametes contain 23 single chromosomes.

<u>Meiosis:</u>



Use the following mitosis and meiosis facts to complete the venn diagram below:



(b) Cellular differentiation

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

I	nterpretation: What does the above sentence mean?
ΙV	Vrite a note above

Stem Cells

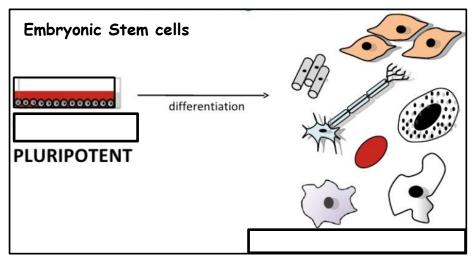
Stem cells are ______ cells that have the ability to ______ to make more stem cells or ______ into specialised cells of one or more types. There are two different types of stem cell:

- Embryonic stem cells
 - Cells in the very early embryo can differentiate into all the cell types that make up the individual and so are **pluripotent**.
 - Pluripotent means _____
- Tissue (adult stem cells)
 - Tissue stem cells are involved in the growth, repair and renewal of the cells found in that tissue. They are **multipotent**.
 - Multipotent means _____

Embryonic Stem Cells

Embryonic stem cells are derived from <u>unspecialised</u> cells found within an <u>embryo</u>. They have the ability to <u>differentiate</u> into all cell types that make up an organism. This is because **most** of their **genes** are still 'switched on' or "**expressed**". All the genes in embryonic stem cells can be switched on so these cells can differentiate into any type of cell.

Embryonic stem cells are said to be **pluripotent**. This means that they have the potential to differentiate into <u>all</u> cell types of the body.

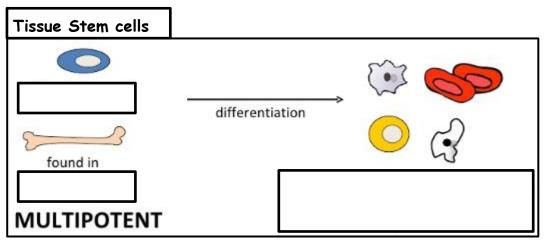


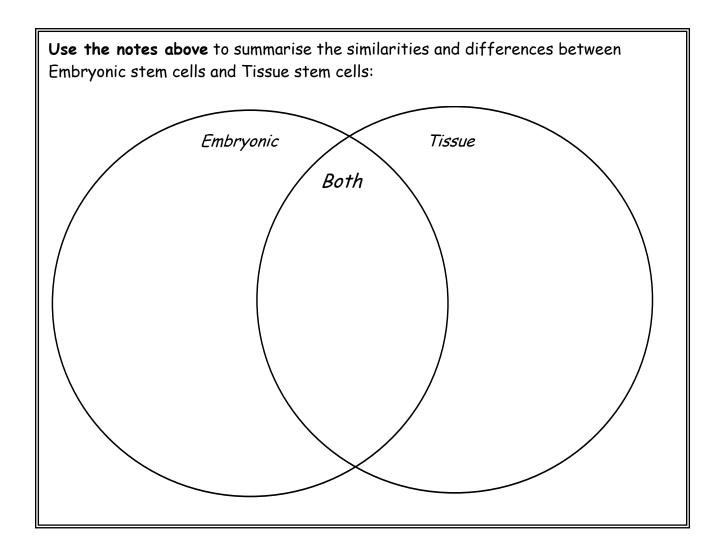
Tissue stem cells

<u>Tissue</u> or adult stem cells are found in various tissues of adults and children, including the <u>brain</u>, **bone marrow**, **skeletal muscle** and **skin**. These cells **replenish** <u>differentiated</u> cells that need replaced through age or damage in the tissues in which they are found. They are able to differentiate into a much more <u>limited</u> range of cell types and will tend to develop into cell types that are closely related to the tissue in which they are found.

Eg tissue stem cells in <u>bone marrow</u> will produce red blood cells, platelets, phagocytes and lymphocytes.

Tissue stem cells are said to be <u>multipotent</u>. This means that they only have the potential to differentiate into a <u>limited</u> number of cell types of the body.





ESSAY 2013: Give an account of cell differentiation under the following headings: (i) Stem cells (4) (ii) Somatic cells (4) (iii) Germline cells (2)

(c) Therapeutic and Research Uses of Stem Cells

Therapeutic uses of stem cells involve the repair of damaged or diseased organs or tissues. Stem cells are said to be "therapeutic" because they can be used to treat or even cure diseases. Stem cells from the embryo can self-renew, under the right conditions, in the lab. These cells can be used to make body tissues that are diseased or damaged.

Two examples of therapeutic uses of stem cells are **corneal repair** and **skin** grafts.

Make notes on how stem cells are used in corneal repair and skin grafts.

Sources: Torrance Pg 10 OR Eurostemcell factsheet

Other therapeutic uses of stem cells include:

- Tissue replacement (in addition to corneal repair and skin grafts):
 - heart valves for heart disease patients
 - brain nerve cells for Parkinson's sufferers
 - blood for transfusions
 - nerve cells for spinal injuries

Stem cell research provides information on how cell processes such as cell growth, differentiation and gene regulation work.

- Model cells: stem cells can be used to study how diseases develop
- Drug testing: stem cells can be used to test new medicines and treatments

Make notes on the advantages of using stem cells in research.

Source: Torrance Pg 9

Ethical issues surrounding stem cell use

Use of embryonic stem cells can offer effective treatments for disease and injury; however, it can involve destruction of embryos.

Carry out a debate on the use of embryonic stem cells. Compare and contrast the views on stem cell research and embryonic stem cells.

Alternatively, use Torrance Pg 11

(d) Cancer cells

Cancer cells divide _____ because they do not respond to _____ signals. This results in a _____ ____ called a tumour. Cells within the tumour may fail to attach to each other, spreading through the body where they may form _____ tumours.

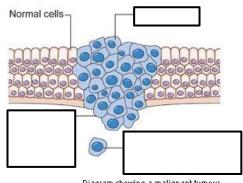


Diagram showing a malignant tumour Copyright © CancerHelp UK

UV light and cancer: How much sun is too much? (SSERC)

http://www.sserc.org.uk/images/Bulletins/228/How_much_sun_is_too_much.pdf

Your teacher will allow you to carry out a practical using UV-sensitive yeast. You will be testing....

- The effects of UV radiation on UV sensitive yeast
- The protection offered by different sunscreens

Background information:

Over the last decade the incidence of skin cancer in Scotland (specifically malignant melanoma) has increased by a staggering 30%. The increased occurrence of this type of cancer can be attributed mainly to the rise in the number of Scots taking holidays abroad each year. A significant factor, particularly amongst younger people, is the use of sun-beds to achieve that healthy glow. When UV radiation hits living cells it may damage the DNA of the cells causing mutations. However, most cells can switch on repair mechanisms to deal with the mutations induced by UV radiation. Repeated damage to the DNA increases the chance of mutations being missed by these cellular repair systems. In humans this failure to repair may result in wrinkles, damage to the immune system and skin cancer.

End of Topic Tasks:

Unit 1 Key Area 1: Division and Differentiation in Human Cells

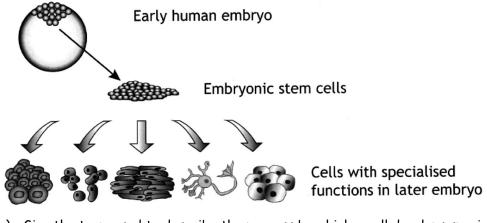
	Cell Type	Type of cell division	Chromosome number in cells produced
Α	somatic	meiosis	diploid
В	somatic	meiosis	haploid
C	germline	mitosis	haploid
D	germline	mitosis	diploid

1. Which line in the table below describes cell division in a specific cell type?

2. In a developing embryo, tissues such as muscle and nerve are produced by

- A somatic cells dividing by meiosis
- B germline cells dividing by meiosis
- C somatic cells dividing by mitosis
- D germline cells dividing by mitosis

3. The diagram below shows the role of embryonic stem cells in the development of a human embryo.

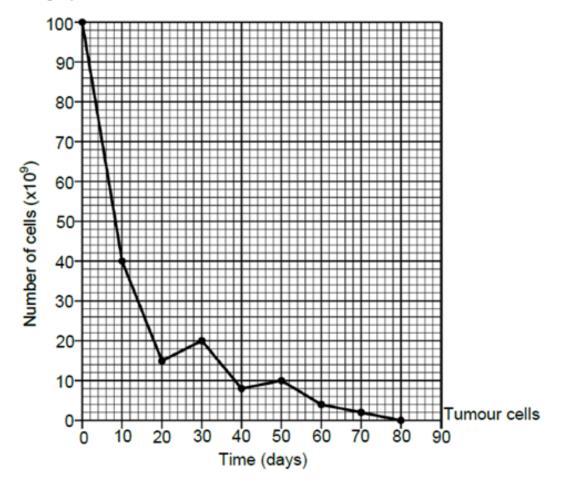


- a) Give the term used to describe the process by which a cell develops specialised functions.(1)
- b) Describe one way in which tissue (adults) stem cells differ from embryonic stem cells.
 (1)
- c) Describe how cancer cells form a tumour and explain how secondary tumours arise. (2)

4. Chemotherapy is used to kill tumour cells. When drugs are given to patients, the number of tumour and normal bone marrow cells are regularly monitored. The results for an individual who received chemotherapy are shown in the table below.

Time	Number of cells (x 10 ⁹)		
(Days)	Tumour cells	Normal bone marrow cells	
0	100	100	
10	40	60	
20	15	32	
30	20	48	
40	8	25	
50	10	38	
60	4	24	
70	2	37	
80	0	50	

(a) Complete the line graph below by plotting the results for normal bone marrow cells. The line graph for tumour cells is shown on the graph.



1

Unit 1: Key Area 1: Glossary

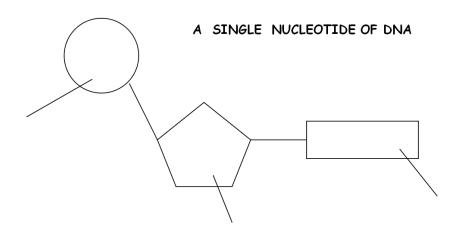
Term	Definition
Cancer	
Carcinogen	
Chromatid	
Chromosome	
Differentiation	
Diploid cells	
Embryonic stem cell	
Ethical issue	
Gametes	
Gene	
Germline cell	
Haploid cells	
Homologous	
Meiosis	
Mitosis	
Multipotent stem cells	
Mutation	
Pluripotent stem cells	
Regulatory signal	
Somatic cell	
Stem cells	
Therapeutic use	
Tissue stem cell	
Tumour	

The small print: Key Area 2 Structure and replication of DNA (a) Structure of DNA • nucleotides (deoxyribose sugar, phosphate and base),
 base pairing (adenine-thymine and guanine-cytosine) by hydrogen bonds double stranded antiparallel structure
 deoxyribose and phosphate at 3' and 5' ends of each strand respectively
 (b) Replication of DNA by DNA polymerase and primers. DNA polymerase adds DNA nucleotides, using complementary base pairing, to the deoxyribose (3') end of the new DNA strand which is forming
 (c) Polymerase chain reaction (PCR) PCR amplifies DNA using complementary primers for specific target sequences

(a) <u>Structure and Function of DNA</u>

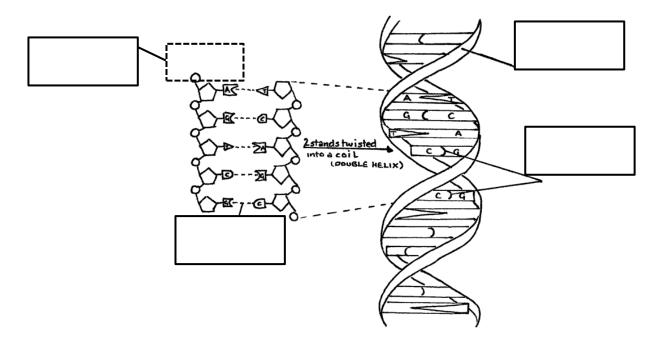
Chromosom	es are thread-like structures fo	und in the	:		of	the	cell.
They are m	ade up of tightly coiled	(deoxy	ribo	onuclei	c acid) ale	ong	with
associated	A	molecule	of	DNA	consists	of	two
strands of .	units calle	d			·		

The basic units of DNA are called nucleotides.



IMPORTANT

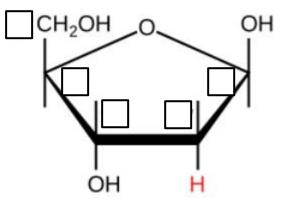
DNA double helix



The two strands stand alongside each other but run in ______ directions, i.e. they run in **<u>opposite</u>** directions. At the end of one strand, the end finishes with a ______ molecule and is named the <u>5-prime</u> (5') end. At the end of the other strand, the end finishes with a ______ sugar molecule and is named <u>3-prime</u> (3') end.

Your teacher will show you how to remember the 3' and 5' ends. Add the numbers to the deoxyribose sugar below:

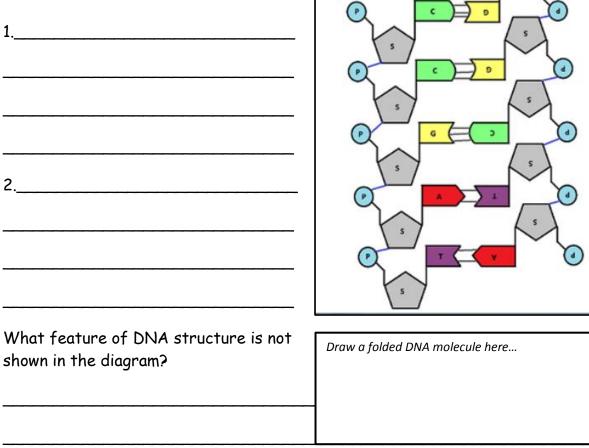
Deoxyribose sugar structure:



H-bonds

Sugar-phosphate backbone

What <u>two</u> features do you notice about the arrangement of the nucleotides in the DNA molecule shown?:





Describe the location and structure of DNA. (8 marks)

(b) **DNA replication**

DNA replication takes place prior to cell division (mitosis and meiosis)

Requirements:-

- DNA to act as a template
- Primers
- Enzymes including DNA polymerase and ligase
- Free nucleotides
- A TP for energy

Before DNA replication can occur, the length of DNA to be copied must be unwound and unzipped to form two separate single strands.

Describe what happens when the DNA is **unwound**:

Describe what happens when the DNA is **unzipped**:

Unwinding and unzipping of DNA forms two template strands. The copying of the template strands requires appropriate enzymes and primers:

<u>DNA polymerase</u> - the enzyme that carries out replication - needs a <u>primer</u> to start replication. A primer is a short strand of nucleotides which binds to the 3' end of the template DNA strand. The function of a primer is to allow DNA polymerase to add DNA nucleotides making a new DNA strand complementary to the template strand.

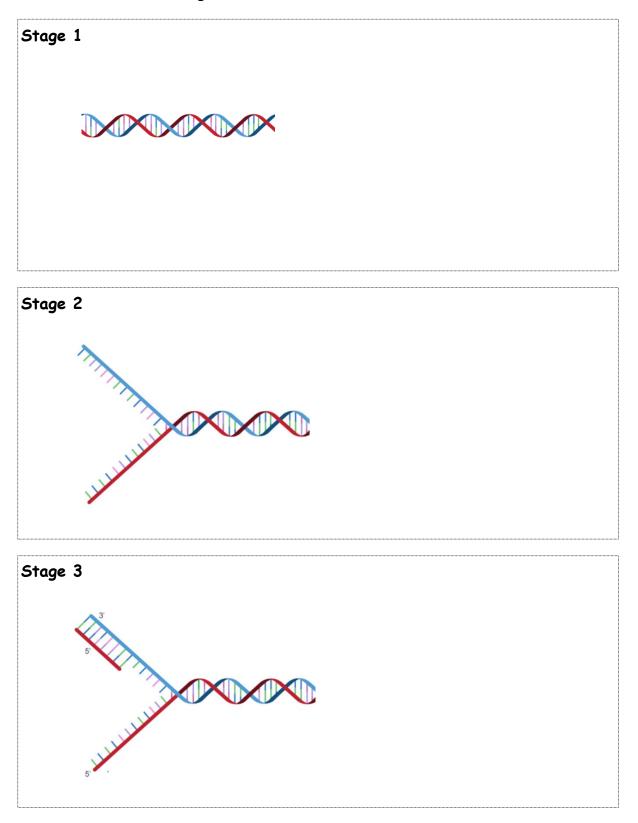
DNA polymerase adds DNA nucleotides using complementary base pairing and is only able to add to the deoxyribose (3') end of the <u>new DNA strand</u> which is forming. This results in one strand (called the leading strand) being replicated continuously and the other strand (called the lagging strand) being replicated in fragments. The fragments of DNA are joined together using the enzyme <u>ligase</u>. As the replication is completed, the two new strands, each consisting of one new strand bonded to one from the original molecule, now fall away from each other and form two separate identical DNA strands. They each <u>coil</u> into their helical shape and the process is complete.

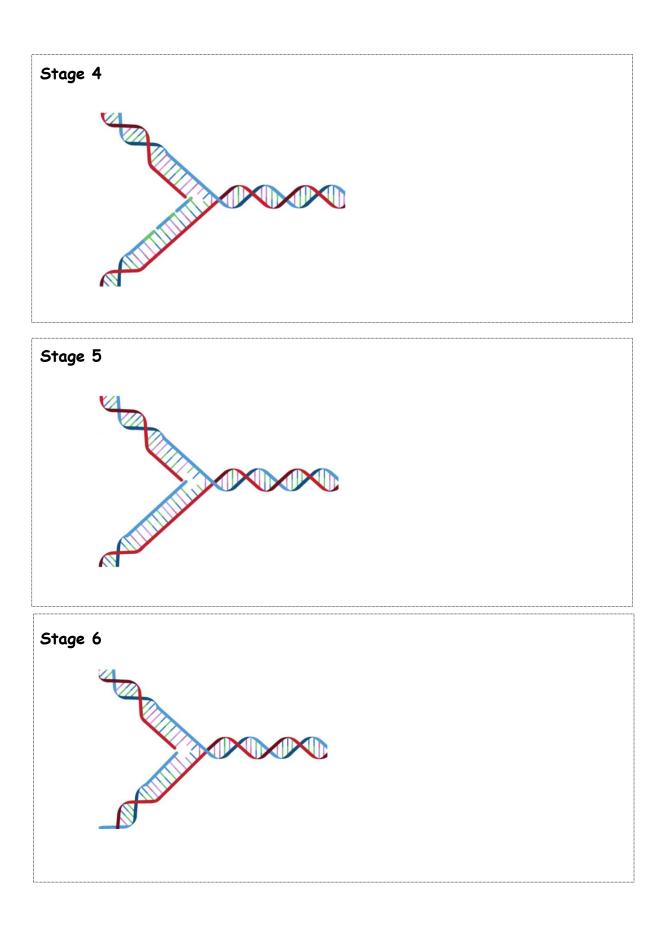
Web site: For DNA replication animation

DNA Replication

http://www.bbc.co.uk/education/guides/z36mmp3/revision/3

Your teacher will show you a step by step guide to DNA Replication. You should use this to label the diagrams below:





What are the differences between the leading strand and the lagging strand?

Leading strand:		 	
5			

Lagging strand: _____

Essay:

Give an account of the replication of DNA. (maximum of 7 marks):

(c) Polymerase Chain Reaction PCR

IMPORTANT	
In PCR, are short strands of nucleotides which are to to at the two ends of the region of DNA to be amplified.	
<u>Repeated cycles</u> of and amplify the target region of DNA.	
Web site - McGraw Hill PCR <u>http://highered.mcgraw-</u> <u>hill.com/olc/dl/120078/micro15.swf</u>	

Stages of PCR

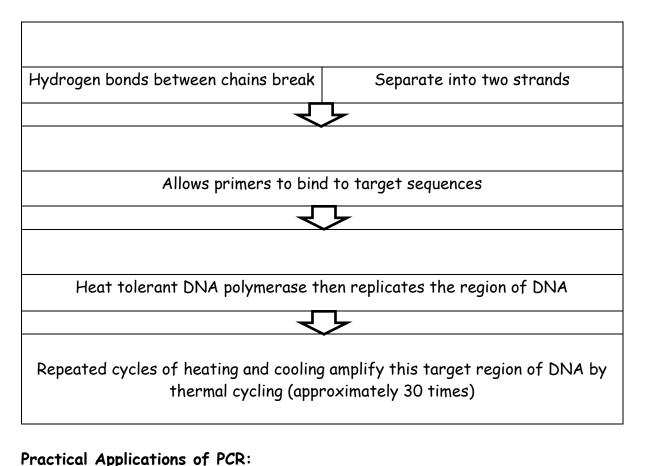
1. DNA is initially <u>heated</u> to between <u>92 and 98°C</u> break the hydrogen bonds between base pairs, separating the two strands.

2. It is then <u>cooled</u> to between <u>50 and 65°C</u> to allow primers to bind to specific target sequences on the DNA strand. Primers are short strands of nucleotides which are complementary to specific target sequences on the DNA to be amplified.

3. The temperature is <u>raised</u> to between <u>70 and 80°C</u>. Heat tolerant DNA polymerase enzymes then add free DNA nucleotides to the primers at the 3' ends of the DNA strands. This amplifies the required region of DNA, meaning we only copy the section of DNA we are interested in.

4. <u>**Repeated cycles**</u> of heating and cooling occurs to create more copies of the DNA. Each cycle doubles the amount of DNA present.

Complete the PCR process summary below, using coloured pencils to show heating and cooling stages



PCR Process Summary

PCR can amplify DNA to help solve _____, settle _____ suits and _____ genetic disorders.

Unit 1: Key Area 2: Glossary

Term	Definition
Anti-parallel	
Complementary	
Deoxyribose sugar	
DNA	
DNA amplification	
DNA polymerase	
DNA primer	
Double helix	
Heat tolerant DNA polymerase	
Hydrogen bonds	
Lagging strand	
Leading strand	
Ligase	
Nucleotide	
Paternity suits	
PCR	
Phosphate	
Replication	
Sugar-phosphate backbone	
Target sequence	
Template strand	

The small print: Key Area 3 Gene Expression

(a) Gene expression involves the transcription and translation of DNA sequences. Transcription and translation involves three types of RNA (mRNA, tRNA and rRNA). Messenger RNA (mRNA) carries a copy of the DNA code from the nucleus to the ribosome. Transfer RNA (tRNA) folds due to complementary base pairing. Each tRNA molecule carries its specific amino acid to the ribosome. Ribosomal RNA (rRNA) and proteins form the ribosome.

(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.

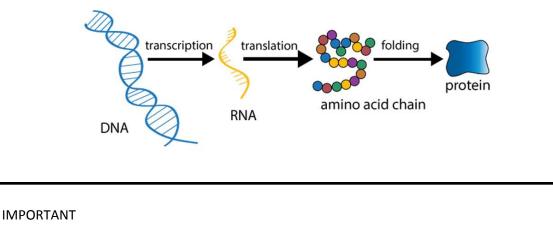
(c) tRNA is involved in the translation of mRNA into a polypeptide at a ribosome. Translation begins at a start codon and ends at a stop codon. Anticodons bond to codons by complementary base pairing, translating the genetic code into a sequence of amino acids. Peptide bonds join the amino acids together. 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 determines their functions.

Phenotype is determined by proteins produced as the result of gene expression.

Gene expression



Although a specialised cell has a complete set of the organism's genes, only those genes needed for its specialised functions are ______. All others are switched off.

Gene _______ is the activation of a gene that results in the formation of a _______. Gene expression is controlled by the regulation of both transcription and translation in protein synthesis. Only a fraction of the _______in a cell are expressed. An organism's appearance - its _______ - is determined by the proteins produced as a result of gene expression. Environmental factors can also influence phenotype.

Gene expression involves the **transcription** and **translation** of DNA sequences.

Transcription...

Translation...

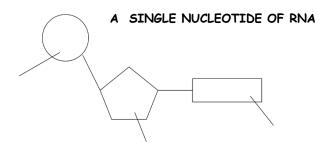
Both transcription and translation involve three types of _____:

MRNA TRNA PRNA

We will consider the role that each type of RNA performs in more detail later. First, we must look at the basic structure of RNA.

Structure of RNA

All types of RNA are also made from nucleotides.

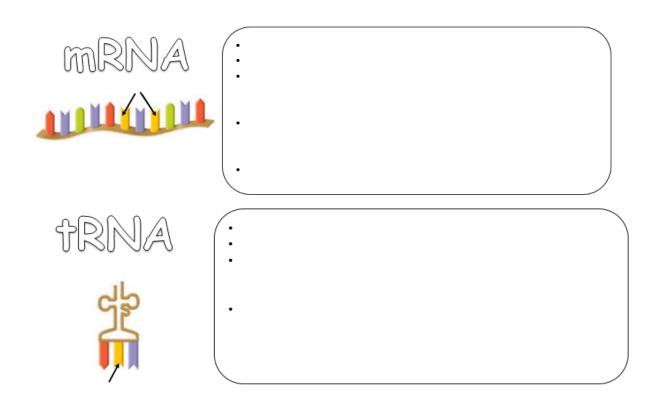


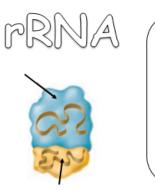
The table overleaf summarises the differences between the structure of DNA and **RNA**.

	DNA	RNA
SUGAR		
BASES PRESENT		
COMPLEMENTARY PARTNER OF ADENINE		
NUMBER OF STRANDS		

There are **<u>three</u>** different types of RNA. These are:

1.	mRNA	messenger RNA which carries the genetic code from DNA	
		in the nucleus to the	
2.	tRNA	transfer RNA folds due to complementary base pairing -	
		each tRNA carries its specific to the	
		ribosome.	
3.	rRNA	(ribosomal RNA. rRNA and proteins form the)





Transcription (of DNA into mRNA)

As DNA is too ______ to leave the nucleus through pores in the nucleus, a copy of it is made, called _____. This process is called transcription and requires:

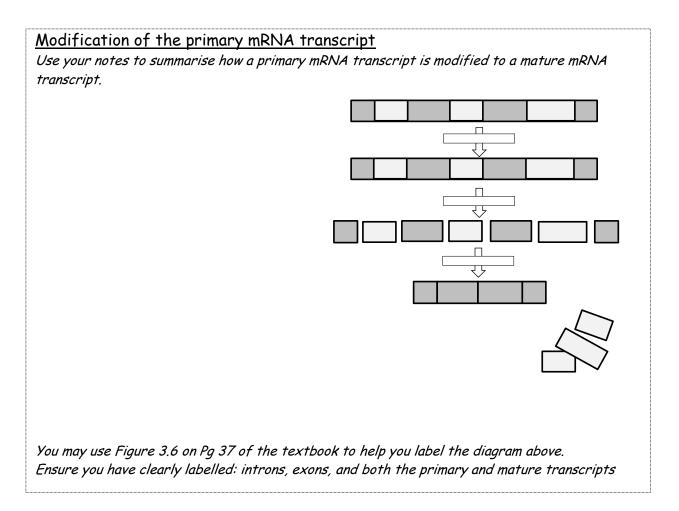
- _____ to act as template
- Free **RNA**_____
- Enzymes including RNA _____
- **ATP** for _____

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 ______ and codes for a specific amino acid. The enzyme RNA polymerase moves along DNA unwinding the ______ and breaking the ______ and breaking the ______ bonds between the bases. This separates the DNA strands to expose the required gene. RNA polymerase synthesises a **primary transcript** of mRNA from free RNA nucleotides in the nucleus. These match up along the strand by ______ base pairings. The hydrogen bonds between bases then reform and the DNA coils back up into a double helix.

What is the primary transcript?		
Work out the primary tr that in RNA <i>uracil</i> is com	anscript for the following DNA sequence. Remember nplementary to adenine:	
DNA sequence:	TAC GAA TAA CAA CCG TTG ATA CGA ACT	
Primary transcript:		

RNA Splicing

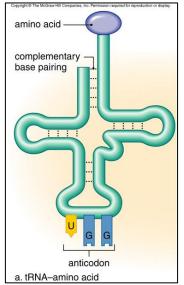
The primary transcript produced is a long straight chain of bases which contains non-coding regions in addition to coding regions that correspond to amino acids. RNA splicing is the process that removes non-coding regions of the primary transcript to form a mature transcript. Regions of the mRNA primary transcript that do not code for an amino acid are called <u>introns</u>. Regions of the mRNA primary transcript that code for amino acids are called <u>exons</u>.



The mature transcript with all of the non-coding introns removed passes out of the nucleus to the cytoplasm, ready for the next stage of protein synthesis:

Translation (of mRNA mature transcript into a polypeptide chain)

The ribosome is the site inside a cell where translation of mRNA into polypeptides occurs. tRNA transfers specific amino acids to the ribosome according to the sequence of codons on the mature mRNA transcript.



tRNA molecules required for ______ are found in the cytoplasm. They contain an attachment site to carry a specific ______ to the ribosome. As there are 20 different amino acids, _____ different tRNA molecules exist. They also have a site where a triplet of 3 bases are exposed, called an _____.

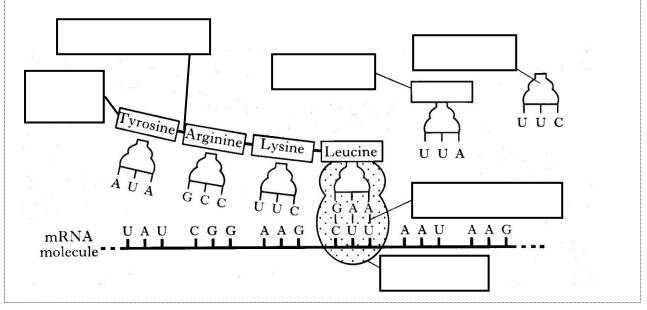
What would be the complementary codon on mRNA for the tRNA anticodon shown?

During translation, the mRNA strand passes through the ribosome which 'reads' the mRNA as it goes through. Certain mRNA codons act as '_____' codons to tell the ribosome where to start reading the strand. Translation begins at a start codon and ends at a stop codon. The ribosome identifies each mRNA codon and then matches it up with the correct tRNA ______ according to complementary base pairs (A-U, C-G). The appropriate tRNA brings its amino acid to the ribosome as it moves along the mRNA. Adjacent amino acids then join with a peptide bond to form a polypeptide. Each tRNA then leaves the ribosome as the polypeptide is formed. This process continues until a _______ 'codon' is reached which tells the ribosome where to end the polypeptide. The

is finally released.

Summary of translation:

Complete the diagram to show 'hydrogen bond', 'peptide bond', tRNA, 'amino acid', ribosome, 'growing polypeptide', 'asparagine*' Use highlighters to show the codons and anticodons.



One gene, many proteins

From one gene, many different proteins may be expressed. This is due to _____.

Different mature transcripts of mRNA may produced from the same primary transcript of mRNA **depending on which exons are retained**. An example of this in the human body is in antibody production. One gene may code for two slightly different antibody structures. Products of alternative RNA splicing: Antibodies

Polypeptides to Proteins

During translation, amino acids are joined together in sequence, linked by peptide bonds to form ________. Proteins are long polypeptide chains, folded to form a specific three dimensional shape. These polypeptide chains are held in a three dimensional shape by hydrogen bonds and other interactions between individual amino acids. Such interactions between amino acids results in proteins having specific shapes:

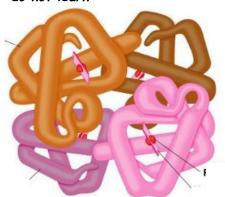
IMPORTANT



<u>Phenotype</u> is determined by proteins produced as a result of gene expression. Environmental factors also influence phenotype.

An example of a protein in the human body is haemoglobin which transports oxygen in the bloodstream. The diagram below shows how the polypeptides in haemoglobin are arranged. The interactions between amino acids result in the haemoglobin protein having a structure which is perfect for the function of transporting oxygen.

Haemoglobin molecule do not learn



Extra challenge: What determines the shape of haemoglobin? What do you think might happen if someone had an error in the gene that codes for haemoglobin?

End of Key Area 3 Activities

ESSAY

2014: Describe protein synthesis under the following headings:

- (i) Transcription of DNA (4)
- (ii) Translation of mRNA (6)
- Test your knowledge: textbook page 37 qn's 1-4 and page 43 qn's 1-4 (not 4 e,f)
- Summary completion note
- Produce a summary poster of gene expression (textbook page 34 has a good overview)
- Use textbook page 39 to make a codon sequence for your partner to translate.
- Do a card sort to make a glossary

Unit 1: Key Area 3: Glossary

Term	Definition
Alternative RNA splicing	
-	
Exons	
Gene expression	
Introns	
Mature mRNA transcript	
mRNA	
Peptide bonds	
Polypeptide	
Тотурерное	
Primary mRNA transcript	
Ribosome	
RNA splicing	
RNA polymerase	
rRNA	
Transcription	
Translation	
†RNA	

The smo	all print: Key Area 4
Mutatio	
	tations are changes in the DNA Mutations are changes in the DNA
•	Mutations can result in no protein or an altered protein being synthesised
(b) Sin •	gle gene mutations Single gene mutations involve the alteration of a DNA nucleotide sequence as a result of the substitution, insertion or deletion of nucleotides
•	Nucleotide substitution mutations include missense, non-sense 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 \Box
•	Nonsense mutations result in a premature stop codon being produced which results in a shorter protein \Box
•	Splice-site mutations result in some introns being retained and/or some exons not being included in the mature transcript
•	Nucleotide insertions or deletions result in frame-shift mutations \Box
•	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
(c) Chr •	omosome structure mutations Duplication is where a section of a chromosome is added from its homologous partner.
•	Deletion is where a section of a chromosome is removed
•	Inversion is where a section of a chromosome is reversed
•	Translocation is where a section of chromosome is added to a chromosome, not its homologous partner
•	The substantial changes in chromosome mutations make them lethal \Box

Key Area 4: Mutation

Mutations are changes to the DNA that can result in no protein or an altered protein being synthesised. In this topic we will look at different types of mutation. The two categories of mutation are:

Single gene mutations:	
Single gene mututions.	

Chromosome structure mutations: _____

Single gene mutations

Mutations involving only one gene are called ______ gene mutations. There are different types of single gene mutations:

- <u>nucleotide substitution</u>: resulting in missense, nonsense and splice-site mutations
 <u>nucleotide insertion</u>
- L. <u>nucleoride insertion</u>

Resulting in frame-shift mutation

3. <u>nucleotide deletion</u>

1. Substitution mutations

A single DNA nucleotide in a gene is either substituted, inserted or deleted. This changes the nucleotide sequence in an mRNA codon and can result in alterations in one or more amino acids.

Different types of substitution mutations occur, depending on where the substitution occurs:

a) Missense - a nucleotide in a codon is substituted, causing a different amino acid to be produced. This may lead to the final protein not functioning e.g.

b) Nonsense –a nucleotide in a codon is substituted, changing it from coding for an amino acid coding for a stop codon. This causes a shorter (usually non functioning) protein e.g. _____.

c) Splice-site - the nucleotide at which mRNA splicing occurs is substituted, causing the location of the splice site to change. This leaves non coding regions (introns) left in and coding regions (exons) removed from the mature mRNA, thus producing non functioning proteins e.g.

2. Insertion mutations

A nucleotide is added to the DNA.

Nucleotide insertions cause a ______. This is where adding a nucleotide causes that mRNA codon and ______ of the codons that follow on the mRNA to change and thus produce the wrong amino acids. This leads to a very different and generally ______ protein. E.g. Tay-Sachs syndrome

3. Deletion mutations

A nucleotide is ______ from the DNA. Nucleotide deletions also cause ______ mutations. E.g. cystic fibrosis

Single gene mutations that involve an insertion or a deletion of a nucleotide are called _________ mutations. After the point of mutation, every subsequent codon will be altered along the length of the gene. This results in every amino acid after that point being altered and ultimately leads to expression of a non-functional protein.

Case studies: Group activity

There are six diseases described in the textbook that are caused by mutation single gene mutation (pages 55-59). Within your group, you should make sure you have information on all six of the following diseases:

Sickle-cell disease, Phenylketonuria (PKU), Duchenne Muscular Dystrophy (DMD), Beta thalassemia, Tay-Sachs disease, Cystic Fibrosis

You should include:

- Name of condition
- Type(s) of mutation
- Symptoms of disease/other information

Use what you have learned in your group to complete the summary table below:

Condition	Type(s) of mutation	Symptoms /information
Sickle-cell disease		
Phenylketonuria (PKU)		
(FKO)		
Beta (β) thalassaemia		
Duchenne muscular dystrophy (DMD)		
Tay-Sachs syndrome		
Cystic fibrosis		

ESSAY

Describe the process of mutations in terms of:

- i) single gene mutations
- ii) effect on protein production

(10)

Chromosome Structure Mutations

There are different types of chromosome mutations. The substantial changes which occur often make them ______.

1. *Duplication* - a section of a chromosome is <u>added</u> from its homologous partner

Eg. _____

2. Deletion - a section of a chromosome is removed.

E.g. _____

3. *Inversion* - a section of chromosome is <u>reversed</u>.

E.g. _____

4. *Translocation* - a section of a chromosome is <u>added</u> to another chromosome that isn't its homologous partner.

E.g. _____

End of Key Area 4 Activities:

Case studies: Chromosome structure mutations (Cri-du-chat syndrome, Chronic Myeloid Leukaemia OR Familial Down's syndrome)

Test your knowledge: Page 62 qn's 1-4

Term	Definition
Chromosome structure mutation	
Chromosome deletion	
Chromosome duplication	
Chromosome inversion	
Chromosome translocation	
Frame-shift mutation	
Homologous partner	
Mutation	
Nucleotide deletion	
Nucleotide insertion	
Nucleotide substitution - missense	
Nucleotide substitution - nonsense	
Nucleotide substitution - splice-site	
Single gene mutation	

Unit 1: Key Area 4: Glossary

The small print: Key Area 5 Human Genomics		
 (a) The genome of an organism is its entire hereditary information encoded in DNA		
 (b) An individual's genome can be analysed to predict the likelihood of developing certain diseases		

Human Genomics

A genome is an organisms entire set of DNA. Genomics is the study of _____.



_____ is the use of computer technology to map genomes and identify DNA sequences.

Studying an individual's	_ may enable doctors to more accurately
a disease and then pre	escribe the correct, at
the correct dosage (level) and at the co	rrect It may reduce
the possibility of the patient suffering ac	lverse reactions. It may further help to
identify an individual's susceptibility to	diseases and enable
steps to be taken to avoid or reduce t	he of developing that
disease.	

ESSAY

Describe the benefits of genomics to the human population. (10)

End of Key Area 5 Activities

Case study:	Human Genome Project OR Alzheimer's disease - assessing the risk using bioinformatics
Test your knowledge:	Page 70 Qn's 1, 3, 4a, 4d only

Unit 1: Key Area 5: Glossary

Term	Definition
Bioinformatics	
Genome	
Genomic sequencing	
Hereditary information	
Personalised medicine	
Pharmacogenetics	

The small print: Key Area 6
Metabolic pathways
 (a) Metabolic pathways are integrated and controlled pathways of enzyme-catalysed reactions within a cell
 (b) Metabolic pathways are controlled by the presence or absence of particular enzymes and the regulation of the rate of key enzymes
 Competitive inhibition can be reversed by increasing substrate concentration

Cell Metabolism

Metabolism describes all enzyme-catalysed reactions which occur within a cell. Metabolic pathways involve:



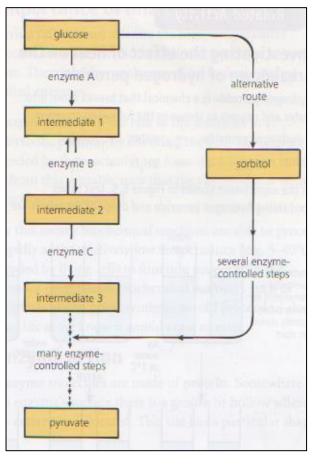
Some metabolic pathways can be reversible, others irreversible. Some metabolic pathways may have alternative routes:

- pyruvate to lactate in muscle cells is reversible
- pyruvate to ethanol and CO2 in yeast cells is irreversible.
- Some pathways have more than one route. An example of an **alternative route** is shown:

The irreversible parts of a pathway are shown with arrows pointing in only one direction. Reversible parts of the pathway are shown with arrows pointing in both directions. Use highlighters to show the reversible steps, irreversible steps and alternative routes.

Control of metabolic pathways

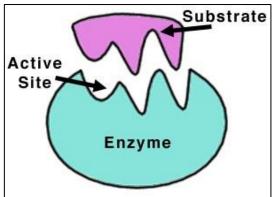
Metabolic pathways can be controlled by the presence or absence of particular enzymes. For example, if an enzyme is absent, the reaction it catalyses will occur much more ______ or not at all. If the appropriate enzyme is present, the reaction will occur much more ______. The regulation of the rate of reaction of key enzymes will have an effect on the metabolic pathway.



What factors might regulate (increase/decrease) the rate of a reaction?

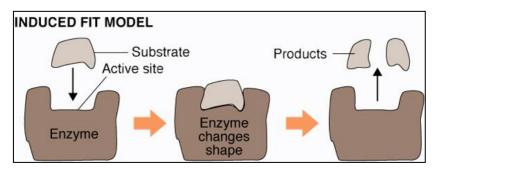
Enzyme action

Made of _____, enzymes possess a region called the active site where the reaction occurs. It has a shape that is to the shape of its substrate.



Induced Fit

The enzyme's active site changes shape to better fit the substrate after the substrate binds to it. Enzymes are not directly involved in the reaction, therefore at the end. they remain _



Activation Energy

The energy required to without enzyme break chemical bonds in the reacting chemicals and to start the reaction is called enzyme rith Energy the reactants energy. e.g. CO₂ + H₂O Enzymes the activation energy required. Reaction coordinate



activation energy without

overall energy released during

enzyme

reaction

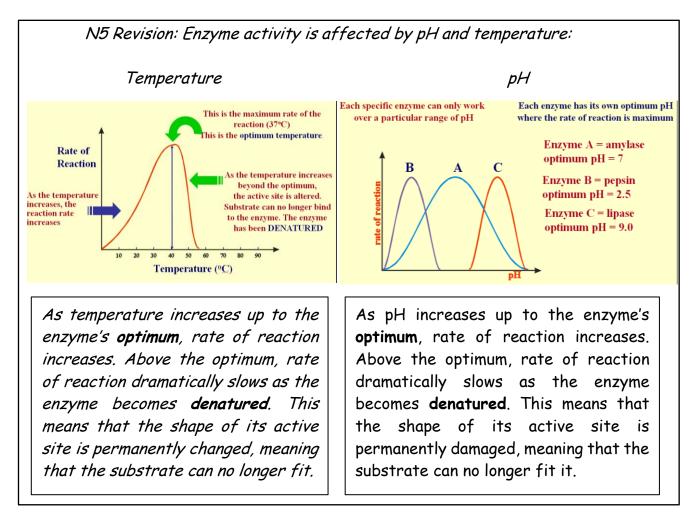
products

H₂CO₃

activation energy with

enzyme

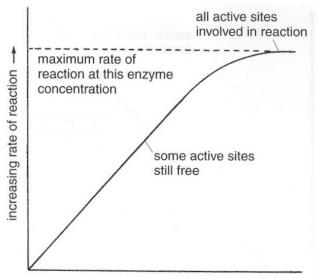




In addition to temperature and pH, enzyme activity is also affected by the concentrations of the ______ and the _____.

Substrate concentration

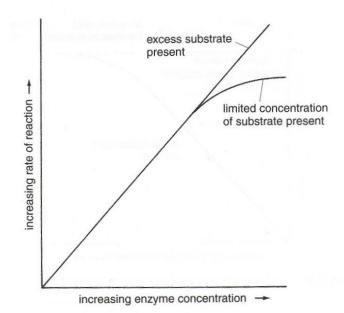
Increasing substrate concentration _______ rate of reaction as there as more active sites become occupied by _______. This is only until the point where all _______ are filled and so rate of reaction levels off. As there are no more enzymes to react with more substrates, *enzyme concentration* becomes the limiting factor.



increasing substrate concentration ----

Enzyme concentration

Increasing **enzyme concentration** increases rate of reaction as there are more _______ to join with substrates. This is only until the point where all ______ are used up and so rate of reaction levels off. As there are no more substrates to react with enzymes, substrate concentration becomes the limiting factor.

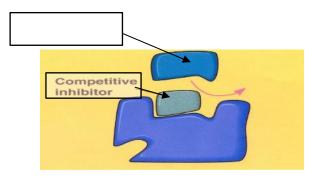


Regulation (control) of metabolic pathways

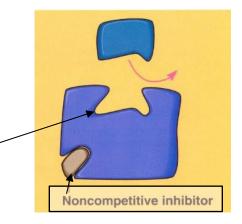
An enzyme inhibitor is a molecule that can affect the rate of a reaction.

Control of metabolic pathways can be achieved in three different ways:

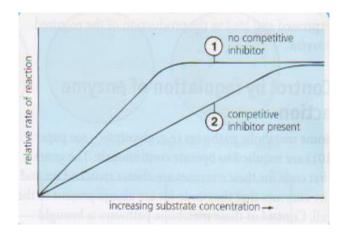
- competitive inhibition
- non-competitive inhibition
- feedback inhibition
- <u>Competitive inhibitors</u> bind to the active site and prevent the ______ from binding.



<u>Non-competitive inhibitors</u> bind to a point on the enzyme other than the ______. They alter the shape of the active site so that the substrate can no longer fit in.



Graphing Competitive and Non-competitive Inhibitors

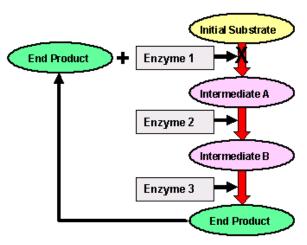


When a competitive inhibitor is present the rate of the reaction is ______ compared to a reaction when no inhibitor is present.

You can tell from a graph if an inhibitor is a competitive or non-competitive inhibitor. With a competitive inhibitor, when the substrate concentration is increased, the rate of the reaction ______. This does not happen with a non-competitive inhibitor.

e.g. _____.

3. Feedback inhibition



ESSAY

Give an account of the factors affecting enzyme activity. (10)

End of Key Area 6 Activities

Experiment write-up: Substrate Concentration:
Look at Figure 6.13 on Pg 84. Suggest a suitable aim for this experiment and write a conclusion that addresses this aim.
Feedback inhibition:
Look at Figure 6.32 on Pg 95. Suggest a suitable aim for this experiment and write a conclusion that addresses this aim.

Test your knowledge: Answer qn's 1-4 on Page 85 & qn's 1-4 on Page 96

Term	Definition
Activation energy	
Active site	
Affinity	
Alternative route	
Anabolic	
Catabolic	
Competitive inhibition	
Enzyme	
Feedback inhibition	
Induced fit	
Inhibitor	
Irreversible step	
Metabolic pathway	
Non-competitive inhibition	
Products	
Reversible step	
Substrate	

Unit 1: Key Area 6: Glossary

The small print, Kay Area 7	
The small print: Key Area 7	
Cellular Respiration	
 (a) Metabolic pathways of cellular respiration Glycolysis is the breakdown of glucose to pyruvate in the cytoplasm	
 (b) ATP synthesis Electrons are passed along the electron transport chain releasing energy The electron transport chain is a series of carrier proteins attached to the inner mitochondrial membrane Energy released in electron transport chain allows hydrogen ions to be pumped across the inner mitochondrial membrane Flow of hydrogen ions back through the membrane protein ATP synthase results in production of ATP 	🗌 🗍 n the
Hydrogen ions and electrons combine with oxygen to form water	

(c) The role of ATP in the transfer of energy

• ATP is used to transfer energy to cellular processes which require energy.....

Cellular Respiration

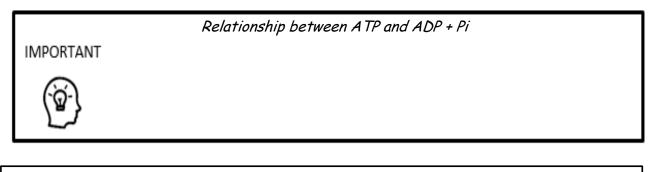
Respiration involves a series of enzyme controlled metabolic steps which release the chemical energy in food for activities such as ______ transport, _____ replication and protein synthesis. Cells use energy in the form of a chemical called adenosine triphosphate (_____) which is produced during respiration.

ATP Generation

ATP is formed from a molecule of adenosine diphosphate (ADP) joined with another inorganic phosphate (Pi). The addition of phosphate to a molecule is called ______. Energy is **required** to generate ATP.

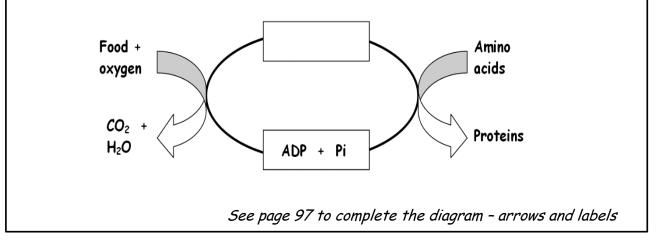
<u>ATP Breakdown</u>

When the last phosphate is broken off the ATP, it forms ADP + Pi and chemical energy is **released** for cell processes. Levels of ATP in the human body remain fairly constant as it is produced at the same rate that it is used up.



Respiration key message

Food contains stored chemical energy. This is released through a series of metabolic pathways and regenerates the high energy compound ATP. ATP can then be broken down to release energy when it is needed for cellular processes.



Respiration consists of 3 stages:

- GLYCOLYSIS
- CITRIC ACID CYCLE
- ELECTRON TRANSPORT CHAIN

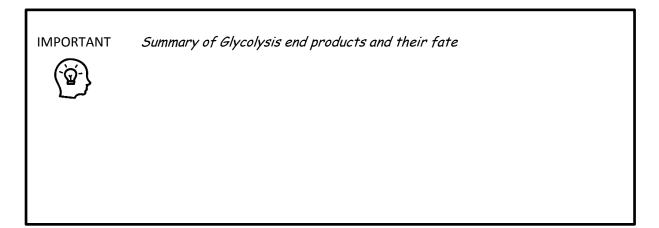
1. Glycolysis

During glycolysis, **glucose** (a 6 carbon compound), is broken down into 2 molecules of **pyruvate** (a 3 carbon compound) in the **cytoplasm**.

Glycolysis Energy investment phase For glucose (and other intermediates) to be 2 ATP broken down it must first be phosphorylation 2 ADP + 2Pi phosphorylated (phosphate is added glucose). The phosphate is intermediate provided by ATP. As two ATP are required for this, it is 4 ADP + 4Pi called the energy investment stage. 4 ATP Pay-off phase of Glycolysis Glycolysis also produces an additional four ATP - this is called the pay-off phase. Therefore a **net gain of 2 ATP** occurs in the reaction.

Dehydrogenase enzymes remove hydrogen ions and electrons and pass them to the **coenzyme NAD** forming **NADH**. NADH is passed onto the electron transport chain on the inner mitochondrial matrix.

As oxygen is not required, glycolysis can occur with oxygen (called **aerobic** respiration) or without oxygen (called fermentation).



2. Citric acid cycle

The Citric acid cycle takes place in the matrix of the mitochondria.

If aerobic conditions, (oxygen is available), the pyruvate from glycolysis is broken down into an acetyl group which combines with co-enzyme A to form acetyl coenzyme A. The carbon lost forms carbon dioxide.

In the citric acid cycle, acetyl coenzyme A combines with **oxaloacetate** to form **citrate**.

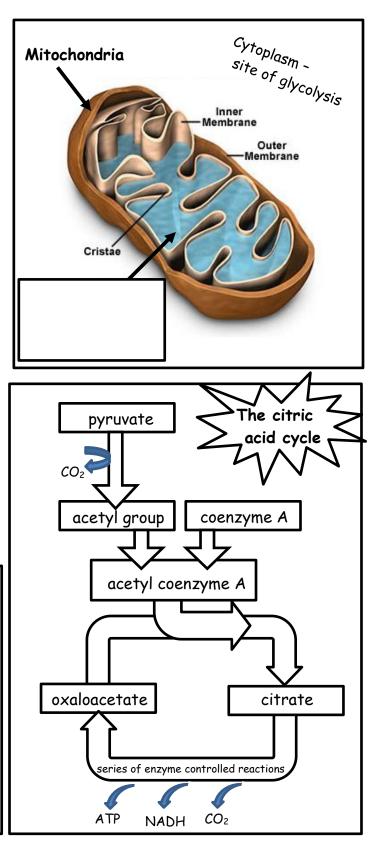
During a series of **enzyme controlled steps**, citrate is gradually converted back into oxaloacetate which results in the generation of **ATP** and release of **carbon dioxide**.

Dehydrogenase enzymes remove **hydrogen ions** and **electrons** and pass them to the **coenzyme NAD**, forming **NADH**. NADH is passed onto the electron transport chain on the inner mitochondrial matrix.

IMPORTANT



Summary of citric acid cycle end products and their fate

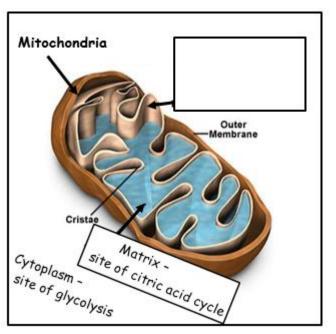


3. ATP Synthesis: Electron transport chain

The electron transfer chain occurs on the inner membrane of the mitochondria.

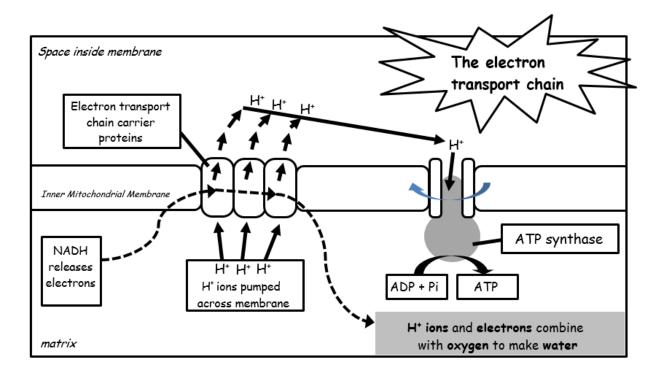
The electron transport chain is a **series of carrier proteins** attached to the inner mitochondrial membrane. The electrons and hydrogen ions from the NADH produced in Glycolysis and the Citric Acid Cycle are used to generate ATP in the electron transport chain. When electrons are passed along the carrier proteins of the electron transport chain, energy is released.

This energy allows **hydrogen ions** to be **pumped** across the inner mitochondrial membrane. The flow of



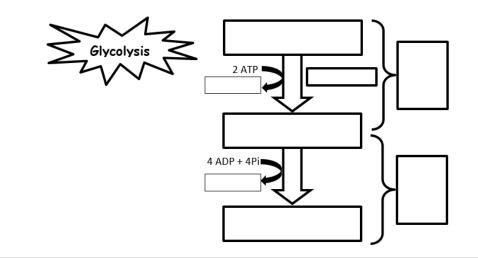
these ions back through the membrane protein **ATP** synthase produces ATP.

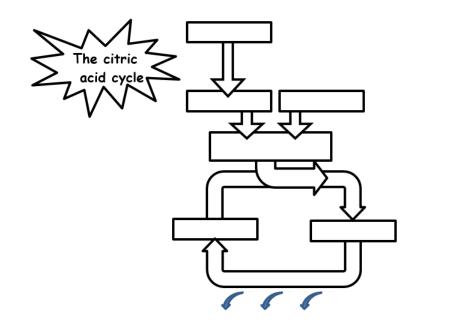
FINALLY, Hydrogen ions (H⁺), electrons and oxygen combine to form water.

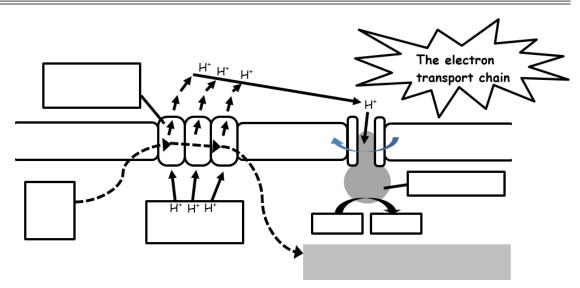


The ATP generated in cellular respiration is used to transfer energy to cellular processes that require energy.

You should now bring together all three stages of cellular respiration:







End of Key Area 7 Activities

ESSAY

2014: Describe aerobic respiration under the following headings

(i) The citric acid cycle (5)

(ii) The electron transport chain (5)

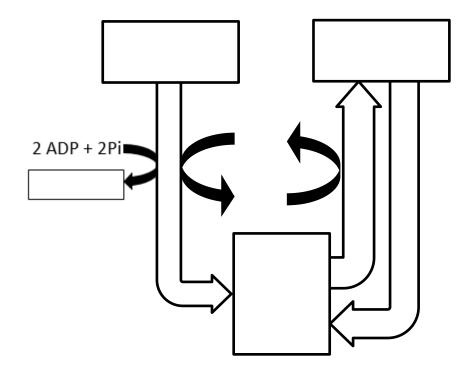
Term	Definition
Acetyl group	
Acetyl coenzyme A	
Aerobic conditions	
ATP synthase	
Citrate	
Citric acid cycle	
Coenzyme A	
Coenzyme NAD	
Dehydrogenase	
Electron transport chain	
Energy investment	
Energy pay-off	
Glycolysis	
Matrix of mitochondria	
Mitochondria	
Oxaloacetate	
Phosphorylation	
Pyruvate	

Unit 1: Key Area 7: Glossary

The small print: Key Area 8
Energy systems in muscle cells
 (a) Lactate metabolism During vigorous exercise, the muscle cells do not get sufficient oxygen to support the electron transport chain
 (b) Types of skeletal muscle fibres There are two types of muscle fibre: slow twitch and fast twitch
 running, cycling or cross-country skiing. Fast twitch muscle fibres contract relatively quickly, over short periods. Fast twitch muscle fibres are useful for activities such as sprinting or weightlifting. Fast twitch muscle fibres can generate ATP through glycolysis only and have fewer mitochondria and a lower blood supply compared to slow twitch muscle fibres. The major storage fuel of fast twitch muscle fibres is glycogen. Most human muscle tissue contains a mixture of both slow and fast twitch muscle fibres.
Athletes show distinct patterns of muscle fibres that reflect their sporting activities

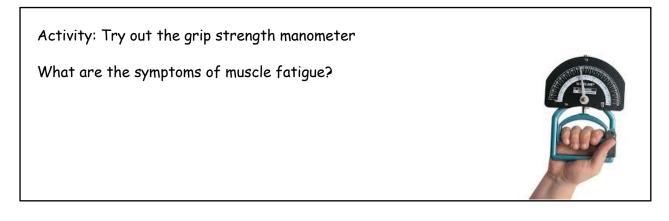
Lactate Metabolism

During ______ exercise, the muscle cells do not get sufficient oxygen to support the electron transport chain. Under these conditions, pyruvate is converted to lactate.



This conversion involves the transfer of hydrogen from the NADH produced during glycolysis to pyruvate in order to produce lactate. This regenerates the NAD needed to maintain ATP production through glycolysis.

Lactate accumulates in the muscle tissues and muscle fatigue occurs.



When exercise is complete, the oxygen debt is repaid. This allows respiration to provide the energy to convert lactate back to pyruvate and glucose in the liver.



Types of skeletal muscle fibres

There are two different type of skeletal muscle fibres:

- 1. **slow** twitch (type 1)
- 2. fast twitch (type 2)

Most human muscle contains a mixture of the two different types of fibres. Athletes show disctinct patterns of muscle fibres that reflect their sporting activities.

1. Slow twitch (Type 1) muscle fibres co	ntract more, but can
sustain contractions for	and so are good for
activities. Endurance activities include la	ong distance running, cycling or cross
country skiing. Slow twitch fibres rely	on respiration to
generate ATP and have many	, a large blood supply and a
high concentration of the oxygen storin	g protein The
major storage fuel of slow twitch muscles	fibres is

2. *Fast twitch* (Type 2) muscle fibres contract more ______, over short periods, so are good for ______ bursts of activity. They are useful for activities such as sprinting or weightlifting. Fast twitch fibres can generate ATP through ______ only and have few ______ and a lower ______ supply than slow twitch fibres. The major storage fuel of fast twitch muscles fibres is ______.

Feature	Type of skeletal fibre	
	Slow twitch	Fast twitch
Speed of contraction		
Length of contraction		
Speed at which fibres become		
fatigued		
Respiratory pathway(s)		
normally used to generate ATP		
Number of mitochondria		
Density of blood capillaries		
Concentration of myoglobin		
Major storage fuel used	Fats	Glycogen

End of Key Area 8 Activities

ESSAY

2013: Give an account of skeletal muscle cells under the following headings:

(i) Lactic acid metabolism (4)

(ii) Slow twitch muscle fibres (3)

(iii) Fast twitch muscle fibres (3)

Term	Definition

Unit 1: Key Area 8: Glossary