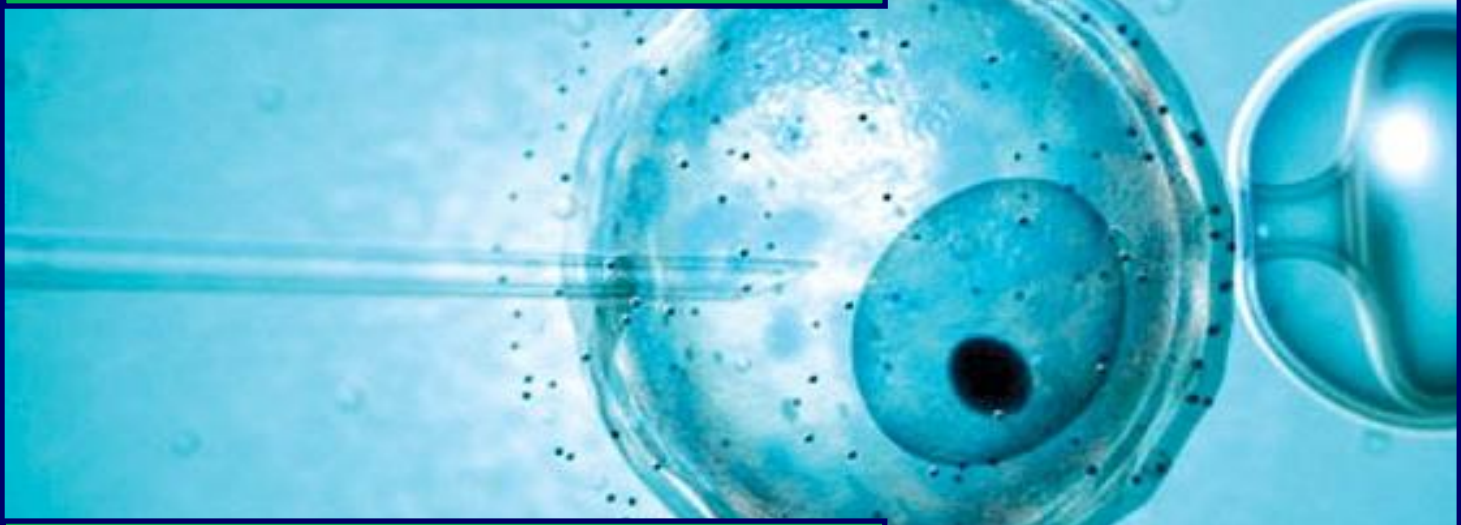
The **Scholars** Programme

Do I Look Good In These Genes?

Autumn 2015

Tutor Name: Dr. R Hoque



Pupil Name:

The Scholars Programme – Autumn 2015 – Pupil Feedback Report

Grade	Marks	What this means
1st	70+	Performing to an excellent standard at first year undergraduate level
2:1	60-69	Performing to a good standard at first year undergraduate level
2:2	50-59	Performing to an excellent standard at GCSE level
3rd	40-49	Performing to a good standard at GCSE level
Workings towards a pass	0-39	Workings towards a pass
Did not submit	DNS	No assignment received by The Brilliant Club

Lateness	
Any lateness	10 marks deducted
Plagiarism	
Some plagiarism	10 marks deducted
Moderate plagiarism	20 marks deducted
Extreme plagiarism	Automatic fail

Name of PhD Tutor			
Title of Assignment			
Name of Pupil			
Name of School			
ORIGINAL MARK / 100		**FINAL MARK / 100**	
DEDUCTED MARKS		**FINAL GRADE**	

If marks have been deducted (e.g. late submission, plagiarism) the PhD tutor should give an explanation in this section:

Learning Feedback Comment 1 - ***Enter Key Learning Priority Here***	
What you did in relation to this Key Learning Priority	**How you could improve in the future**
Learning Feedback Comment 2 – ***Enter Key Learning Priority Here***	
What you did in relation to this Key Learning Priority	**How you could improve in the future**
Learning Feedback Comment 3 – ***Enter Key Learning Priority Here***	
What you did in relation to this Key Learning Priority	**How you could improve in the future**
Resilience Comment	
How you showed learning resilience during the course	**How you could build learning resilience in the future**

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Course Rationale

The function of DNA in the living world is at both confoundingly complex and yet surprisingly simple. The genetic code carried in molecules of DNA is responsible for a stunning variety of life forms. Inside every living cell, DNA directs vital activities, such as growth, division and even death. It does this by providing the instructions that cells use to build proteins.

DNA gives rise to the physical and behavioural adaptations that make each organism unique. And yet, the chemical language in which DNA's instructions are written is stunningly simple. It consists of just four letters, called nucleotides, from which DNA is built: adenine (A), thymine (T), cytosine (C), and guanine (G). Specific sequences of these four molecules function as genes by providing the instructions required to build proteins.

In the duration of this course, students will learn what happens when this process goes wrong and how errors give rise to genetic variation. They will explore how genetic mutations are beneficial for human survival and how it can form the basis of disease such as cystic fibrosis and sickle cell anaemia.

Scientific research on finding cures to genetic diseases often centres on genetic testing. Students will learn the different types of testing available and discuss the ethical issues surrounding genetic tests. The final assessment will focus on students producing an essay to argue their view on a controversial case study.

By the end of this course, students will have learnt how to present information in a university style setting, formulate scientific opinions and enhance their analytical thinking.

Mark Scheme

Key Skill	1st	2.2
Language and Communication (Use of appropriate specialist vocabulary and quality of written communication)	☐ New and technical vocabulary is correctly and appropriately used. ☐ Writing is well structured and of an appropriate style for an academic document. ☐ Writing is clear, concise, confident and in an engaging style. ☐ Spelling, grammar and punctuation are almost perfect (<5). ☐ Opinions and arguments are articulately made.	☐ Some technical vocabulary is used correctly but some minor mistakes. ☐ Writing is poorly structured at times, and the style used is not always appropriate for an academic document. ☐ Writing lacks clarity and not always concise. ☐ Spelling, grammar and punctuation are weak in places. ☐ Arguments are weak in places.
Research Skills (Use of sources and acknowledgement of this by Vancouver style referencing)	☐ A wide range of relevant and reliable sources has been used. ☐ Sources are referenced correctly and in the format explained in the course handbook. ☐ Logical interpretations are made. ☐ Figures are relevant, used appropriately and labelled correctly.	☐ A limited range of sources used; not all sources are reliable. ☐ Sources are not always referenced correctly. ☐ Some effort has been made to give logical interpretations or conclusions but these may lack confidence or rigour. ☐ Figures may be irrelevant, inappropriately used, or not labelled correctly.
Application (Recognise, recall and show understanding of content knowledge)	☐ Uses all sources of evidence to support a strong conclusion. ☐ Good, clear and effective presentation of data. ☐ High levels of insight and creativity with a clear understanding. ☐ The course concepts and explanations are coherently applied to the situations specified. ☐ Arguments are well-balanced, aptly linking theory to case study.	☐ Uses some sources of data/evidence to support a conclusion. ☐ Some effort made to effectively present data. ☐ Some evidence of creative qualities. The topic is explored in quite a superficial manner. ☐ Attempt made to link theory learnt on course to case study but some steps in the reasoning may be missing and/or irrelevant points have been included.
Critical analysis (Validity, reliability and credibility of scientific information and of their conclusions. Suggested improvements including ideas for future work)	☐ Shows a good understanding of what a hypothesis is and how data presented supports or refutes it. ☐ Stages of method covered in tutorials are correctly written and easy to follow. ☐ Has considered the strengths and limitations of method/data where appropriate. ☐ Appropriate suggestions are made for improvement of the experimental design to increase the reliability of the results. ☐ The argument towards the conclusion is well developed and logical. ☐ Simple calculations are consistently correct and well presented	☐ Some understanding of what a hypothesis is but this might be confused with a theory. ☐ Stages of method covered in tutorials are correctly used but there may be difficulty in following due to it being too detailed/simplified. ☐ No or little appreciable recognition of limitations of methods. ☐ The suggestions for experimental improvements can be unrealistic and would not have the desired effect. ☐ The argument towards the conclusion is unsupported. ☐ Simple calculations are attempted.

Glossary of Keywords

Term	Definition / Meaning
Amino acid	Organic compounds that combine to form proteins; building block of proteins
DNA (deoxyribonucleic acid)	Ladder-like molecule that stores genetic information, including information needed to build proteins
Nucleotide	The basic unit of DNA consisting of a base (A, C, T, or G), a sugar, and a phosphate
Polypeptide	A chain of amino acids joined together by peptide bonds; proteins are made up of one or more polypeptides
Protein	Long-chained molecules used to build and repair cells; made of one or more chains of amino acids
Ribosomes	Protein-making sites within cells
RNA (ribonucleic acid)	Substance that reads the genetic information carried by DNA; the same as DNA except that it is made up of only one strand, contains the base uracil (U) instead of thymine (T), and contains the sugar ribose instead of deoxyribose
Genetic Testing	Analysis of a person's DNA. Usually done to determine whether an individual carries mutations of genes that make them more susceptible to a disease
Clinical Validity	How accurately a test predicts whether a person will get a particular disease or symptom (known as the "clinical outcome").
Effective Treatment Availability	Whether there are treatment options available for a particular disease or condition
Penetrance	The probability that individuals with a specific genotype will express a specific phenotype. For example, 85% of women with particular BRCA1 mutations will develop breast or ovarian cancer.
Polygenic	A polygenic trait is one that involves multiple genes. For example, cat coat colour is known to involve multiple genes, including ones for white, orange, and black colouring. Many common diseases, like heart disease and diabetes, are thought to be polygenic.

Glossary of Keywords or Notes

Term	Definition / Meaning

Tutorial 1 – DNA

What are the objectives of Tutorial 1?

- Introduce tutor and provide VLE details
- Be able to describe DNA structure
- Understand how DNA replicates

Key words

Genome
Base pairs
Complementary
Double helix

Activity 1: What is DNA?

Use the space below to write down five things you know about DNA.

1.

2.

3.

4.

5.

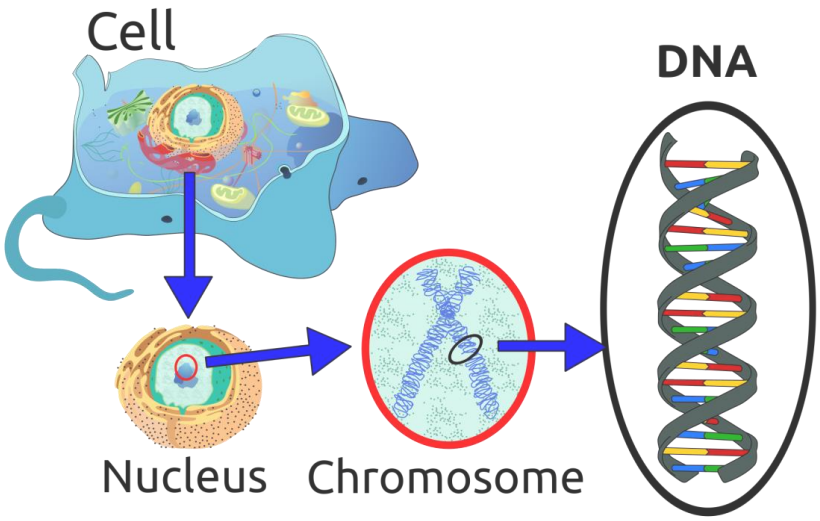


Figure 1. Where is DNA found

DNA is a long molecule consisting of **Deoxyribose, Phosphate,** and a **nitrogenous base**. One of each of these components bonded together correctly forms a nucleotide. The whole DNA molecule has two strands, and twists together to form a _____ _____.

There are __ different nitrogenous bases, _____, _____, _____, _____.

These are shortened to their respective letters, ___, ____, ___, ___. The bases always occur in pairs:

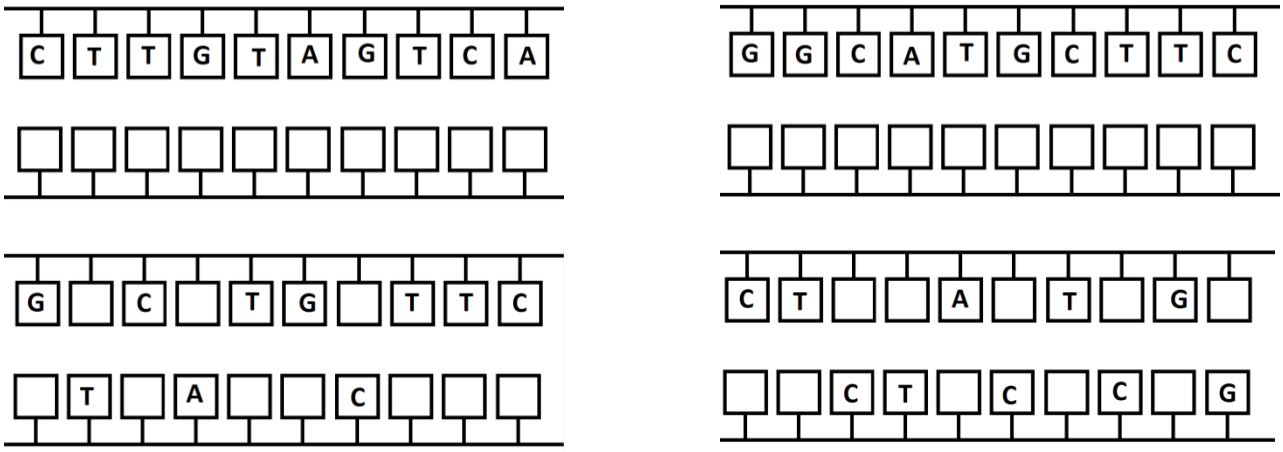
Adenine will always bond with _____

Cytosine will always bind with _____

Therefore the bases **complement** each other. The bases form weak bonds between the two strands, which hold the whole structure together.

Activity 2: Copy the DNA strand

With your knowledge of base pairing, correctly work out the complementary strand for the given DNA molecules:



DNA Replication

DNA is used to store information because it is:
a) Durable
b) Easily copied

It is easy to copy DNA due to the base pairing rule, as any free cytosine will bind to a free guanine and any free adenine will bind to a free thymine.

When DNA replicates, the **double helix strand is split in half** by DNA Helicase (an enzyme which 'unzips' the molecule).

Free nucleotides rush in and bond to their complementary bases, creating a new complementary DNA strand. As you can see, when one DNA is replicated, half of the original DNA is in one new strand and half is in the other. This process is known as **semi-conservative replication**.

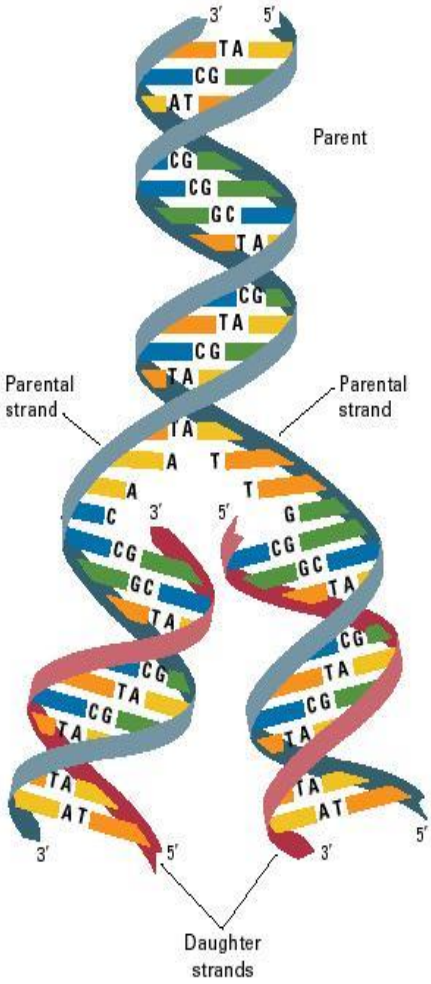
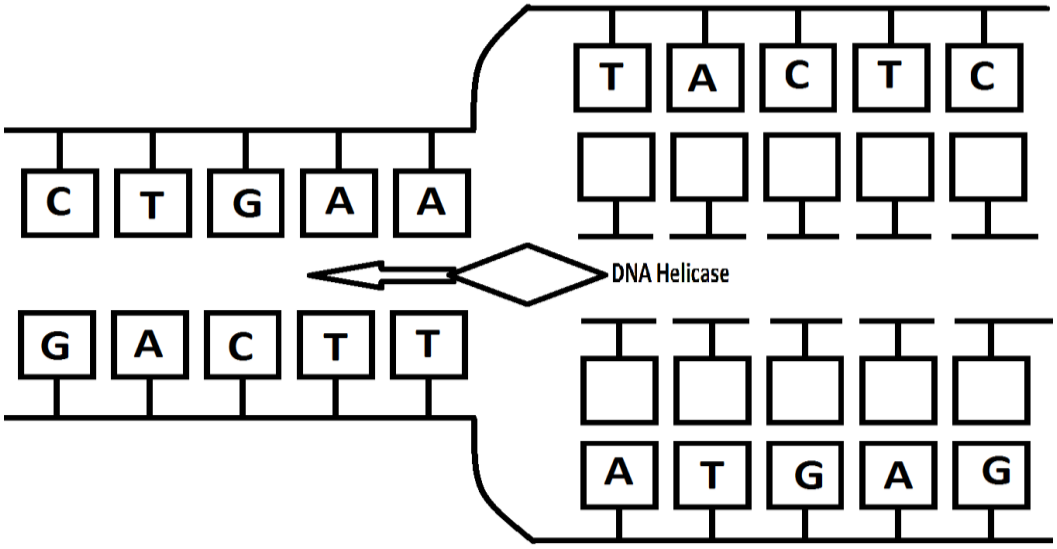


Figure 2: depiction of semi-conservative replication

Activity 3: Fill in the bases of the new nucleotides.



What do you notice about this new chain?

Tutorial 1 – Baseline Test

The homework assignment for the first tutorial is a baseline test to see your initial level of attainment in this subject area. The assignment will test for some or all of the subject specific skills that are required later in the final assignment. However, it is shorter than the final assignment and is will be an introduction to the subject as well as a challenge!

Do not worry too much about doing 'well' or 'badly' on the baseline test, it takes into account the fact that you may not be familiar with the subject area. It is designed to help you and your PhD tutor identify where you are at the start of the programme and to help you measure your progress along the way.

Read the following two articles and answer the questions the end.

Biological polymers

Look at the words on this page. Although every word is different – different length and using different letters – they are all made up of the same 26 letters strung together in different orders. Lots of molecules in biology are very much like words on a page – they are made up of small units strung together in different orders to make a very long molecule. The scientific word for something that is made up of short units strung together is a **polymer**.

One example of a polymer is DNA. DNA is made up two strands, circling around each in a shape called a helix. A single strand of DNA is made up of lots of bases, strung together in a particular order. There are only 4 types of base – ADENINE, THYMINE, GUANINE and CYTOSINE, often abbreviated to A, T, G, and C. Just like the words on the page can communicate any idea you can think of, by putting the bases A, T, G, and C in a certain order, information about how to make a living organism can be stored. This is the job of DNA, which is stored in the nucleus of a cell.

There are other polymers too. A very similar molecule to DNA is called RNA. Unlike DNA, RNA only has one strand, but it is also made up of 4 bases strung together, In RNA, these bases are adenine, uracil, guanine and cytosine – A, U, G and C – but otherwise it is very similar to DNA. RNA is made in the nucleus of the cell, but it is quickly transported in to the cytoplasm.

Another type of polymer is protein. Proteins are very different to DNA or RNA but because they are polymers, they are also made up of short units strung together. The short units that make up proteins are called amino acids. There are 20 different types of amino acids, so proteins are a lot more complicated than DNA or RNA, and can fold into very complicated shapes.

What is a gene and what is a protein?

The bases of DNA make up a long string of letters. AGCTGAGTCACAAATAGCTAGCTGTTTCAG... it goes on and on and on. How does a cell know what to do with all these letters, how to follow the instructions to make a new cell? The key to understanding this is that DNA tells the cell how to make proteins. Proteins are complicated molecules that make up most of the cell. By making the right proteins, everything else follows.

It turns out that DNA is organised into units called **genes**. A single gene contains the instructions to make a single protein. It is a bit like a recipe book, where the different recipes in the book are the same as the different genes in the DNA. The cell follows the instructions in each gene to make each protein.

How does the cell do this? One problem is that the DNA is stored in the nucleus and proteins are mostly needed in the cytoplasm. The cell gets around this problem by making a copy of the gene before making it. Again, if DNA were a recipe book, it would be like photocopying the recipe you need so that you can take it somewhere else. The copy that the cell makes is made of RNA, and the process of making this RNA copy of a gene is called **transcription.** Once the RNA copy has been made in the nucleus, the RNA moves out of the nucleus and in to the cytoplasm.

In the cytoplasm, the RNA meets a machine called ribosome. This is the cook! The ribosome reads the RNA code and makes a protein, by joining amino acids together in the right order. The scientific term for making a protein by following the RNA code is **translation.**

The answers to the questions should be submitted as an attachment on the VLE

1. How are DNA, RNA and proteins similar to each other? And how are they different? Write a paragraph describing as many similarities and differences as you can.

2. What is transcription? Write one or two sentences describing transcription in your own words.

3. What is translation? Write one or two sentences describing translation in your own words.

4. Write down 1 question that you would like answered in Tutorial 2.

Tutorial 2 – From DNA to protein

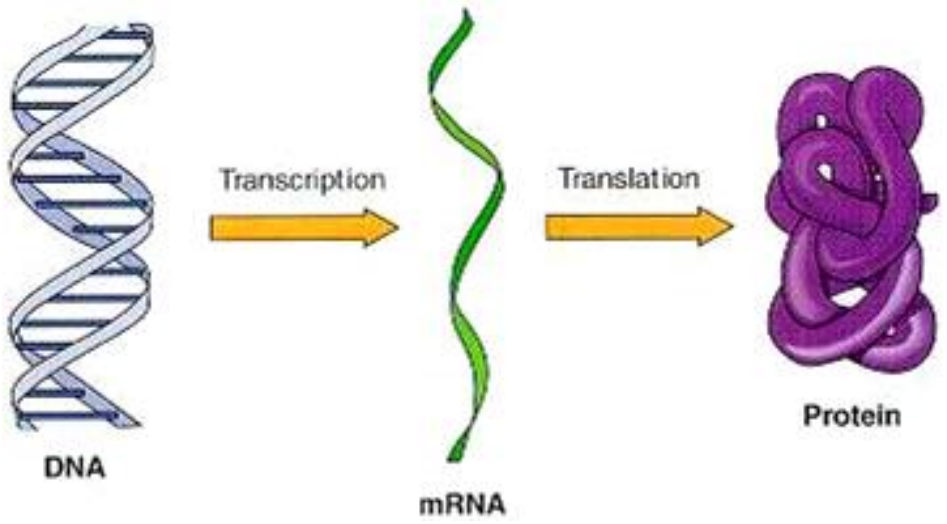


Figure 4: The central dogma: DNA makes RNA which makes protein. It is called this because it a fundamental idea that underpins how all life is created and all further advancements in biology are built upon this idea.

What is the Purpose of Tutorial 2?

- Analyse the stages of protein synthesis
- Understand how DNA is transcribed to RNA
- Understand how RNA code is translated into protein

Key words

Transcription
Translation
RNA
Protein

Activity 1: DNA vs RNA

Compare DNA and RNA

Feature	DNA	mRNA
Shape	Double	
Sugar		
Bases	A, C, G, T	
Stability	Very stable	

Genes are instructions to build a protein. However, DNA that contains the genes can't leave the nucleus so a copy of the gene must be made. This copy is known as RNA.

RNA is very similar to DNA but it contains only one strand. In RNA the thymine (T) is replaced with by a very similar base called Uracil (U) so the bases in RNA are A, C, G, U.

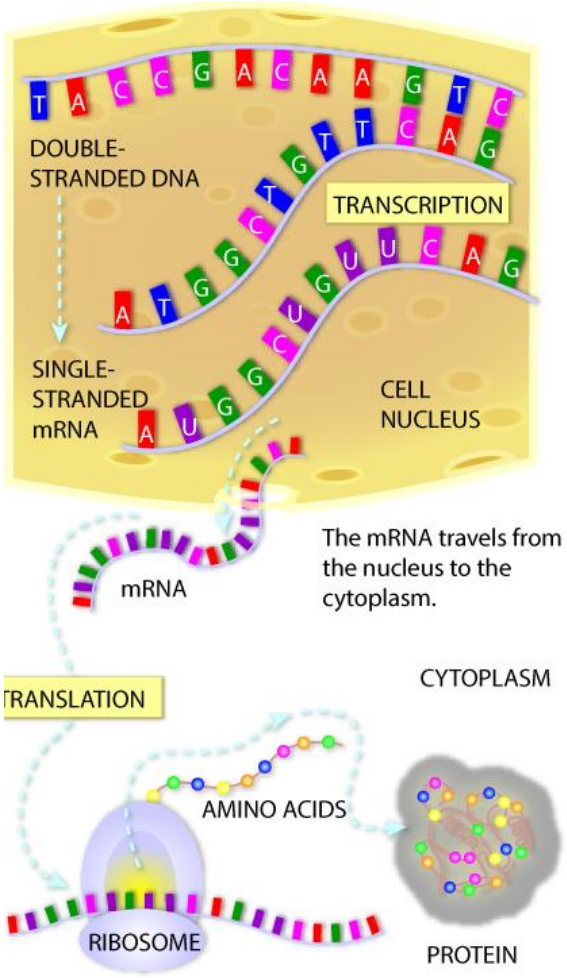
RNA has several jobs in the cell, but we are going to focus on ONE job – its role as a messenger as **messenger RNA (mRNA)**. The process of copying information from DNA to RNA is called transcription.

Transcription

To transcribe something, means to **copy**. Transcription takes place in the nucleus where the DNA double helix unwinds by DNA helicase and the hydrogen bonds are broken. Once the strands are separated, using DNA as a template, complementary RNA nucleotides are joined to make mRNA. The completed mRNA is small enough to exit the nucleus through a nuclear pore in to the cytoplasm to a ribosome. This is where the second stage takes place called translation.

Translation

To translate means to interpret. The ribosome moves along the RNA reading three bases at a time, known as a **triplet codon** that code for a particular **amino acid** which is brought to the ribosome by transfer RNA (tRNA)molecules. Peptide bonds form between the amino acids. A long chain of amino acids is called a **polypeptide.** When a stop codon is reached, translation is finished and the resulting polypeptide chain folds in to a **protein**.



TRANSCRIPTION: In the nucleus, the cell's machinery copies the gene sequence into messenger RNA (mRNA), a molecule that is similar to DNA. Like DNA, mRNA has four nucleotide bases - but in mRNA, the base uracil (U) replaces thymine (T).

TRANSLATION: The protein-making machinery, called the ribosome, reads the mRNA sequence and translates it into the amino acid sequence of the protein. The ribosome starts at the sequence AUG, then reads three nucleotides at a time. Each three-nucleotide codon specifies a particular amino acid. The "stop" codons (UAA, UAG and UGA) tell the ribosome that the protein is complete.

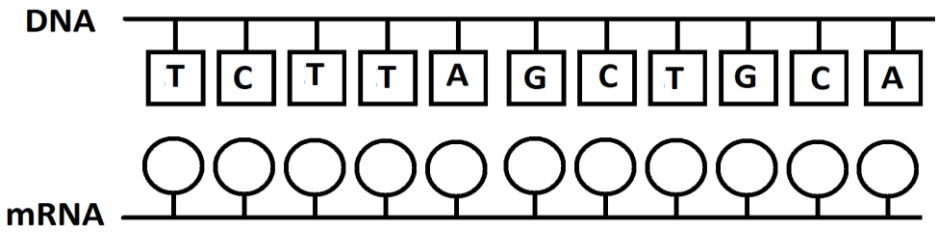
Figure 5: a summary of transcription and translation processes

Video

Watch the video summarising the entire process of protein synthesis. Use the space below to make notes to remind yourself how this process works and anything else you find useful or interesting.

Activity 2 – Transcribe and translate

1. Use your knowledge of the base pair rule to **transcribe** this single DNA strand into mRNA (remember Uracil…).



Become a ribosome

2. Translate the following sequence in to a protein. The universal genetic code can be found at the back of the book (Appendix 1).

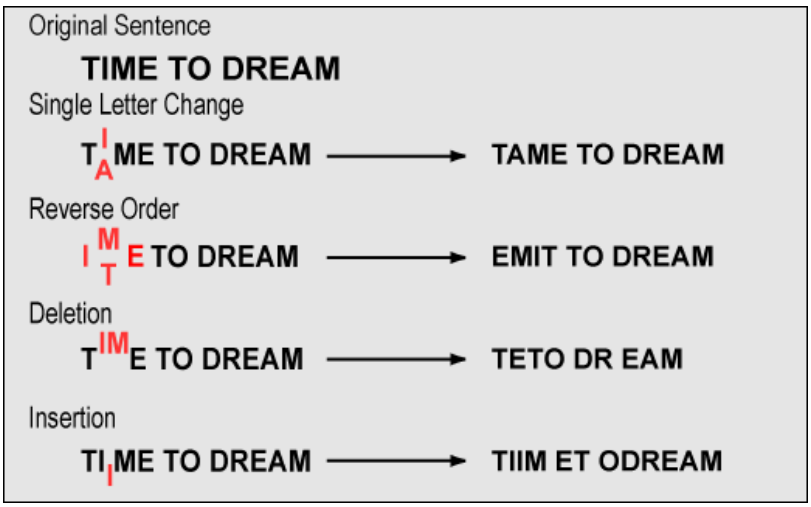
ATG TCC TCC CCA GAG GAG CCA ATA GGC TAT GGG CAT AAT ATT GGC CTG GTC TAA TGG GCA ATT

3. Write your own randomly chosen DNA strand and work out its protein sequence.
(Optional: swap your DNA strand with a partner, they should be roughly the same length and take an attempt at each other's DNA. Fastest one wins!).

Mutation

Because A, T, G, and C are letters in the genetic alphabet, changing one letter for another can change the meaning of a gene. Just as "time" changes to "tame" when one letter changes, a single nucleotide change in a gene may sometimes cause disease.

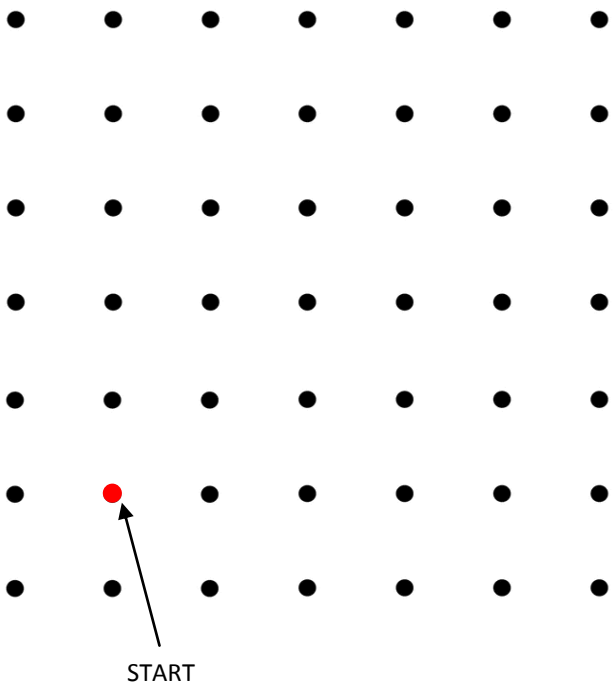
Since many genes govern the fundamental structure and chemical processes of life, defects in their instructions can potentially interfere with the chemical interactions that control an organism's growth and health. However, most mutations that are passed on from generation to generation occur in the long stretches of DNA in between the parts of genes that carry instructions, where they do not harm the person.



Activity 3 – Follow the code

Can you follow the code?

ATG TCC TCC CCA GAG GAG CCA CCA GGC GGC CAC CGA ATA ATA TAG



ATG	Start instructions. Out your pen on the start dot!
ATA	Go down 1 dot
GGG	Go left one dot
CCA	Go right one dot
TCC	Go up one dot
CAC	Go diagonally up and right 1 dot
GGC	Go diagonally up and left 1 dot
CGA	Go diagonally down and right 1 dot
GAG	Go diagonally down and left 1 dot
TAG	End instructions. Take your pen off the paper

What would happen if the instructions were wrong?

This is what a **mutation** is.

Tutorial 2: Homework

1. Look up examples of short lab reports. Bring one with you next class if you can. Come with any questions that you have and we will try to answer these in the next tutorial.

2. Read next week's practical. Familiarise yourself with the protocol and questions.

Tutorial 3 – Practical Session: Extracting your own DNA!

What is the Purpose of Tutorial 3?

- Explain why it is important for scientists to be able to extract DNA from organisms.
- Analyse and evaluate the use of extracted DNA.

Ever wish you could see the strands of genetic material that make you...you? Well now you can!

In this practical, students will extract DNA from their cheek cells. Students will learn key concepts about the function of DNA during the intervals required for the extraction procedure. In any extra time, we will discuss the layout for the lab report (Tutorial 3 assignment).

Protocol

DNA carries the genetic information in all living organisms. Each cell in your body has a nucleus with multiple chromosomes. Each chromosome contains a DNA molecule with multiple genes.

To extract DNA from human cells, you will need to separate the DNA from the other types of biological molecules in human cells. You will be using the same basic steps that biologists use when they extract DNA (e.g. to clone DNA or to make a DNA fingerprint). You will follow these 3 easy steps to extract the DNA:

Detergent
e**N**zymes
Alcohol

Getting Your Sample of Cells
Cells from the lining of your mouth come loose easily, so you will be able to collect cells containing your DNA by swishing a liquid around in your mouth. Obtain a cup with sports drink. You will need to get thousands of your cheek cells in the sports drink in order to extract enough DNA to see. Therefore you should swish the sports drink around in your mouth vigorously for at least 1 – 2 minutes. Then spit the drink back into the cup.

1: Detergent
Add a small amount of detergent to a test tube (0.25 l). Put a glove on the hand you will use to hold your test tube, not the hand you will use to pour. Now carefully pour the drink containing your cheek cells into the test tube with detergent until the tube is **half full**.

Why am I adding detergent?
To get the DNA out of your cheek cells you need to break open both the cell membranes and the nuclear membranes. Cell membranes and nuclear membranes consist primarily of lipids. Dishwashing detergent, like all soaps, breaks up clumps of lipids. This is why you use detergents to remove fats (which are lipids) from dirty dishes. Adding the detergent to you cheek cell solution will break open the cell membranes and nuclear membranes and release your DNA into the solution.

2: Enzymes
Add a pinch of enzyme (meat tenderizer) to your test tube. With your gloved thumb covering the top of the test tube; gently invert the tube five times to mix. Remove your glove. Let the mixture sit for at least 10 minutes. *While you are waiting, answer **Question 1**.*

Why am I adding enzymes?

The nucleus of each of your cells contains multiple long strands of DNA with all the instructions to make your entire body. To fit all of this DNA inside a tiny cell nucleus, the DNA is wrapped tightly around proteins. The enzyme in meat tenderizer is a protease, which is an enzyme that cuts proteins into small pieces. As this enzyme cuts up the proteins, the DNA will unwind and separate from the proteins.

3: Alcohol

Using a pipette, slowly add ice cold alcohol; let the alcohol run down the side of the test tube so it forms a layer on top of the soapy liquid. Add alcohol until you have about 2 cm of alcohol in the tube. Alcohol is less dense than water, so it floats on top. **Do not mix or bump the test tube for 10 minutes.** DNA molecules will clump together where the soapy water below meets the cold alcohol above, and you will be able to see these clumps of DNA as white strands.

While you are waiting for the DNA to become visible, read the next paragraph and answer question 2.

Why am I adding alcohol?

The cold alcohol reduces the solubility of DNA. When cold alcohol is poured on top of the solution, the DNA precipitates out into the alcohol layer, while the lipids and proteins stay in the solution.

Question 1. Which cells in your body contain DNA? Why do these cells need DNA?

Question 2. For most people, when they are injured, the clotting proteins in their blood can produce a blood clot to prevent excessive bleeding. However, some people have a defective version of one type of clotting protein, so clots do not form normally. As a result, they tend to bleed excessively after even minor injuries. These people have haemophilia. Explain how differences in DNA molecules can determine whether or not a person has hemophilia.

4: By now your DNA should be visible as clumps of white strands floating in or just at the bottom of the alcohol layer. There may be air bubbles attached to the strands.

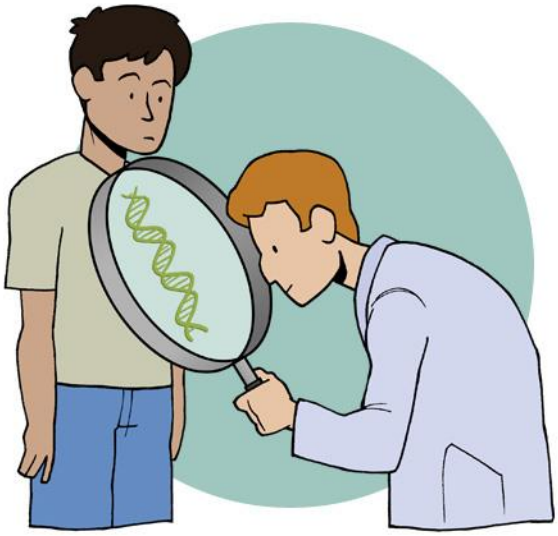
Now you are ready to transfer your DNA from the test tube to your necklace. Be sure to <u>squeeze the air out of your pipette before</u> you put the pipette in the test tube; then gently suck up your DNA. Transfer the DNA to the small capped tube and fill it the rest of the way with alcohol. Close the cap of the tube around a piece of string. Now you have a necklace with your very own DNA!

Tutorial 3: Assignment

Write a lab report on the DNA extraction practical. This will contribute 25% towards your final grade.
Reports should be in a scientific format, Vancouver (numbering) referencing and no longer 2 A4 pages. Font should be Times New Roman, point 11 , 1.15 spacing.

The submission deadline for the assignment is: _____

Tutorial 4 – Genetic testing



What if you could go to the doctor and present a card that contained all your genetic information? Your doctor could scan the card and see whether you were more likely to have an allergic reaction to a particular drug, or whether you were at increased risk for a disease or disorder.

Every year, scientists come closer to making this scenario a reality. Genetic testing companies are making it increasingly possible for consumers to have portions of their DNA sequenced, and to receive some information about what the results might mean. The growth of these "direct-to-consumer" testing companies has resulted in the ability of individuals to learn unprecedented amounts of information about their own genes.

What is the Purpose of Tutorial 4?

- Genetic tests can be characterized by their clinical validity and usefulness
- There are some conditions for which there are genetic tests but no effective treatment
- **Compare and contrast** genetic tests with respect to their clinical validity and the effectiveness of available treatments

Key words

Genetic Testing
Clinical Validity
Effective Treatment Availability
Penetrance
Polygenic

There are a variety of genetic tests available. For example, some tests are performed on early embryos formed by ***in vitro fertilization*** (in which egg cells are fertilized by sperm outside the body) to determine whether they are disease-free and should be implanted in a woman (**Pre-implantation Genetic Diagnosis**, or PGD). **Prenatal** genetic tests test cells from a developing foetus before birth. Newborn screening tests are provided to children immediately upon birth. **Diagnostic tests** are used to identify (or rule out) a particular genetic condition and **carrier tests** are used to identify whether an individual who may not show any condition him/herself carries a particular gene.

This framework helps to evaluate whether a particular genetic test should be recommended for approval for use in patients. It can also help genetic counsellors and patients make decisions about the validity of genetic tests. In addition, different ethical issues arise in each quadrant.

Activity 1 – Place the disease

Using the information provided for each disease, place the disease in what you think is the most appropriate quadrant on the graph. Be ready to explain why.

Alzheimer's disease: A brain disease that causes problems with memory (such as difficulty remembering people and events), thinking, and behaviour. It is most common in people over 65, but up to five percent of people with the disease have early-onset Alzheimer's (also known as younger-onset), which can appear in the patient's 40s or 50s. Alzheimer's worsens over time, and there is no cure, though some treatment options are available that appear to reduce the speed with which the condition worsens.

Hemochromatosis: An autosomal recessive genetic disease that results in the body absorbing too much iron from food. This extra iron is stored in the body, including in organs like the liver and pancreas. The extra iron results in pain, organ damage, cancer, heart problems, and in some cases, death. Symptoms usually begin around age 30 to 40, but may begin in childhood. There is no cure, but the condition can be controlled with a specific diet, removal of blood (to remove the extra iron), and medication.

Huntington's disease (HD): An autosomal dominant genetic disease causing nerve cells to waste away gradually, resulting in uncontrolled movements, severe problems with balance, clumsiness, emotional distress, problems swallowing, and loss of mental function. The condition usually begins when a person is in their 40s, and gets worse with age. The condition is ultimately fatal, and there is no cure.

Phenylketonuria (PKU): A rare autosomal recessive genetic disease in which the body does not make an enzyme necessary to convert the amino acid phenylalanine to the amino acid tyrosine, causing phenylalanine to build up in the body to unsafe levels. This can cause mental retardation, brain damage, and seizures during infancy and early childhood. While a modified diet, including special protein supplements, can reduce the severity of PKU's symptoms, new research suggests that diet alone is not enough to prevent symptoms.

Class discussion

Slightly more complicated: BRCA1 and BRCA2.

Activity 2 – Weighing the risks and benefits – Present a balanced argument

Consider the risks and benefits associated with genetic testing for the following. Would you carry out a genetic test? Give your reasons.

Prenatal testing

In the late 1960s, long before the advent of the technology for DNA sequencing, an early form of genetic testing became available to pregnant women. In amniocentesis, fluid drawn from the amniotic sac surrounding the foetus is analysed for signs of serious chromosomal and genetic disorders, most commonly Down syndrome, Spina bifida, and severe forms of anaemia. In Down syndrome, the sign is an extra copy of chromosome 21, and foetuses with this abnormality are almost certain to develop some form of the disease.

Another prenatal test, chorionic villus sampling (CVS), gathers similar information from cells of the placenta. Doctors can order a CVS test as early as 10 weeks into a pregnancy, 15 weeks for an amnio. Both tests carry a small risk of miscarriage, about 1% for CVS and 0.5% for amnio.

Gene sequencing

Most diseases involve a complex interplay of genetics, environmental influence, and random bad luck. But other conditions, such as Huntington's and cystic fibrosis, are primarily genetic; if you inherit certain variants of certain

genes, you will develop the disease. Starting in the 1980s, tests became available to look specifically for such variants using gene sequencing.

In some instances, these tests don't provide clear-cut results. A case in point is the test for the BRCA1 gene associated with breast and ovarian cancer. Women with certain mutations in the BRCA1 gene have a greater chance of developing cancer - about 85% - compared to women in the general population, who face a 13% lifetime risk. In addition to not having 100% certainty, the test also cannot predict how virulent the cancer will be.

Genome sequencing

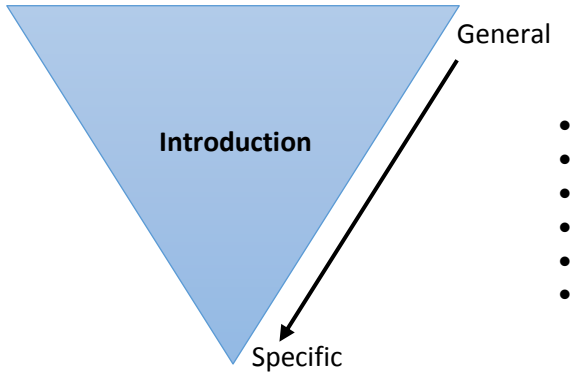
In 2003, researchers from the Human Genome Project (HGP) released the first complete sequence of the human genome—the roughly six billion nucleotides that make up human DNA. Scientists worldwide rely on this information for medical research and the study of human biology and origins.

But the HGP sequence does not account for genetic variation among human beings, and many advances, particularly in the budding area of "personalized medicine," will require a much larger database of genomes that can be compared to one another. With such a database, researchers should be able to better understand the genetic factors underlying countless health conditions as well as to devise personalized treatments.

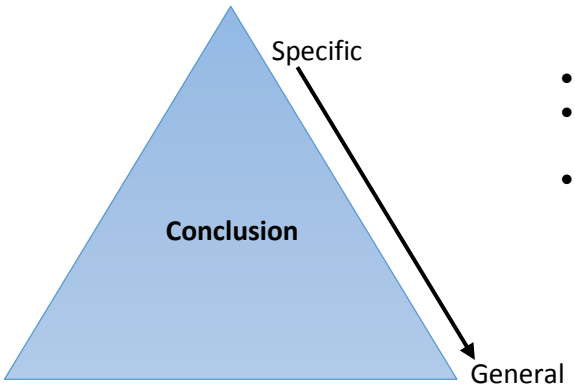
To gather this invaluable data, the Personal Genome Project aims to recruit 100,000 people willing to have their genomes sequenced as well as to share personal information about their health and lifestyles. All this information will be public, available over the Internet.

Tutorial 4 – Homework: Draft Assignment

Start your draft for your final assessment.



- general statement about issue or topic
- necessary background
- focus on problem you will consider
- specific thesis or position
- outline of structure for body of essay
- develop through the essay



- restate your main argument
- emphasise or reinforce the main points of your argument in a concise way
- included final broad statements (about possible implications, future directions for research, limitations, to qualify the conclusion)

Tutorial 5 – Genetic testing dilemmas: Bioethics

What is the Purpose of Tutorial 5?

- Recognise the social and ethical implications of genetic testing
- Weigh some of the harms and benefits of direct-to-consumer genetic testing
- Essay planning

Activity 1 – Game show skit

Class Activity: Appendix 2

The environment plays a large part in diseases, even genetic diseases. In the play, Chris and the host have this interaction:

Dr Chris: That gene suggests you have a predisposition towards breast cancer.
Host: What's a predisposition?
Dr Chris: Well, it doesn't mean you will get the disease, but you might.
Host: Anybody might.
Dr Chris: Yes, but you are at greater risk.
Host: How much greater?
Dr Chris: That would be hard to say.
Host: So what's the point in telling me? It's in my genes, so there's nothing I can do.
Dr Chris: There are lots of things you can do to reduce the risk: diet, exercise, self examination

Questions from skit:

1. What genetic traits are mentioned in the play?

2. Are genetic tests for all of the traits mentioned in the play equally valuable? Are tests for some traits more important than others? Why?

Activity 2 – Debate

1. Would you want to know the results from a genetic test if you knew the contribution to the disease was about 50% genetic and 50% environment? Why or why not?

2. What are some harms that could come from genetic testing?
   What are some benefits that could come from genetic testing?

Activity 3 – Essay planning

Tutorial 5 – Homework: Final Assignment

Genetic testing: power or paranoia?

Choose one of the scenarios from **Appendix 3** (back of the book) and discuss what you would do in the situation. You might find it helpful to address the following points:

- How does science define/explain the scenario?
- What are the main issues surrounding the question and how might genetic testing help/hinder this?
- Which test would be used and the clinical validity of such a test?
- What is your view and why might others feel differently?

Aim to write approximately 1500 words. You can include diagrams/pictures if you wish but remember to label these. Use references in text and don't forget to include a bibliography at the end (this will not be included in your word count). The referencing style is **Vancouver** (references are numbered in text). You will be given a journal (Appendix 4) as an example of how to use this type of referencing. Use font size 11 in Times New Roman with 1.15 spacing.

Good luck!

The submission deadline for the final assignment is: _____

Tutorial 6 – Feedback

What is the Purpose of Tutorial 6?

- Review feedback on final assignment
- Share best examples of academic work among peers
- To reflect on skills learned on the programme with a view to encouraging resilience
- To get feedback from students on the course and their experiences

Activity 1: What are the key questions?

- What strengths have I demonstrated in my work and what areas of development are there for me to address?
- What next steps do I need to take to improve my academic output?

Use the mark scheme and your feedback to help with this as well as reflecting on the entire course.

What I did well?

1.

2.

3.

What I could have improved?

1.

2.

3.

My target for future work is?

(Acknowledgement: The structure and the activities in this feedback tutorial were first designed by Hannah Manktelow in January 2015. We have repeated them here as they demonstrate thoughtful and excellent planning, and can be used by all tutors to ensure a successful and productive feedback tutorial.)

Notes

Appendix 1 - The universal genetic code

2nd base in codon

1st base in codon	U	C	A	G	3rd base in codon
U	Phe	Ser	Tyr	Cys	U
	Phe	Ser	Tyr	Cys	C
	Leu	Ser	STOP	STOP	A
	Leu	Ser	STOP	Trp	G
C	Leu	Pro	His	Arg	U
	Leu	Pro	His	Arg	C
	Leu	Pro	Gln	Arg	A
	Leu	Pro	Gln	Arg	G
A	Ile	Thr	Asn	Ser	U
	Ile	Thr	Asn	Ser	A
	Ile	Thr	Lys	Arg	C
	Met	Thr	Lys	Arg	G
G	Val	Ala	Asp	Gly	U
	Val	Ala	Asp	Gly	A
	Val	Ala	Glu	Gly	C
	Val	Ala	Glu	Gly	G

Ala	Alanine
Arg	Arginine
Asn	Asparagine
Asp	Aspartic acid (Aspartate)
Cys	Cysteine
Gln	Glutamine
Glu	Glutamic acid (Glutamate)
Gly	Glycine
His	Histidine
Ile	Isoleucine
Leu	Leucine
Lys	Lysine
Met	Methionine
Phe	Phenylalanine
Pro	Proline
Ser	Serine
Thr	Threonine
Trp	Tryptophan
Tyr	Tyrosine
Val	Valine
Asx	Asparagine or Aspartic acid (Aspartate)
Glx	Glutamine or Glutamic acid (Glutamate)

Appendix 2 – The Gene Machine

Modified from "Meet the Gene Machine," written by Laura Strieth, Karen Bultitude, Frank Burnet, and Clare Wilkinson. "Meet the Gene Machine" was devised by the Science Communication Unit at the University of the West of England, Bristol, and is funded by the Wellcome Trust (adapted with permission).

Host	Welcome back; so good to see you all again. Now, stop what you're doing; sit down and pay attention, because we are about to make television history. Ladies and Gentlemen, please meet the Gene Machine.... (LEADS THE APPLAUSE)
Applause	
Host	And with me today is scientist, Dr. Chris Taylor.
Chris	Hello.
Host	Chris, we are all just so excited about having you and the Gene Machine here with us today.
Chris	(STILL HOLDING THE COTTON SWAB) It's great to be here.
Host	Now Chris and other scientists have invented a fantastic new device that can read all your genes.
Chris	Yes, It's called the Microarray...
Host	The Gene Machine, that's right Chris. Now as I understand it Chris, we all have genes inherited from our parents.
Chris	Yes, we actually have two full sets of genes, one from each parent. One from the sperm and one from the egg.
Host	OK, OK, keep it clean doctor, this is a family show. Now, these genes can tell us a lot of things, can't they?
Chris	Yes, our genes determine many things about us, many of our key characteristics.
Host	So the Gene Machine can tell us much more than, for instance, a lie detector?
Chris	Oh yes, a lie detector will only tell you if somebody is lying.
Host	Whereas we will know the truth.
Chris	(SEES DISASTER LOOMING) The output from the machine is true, yes. But it can only tell us about a person's genes....
Host	You bet. In fact, this will change the whole future of reality television. With this new machine we can get closer to the truth than ever before.
Chris	Errm, it's not just for television.
Host	(WORRIED) Oh? Who else has a Gene Machine?
Chris	Well, medicine obviously.
Host	Really?
Chris	Yes, many common diseases have genetic components. Identifying those genes will mean doctors can treat people more effectively.
Host	Isn't that great!
Chris	Obviously the police and others would like to use the machine to assist in identifying people...

Host But the biggest impact will be here on TV. And guess who will be the very first celebrity to Meet the Gene Machine…

Chris You?

Host (LAUGHS) As we like to say in television "Here's one I prepared earlier." (INDICATES MACHINE)

Chris Oh yes, we did. I have it here.

Operates machine. The swab goes in one side and a large printout comes out the other side.

Host Wow, that's a lot of paper. More is always better, right?

Chris (TRYING TO HOLD THE PRINTOUT) Well, we all have the same amount of genetic information, but there is a lot of it. Even if genetic tests only look a small part of each gene, that is still quite a bit of information.

Host So what does it say?

Chris Well first of all, it confirms that that is your natural hair color, and will be until you are in your 50s. However long that may be.

Host Does it tell you my age?

Chris No, your genes stay more or less the same all through your life.

Host So I have the genes of a teenager? (CHUCKLE) What else can you see there?

Chris (JUGGLING THE PAPERS) Ah, you have a number of gene variants associated with being musical.

Host I knew it!! I always said I had talent. My mom said I was too lazy to have lessons.

Chris You have some versions of some genes that are associated with musical ability. But, yes, hard work and practice are still very important.

Host I could have been a superstar – wait a minute, I AM.

Chris (TRIES TO ENJOY THE JOKE)

Host OK, so what else have you got there?

Chris Well, it says here that you are likely to be quite tall.

Host (BECOMING IMPATIENT) I can see that for myself; let me have a look. (TAKES THE PRINTOUT)

Host What's this?

Chris That says that you produce a lot of red blood cells.

Host So?

Chris So you should have high levels of fitness and stamina.

Host OK. Tell me something I don't know. What's this?

Host	I'm happy to share this with my public.
Chris	That says you carry the gene variant that causes cystic fibrosis.
Host	Now you're wrong there, I'm as fit as a fiddle, you just said so.
Chris	No, it's not like that. I said earlier that genes come in different varieties, and some varieties cause harm. If you had inherited this variety from each parent, then you would have the disease.
Host	So I'm OK?
Chris	Yes, but you could pass the gene on to your own children, and the disease too, if your partner also has this version of the gene.
Host	How would I know?
Chris	Ask your partner to use the machine.
Host	Oh boy, our lawyers are gonna love you. OK, what does this one mean?
Chris	Ah, now that's a really interesting gene.
Host	Uh huh.
Chris	Well, it's pretty technical.
Host	You don't think my audience is stupid, do you?
Chris	No, no, no, it's just…
Host	Yes?
Chris	(DEEP BREATH) That gene suggests you have a predisposition towards breast cancer.
Host	What's a predisposition?
Chris	Well, it doesn't mean you will get the disease, but you might.
Host	Anybody might.
Chris	Yes, but you are at greater risk.
Host	How much greater?
Chris	That would be hard to say.
Host	So what's the point in telling me? It's in my genes, so there's nothing I can do
Chris	There are lots of things you can do to reduce the risk: diet, exercise, self examination. How often do you examine your breasts?
Host	(NO LONGER HAVING FUN) You scientists really are quite blunt aren't you? We've got time for one more result before the break. How about this one?
Chris	That one?
Host	Yeah.
Chris	(INDICATES ANOTHER) This one's interesting.
Host	That one.
Chris	It's sort of private.

Host	I have nothing to hide.
Chris	Are you sure?
Host	It's a disclosure show – get disclosing.
Chris	You are at very high risk of alcohol dependence.
Host	What is that supposed to mean?
Chris	Alcohol dependence is a condition in which a person…
Host	(STANDS UP) I know what an alcoholic is.
Chris	(STANDING UP) Look, you asked me to tell you.
Host	Are you trying to ruin my reputation?
Chris	Of course not, in fact I tried to warn you about revealing this information in public.
Host	You didn't tell me it would be like this. This is terrible.
Chris	The information in our genes is very powerful, and valuable.
Host	Yes, to blackmailers and paparazzi.
Chris	No, to all of us.
Host	You want to do this to everyone?
Chris	Of course not, but we can't keep this information locked away forever. Gene testing is here to stay.
Host	People have a right to privacy!
Chris	People have a right to know!
Host	To know how much? To know about who? Who decides?
Chris	The American public of course, but do you have the nerve to ask them what they think?
Host	Yes, I do.

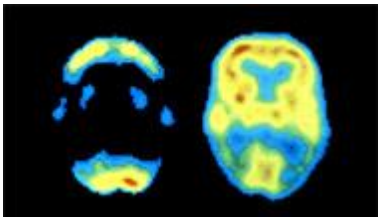
Appendix 3 - What would you do?

SCENARIO 1

Should you test your embryos for gene mutations or other abnormalities before having a baby?

SCENARIO 2

Should you ask your doctor for a genetic test, or order a direct-to-consumer test, that can offer some idea of your risk for contracting Alzheimer's later in life?



SCENARIO 3

You have a family risk of breast cancer. Should you ask your doctor about getting your genes tested for mutations that increase your risk?

1. WHAT IS REFERENCING?

It is a method used to demonstrate to your readers that you have conducted a thorough and appropriate literature search, and reading. Equally, referencing is an acknowledgement that you have used the ideas and written material belonging to other authors in your own work. As with all referencing styles, there are two parts: citing, and the reference list.

2. WHY SHOULD I REFERENCE?

Referencing is crucial to you to carry out successful research, and crucial to your readers so they can see how you did your research. Knowing why you need to reference means you will understand why it is important that you know how to reference.

a) Accurate referencing is a key component of good academic practice and enhances the presentation of your work: it shows that your writing is based on knowledge and informed by appropriate academic reading.
b) You will ensure that anyone reading your work can trace the sources you have used in the development of your work, and give you credit for your research efforts and quality.
c) If you do not acknowledge another person's work or ideas, you could be accused of plagiarism. Plus your lecturers are very keen to see good reference lists. Impress them with the quality of the information you use, and your references, and you will get even better marks.

3. WHAT SHOULD I REFERENCE?

You should include a reference for all the sources of information that you use when writing or creating a piece of your own work.

4. WHAT IS A CITATION?

When you use another person's work in your own work, either by referring to their ideas, or by including a direct quotation, you must acknowledge this in the text of your work. This acknowledgement is called a citation.

5. HOW DO I WRITE A REFERENCE LIST?

This is your list of all the sources that have been cited in the text of your work. The list is inclusive showing books, journals etc. listed in one list, not in separate lists according to source type.

- When using the Vancouver style, the reference list should be in numerical order and each number matches and refers to the one in the text.
- The list should be at the end of your work.
- Books, paper or electronic journal articles, etc., are written in a particular format that must be followed.

6. EXAMPLE OF A BIBLIOGRAPHY

This list includes examples of books, journals and websites.

(1) Arrami M, Garner H. A tale of two citations. *Nature*. 2008; 451(7177):397–399.
(2) Watkins PJ. *ABC of Diabetes*. 5th ed. London: Blackwell Publishing; 2003.

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(6) Pears R, Shields G. *Cite them right: the essential referencing guide*. 3rd ed. Durham: Pear Tree Books; 2008.
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EXEMPLAR

Quercetin Inhibits Intestinal Iron Absorption and Ferroportin Transporter Expression *In Vivo* and *In Vitro*

Marija Lesjak[1,2], Rukshana Hoque[3], Sara Balesaria[1], Vernon Skinner[1], Edward S. Debnam[4], Surjit K. S. Srai[1]*, Paul A. Sharp[3]*

Introduction

Iron is an essential transition metal that plays an important role in all mammalian organisms. It is incorporated into a diverse array of proteins, including the oxygen carriers haemoglobin and myoglobin, cytochrome complexes involved in electron transfer in the mitochondria, and enzymes participating in nucleic acid processing such as ribonucleotide reductase [1,2]. Balancing systemic iron levels within narrow limits is critical for human health, as both iron deficiency and iron overload leads to serious haematological, metabolic and neurodegenerative disorders. In mammals there are no known pathways to eliminate excess iron from the body and therefore iron homeostasis is maintained by modifying dietary absorption so that it matches daily obligatory losses [1].

There are two forms of dietary iron, haem and non-haem iron. Non-haem iron is the major dietary form but its bioavailability depends on presence of other dietary factors which either enhance or inhibit absorption in the duodenum [3,4]. Non-haem iron is present almost entirely in the ferric (Fe^{3+}) form; however, to be bioavailable it must first be reduced to ferrous (Fe^{2+}). This is achieved by the combined action of duodenal cytochrome b (Dcytb), a ferri-reductase which resides on the apical membrane of duodenal enterocytes [5,6], or exogenous dietary reducing agents, such as ascorbic acid (reviewed in [4]). Reduced iron is then transported across the membrane into the enterocyte via the apical iron transporter, divalent metal transporter-1 (DMT1) [7]. Subsequently, ferrous iron is transferred across the basolateral membrane of the enterocyte via the iron exporter, ferroportin (FPN) [8–10] and re-oxidized by ferroxidase hephaestin [11] on the basolateral surface, prior to loading onto transferrin.

In addition to enhancers of iron bioavailability there are a number of dietary components that act as potent inhibitors of intestinal iron absorption, including phytic acid and polyphenolic

References

1. Sharp P, Srai SK (2007) Molecular mechanisms involved in intestinal iron absorption. World J Gastroenterol 13:4716–4724.
2. Evstatiev R, Gasche C (2011) Iron sensing and signalling. Gut. 61:933–952.
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4. Sharp P (2010) Intestinal iron absorption: regulation by dietary and systemic factors. Int J Vitamin Nutr Res 80:231–242.
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University Applications Guidance

For course choices and careers information: www.brightknowledge.org.uk

For information and statistics on universities and course: www.unistats.direct.gov.uk

For comprehensive information on universities, including rankings: www.thecompleteuniversityguide.co.uk

PLEASE NOTE – The Scholars Programme is designed to support the university applications process at your school. The work you complete for the programme should add to your schoolwork rather than detract from it. Please be aware of deadlines and try to manage your workload appropriately. If you are worried about the Scholars Programme interfering with your schoolwork then please speak to your teacher.

Subject Specific Top Tips for strengthening your application

Do you enjoy Chemistry but think becoming a Chemist is the only thing it leads to? It most certainly does not! Chemistry overlaps with many other subjects so you will find that you pick up skills that are useful in biology and medicine, physics and engineering, geology and earth science. Chemistry is also studied in an environmental and social context, so you'll gain awareness of its ethical implications and issues relating to environmental impact and sustainability. It involves math so you will develop strong mathematical/numerical ability. Find out what it is that interests you and read up on it! What would a degree encompass? What different career paths does it lead to?

Reading books on the subject can help but reading a book isn't enough on its own. You need to enjoy the material and more importantly, be able to talk about it. So for example, something like 'A brief history of time' by Stephen Hawking may not have a lot of value, because, relatively speaking, it isn't that accessible or discussable.

For interviews, it is important that you can wow the interviewer with your enthusiasm, curiosity and knowledge for the subject, so try to find a book that stretches you on something that you're genuinely interested in and absorb that. Here's a top tip: If you can spend the interview discussing with them in detail about something you do understand, it will limit the time where they can ask you about things you might not. (Sshhhhh don't tell anyone I told you).

Additionally, it is always useful to get a taste for what you want to do. Think you might want to be a doctor? Ask your GP if you can shadow him or her for the day. Want to be a chemist? Ask university labs if you can come in for a few days and watch them perform some experiments. Dentist? Visit a dental clinic and learn about the all the procedures they do as you go around with a dental nurse. These sorts of experiences can be an invaluable tool and really make an interesting story for a personal statement as well as showing how committed you are to the field. If there are no places for you to visit, ask teachers or other brilliant club tutors to help you get in touch with professionals; all you have to do is ask around!

Possible things to read or watch

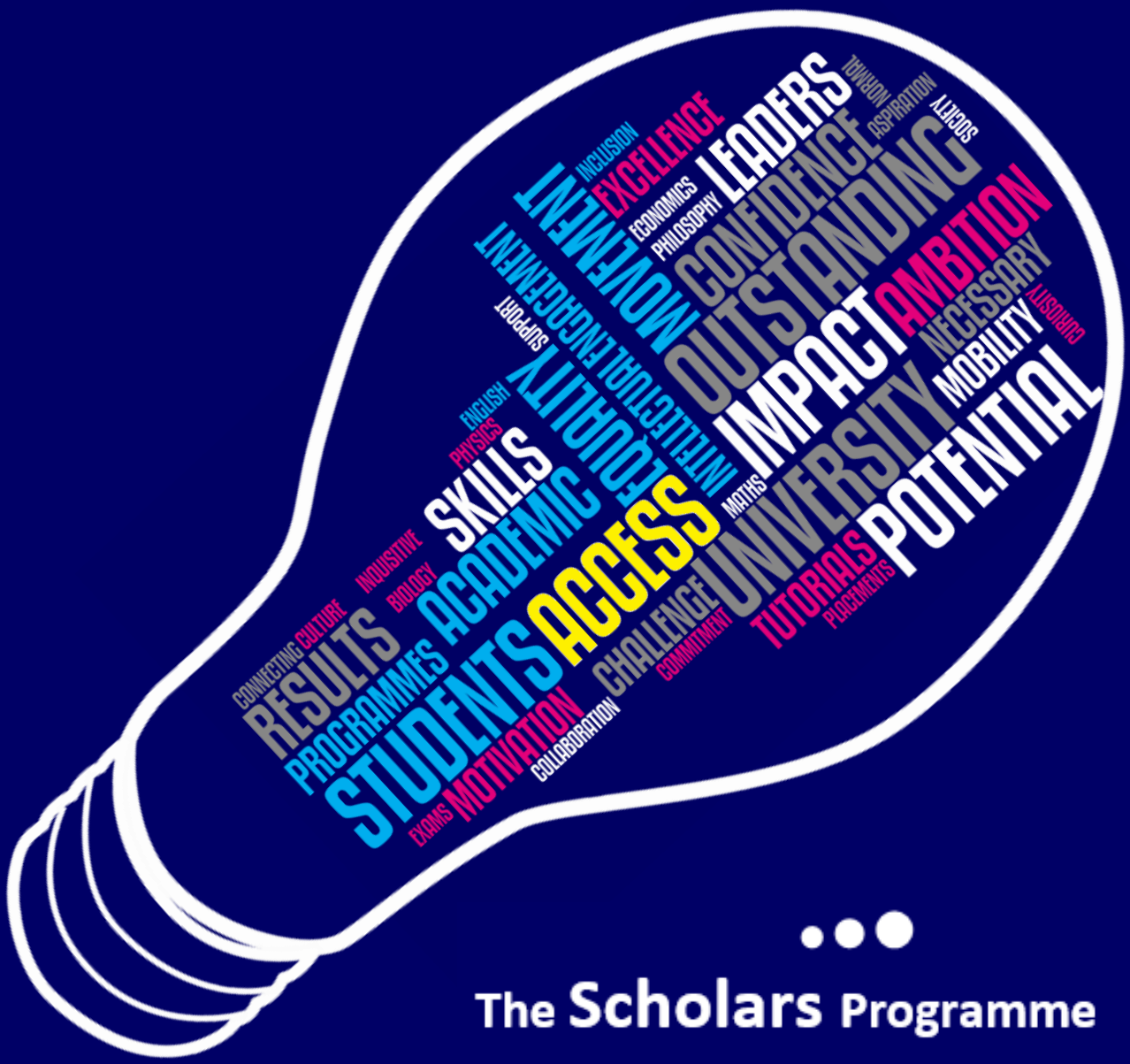
Popular science book – *A short history of everything* by Bill Bryson.

Neuroscience/Medicine - *The man who mistook his wife for a hat* by Oliver Sacks

Chemistry - *The Disappearing Spoon: And Other True Tales of Madness, Love, and the History of the World from the Periodic Table of the Elements* by Sam Kean (I apologise for the American terminologies used)

Physics - This MIT lecture course on electricity and magnetism is very clear and a 1st year university standard: https://www.youtube.com/watch?v=3omwHv3Cmog
'Parallel Worlds, Parallel Lives' – a documentary (6 parts - 10 minutes each) by a rock star about his deceased father - a quantum mechanic, follows his quest to try and understand his father's theories with no maths/physics background. Available on YouTube if you type in the title.

Reading/watching things like these can help make you more interested and passionate about a subject, and that will shine through on your personal statement and interview.



The Scholars Programme