SURFING

WACE HUMAN BIOLOGY

Unit 2 Reproduction and Inheritance

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Introduction

Each book in the *Surfing* series contains a summary, with occasional more detailed sections, of all the mandatory parts of the syllabus, along with questions and answers.

All types of questions – multiple choice, short response, structured response and free response – are provided. Questions are written in exam style so that you will become familiar with the concepts of the topic and answering questions in the required way.

Answers to all questions are included.

A topic test at the end of the book contains an extensive set of summary questions. These cover every aspect of the topic, and are useful for revision and exam practice.

Words To Watch

account, account for State reasons for, report on, give an account of, narrate a series of events or transactions.

analyse Interpret data to reach conclusions.

annotate Add brief notes to a diagram or graph.

apply Put to use in a particular situation.

assess Make a judgement about the value of something.

calculate Find a numerical answer.

clarify Make clear or plain.

classify Arrange into classes, groups or categories.

comment Give a judgement based on a given statement or result of a calculation.

compare Estimate, measure or note how things are similar or different.

construct Represent or develop in graphical form.

contrast Show how things are different or opposite.

create Originate or bring into existence.

deduce Reach a conclusion from given information.

define Give the precise meaning of a word, phrase or physical quantity.

demonstrate Show by example.

derive Manipulate a mathematical relationship(s) to give a new equation or relationship.

describe Give a detailed account.

design Produce a plan, simulation or model.

determine Find the only possible answer.

discuss Talk or write about a topic, taking into account different issues or ideas.

distinguish Give differences between two or more different items.

draw Represent by means of pencil lines.

estimate Find an approximate value for an unknown quantity.

evaluate Assess the implications and limitations.

examine Inquire into.

explain Make something clear or easy to understand.

extract Choose relevant and/or appropriate details.

extrapolate Infer from what is known.

hypothesise Suggest an explanation for a group of facts or phenomena.

identify Recognise and name.

interpret Draw meaning from.

investigate Plan, inquire into and draw conclusions about.

justify Support an argument or conclusion.

label Add labels to a diagram.

list Give a sequence of names or other brief answers.

measure Find a value for a quantity.

outline Give a brief account or summary.

plan Use strategies to develop a series of steps or processes.

predict Give an expected result.

propose Put forward a plan or suggestion for consideration or action.

recall Present remembered ideas, facts or experiences.

relate Tell or report about happenings, events or circumstances.

represent Use words, images or symbols to convey meaning.

select Choose in preference to another or others.

sequence Arrange in order.

show Give the steps in a calculation or derivation.

sketch Make a quick, rough drawing of something.

solve Work out the answer to a problem.

state Give a specific name, value or other brief answer.

suggest Put forward an idea for consideration.

summarise Give a brief statement of the main points.

synthesise Combine various elements to make a whole.

1 Assumed Knowledge

1. The diagram shows two types of cell division.

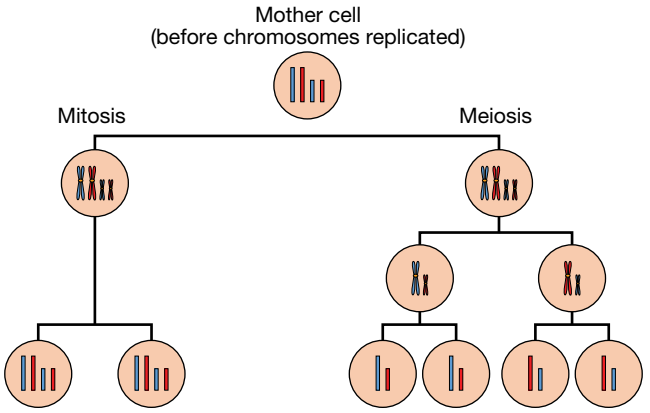


Figure 1.1 Mitosis and meiosis.

   (a) Define mitosis.
   (b) What is meiosis?
   (c) Identify a difference in the daughter cells produced by mitosis and meiosis.

2. How is information transferred when cells reproduce themselves?
3. What does DNA stand for?

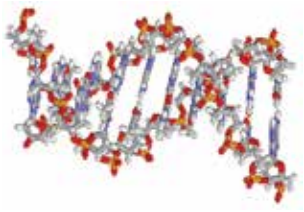


Figure 1.2 DNA.

4. Name the basic unit of DNA.
5. Where is DNA located in cells?
6. Outline the structure of the DNA molecule.
7. What is the relationship between genes and DNA?
8. Explain the advantages of DNA replicating exactly.
9. What is a mutation?
10. What is a pedigree?
11. The diagram shows a body system.

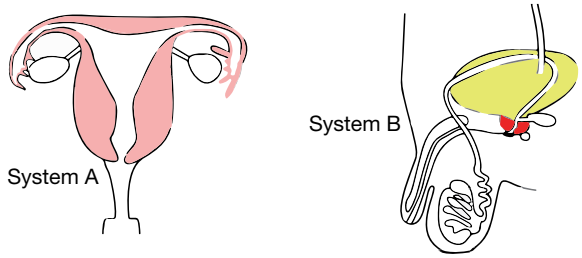


Figure 1.3 Two body systems.

   Identify system A and system B.

12. What is biotechnology?
13. What is a somatic cell?
14. Define a stem cell.
15. What is a germ layer?
16. The diagram shows different stages of pregnancy.

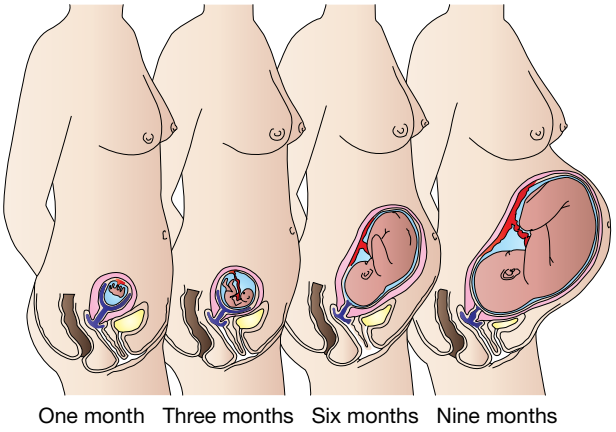


Figure 1.4 Different stages of pregnancy.

   How long is human gestation?

17. What is parturition?
18. The diagram shows a type of human cell. What is the name of this cell and what is its function?

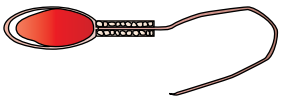


Figure 1.5 Type of human cell.

19. Distinguish between an embryo and a foetus.
20. Define a mutagen.
21. Why is Gregor Mendel often referred to as the 'father of genetics'?
22. Identify the factors that determine the features of an organism.
23. Use an example to show how environmental influences can affect the appearance of a person.
24. Use an example to show how genes determine the features of an organism.
25. What is the 'Watson-Crick' model of DNA?
26. Define genome.
27. What is a chromosome?
28. What is meant by genotype?
29. Define fertilisation.
30. Why is it important for gametes to have half the number of chromosomes of the species?
31. The diagram shows the structure of a female reproductive system.

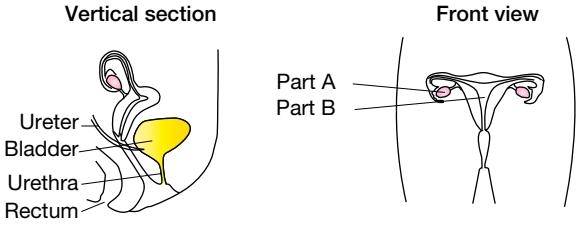


Figure 1.6 Female reproductive system.

   State the function of part A and part B.

2 Discovering DNA

The discovery of DNA, its importance in the transmission of information from one generation to the next and its structural properties involved the findings of many scientists who shared their data, experimental evidence and theories. Multiple scientists built their work on the findings of their predecessors.

Friedrich Miescher (1844-1895)

In 1869 Miescher isolated some phosphate-rich chemicals that came from the nuclei of white blood cells. He called these chemicals **nuclein**. He researched the properties of these chemicals but did not know their function. Nuclein is now called nucleic acid.

Figure 2.1 Friedrich Miescher (left), Phoebus Levene (centre) and William Astbury (right).

Frederick Griffith (1879-1941)

In 1928 Griffith found that *Streptococcus pneumoniae* bacteria could change its form in a phenomenon known as **bacterial transformation**. He killed a pathogenic strain of bacteria with heat and then mixed the remains of the dead cells with living non-pathogenic bacteria. Some of the living cells transformed and became pathogenic and their offspring inherited the pathogenicity. The transforming factor that caused this heritable change was unknown.

Phoebus Levene (1869-1940)

In 1909 Levene discovered ribose and in 1929 discovered deoxyribose. He showed that DNA consisted of phosphate-sugar base units that he called **nucleotides** and in his **tetranucleotide hypothesis** he proposed that there were only four nucleotides per molecule of DNA. He also believed that the structure of DNA was too simple to hold the genetic code at a time when many believed that the protein component of chromosomes for inheritance. His work with nucleotides became the basis for the final determination of the structure of DNA.

William Astbury (1898-1961)

Astbury used X-ray diffraction to study the structure of many biological molecules. In 1938 he reported that the structure of DNA repeated every 2.7 nanometres and that the bases were arranged in flat stacks that were 0.34 nanometres apart.

Linus Pauling (1901-1994)

Pauling studied the structure of many biological molecules and found that many proteins had helical shapes. After reviewing the results obtained by Astbury he proposed that DNA was a triple helix. In 1949 he provided proof that a specific mutation in haemoglobin caused sickle cell anaemia and showed that diseases could be understood on a molecular level.

George Beadle (1903-1989) and Edward Tatum (1909-1975)

In 1940 Beadle and Tatum carried out an experiment using the bread mould *Neurospora crassa* showing there was a direct link between genes and enzymes with one gene responsible for the production of a single enzyme. They proposed the one gene-one enzyme hypothesis.

Figure 2.2 Linus Pauling (left),Oswald Avery (centre) and Colin McLeod (right).

Oswald Avery (1877-1955), Colin McLeod (1909-1972) and Maclyn McCarty (1911-2005)

In 1944 Avery-McLeod-McCarty reported that their experiments with pneumococcal bacteria showed that DNA was responsible for bacterial transformation. They had tested all the classes of molecules found in the remains of the dead pathogenic bacteria for transforming ability and found that only DNA could change non-pathogenic bacteria into pathogenic bacteria. Their discovery was met with interest and scepticism. At the time there was debate about the molecule that carried the genetic information with many believing that the protein was the heredity material.

Erwin Chargaff (1905-2002)

Chargaff analysed the base composition of DNA from a number of different species. In 1947 he showed that the number of units of adenine, thymine, cytosine and guanine units is different for different organisms. He also showed that in natural DNA the number of guanine units is the same as the number of cytosine units and the number of adenine units is the same as the number of thymine units. His findings became known as *Chargaff's rules*.

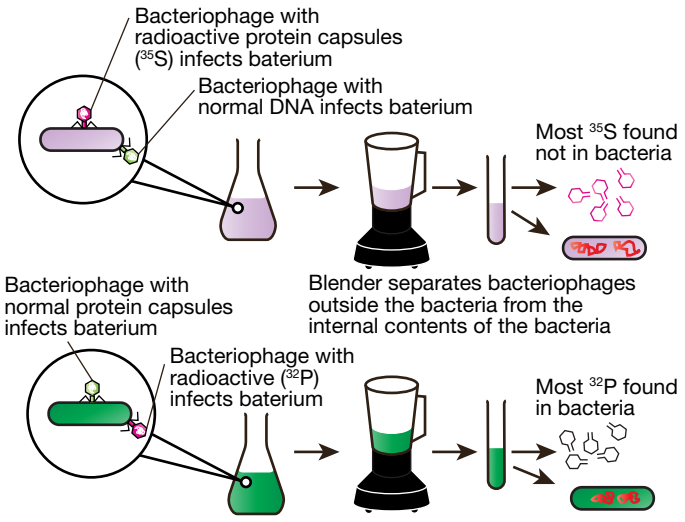


Figure 2.3 Hershey-Chase experiment.

Alfred Hershey (1908-1997) and Martha Chase (1927-2003)

In 1952 Hershey and Chase carried out experiments that confirmed DNA as the genetic material. **Bacteriophages** are viruses that can infect bacteria. Hershey and Chase used bacteriophages (phages) to infect *Escherichia coli* which is a bacteria that lives in the intestines of mammals. They used two sets of bacteriophages, one with proteins labelled with radioactive sulfur (using ^{35}S) and one with DNA radioactively labelled with phosphorus (using ^{32}P) and found that the radioactively labelled DNA entered the bacteria cells while the radioactively labelled proteins did not enter the bacteria cells. When the results of the Hershey-Chase experiment were published most of the scientific community accepted DNA as the heredity material.

Rosalind Franklin (1920-1958)

Franklin took X-ray diffraction images of DNA which showed the helical structure of DNA. Her photograph was used by Watson and Crick to determine the double helix structure of DNA.

(a) (b)

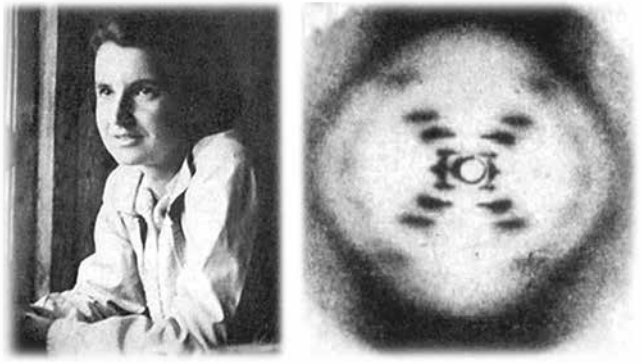


Figure 2.4 (a) Rosalind Franklin. (b) Franklin's X-ray diffraction pattern of DNA.

Maurice Wilkins (1916-2004)

Wilkins produced the first clear X-ray images of DNA with his work showing there was a repeating pattern in the DNA structure. As a friend of both Watson and Crick, Wilkins took a clear X-ray diffraction photograph of DNA taken by Rosalind Franklin and showed it to Watson who then shared the information shown in the photograph with Crick. This information was vital in the final determination of the structure of DNA. Wilkins shared the 1962 Nobel Prize for Physiology or Medicine with Watson and Crick.

James Watson (1928-) and Francis Crick (1916-2004)

In 1953, James Watson and Francis Crick suggested the double helix structure of DNA with two phosphate-sugar strands winding around the outside of nitrogenous bases with the pairs A-T and C-G. Watson and Crick based this structure on X-ray diffraction photographs of DNA taken by Rosalind Franklin. Maurice Wilkins was Franklin's colleague and showed her X-ray photographs to Watson. From the photographs, Watson and Crick were able to work out the distance between atoms, the angles between bonds and the size of atoms. They could then construct a three-dimensional model of DNA.

Figure 2.5 Watson and Crick.

Collaboration and competition

The accumulation of knowledge about the carrier of genetic information occurred over several decades and involved the cumulative work of multiple scientists. The final determination of the structure of DNA relied on experimental evidence and shows how collaboration and effective communication can lead to great scientific discoveries.

The collaboration between molecular biochemist Crick and biochemist Watson produced the final double helix structure. Wilkins collaborated when he showed the X-ray diffraction photographs to Watson. For this joint effort the three men were awarded the 1962 Nobel Prize for Physiology.

When Watson and Crick published their proposal for the double helix structure of DNA they did not credit Franklin with supplying the critical photograph or acknowledge that they did have permission from Franklin to use her work. This goes against scientific ethics and in later interviews Watson admitted that their work would not have been possible without the photograph taken by Franklin. There was also friction between Franklin and her colleague Wilkins. Franklin died of cancer in 1958 and could not be given credit posthumously for the Nobel Prize.

QUESTIONS

1. When were nucleic acids first identified and what were they called at this time?
2. Describe the bacterial transformation observed by Frederick Griffith.
3. (a) What is a nucleotide?
   (b) Who named nucleotides?
   (c) When nucleotides were first named, what was the common belief about the carrier of genetic information?
4. Explain why the work of William Astbury was important in determining the structure of DNA.
5. Outline how the work of Beadle and Tatum increased scientific understanding about genes and DNA.
6. Describe Chargaff's two rules for the composition of DNA.
7. Describe the Hershey-Chase experiment.
8. Outline the contribution made by Rosalind Franklin to our knowledge about the structure of DNA.
9. (a) Briefly describe how the work of James Watson, Francis Crick, Rosalind Franklin and Maurice Wilkins was interlinked and led to the final determination of the structure of DNA.
   (b) Using the discovery of the double helix structure, explain how the role of collaboration and effective communication is necessary in scientific research.
10. In the early 1950s Wilkins and Franklin were working at the University of London, Pauling was working at Caltech and Watson and Crick were working at Cambridge University. How does the work of these three groups show competition between scientists?
11. The diagram shows a bacteriophage.
    (a) What is a bacteriophage?
    (b) Explain how bacteriophages were used to help collect experimental data about the DNA molecule.

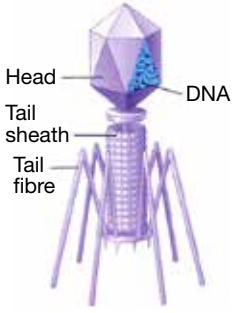


Figure 2.6 Bacteriophage.

12. The diagram shows the structure of a molecule found in living organisms.

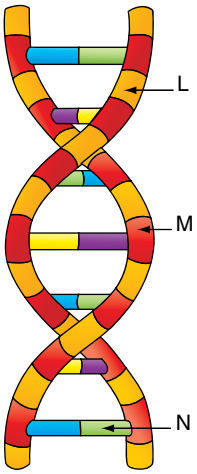


Figure 2.7 Molecule found in living things.

    (a) What is this molecule?
    (b) Identify the parts L, M and N.
    (c) Who discovered the structure of this molecule?
    (d) Discuss three aspects of the structure of this molecule that make it suitable for its function.
13. In 1868 Friedrich Miescher isolated a mixture of DNA and proteins from pus-soaked bandages of wounded soldiers. Later work proved the mixture contained the material of chromosomes. For many years scientists could not work out how information was coded for inheritance. Who proposed, in 1953, the double helix model of DNA?
    (A) Franklin and Wilkins.
    (B) Wilkins and Crick.
    (C) Watson and Crick.
    (D) Watson and Franklin.
14. What was Rosalind Franklin's major contribution to our understanding of the structure of DNA?
    (A) Took X-ray diffraction photographs of DNA.
    (B) Showed there were covalent bonds in nucleic acid polymers.
    (C) Suggested the double helix structure of DNA.
    (D) Deduced there is a purine-pyrimidine pair for nitrogenous bases.
15. Who determined that there were equal number of units of adenine and thymine and equal number of units of cytosine and guanine in DNA?
    (A) Levene.
    (B) Chargaff.
    (C) Avery.
    (D) Franklin.
16. Which of the following was named by Levene?
    (A) Nuclein.
    (B) Nucleotide.
    (C) Bacteriophage.
    (D) Bacterial transformation.

3 The Structure of DNA

Early in the 20th century scientists tried to determine if hereditary information was passed on through proteins or deoxyribose nucleic acid (DNA).

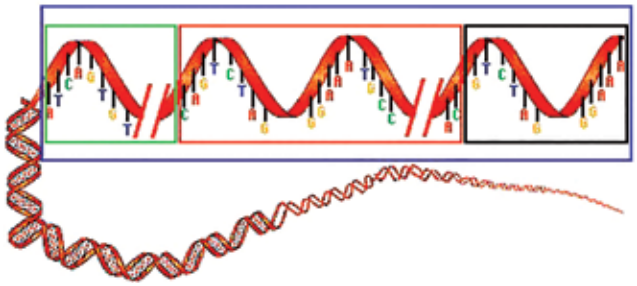


Figure 3.1 DNA.

Eventually enough evidence was gathered to show that DNA carried the genetic material from one generation to the next. In 1944 **Oswald Avery** demonstrated that genes were made of DNA. In 1952 **Rosalind Franklin**, with **Maurice Wilkins**, showed that the DNA molecule was helical using X-ray diffraction crystallography. In 1953 **James Watson** and **Francis Crick** proposed the double helix structure for DNA.

Each chromosome consists of many genes, i.e. each **gene** is a certain length of the DNA molecule. When working out the chemical structure of genes scientists knew that the substance would have to be able to encode a large amount of information, that it must be chemically stable, that it must be able to make an accurate replication of itself, that occasional errors (mutations) could occur and that the substance controls and directs protein synthesis.

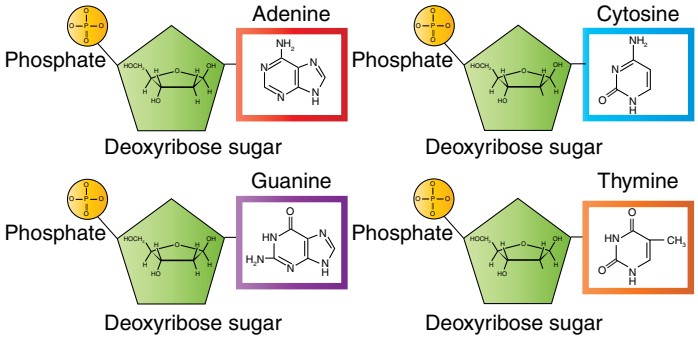


Figure 3.2 Four different nucleotides that make up DNA.

Nucleotides

The basic unit of DNA is the **nucleotide**. A nucleotide consists of a sugar, a phosphate and one of four nitrogenous bases – either adenine (A), guanine (G), cytosine (C) or thymine (T). To form the double helix, the sides of the ladder are made up of alternating sugar and phosphate molecules and the rungs consist of paired nitrogenous bases. Adenine always pairs with thymine and guanine always pairs with cytosine.

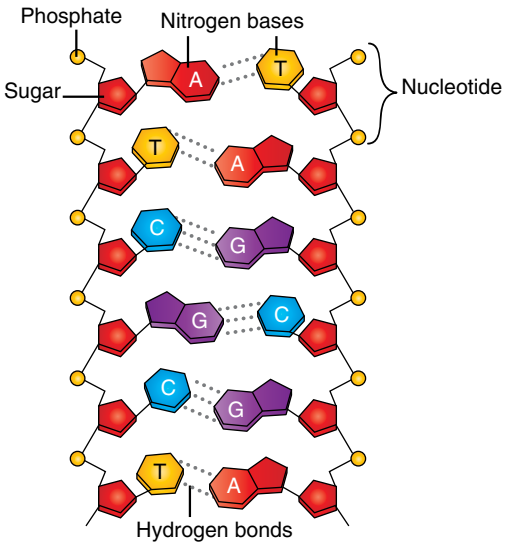


Figure 3.3 Base pairs of DNA.

The double helix is a structure where two spirals coil around each other keeping a constant diameter of the coil. It is a right-handed spiral polymer.

Bonding in DNA

The base pairs on the rungs of the ladder are held together by hydrogen bonds and by van der Waals interactions between the stacked bases. These hydrogen bonds break during DNA replication.

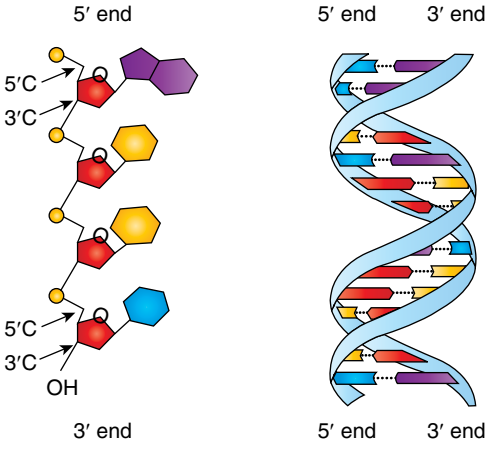


Figure 3.4 Four different nucleotides that make up DNA.

The nucleotides are joined together by covalent bonds between the phosphate group of one nucleotide and the sugar of the next. These bonds are called **phosphodiester linkages** and occur between the –OH group on the 3′ carbon of one nucleotide and the phosphate on the 5′ carbon of the next. This means that the free ends of the DNA molecule are different to each other with one side having a phosphate attached to a 5′ carbon and the other side having a hydroxyl (OH) group on a 3′ carbon. Since the two strands of the double helix run in opposite directions, they are said to be **antiparallel** to each other. The phosphate groups give the nucleic acids their acidic properties.

QUESTIONS

1. What is a gene?
2. What does 'DNA' stand for?
3. Use an example to show how major advances in scientific understanding have directed future biological research.
4. Describe the shape of DNA.
5. What is a nucleotide?
6. Which part of the nucleotide gives the nucleic acid its acidic properties?
7. Purines, e.g. adenine and guanine, are double-ringed nitrogenous bases and pyrimidines, e.g. cytosine and thymine, are single-ringed nitrogenous bases. How do these bases form complementary pairs?
8. List five features of DNA that make it a suitable genetic medium.
9. What type of bond holds the sugar of one nucleotide to the phosphate group of the next nucleotide?
10. What type of bond holds the nitrogenous bases on the 'rungs' of the ladder together?
11. Explain why the arrangement of the arrangement of the two strands of DNA are called antiparallel.
12. Which bonds break during DNA replication?
13. Outline the role of DNA in the transmission of genes from one generation to the next.
14. Explain the significance of DNA replication.
15. The diagram shows a short section of part of a DNA molecule.

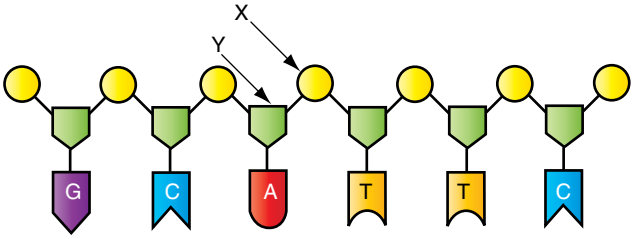


Figure 3.5 Part of a DNA molecule.

(a) Identify parts X and Y.
(b) Draw the complementary strand of DNA.
(c) Using the given section of DNA, describe the process by which this segment controls the production of proteins and/or polypeptides.

16. Since the 1940s it has been known that genes consist of which chemical?
(A) Ribose nucleic acid.
(B) Deoxyribose nucleic acid.
(C) Dinitroadenine acid.
(D) Adenosine triphosphate.

17. Which of the following identifies the basic components of a nucleotide?
(A) Sugar, phosphate, nitrogenous base.
(B) Adenine, thymine, cytosine, guanine.
(C) Glucose, nitrate, phosphate base.
(D) Sugar, amino acid, nitrogenous base.

18. What is shown by an analysis of the DNA molecule?
(A) The 'rungs' of the 'ladder' are made of phosphate-sugar bonds.
(B) The 'sides' of the 'ladder' are made of phosphate-sugar bonds.
(C) Nucleotides of one strand are identical to the nucleotides of the opposite strand.
(D) DNA with 40% thymine will have 20% guanine.

19. A certain chromosome has 20% guanine in its structure. What would you expect to be the amount of adenine present?
(A) 20%
(B) 30%
(C) 60%
(D) 80%

20. The diagram shows a short section of DNA.

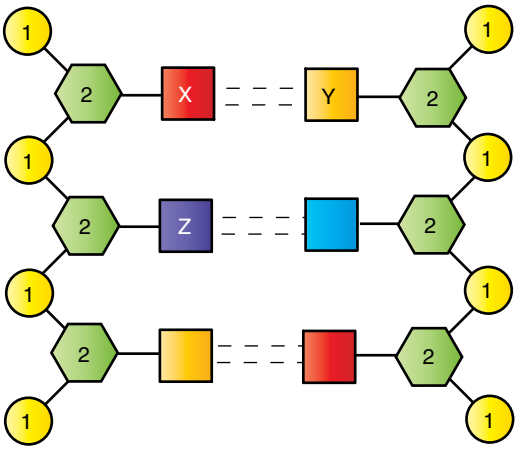


Figure 3.6 Short section of DNA.

If X represents adenine then what could Z represent?
(A) Thymine.
(B) Sugar.
(C) Phosphate.
(D) Cytosine.

21. The figure below represents a single strand of DNA. What is its complementary strand?

A A C T G G

(A) AACTGC
(B) GGTCAA
(C) TTGACC
(D) CCAGTT

22. What type of bonds would you expect to find between the nitrogenous bases on the rungs of the DNA ladder?
(A) Hydrogen bonds.
(B) Covalent bonds.
(C) Phosphodiester linkages.
(D) Peptide bonds.

4 Activity – Making a Model of DNA

In eukaryotic cells a chromosome is one extremely long linear DNA molecule. In prokaryotic cells there is a circular DNA molecule.

There are many ways you can construct a model to show the structure of DNA. Models can be made of paper, cardboard, pipecleaners, plastic sticks, coloured balls or created using multimedia graphics. The model needs to be able to show the triplet code of DNA which is the basic instruction of the genetic code.

(a) (b) (c)

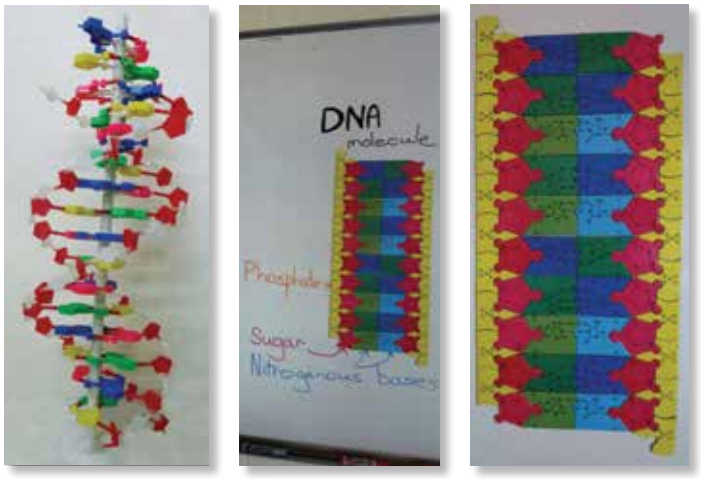


Figure 4.1 (a) Three-dimensional model of the DNA molecule. (b) Building a 2-D model of DNA on a classroom whiteboard using cardboard cutouts that have magnetic strips which stick to the whiteboard. (c) 2-D jigsaw model of DNA using a purchased DNA kit.

The **triplet code** is three bases next to each other in a molecule of DNA and specifies a particular amino acid. Reading the sequence of nucleotides in the triplet code in DNA means that you are reading the sequence of amino acids which will be joined together to form a polypeptide. There are three codes that are 'stop' signals which stop the process of gene expression and the translation of the code into a polypeptide. The DNA codes ATT, ATC and ACT signal 'stop'. The DNA code TAC is the code for the amino acid methionine but also acts as a 'start' signal for translation. Note that these start and stop codes are usually given as the mRNA code (e.g. stop codes UAA, UAG and UGA) as it is the **mRNA code** that is used in definitions and used to identify different amino acids. The mRNA code is complementary to the DNA code with uracil substituting for thymine. In gene expression the mRNA code is read in the cytoplasm when the code is translated to produce a polypeptide.

In gene expression when the DNA code is translated from the mRNA code into a polypeptide it is important to read the three nucleotide bases of an amino acid in the correct **reading frame**. Any alteration of the code will affect the ability to make a particular polypeptide.

Thus it is important when making a model of DNA to know what the code means when it is translated to produce a polypeptide.

3-D models

You can use molecular model kits with sticks and balls and make a model similar to the one built by James Watson and Francis Crick. A model using plastic sticks, coloured spheres and or pipecleaners has the benefit that it gives a three-dimensional vision of DNA to show the double helix structure.

Another way of showing this three-dimensional structure is to make an 'edible' DNA, e.g. using different coloured jellybeans to represent the nitrogenous bases, the phosphate and the sugar. The jellybeans can be joined with toothpicks to represent the bonds holding the nucleotides together.

2-D models

Many models of DNA use jigsaw cut-outs of coloured paper or cardboard and align the pieces to give a two-dimensional view of the ladder and rung structure of DNA. There are DNA jigsaw kits that can be purchased to use in class or you can create your own jigsaw. The jigsaw pieces can also be used to show the process of DNA replication and protein synthesis.

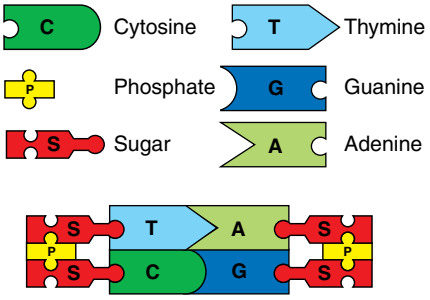


Figure 4.2 DNA jigsaw.

Evaluating a model of DNA

When evaluating a model the model must be consistent with known information. For example, one nucleotide of DNA must contain one phosphate, one deoxyribose sugar and one nitrogenous base, e.g. adenine (A), thymine (T), cytosine (C) or guanine (G). The nitrogenous bases must also be in complementary pairs, e.g. A-T and C-G.

An evaluation also needs to consider the advantages and the limitations of the model. Models of DNA are very useful in showing how a phosphate, a sugar and a nitrogenous base form a nucleotide. They also show how the nucleotides link together to form a ladder structure with the sugar-phosphates forming the sides of the ladder and the nitrogenous bases forming the rungs of the ladder. 3-D models have the added advantage that they can show how the ladder twists to form the double helix. 2-D cutout jigsaw models have the advantage that the two strands of the model can be easily separated to show the process of DNA replication.

Thus making a model of a section of DNA is an important tool in making the structure of DNA easier to understand giving a good visualisation of how each component fits into the molecule. The model can then be used as the bases for further knowledge about DNA, e.g. how DNA can replicate to form two identical double helix molecules and the model shows the genetic coding which is used to manufacture a particular polypeptide and protein.

Since proteins determine the different appearance and type of cells found in the body and also determine the reactions that occur in cells (enzymes are proteins), a section of DNA (gene) determines your appearance and phenotype.

QUESTIONS

1. Compare the structure of the DNA molecule in prokaryotic and eukaryotic cells.
2. Identify some ways in which DNA can be represented in models.
3. What are some features that must be shown in a model of DNA?
4. What is the triplet code?
5. Do all triplet codes specify a particular amino acid? Explain your reasoning.
6. (a) Which code is used in genetics in definitions and to identify amino acids?
   (b) What is the relationship between this code and the DNA code?
7. Draw a simple, fully labelled diagram of a part of a DNA molecule showing eight base pairs.
8. The table shows the percentage of adenine, guanine, cytosine and thymine in a series of DNA samples.

DNA source	Adenine	Guanine	Cytosine	Thymine
Yeast	33.2	16.1	16.9	33.8
Bacteria	26.9	22.7	23.0	27.4
Invertebrate	31.5	18.5	18.6	31.4

   Discuss how this data shows an important feature of the DNA molecule.
9. It has been found that cells from turtles contain, on average DNA with 30% adenine. From this information, how much guanine would you expect to find in the average turtle cell?
10. Give an advantage and a disadvantage of making 3-D models of DNA.
11. Explain why scientists construct models such as the model of DNA constructed by Watson and Crick.
12. DNA is a very regular polymer of nucleotides. How many different basic nucleotides are in DNA and what are they?
13. The diagram represents one nucleotide.
    (a) Identify the components X, Y and Z.
    (b) Draw a diagram to show how two more nucleotides would connect with this nucleotide.

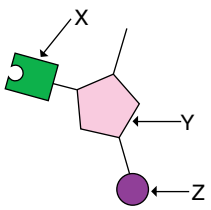


Figure 4.3
One nucleotide.

14. Describe a model you could construct to show the structure of the DNA molecule and assess the uses and limitations of this model.
15. What is a codon?
16. Explain why the coding of the base pairs in a section of DNA is important.
17. The diagram shows a section of DNA.

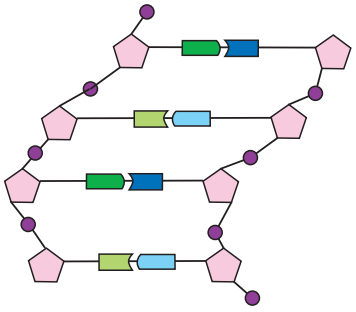


Figure 4.4 A section of DNA.

    Copy this diagram and circle one nucleotide.
18. A biology student needed to evaluate a model of DNA made by another student. For the model to be consistent with known information, where should the phosphates be located in the model?
    (A) On the left-hand side of the ladder.
    (B) Alternating with sugars down both sides of the ladder.
    (C) Alternating with nitrogenous bases down both sides of the ladder.
    (D) On the rungs of the ladder.
19. Watson and Crick deduced the double helix structure of DNA and created a model of the DNA molecule. Many scientists went to see this model and their model is on display in the Science Museum, London. What is the benefit of creating models?
    (A) Models help visualisation of scientific concepts.
    (B) Models can be used to make further predictions.
    (C) Models aid communication.
    (D) All of the above.

5 DNA in the Nucleus and Organelles

DNA is deoxyribose nucleic acid and is a double helix molecule that carries genetic information. Some organelles in eukaryotes contain DNA. The genes in these organelles are called **extranuclear genes**. These organelles can reproduce themselves. In humans only mitochondria contain extracellular genes.

There are significant differences between the DNA in the nucleus and the DNA in cellular organelles, although both follow the **universality of the genetic code** with adenine pairing with thymine and cytosine pairing with guanine as complementary pairs. The triplet codes in DNA for amino acids are also the same in both types of DNA.

Nuclear DNA (nDNA)

Nuclear DNA has two sets of each chromosome – one paternal from the sperm and one maternal from the egg. The DNA is linear and has both exons and non-coding introns in its coding.

Chromatin is the name given to the complex of DNA and protein that makes up chromosomes in the nucleus of eukaryotic cells.

Nuclear DNA has a bead-like structure called **nucleosomes** that each consist of a protein core of histones with DNA wound twice around the group of histones. The histone tails extend outward from each nucleosome core.

Histones are important in the structure and packaging of chromatin in the nucleus. Without histones the DNA of human cells (eukaryotic cells) would be very long compared to the size of the cell and histones also aid the ability for certain genes to be expressed in the cell while other genes stay dormant. The histone tails are accessible to the enzymes that begin gene expression and transcription to start protein synthesis.

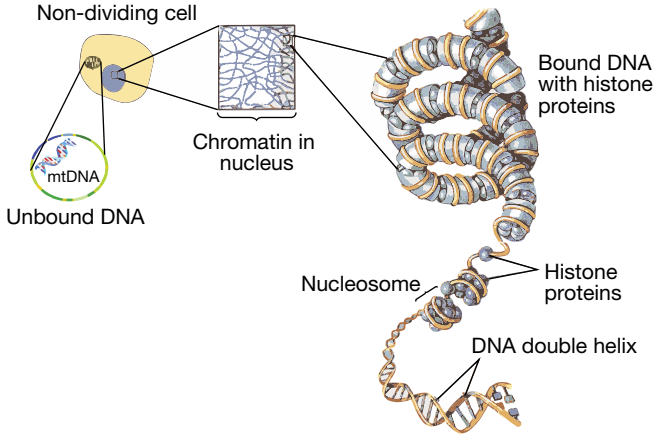


Figure 5.1 Bound DNA in the nucleus and unbound DNA in mitochondria.

Mitochondrial DNA (mtDNA)

Mitochondria are a cellular organelle that have their own DNA (mtDNA). mtDNA is circular, not bound to histone proteins and only encodes 37 genes. It is inherited with the egg which means it is only inherited from the mother.

Unlike nuclear DNA mtDNA does *not* have non-coding exons so that all the code is used in transcription and translation in protein synthesis.

Mitochondria are cylinder shaped organelles that vary in size from 2 to 8 micrometres in length. They are the site of aerobic respiration and generate most of the ATP in human cells. This means the number of mitochondria present in a cell depends on the energy needs of the cell.

Mitochondria have a double membrane with the inner membrane infolded to form **cristae**.

A mitochondrion can divide to form two mitochondria due to their circular DNA which resembles the DNA of prokaryotes. This means a mitochondrion can replicate itself even when the cell is not dividing. The dimensions of mitochondria are also similar to the size of prokaryotes.

Since the inheritance of mtDNA from one generation to the next does not involve both parents it has been used to track ancestry and evolutionary trees. In humans the analysis of mtDNA has been used to track the migration of different human populations following the female line back through many generations.

Endosymbiotic theory

The prokaryotic **endosymbiotic theory** suggests that mitochondria arose as a symbiotic relationship between a free living heterotrophic bacterium with aerobic respiration that entered a larger cell – either a larger prokaryote or an eukaryote and both then existed in a mutualistic partnership.

The endosymbiotic theory also explains the presence of unbound circular DNA in mitochondria in both plants and animals, and also the presence of DNA in plant plastids, e.g. chloroplasts.

The size of mitochondrial ribosomes is also the same size as ribosomes in prokaryotes rather than the same size as the ribosomes in the cytoplasm in human cells.

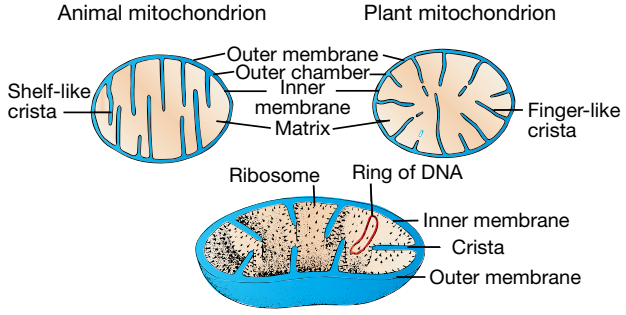


Figure 5.2 Plant and animal mitochondria.

QUESTIONS

1. What are extranuclear genes?
2. What is meant by the universality of the genetic code?
3. How is nuclear DNA inherited?
4. What is chromatin?
5. Describe a nucleosome.
6. Describe a benefit of DNA being bound to histones in nDNA.
7. Describe mitochondria.
8. Describe mitochondrial DNA.
9. How does the endosymbiotic theory apply to mitochondria?
10. Explain how human mtDNA has been used to trace human migration across the Earth.
11. Construct a table to compare mitochondrial DNA and nuclear DNA.
12. The diagram shows one proposal about the origin of mitochondria and chloroplasts.

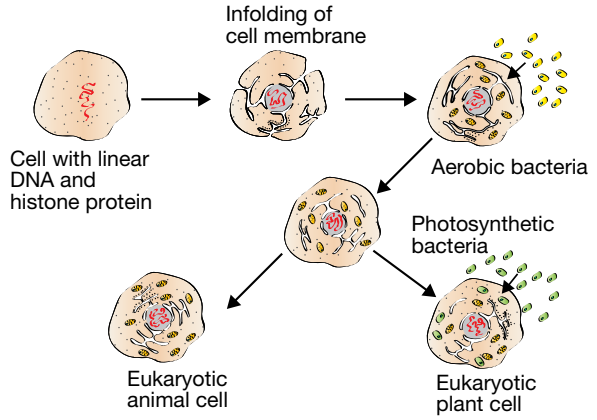


Figure 5.3 Origin of mitochondria and chloroplasts.

(a) Use this diagram to write out the sequence of events that has been proposed to explain the origin of human cells (eukaryotic animal cells).
(b) Explain how mitochondrial DNA and chloroplast DNA supports the endosymbiotic theory.

13. The diagram shows a DNA structure.

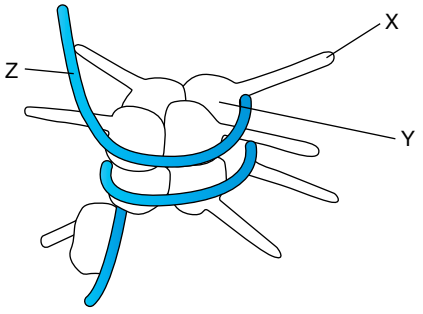


Figure 5.4 Origin of mitochondria and chloroplasts.

(a) Name the structure.
(b) Identify the parts X, Y and Z.

14. Which of the following has its own DNA?
    (A) Golgi apparatus.
    (B) Endoplasmic reticulum.
    (C) Mitochondria.
    (D) Lysosomes.
15. What is the structure of mitochondrial DNA?
    (A) Small circular ring.
    (B) Large circular ring with histone protein.
    (C) Linear chromatin.
    (D) Small circular ring linked with lipids.
16. Which of the following is the best description of chromatin?
    (A) Small protein with large number of negatively charged amino acids found in the nucleus.
    (B) Complex of DNA and protein that makes up chromosomes in the nucleus.
    (C) Circular DNA with few genes found in mitochondria.
    (D) One of two thread-like strands into which a chromosome divides longitudinally during cell division.

Use the diagram for the next TWO questions.

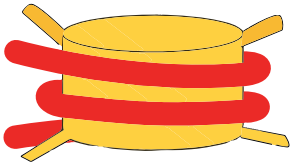


Figure 5.5 Biological structure.

17. What is the name of this structure?
    (A) Histone protein. (B) Chromatid.
    (C) Gene. (D) Nucleosome.
18. Where would you expect to find this structure in human cells?
    (A) Only in the nucleus.
    (B) Only in the mitochondria.
    (C) Only in Golgi apparatus.
    (D) In the nucleus and in mitochondria
19. In the cell cycle histones briefly leave DNA for a very short time. At which of the following steps would you expect this separation to occur?
    (A) During cytokinesis and division of the cytoplasm.
    (B) During DNA replication.
    (C) During metaphase when chromatids line up at the equator of the cell.
    (D) During anaphase when chromatids separate to become chromosomes going to separate poles.
20. What is meant by bound DNA?
    (A) DNA is restricted to being inside the nucleus.
    (B) DNA is in the process of protein synthesis.
    (C) DNA is wound around a protein core.
    (D) A protein is wound around a DNA core.

6 Genome, DNA and Bioinformatics

The genome can be considered to be the sum total of an organism's DNA measured in the number of base pairs contained in a haploid set of chromosomes.

DNA stands for deoxyribose nucleic acid.

The basic unit of DNA is the **nucleotide**. A nucleotide consists of a sugar, a phosphate and one of four nitrogenous bases – either adenine (A), guanine (G), cytosine (C) or thymine (T). To form the double helix, the sides of the ladder are made up of alternating sugar and phosphate molecules and the rungs consist of paired nitrogenous bases. Adenine always pairs with thymine and guanine always pairs with cytosine.

A **genome sequence** is the order of the As, Ts, Cs and Gs in the DNA code. The human genome has over 3 billion of these genetic letters in its coding. A **genome map** is less detailed than a genome sequence identifying short DNA sequences as markers in a genome.

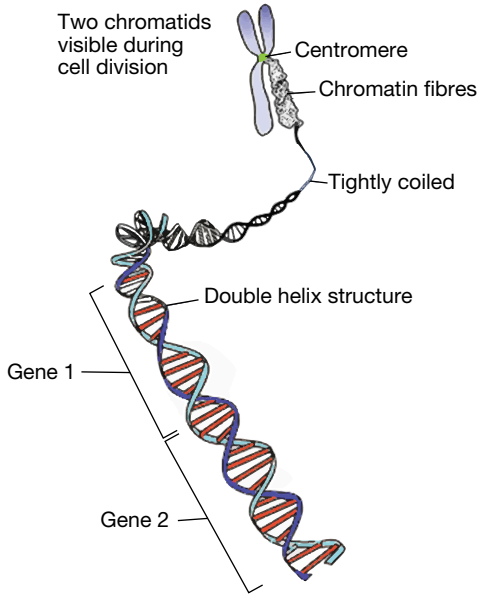


Figure 6.1 DNA.

DNA has a **double helix** structure where two spirals coil around each other keeping a constant diameter of the coil.

Bioinformatics

Bioinformatics uses computers, software and mathematical models to process and integrate biological information from large data sets. Bioinformatics is used to study and analyse genome sequences to help determine the genetics of disease, to understand gene expression and protein synthesis, to determine the effects of mutations, to understand the processes involved in the formation of tumours and cancers and to trace evolutionary changes in DNA and determine evolutionary relationships between different species.

QUESTIONS

1. What is the genome?
2. What does 'DNA' stand for?
3. Describe the shape of DNA.
4. What is a nucleotide?
5. The diagram shows a short section of part of a DNA molecule.

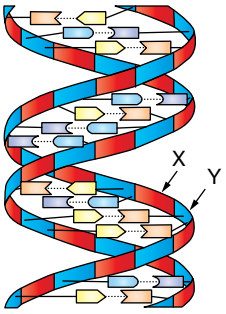


Figure 6.2 Part of a DNA molecule.

Identify parts X and Y.

6. Identify the base pairs of DNA.
7. Distinguish between a genome sequence and a genome map.
8. What is bioinformatics?
9. How is bioinformatics used with genome sdquences?
10. Which of the following identifies the basic components of a nucleotide?
    (A) DNA and protein.
    (B) Adenine, thymine, cytosine, guanine.
    (C) Glucose, nitrate, phosphate base.
    (D) Sugar, phosphate, nitrogenous base.
11. What forms the 'rungs' of the DNA molecule?
    (A) Phosphate and sugar molecules.
    (B) One phosphate and one nitrogenous base.
    (C) Paired nitrogenous bases.
    (D) One phosphate and one sugar.
12. The diagram shows a short section of DNA.

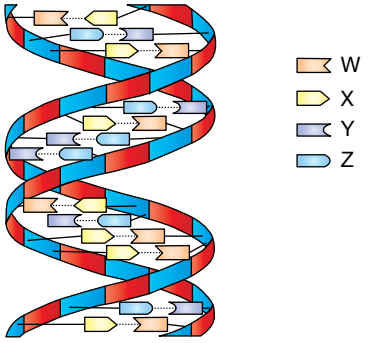


Figure 6.3 Short section of DNA.

What could be represented by X and Y?
(A) Thymine and cytosine.
(B) Adenine and thymine.
(C) Phosphate and sugar.
(D) Cytosine and phosphate.

7 DNA Replication

Watson and Crick predicted that DNA replication involved the **semiconservative model** which involves each of the two daughter molecules having one old strand from the parental molecule and one newly made strand.

In the 1950s there were three models proposed for DNA replication, the conservative model where the parental strands reassemble after forming a template that forms a new double helix, the semiconservative model as proposed by Watson and Crick and the dispersive model where both the daughter molecules contain a mixture of old and newly synthesised DNA.

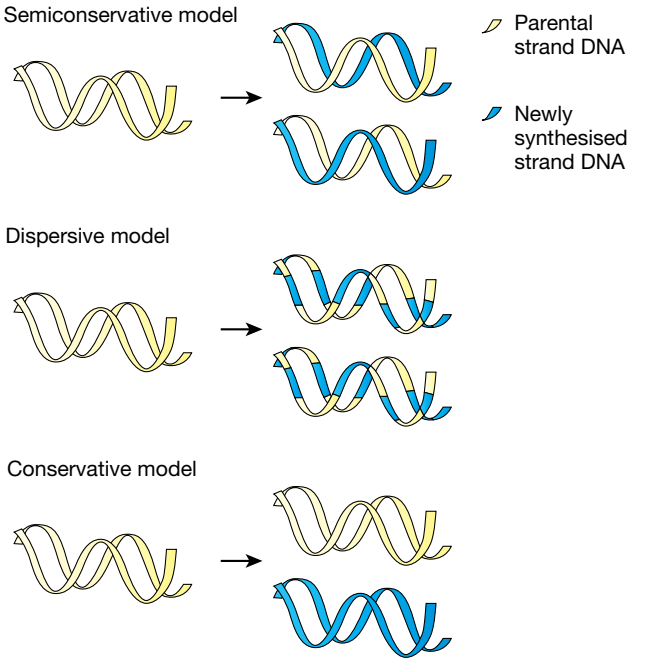


Figure 7.1 Three proposed models for DNA replication.

Meselson-Stahl experiment

In 1958 Matthew Meselson and Franklin Stahl carried out an experiment with the bacteria *Escherichia coli* labelled with radioactive nitrogen (^{15}N) to investigate the three models of DNA replication. They found that the amount of ^{15}N in the each generation after cell division and replication over several generations was consistent with the semiconservative model and confirmed the model of semiconservative replication proposed by Watson and Crick.

Process of replication

During DNA replication the DNA molecule unwinds and the double helix separates into two single strands. Each single strand is a template for the synthesis of a complementary strand so that adenine is opposite thymine and guanine is opposite cytosine.

The process of replication begins at **origin of replication** sites along a chromosome. These sites are sections that coded with a specific sequence of nucleotides. Proteins that start DNA replication detect and attach to these sections causing the two strands to separate to form a replication 'bubble'. There is a **replication fork** at the end of each bubble where the unwinding process begins and the bubble opens up further in both directions. **Helicase enzymes** untwist the double helix at the replication forks and break the bonds between the two strands of DNA. The separated sections in the bubble act as templates for new complementary strands.

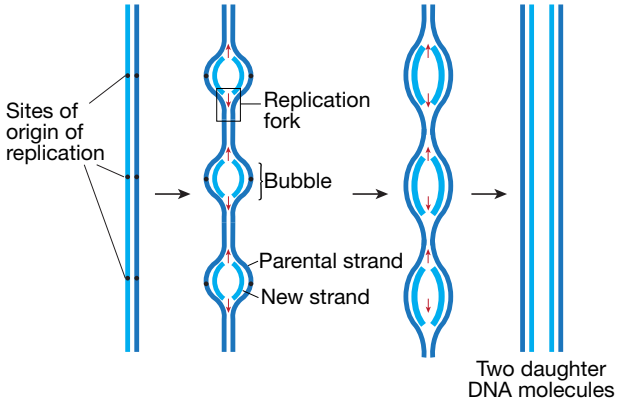


Figure 7.2 DNA replication begins at the sites of origin of replication.

The beginning nucleotide chain is a short RNA chain called a **primer** and is synthesised by the enzyme **RNA primase**. Once the primer has formed **DNA polymerase enzymes** add nucleotides to the end of the existing chain. Nucleotides can only be added to the 3′ end of the growing DNA strand. This means the new strand always grows in a 5′ → 3′ direction.

The **leading strand** is the new DNA strand that is **continuously** synthesised in the 5′ → 3′ direction. The other strand is called the **lagging strand** and is also synthesised in the 5′ → 3′ direction but grows **away** from the replication fork and forms **discontinuously** in a series of segments. These segments are called **Okazaki fragments**. DNA ligase enzymes join the sugar-phosphate backbones of the Okazaki fragments together to complete the new single DNA strand.

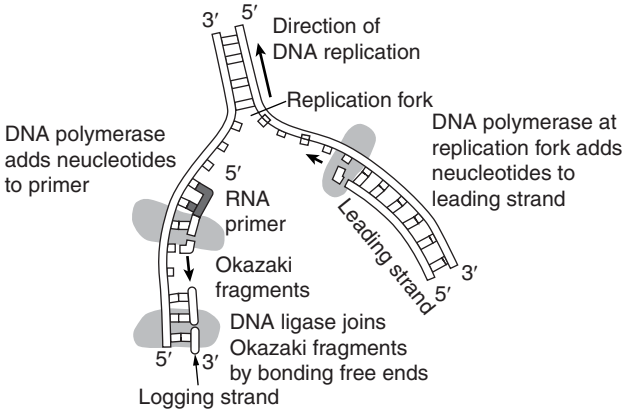


Figure 7.3 DNA replication occurs in 5′ → 3′ direction.

The cytoplasm of the cell contains many free nucleotides which are used during DNA replication. DNA polymerase proofreads each nucleotide against its template removing incorrect nucleotides or catalysing the formation of bonds to join the correct nucleotide to the DNA strand. If DNA polymerase while proofreading misses a set of mismatched nucleotides other repair enzymes can remove and replace the incorrect pair.

The significance of DNA replication is that large amounts of coded information can be copied and passed onto the next generation, providing continuity of a species. The method also allows for some changes and for mutations when the code is incorrectly copied.

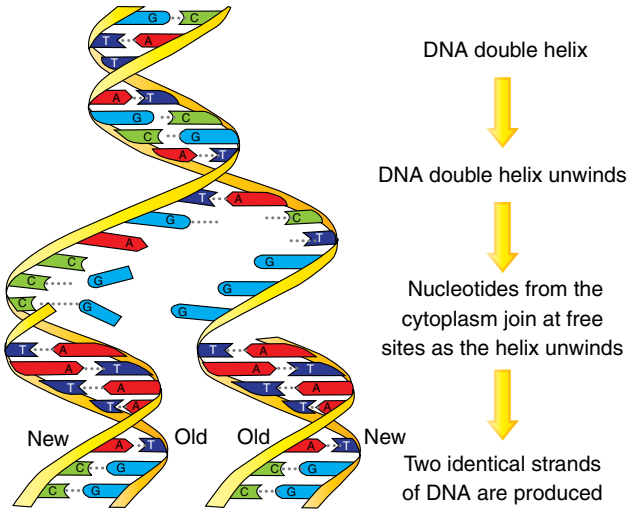


Figure 7.4 DNA replication.

QUESTIONS

1. Which model of DNA replication was proposed by Watson and Crick?
2. Construct a table to summarise the three models of DNA replication that were proposed in the early 1950s.
3. Describe the experiment that eventually showed the how DNA replicated.
4. The diagram shows a simple summary of the process of DNA replication.

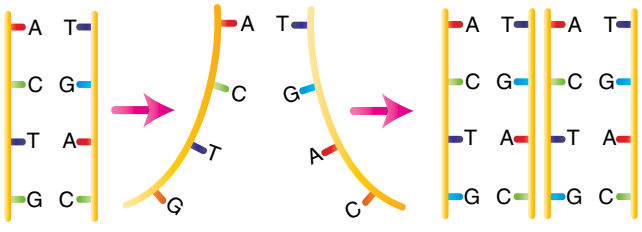


Figure 7.5 Simple summary of DNA replication.

Briefly explain what is happening in this diagram.

5. What is an origin of replication site?
6. What is a replication 'bubble'?
7. Outline the function of helicase enzymes.
8. (a) What is a primer?
   (b) What is the function of RNA primase?
9. Outline the function of DNA polymerase enzymes.
10. Distinguish between the leading strand and the lagging strand in DNA replication.
11. What are Okazaki fragments?
12. Outline the function of DNA ligase enzymes.
13. Explain the significance of DNA replication.
14. The diagram shows several stages of DNA replication.

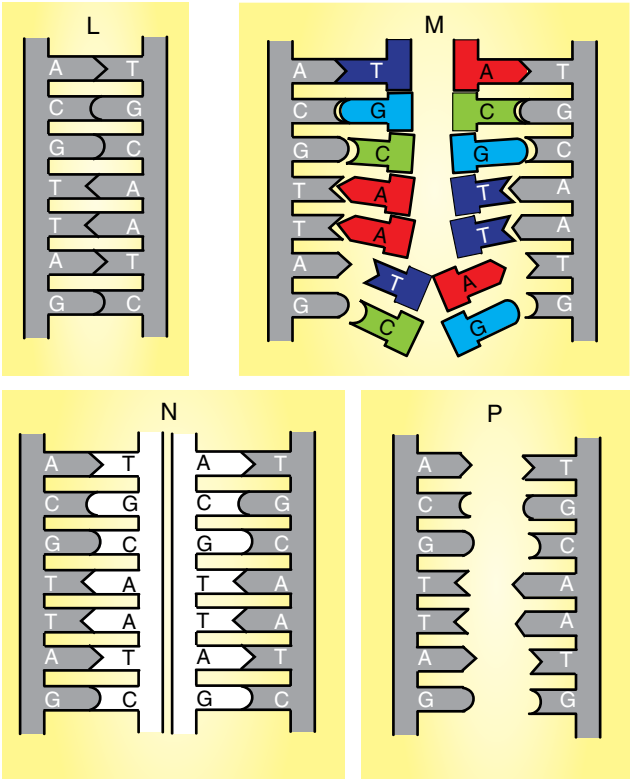


Figure 7.6 Stages of DNA replication.

Identify the correct sequential order.

(A) L, P, N, M
(B) L,M, N, P
(C) N, P, M, L
(D) L, P, M, N

15. The diagram shows a DNA replication bubble.

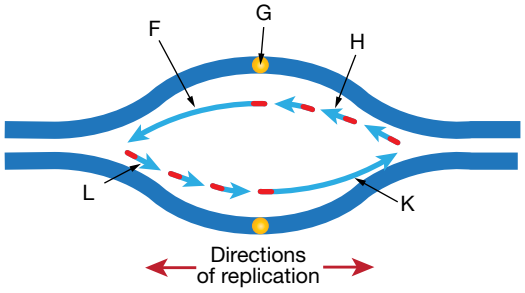


Figure 7.7 DNA replication bubble.

Which arrow(s) point to the leading strand in DNA replication?

(A) F only.
(B) H only.
(C) H and L.
(D) F and K.

Answers

1 Assumed Knowledge

1. (a) Mitosis is a process during cell division in which the cell nucleus divides into two.
   (b) Meiosis is cell division to produce haploid daughter cells.
   (c) Meiosis produces four daughter cells.
2. Information is transferred as DNA on chromosomes when cells reproduce themselves.
3. DNA stands for deoxyribose nucleic acid.
4. The basic unit of DNA is the nucleotide.
5. Most DNA is located in the nucleus. DNA is also found in mitochondria.
6. The structure of the DNA molecule is a double helix.
7. A gene is a certain length of DNA that has the code for one characteristic.
8. DNA needs to be able to replicate itself exactly so that cell division can form identical new cells for growth, repair and maintenance of the body of a multicellular organism. Exact replication is also needed to maintain the genetic code for a species and hence keep its integrity as a distinct unit in nature.
9. A mutation is a change in the chemical structure of the DNA.
10. A pedigree is a graphical way of picturing the ancestry of living things. It shows genetic history.
11. System A is the female reproductive system and system B is the male reproductive system.
12. Biotechnology is the use of biological processes by industry or agriculture to change organisms in order to produce useful products or provide services.
13. A somatic cell is a body cell.
14. A stem cell is a relatively unspecialised cell that can divide by cell division to produce identical daughter cells and to differentiate to form different specialised cells.
15. A germ layer is a group of cells in an embryo that will give rise to tissues and organs of the body.
16. Human gestation is on average 266 days (approximately 38 weeks). Since timing is related to the menstrual cycle it is 40 weeks since the start of the last menstrual cycle.
17. Parturition means giving birth to a baby.
18. It is a sperm and it is the male reproductive sex cell (gamete).
19. An embryo is the early stage of development and in humans after 6 to 8 weeks when all the organs have formed the embryo is called a foetus.
20. A mutagen is a chemical or physical agent that causes a mutation changing DNA and the genetic information.
21. Gregor Mendel experimented with pea plants and worked out the basic laws of inheritance. His work led to the study of genetics and hence he is often referred to as the 'father of genetics'.
22. Both genes and environmental factors determine the features of an organism.
23. Many different examples can be used to show how the environment influences the appearance of a person. If a person changes their diet, the amount and type of exercise they do every day and their exposure to UV sunlight their appearance can change with body shape, weight, muscle tone and skin colour with sunburn and tanning.
24. Studying eye colour with two alleles for a particular gene – brown eyes (B) and blue eyes (b). Given that all other environmental factors are the same, a person with the genetic code BB or Bb will have brown eyes, while a person with the code bb will have blue eyes.
25. Watson and Crick discovered that DNA had a double helix structure.
26. The genome is the complete genetic information of an organism.
27. A chromosome is a cellular structure that holds genetic information in the coding of the DNA molecule.
28. Genotype is the genetic make-up of an organism, or a set of alleles of an organism.
29. Fertilisation is the union of two gametes.
30. Gametes fuse to form a zygote. It is essential that gametes contain only half the number of chromosomes to maintain the chromosome number of the species. Otherwise the number of chromosomes would double every generation.
31. Part A is the ovary which produces eggs (female gamete) and part B is the uterus which is where the embryo/foetus will grow and develop until birth.

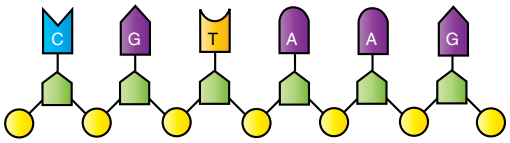
2 Discovering DNA

1. Nucleic acids were first identified in 1869 and were called 'nuclein' as they came from the nucleus.
2. When Frederick Griffith mixed the remains of dead pathogenic (disease-causing) bacteria with living non-pathogenic bacteria he found that some of the living bacteria transformed into pathogenic bacteria and that their offspring were pathogenic. He called the phenomena bacterial transformation.
3. (a) A nucleotide is the basic unit of DNA and consists of a phosphate, sugar and base.
   (b) Phoebus Levene named nucleotides.
   (c) When nucleotides were first named it was commonly believed that the carrier of genetic information was proteins.
4. William Astbury used X-ray diffraction to study DNA molecules and found that the structure repeated every 2.7 nanometres and that the bases were arranged in flat stacks that were 0.34 nanometres apart. Data like this is crucial in determining the structure of a molecule as it provides the dimensions for any proposed model.
5. Beadle and Tatum carried out experiments with the bread mould *Neurospora* and identified the link between genes and enzymes proposing the one gene – one enzyme hypothesis.
6. Chargaff's rules for the composition of DNA are: 1. In natural DNA the number of guanine units is the same as the number of cytosine units and the number of adenine units is the same as the number of thymine units. 2. The number of units of adenine, thymine, cytosine and guanine units is different for different species.
7. The Hershey-Chase experiment used two sets of bacteriophages, one with proteins labelled with radioactive sulfur (using ^{35}S) and one with DNA radioactively labelled with phosphorus (using ^{32}P) to infect *Escherichia coli* which is a bacteria that lives in the intestines of mammals. They found that the radioactively labelled DNA entered the bacteria cells while the radioactively labelled proteins did not enter the bacteria cells. This confirmed that DNA and not protein was the genetic material.
8. Rosalind Franklin took X-ray diffraction images of DNA that clearly showed the helical structure of DNA. The evidence in her photograph was used by Watson and Crick to finally determine the double helix structure.
9. (a) In 1953, Watson and Crick suggested the double helix structure of DNA with two phosphate-sugar strands winding around the outside of nitrogenous bases with the pairs A-T and C-G. Rosalind Franklin took X-ray diffraction photographs of DNA. Maurice Wilkins was Franklin's colleague and showed her X-ray photographs to Watson. The work of all these scientists needed to be put together as each piece of information and evidence contributed to the final determination of the structure of DNA.
   (b) Watson and Crick collaborated to produce the final double helix structure and Wilkins collaborated when he showed the X-ray diffraction photographs to Watson which led to an understanding of the physical shape of the molecule. For this joint effort the three men were awarded the 1962 Nobel Prize for Physiology. Franklin took the photographs but there was friction between her and her colleague Wilkins. She died of cancer in 1958 and could not be given credit posthumously for the Nobel Prize.

10. In the early 1950s the three groups – Wilkins and Franklin at the University of London, Pauling at Caltech and Watson and Crick at Cambridge University – were all trying to determine the structure of DNA. They were competing to be the first to unravel the mystery about how genetic information is coded and able to be passed from one generation to the next. It was a scientific race. A number of scientists had collected experimental evidence and information about the structure of DNA, e.g. Astbury had determined the dimension of the repeating structure and Chargaff's rules showed there were equal amounts of adenine and thymine and equal amounts of cytosine and guanine. The three groups were trying to collect the final piece of evidence that would show how all the pieces of information fitted together. There was competition but there was also collaboration, e.g. Wilkins showed Watson and Crick Franklin's photograph and the structure of DNA was able to be finally determined.
11. (a) A bacteriophage is a virus that can infect bacteria.
    (b) Bacteriophages were used in the Hershey-Chase experiment to determine that DNA was the carrier of genetic information and not protein. Bacteriophages labelled with radioactive DNA ie with radioactive phosphorus (^{32}P) and other bacteriophages labelled with radioactive protein ie with radioactive sulfur (^{35}S) were allowed to infect bacteria. Since bacteriophages can inject their DNA into bacteria and their protein coat stays outside the bacteria, the experiment with bacteriophages proved that it was the DNA that was injected into the bacteria to cause bacterial transformation and not the protein.
12. (a) Molecule is DNA – deoxyribose nucleic acid.
    (b) L = phosphate, M = sugar, N = nitrogenous base.
    (c) Watson and Crick.
    (d) DNA encodes a large amount of hereditary information in the sequences of the nitrogenous bases. It is able to replicate itself with the complementary pairs AT and CG. Mutations or errors can occur during replication, e.g. base-pair substitution, which is a source of genetic variation.
13. C
14. A
15. B
16. B

3 The Structure of DNA

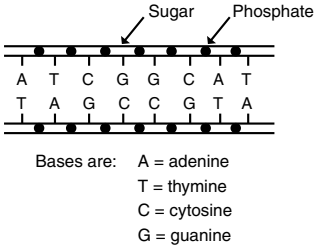
1. A gene is a certain section of DNA in a chromosome that represents a particular characteristic.
2. DNA stands for deoxyribose nucleic acid.
3. A major advance in scientific understanding occurred when in 1944 Oswald Avery demonstrated that genes were made of DNA. This directed future biological research into the structure of DNA, leading to Rosalind Franklin, with Maurice Wilkins, using X-ray diffraction crystallography in 1952 to show that the DNA molecule was helical. This major advance then led to James Watson and Francis Crick proposing in 1953 that DNA molecules had a double helix structure. The work of each scientist built upon the work of another scientist to unravel the mystery of the structure of DNA.
4. DNA is a double helix or twisted ladder shape.
5. A nucleotide is the basic unit of DNA. It consists of a sugar, a phosphate and a nitrogenous base.
6. The phosphate group gives the nucleotide its acidic properties.
7. Adenine pairs with thymine and cytosine pairs with guanine.
8. 1. DNA is able to encode a large amount of information. 2. It is chemically stable. 3. It is able to make an accurate replication of itself. 4. It controls and directs protein synthesis. 5. Occasional errors (mutations) occur.
9. Covalent bonds called phosphodiester linkages hold the sugar of one nucleotide to the phosphate group of the next nucleotide.
10. Hydrogen bonds hold the nitrogenous bases on the rungs of the ladder together.
11. The two stands of DNA are called antiparallel a one sugar-phosphate backbone runs 5′ → 3′ direction while the other strand runs 3′ → 5′ direction.
12. The hydrogen bonds holding the nitrogenous bases together on the rungs of the ladder break during DNA replication.
13. Each gene is a certain section of a chromosome which means it is a certain length of the DNA molecule. The coding of the DNA molecule passes hereditary information from one generation to the next.
14. The significance of DNA replication is that large amounts of coded information can be copied and passed onto the next generation, providing continuity of a species. The method also allows for some changes (mutations).
15. (a) Part X is phosphate, Part Y is sugar.
    (b)



    (c) The segment of DNA has the base code GCATTC. The messenger RNA uses the DNA as a template, with uracil replacing thymine, forming the code CGUAAG. mRNA moves from the nucleus to the cytoplasm and attaches to a ribosome. The sequence of codons along mRNA is translated into amino acids by tRNA, e.g. CGU forms one amino acid and AAG forms the next amino acid.
16. B
17. A
18. B
19. B
20. D
21. C
22. A

4 Activity – Making a Model of DNA

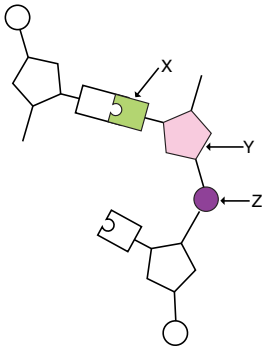
1. In eukaryotic cells each chromosome is a long linear DNA molecule. In prokaryotic cells there is a single circular DNA molecule.
2. There are many ways you can construct a model to show the structure of DNA. Models can be made of paper, cardboard, pipecleaners, plastic sticks, coloured balls or created using multimedia graphics.
3. A model of DNA must show: 1. One nucleotide of DNA must contain one phosphate, one deoxyribose sugar and one nitrogenous base, e.g. adenine (A), thymine (T), cytosine (C) or guanine (G). 2. The nitrogenous bases must also be in complementary pairs, e.g. A-T and C-G. 3. The nucleotides need to link together to form a ladder structure with the sugar-phosphates forming the sides of the ladder and the nitrogenous bases forming the rungs of the ladder.
4. The triplet code is three bases next to each other in a molecule of DNA and specifies a particular amino acid or instruction.
5. Not all triplet codes specify an amino acid. There are three codes that are 'stop' signals – DNA triplet code ATT, ATC and ACT. The DNA code TAC is the code for methionine but also a 'start' signal.
6. (a) The mRNA code is used in definitions and to identify amino acids.
    (b) The mRNA code is complementary to the DNA code.
7. A fully labelled diagram of DNA with eight base pairs:

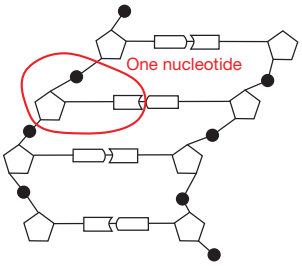


8. The data shows the pairing of the nitrogenous bases in the DNA molecule. The percentage of the bases shows that adenine only pairs with thymine and cytosine only pairs with guanine.
9. If the DNA from a turtle cell contains 30% adenine, then it must contain 30% thymine since A-T are complementary pairs. This totals 60% DNA. The rest of the DNA (40%) must be C-G. Thus guanine would be 20% DNA of a turtle cell.
10. An advantage of making a 3-D model of DNA is that you can see the double helix structure as the two strands twist around each other. A disadvantage of making a 3-D model of DNA is that it is not particularly easy to use to show DNA replication and proteins synthesis as when you try to separate the complementary nitrogenous bases to separate the two strands the structure becomes unstable and tends to fall apart.
11. Scientists construct models, e.g. Watson and Crick constructed a 3-D model of DNA as a visual representation of a molecule that could not be directly seen at that time. The model made it easier for Watson and Crick to explain their ideas to other people and to show how DNA could hold a vast amount of genetic information and could also replicate to pass information to the next generation. A 3-D model of DNA shows how each component of a nucleotide links to each other component and the model could be used to explain other processes, e.g. DNA replication and protein synthesis.
12. There are four basic nucleotides in DNA.

Nucleotide	Description
1	Phosphate – sugar – adenine
2	Phosphate – sugar – thymine
3	Phosphate – sugar – cytosine
4	Phosphate – sugar – guanine

13. (a) X = nitrogenous base, Y = sugar, Z = phosphate
    (b) Diagram with two other nucleotides attached:



14. To show the structure of DNA, you can use jigsaw cardboard cut-outs with different shapes and colours for the nitrogenous bases, adenine, thymine, cytosine and guanine and a different shape for the sugar and for the phosphate. This model is useful as it shows the pairing of the DNA bases (A-T and C-G) to form the rungs of the ladder of the double helix and it also shows how the arrangement of the sugars and phosphates forms the sides of the ladder. The model has limitations as it is only a two-dimensional model of a molecule that is three-dimensional and it does not show how the structure is twisted to form the double helix. Making a model is important as it means a complex structure, too small to be seen by the naked eye, can be simplified and studied more easily. However, relative size can be misrepresented and there is no information about the bonding between nucleotides and within the nucleotides.
15. A codon, or triplet, is the basic instruction of the genetic code consisting of three bases next to each other in a molecule of DNA or mRNA and specifying a particular amino acid.
16. The sequence of base pair coding in a section of DNA is important because the sequence of codons gives instructions for the sequence of amino acids which will be joined together to form a polypeptide. Since proteins determine the different appearance and type of cells found in the body and also determine the reactions that occur in cells a section of DNA is very important as it determines your appearance and phenotype.
17. 

18. B
19. D

5 DNA in the Nucleus and Organelles

1. Extranuclear genes are genes found in organelles in the cytoplasm of eukaryotes.
2. The universality of the genetic code means that DNA is similar in its coding of information with the nucleotide complementary pairing – adenine pairing with thymine and cytosine pairing with guanine. The triplet codes in DNA for amino acids are also universal
3. A person inherits nuclear DNA with one set of chromosomes from their father and one set of chromosomes from their mother.
4. Chromatin is the name given to the complex of DNA and protein that makes up chromosomes in the nucleus of eukaryotic cells.
5. Nucleosomes are bead-like structures with a protein core of histones and DNA wound twice around the group of histones.
6. Histones are important in the structure and packaging of chromatin in the nucleus. Without histones the DNA would be very long compared to the size of the cell. Histones also aid the ability for certain genes to be expressed in the cell while other genes stay dormant as the histone tails are accessible to the enzymes that begin gene expression and transcription to start protein synthesis.
7. Mitochondria are cylinder shaped organelles that are the site of aerobic respiration. They have a double membrane with the inner membrane infolded to form cristae.
8. mtDNA is a ring of DNA similar to DNA found in prokaryotes.
9. The endosymbiotic theory suggests that mitochondria arose as a symbiotic relationship between a free living heterotrophic bacterium with aerobic respiration that entered a larger cell – either a larger prokaryote or an eukaryote and both then existed in a mutualistic partnership.
10. Human mtDNA is inherited through the female line as the mitochondria are in the egg. This means that scientists have been able to compare the nucleotide sequences of different populations of humans to determine the possible timelines and paths of human migration across the Earth.
11.

Nuclear DNA	Mitochondrial DNA
DNA in the nucleus (nDNA) of human cells (eukaryotic cells) is bound to proteins to form bead-like structures called nucleosomes and the nucleosomal chromatin is organised to form chromosomes.	Mitochondrial DNA (mtDNA) is not bound to proteins – it lacks histones.
nDNA has two sets of each chromosome – one paternal and one maternal.	In humans mtDNA is maternal.
nDNA has many more genes in its structure than mtDNA.	mtDNA is relatively small (16 500 bases).
Has non-coding exons.	mtDNA is circular and does not have the non-coding regions (exons) found in nDNA.

12. (a) The diagram suggests that the sequence of events that led to the origin of mitochondria and chloroplasts were: 1. There was a cell with linear DNA and histone protein. 2. There was infolding of the cell membrane which led to the formation of endoplasmic reticulum and the nuclear membrane. 3. Aerobic bacteria were engulfed and retained within the cytoplasm living in a mutualistic relationship with the host cell. 4. Some of these cells went on to become eukaryotic animal cells. 5. Other cells engulfed photosynthetic bacteria which were retained in the cytoplasm living in a mutualistic relationship with the host and the host became eukaryotic plant cells.
    (b) Mitochondrial DNA and chloroplast DNA are both circular rings of DNA that are similar to prokaryotic DNA.
13. (a) The structure is a nucleosome.
    (b) Part X shows a histone tail. Part Y shows the histone protein and part Z is the double helix of the DNA molecule.
14. C
15. A
16. B
17. D
18. A
19. B
20. C

6 Genome, DNA and Bioinformatics

1. The genome is the sum total of an organism's DNA measured in the number of base pairs contained in a haploid set of chromosomes.
2. DNA stands for deoxyribose nucleic acid.
3. DNA is a double helix or twisted ladder shape.
4. A nucleotide is the basic unit of DNA. It consists of a sugar, a phosphate and a nitrogenous base.
5. Part X is phosphate, part Y is sugar.
6. The base pairs of DNA are A-T (adenine always pairs with thymine) and C-G (cytosine always pairs with guanine).
7. A genome map is less detailed than a genome sequence identifying short DNA sequences as markers in a genome. A genome sequence is the complete order of the As, Ts, Cs and Gs in the DNA code.
8. Bioinformatics uses computers, software and mathematical models to process and integrate biological information from large data sets.
9. Bioinformatics is important in genome sequencing as the computers, software and mathematical models can manipulate and analyse the large data sets of the genome sequences to help determine the genetics of disease, to understand gene expression and protein synthesis, to determine the effects of mutations, to understand the processes involved in the formation of tumours and cancers and to trace evolutionary changes in DNA and determine evolutionary relationships between different species.
10. D
11. C
12. B

7 DNA Replication

1. Watson and Crick proposed the semiconservative model of DNA replication.
2.

Model of DNA replication	Process
Semiconservative model	Each of the two daughter molecules have one old strand from the parental molecule and one newly made strand.
Daughter cells are haploid	The parental strands reassemble after forming a template that forms a new double helix.
Daughter cells are different to parent cells	Both daughter molecules contain a mixture of old and newly synthesised DNA

3. The Meselson-Stahl experiment showed that DNA replication followed the semiconservative model. In this experiment *Escherichia coli* labelled with radioactive nitrogen (^{15}N) was used to investigate the three models of DNA replication. Matthew Meselson and Franklin Stahl found that the amount of ^{15}N in the each generation after cell division and replication over several generations was consistent with the semiconservative model and not with the other two models – conservative model and dispersive model.
4. The DNA molecule unwinds and the double helix separates into two single strands. Each single strand is a template for the synthesis of a complementary strand so that adenine is opposite thymine and guanine is opposite cytosine. Two new daughter DNA molecules are produced.
5. An origin of replication site is section of DNA with a specific code where the process of replication begins. There are many origin of replication sites along a chromosome.
6. A replication 'bubble' is a section of DNA where replication has begun and the two strands have separated to form a 'bubble'.
7. Helicase enzymes cause the bonds between the two strands of DNA to break and the double helix to untwist.
8. (a) A primer is a short RNA chain with a free 3′ end that is bound by complementary base pairing to the template strand to which nucleotides are added in DNA replication.
    (b) RNA primase is an enzyme that joins RNA nucleotides to make a primer during DNA replication using the parental DNA strand as a template.
9. DNA polymerase enzymes catalyse the addition of nucleotides to the 3′ end of an existing DNA chain. They proofread each nucleotide pair and remove and replace any incorrect nucleotide.
10. The leading strand is the new DNA strand that is continuously synthesised and goes towards the replication fork. The lagging strand is discontinuously synthesised and grows away from the replication fork.
11. Okazaki fragments are short sections of DNA that are synthesised on the lagging strand and become linked together by ligase enzymes to complete a new single DNA strand.
12. DNA ligase enzymes catalyse the joining of DNA fragments joining the sugar-phosphate backbone of DNA molecule with covalent bonds.
13. The significance of DNA replication is that large amounts of coded information can be copied and passed onto the next generation, providing continuity of a species. The method also allows for some changes (mutations).
14. D
15. D

8 DNA Repairs Itself

1. DNA exposed to mutagens, radiation etc becomes damaged and needs to be repaired.
2. Two causes of DNA damage include: 1. Certain wavelengths of radiation, e.g. X-rays and gamma rays. 2. Chemical mutagens, e.g. some hydrocarbons found in cigarette smoke and nitrogen mustard (the poisonous part of mustard gas).
3. DNA can be damaged in several ways: 1. One of the nitrogenous bases in DNA is modified. 2. Mismatching of normal bases in DNA. 3. Crosslinks between DNA bases either on the same DNA strand or between different DNA strands. 4. Breaks in the backbone.
4. UV radiation can be absorbed by the bases in DNA. The changes in DNA caused by UV can lead to mutations, lesions, and can be lethal if not repaired. People with the hereditary disease xeroderma pigmentosum (XP) have XP cells with defective DNA repair. This means the sections of DNA damaged by UV are not able to be removed and replaced. These people will then have mutations, lesions, and could die if overly exposed to UV light.
5. (a) The thymine dimer distorts the DNA molecule and will block transcription and thus stop DNA replication.
    (b) Enzymes involved in the DNA repair process include a glycosylase to cut the damaged DNA, a DNA polymerase to replace the section and a DNA ligase to seal the section.