



Understanding the basics of biochemistry, DNA, genetics, disease, drugs and immunity for everyone.

An introduction to the basic sciences and new discoveries
that underpin health and disease for all health care
practitioners and students of science.

Dr. Peter H. Kay, PhD

Foreword.

The author of this educational article, Dr. Peter H Kay, spent many years in academia in Australia as a teacher and researcher in a wide range of medical and biological sciences. He has published over 80 articles in world class scientific publication outlets. He has taught students of all ages including high school students, undergraduates, medical students, post-graduate students and PhD students. He has also taught many medical science based subjects to complementary healthcare practitioners worldwide. Dr Kay currently runs the successful Homeovitality Natural Super Health Clinic in Preston, UK.

From his teaching experiences over the years, he has identified key areas with which many students have had difficulties coming to terms with. In this unique publication, he has addressed all these student difficulties. He has introduced the basics of genetics, including pharmacogenetics and nutrigenetics and genetic diseases, biochemistry, cell biology, molecular biology, immunity and immunopathology in an easy to read format.

Because the subject matter of this book is designed to understand more about health and disease, it is of great benefit to all health care practitioners such as nurses, paramedics and other hospital staff as well as all complementary health carers. Because it is science based it is also of great benefit to a wide range of science students from all secondary and tertiary educational institutions.

At the end of each chapter, there is a series of multiple choice questions that enable the reader to carry out their own self-assessment programmes.

THIS SAMPLE BOOK ONLY CONTAINS SELECTED PAGES, QUESTIONS AND ANSWERS. FOR THE FULL BOOK, PLEASE GO TO WWW.HOMEOVITALITYUK.COM/EBOOKS, TO BUY AND READ THE FULL BOOK INSTANTLY.

“Understanding the basics of biochemistry, DNA, genetics, disease, drugs and immunity for everyone” by Dr P H Kay.
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The Purpose.

The genomics revolution has begun. This compact educational book has been developed as a conventional teaching aid or as a self-teaching aid to enable everyone from the age of 5 to 105, including science students of all ages, to learn about the basic sciences that underpin the genomics revolution and understand more about the science of life to any level.

This book is also designed to help healthcare practitioners such as nurses, paramedics and complementary healthcare workers to understand more about the way in which a wide range of scientific discoveries affect health and well-being.

It is now possible to determine the complete sequence of anyone's DNA in a matter of days at a cost within most people's reach. This technological breakthrough is about to have a dramatic impact on everyone's everyday life, not only from a health standpoint but in many other ways. For example, DNA will soon be used to add another layer of computing capability. Those with an interest in information technology will appreciate the benefits of this book. Also, it will soon be possible to have a complete personal genetic blueprint stored in mobile communication systems such as iPads and Android devices. This has important implications for optimisation of personal health care strategies and prediction of social compatibility.

It begins with an introduction to the atom, to molecules, to biochemistry, through DNA (the genetic blueprint as known today) and RNA (how proteins are made), disease genetics, how drugs work, why drugs help some people whilst others are killed or maimed. Breast cancer is used to demonstrate the significance of this point.

It also introduces the reader to the basics of immunity and the evolution of the allergic response. Finally, it touches on the interaction between food and the genetic blueprint. This is important because it begins to explain the old adage "one man's meat is another man's poison".

To develop this booklet, I have drawn upon my many years of teaching and research within the medical sciences at the University of Western Australia, particularly with respect to disease genetics.

It is designed to help everyone learn more about a wide range of basic sciences that can be used to improve quality of life and understand much more about the world at present and what's to come.

Best wishes,

Dr. Peter H Kay, PhD.

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Introduction to atoms, elements, periodic table and molecules.

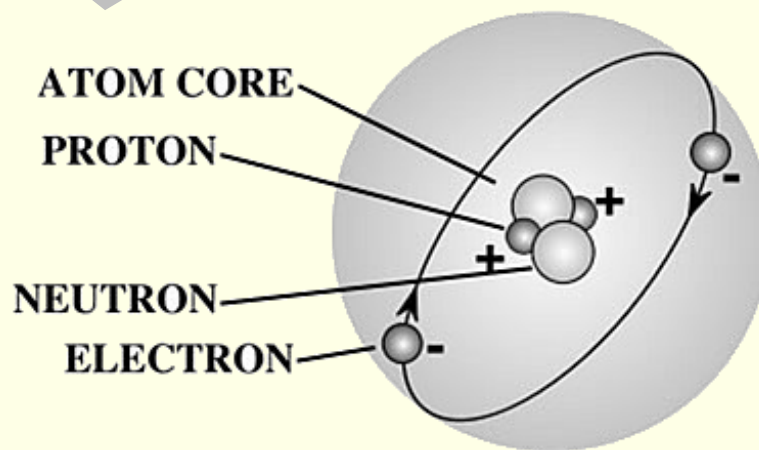
The atom.

The atom is the smallest stable particle of matter. It consists of a central nucleus that is surrounded by very small orbiting particles called electrons.

The nucleus consists of positively charged dense particles called protons and dense uncharged particles called neutrons. The orbiting electrons are negatively charged and do not contribute to the density of the atom. Each atom always has the same number of electrons as it does protons, so the overall electrical charge of an atom is neutral.

Actually, subatomic particles are more complex than described as they are now known to consist of a series of smaller particles. The most recent subatomic particle to be identified is the Higgs boson.

Each of the different atoms or elements is distinguished from one another by the number of protons in their nucleus. The image below illustrates a model of the helium atom:



Isotopes.

Each atom exists in a number of different forms called isotopes. Each of an atom's isotopes contains the same number of protons but a different number of neutrons.

The simplest atom is hydrogen. It is a gas. It mostly exists in a form that has only one proton and one electron. Hydrogen has two isotopes, deuterium, with one neutron as well as a proton in its nucleus and tritium, with two neutrons as well as a proton in its nucleus.

The second atom or element is called helium. It includes two protons and two electrons. Its main isotope has two neutrons in its nucleus.

The third element is called lithium. Lithium has three protons and three electrons. Its main isotope has three neutrons in its nucleus. Although they are rare, lithium also exists in other isotopic forms that include more than three neutrons as well as the three protons in its nucleus.

Subsequent elements increase in atomic number in accord with the number of protons and electrons, rather like having increasing numbers of the basic hydrogen atom. The series of atoms in ascending atomic number is called

3. The element symbol K is short for:-

- a) sodium
- b) potassium
- c) krypton

4) Organic chemistry mainly involves which element?

- a) iron
- b) chlorine
- c) carbon

5. How many neutrons has the hydrogen isotope tritium?

- a) 1
- b) 2
- c) none

6. How many electrons does a chlorine atom have in its outer orbital shell?

- a) 1
- b) 4
- c) 7

7. Which of these elements is not a trace element?

- a) nickel
- b) chromium
- c) cobalt

8. Aluminium is one of the most common elements on earth. Is it-

- a) deleterious
- b) required by
- c) or, one of the top elements in the human body

9. Which of these elements is deleterious to the human body?

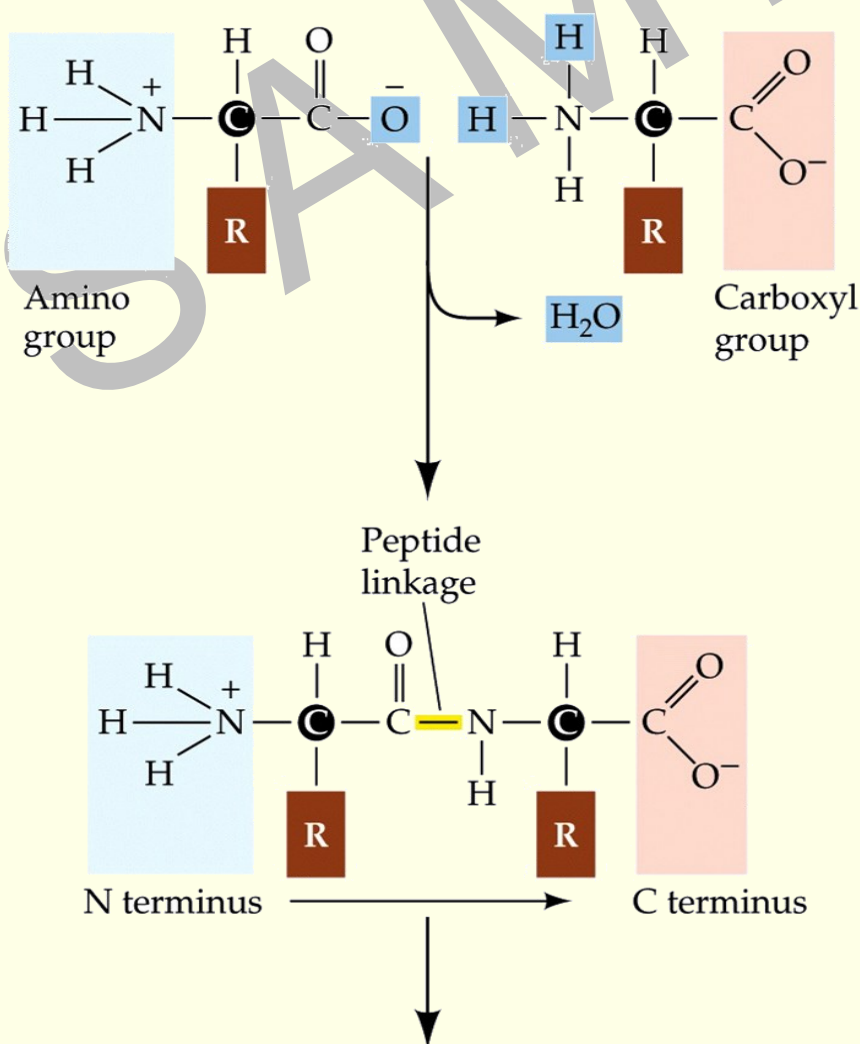
- a) copper
- b) zinc
- c) barium

Non-protein functions of amino acids.

Amino acids are also used to make other important biologically active molecules that are not classified as proteins because they do not have the typical linear chain structure. For example, tryptophan is a precursor of the neurotransmitter serotonin. Tyrosine is a precursor of the neurotransmitter dopamine. Glycine is a precursor of porphyrins such as heme. Arginine is a precursor of nitric oxide and glycine and glutamine are precursors of nucleotides.

How are amino acid chains made?

Amino acid chains or strings are made by combining amino acids through a covalent peptide bonding process. An amino acid is attached to the C terminus of the original amino acid by a condensation reaction. Water, H_2O is removed from the $-COO^-$ end of one amino acid as well as the $-NH_3^+$ of the next amino acid to combine the two amino acids by a peptide bond, $-C-N-$ as shown in the diagram. The chain is lengthened by addition of the next amino acid to the C terminus and so on as shown below.



Protein structural variation.

The final molecular structure of a protein is highly variable and complex. However, its study can be simplified by defining the various levels of structural organization.

IT IS IMPORTANT TO NOTE THAT THE FUNCTION OF A PROTEIN IS DEPENDENT ON ITS FINAL SHAPE OR TERTIARY STRUCTURE. THE SHAPE OF A PROTEIN DEPENDS ON THE NATURE OF THE **R** GROUPS ASSOCIATED WITH EACH OF ITS AMINO ACID CONSTITUENTS. THE TYPE OF **R** GROUP OR AMINO ACID INCLUDED IN THE PROTEIN IS DEPENDENT UPON A CHANGE IN SEQUENCE OF DNA THAT ENCODES THAT AMINO ACID, THEREFORE AS WILL BECOME CLEAR IN LATER MODULES, THE SEQUENCE OF DNA PLAYS A CRITICAL ROLE IN PROTEIN STRUCTURE AND CONSEQUENTLY FUNCTION.

A SINGLE AMINO ACID SUBSTITUTION IN A PROTEIN CAN HAVE A MAJOR IMPACT ON ITS FINAL TERTIARY STRUCTURE AND HENCE ITS FUNCTION.

Primary Structure:

The amino acid sequence is the primary structure. With 20 amino acids and the ability to vary the length of the amino acid chain, the structural possibilities in terms of sequence are endless. All the structural levels of a protein (secondary, tertiary and quaternary) are ultimately determined by the primary structure because all the information

7. Amino acids form protein chains by bonds called:-

- a) hydrophobic
- b) peptide
- c) alpha-helix

8. Which of these amino acids are not essential:-

- a) methionine
- b) leucine
- c) glycine

9. The single letter amino acid code G stands for:-

- a) glycine
- b) glutamic acid
- c) glutamine

10. Which of these amino acids are hydrophilic in nature:-

- a) glutamine
- b) histidine
- c) valine

11. Which group of amino acids contains the most methyl groups, $-CH_3$:-

- a) hydrophilic
- b) neutral
- c) hydrophobic

12. Which of these amino acid substitutions would you most expect to change the function of a protein:-

- a) valine to leucine
- b) isoleucine to lysine
- c) serine to threonine

13. Is the protein haemoglobin considered a:-

- a) glycoprotein
- b) metalloprotein
- c) lipoprotein

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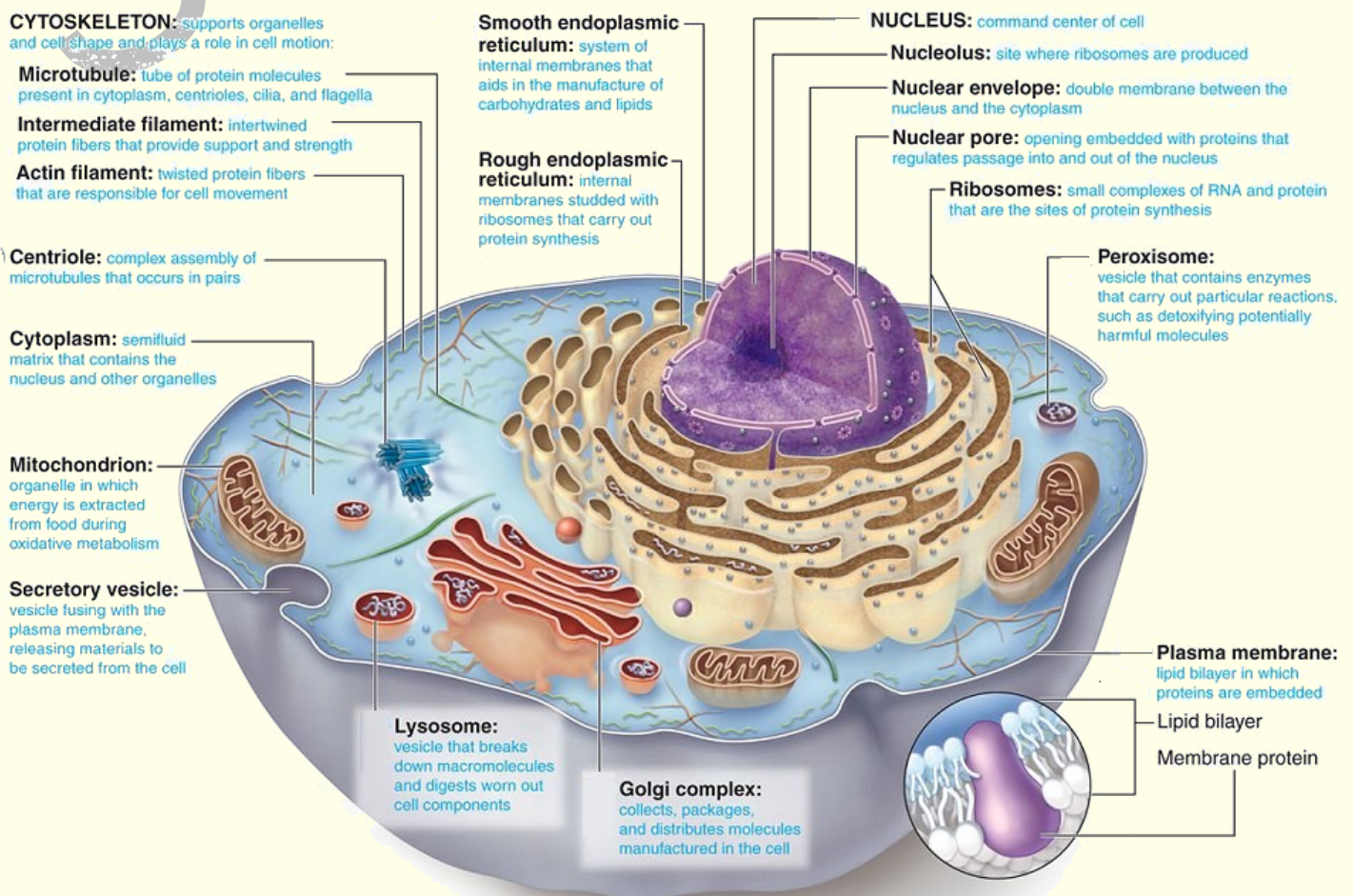
Introduction to the cell.

The cell.

All living things are made of cells. Just as a house is built from the basic building blocks, bricks, an organism is built from the basic building blocks, cells. There are two basic types of cells, prokaryotic cells and eukaryotic cells. Bacteria are prokaryotic. Prokaryotic cells are structurally simple and do not have a nucleus. In prokaryotic cells, DNA is in the form of a loose circle. Eukaryotic cells are more complex. Their DNA is contained within a nucleus and organised in structures called chromosomes. All animal and plant cells are eukaryotic.

A general eukaryotic cell.

In the human body, there are many different types of cells that make up organs such as the kidney, the brain, the heart, muscle, liver and all the other tissues and organs of the body. Even though they all have different organic functions, shapes and sizes, they all have common structural features and intracellular organelles. A general eukaryotic cell and its components are shown.



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How are proteins made?

Proteins are the molecules of life. So, how are they made? They are made by RNA.

What is RNA?

Ribonucleic acid (RNA) is a chain of nucleotides present in the cells of all life forms. RNA has a number of important functions including regulation of gene expression. RNA is also responsible for synthesising a protein or polypeptide chain whose amino acid composition is instructed by the sequence of the nucleotides within a gene, or DNA. The synthetic instructions are transferred from the gene by RNA (ribonucleic acid)

There are 5 basic types of RNA:-

1. mRNA - messenger RNA: encodes the amino acid sequence of a polypeptide.
2. tRNA - transfer RNA: brings amino acids to the ribosomes during translation.
3. rRNA - ribosomal RNA: with ribosomal proteins, makes up the ribosomes, the organelles that translate the mRNA.
4. snRNA - small nuclear RNA: with proteins, forms complexes that are used in RNA processing.
5. RNAi – interference RNA, of which there are two basic types, micro RNA (miRNA) and siRNA (small interfering RNA) involved in controlling gene expression.

To make a protein, or a polypeptide from a gene, there is a series of essential steps that must take place. Firstly, the gene must be transcribed.

Gene Transcription.

Firstly, through a process called gene transcription, an immature or primary RNA molecule is copied from the sequence of DNA. To do this, the gene's DNA must be accessible to a series of RNA synthesising agents called transcription factors and an enzyme called RNA polymerase. DNA that can be readily transcribed must be in a relaxed or unwound form, that is, as euchromatin. (Heterochromatin is a word that describes the condensed or compacted state of DNA that is not accessible to the transcription machinery).

There are many different transcription factors that recognise particular specific sequences in the upstream region of a gene. It is these recognition sequences that determine which of the 22,000 genes are about to be transcribed in any particular cell.

The molecular transcription machinery recognises a gene specific DNA sequence or site at the 5' end or the beginning of a gene. It then proceeds to open the double stranded DNA into two single strands and the RNA polymerase begins to synthesise a primary RNA molecule from the coding strand. The newly forming RNA molecule is built with a nucleotide sequence that is complementary to the coding DNA strand. Where DNA has a C, RNA will have a G, the same as the nucleotide pairing in DNA. Instead of pairing A with a T though, RNA uses uracil or U instead. So, RNA can be distinguished from DNA by having U instead of T.

Splicing of the primary RNA transcript.

The primary RNA molecule contains sequences that are complementary to the sequence of DNA from

4. Angelman's syndrome is associated with alterations in the imprinting of an area of chromosome:

- a) X
- b) 15
- c) Y

5. DNA imprinting usually involves changes in DNA:

- a) sequence
- b) duplication
- c) methylation

6. Alternative health care strategies may have the potential to affect gene:

- a) expression
- b) sequence
- c) copy number

7. Fabry disease is caused by mutations in the:

- a) GLA gene
- b) CFTR gene
- c) LEP gene

8. Cystic fibrosis is a:

- a) co-dominant
- b) dominant
- c) recessive genetic disorder.

9. Expression of the KLOTHO gene is known to decline with:

- a) age
- b) diet
- c) gender

10. Daughters inherit which chromosome from their fathers?

- a) Y
- b) X
- c) neither

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Introduction to:-

The immune system.

The immune system plays a very important role in protection against a wide range of infectious agents including bacteria, viruses and fungi. Subjects with impaired immunity suffer many serious infectious diseases whereas those with heightened immunity may suffer from auto-immune diseases.

There are many parts to the immune system. They are classified as either adaptive immunity or the innate immune system.

The adaptive immune systems are so called because they can respond to most forms of infectious or foreign agents as we are exposed to them. Each of the members of the adaptive immune systems has remarkable structural variability within the genes that they are encoded by. This structural variability is needed in order to respond to the huge number of foreign agents that we are or may be exposed to. For example, an immunoglobulin heavy chain and a light chain gene can re-arrange themselves so that they can make antibodies to perhaps a million different foreign substances. Genes that synthesise receptors on T cells can re-arrange similarly. For further information, there is much literature available which explains the complexity of the remarkable hypervariability of genetic systems that encode components of adaptive immunity.

Members of the innate or non-adaptive (sometimes called natural) immune systems are encoded by genes that are not subject to structural hypervariability.

Adaptive Immune systems.

The adaptive immune system comprises two quite different components referred to as cell mediated immunity and humoral immunity.

Cell mediated immunity.

Cell mediated immunity involves cells called T cells. T cells belong to a group of white blood cells known as lymphocytes. They are called T cells because they have had to undergo an important maturation process early on in life by passage through the thymus gland.

T cells begin their lives in the bone marrow. By a remarkable genetic process, different T cells develop receptor sites on their surface for many different infectious or foreign agents they are likely to come across in their lives.

They then pass through the thymus where those that may attack "self" are removed. The remaining cells undergo a final maturation process and become ready to recognise foreign or abnormal cells such as virus infected cells or tumour cells. When they encounter a foreign or infected cell, they become activated, multiply and kill virus infected cells and tumour cells by a direct contact killing process.

There are two basic types of T cells called helper and suppressor T cells. Helper T cells (also called CD4+ cells) assist in enhancing and stabilising the immune response. Helper T cells also help B cells to synthesise antibodies. CD4+ cells play a very important role on regulation and stabilisation of the immune system. By contrast, suppressor T cells (also called CD8+ cells) are the cytotoxic T cells that actually kill infected cells.

CD4+ helper T cells are destroyed by HIV, the AIDS virus. That is why HIV infection causes immune deficiency.

Answers to the self-assessment questions in the book.

Chapter 1 3,b; 4,c; 5,b; 6,c; 7,a; 8,a; 9,c;

Chapter 2. 7,b; 8,c; 9,a; 10,a and b; 11,c; 12,b; 13,b;

Chapter 6. 4,b; 5,c; 6,a; 7,a; 8,c; 9,a; 10,b.

NOTE: Some questions/answers have been omitted in the sample book.

SAMPLE

Publisher's note.

This book was designed and published by Homeovitality Company Limited in the United Kingdom.

“Understanding the basics of biochemistry, DNA, genetics, disease, drugs and immunity for everyone” (version 1.1)
by Dr P H Kay.

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