



THE UNIVERSITY OF
BUCKINGHAM

MEDICAL SCHOOL

MB ChB

Unit Summary: Molecules, Genes and Diseases

1 *Educational Aims of the Unit*

The unit aims to enable students to make progress towards meeting some of the learning outcomes described in Tomorrow's Doctors (2009) relevant to 'The Doctor as a Scholar and Scientist' and 'the Doctor as a Professional'. The specific aims of this unit are that students will develop an understanding of the general relationship between the processes involved in chromosome behaviour, gene expression and the activity of cells. At the end of the unit students should appreciate the variety of protein structures necessary to carry out the range of cellular processes, including protein synthesis and secretion. Additionally the student will understand the fundamental processes of inheritance and mutation and how these may affect patients. Finally the student should be able to connect the themes and molecular analyses discussed within the unit to a clinical context, and be aware of the ethical issues surrounding it.

2 *Learning Outcomes From Tomorrow's Doctors (2009)*

Outcomes 1: The Doctor as a Scholar and Scientist.

8. The graduate will be able to apply to medical practice biomedical scientific principles.
 - a) Explain normal human structure and functions.
 - b) Explain the scientific bases for common disease presentations.
 - c) Justify the selection of appropriate investigations for common clinical cases.
 - d) Explain the fundamental principles underlying such investigative techniques.
 - g) Make accurate observations of clinical phenomena and appropriate critical analysis of clinical data.
12. Apply scientific method and approaches to medical research.
 - a) Critically appraise the results of relevant diagnostic, prognostic and treatment trials and other qualitative and quantitative studies as reported in the medical and scientific literature.
 - b) Formulate simple relevant research questions in biomedical science, psychosocial science or population science, and design appropriate studies or experiments to address the questions.
 - c) Apply findings from the literature to answer questions raised by specific clinical problems.

Outcomes 3: The Doctor as a Professional

20. The graduate will be able to behave according to ethical and legal principles. The graduate will be able to:
 - e) Recognise the rights and the equal value of all people and how opportunities for some people may be restricted by others' perceptions.
21. Reflect, learn and teach others.
 - b) Establish the foundations for lifelong learning and continuing professional development, including a professional development portfolio containing reflections, achievements and learning needs.
 - c) Continually and systematically reflect on practice and, whenever necessary, translate that reflection into action, using improvement techniques and audit appropriately for example, by critically appraising the prescribing of others.

- d) Manage time and prioritise tasks, and work autonomously when necessary and appropriate.
- e) Recognise own personal and professional limits and seek help from colleagues and supervisors when necessary.

3 Teaching and Learning Strategies

Principles will be introduced in formal lectures, and learning will be reinforced in practical classes and facilitator led small-group work immediately afterwards. Student will work in the same teams throughout Phase I to encourage team-working.

Some concepts will be discussed in more detail in tutorials, and Moodle- based tests and coursework will allow for formative assessment. Students will be provided with workbooks describing structured tasks to direct independent learning throughout the unit, and ongoing use of an e-portfolio will nurture and encourage reflective practice.

4 Unit Outline/Syllabus

Session 1: DNA structure and chromosome organisation

Lecture: Introduction to the module

Nucleotides, DNA structure and chromosomes

Lecture: DNA replication and the cell cycle

Group Work: DNA structure

Session 2: Inheritance of genes

Lecture: Mitosis and meiosis, genotypes and phenotypes

Lecture: Genetic linkage and pedigree analysis

Group Work: Inheritance of genes

Session 3: Transcription and translation

Lecture: What is a gene and transcription

Lecture: The genetic code and translation

Group Work: Transcription and translation

Session 4: Cell and biological molecules

Lecture: Cell structures

Lecture: Amino acids and proteins

Group Work: Amino acids and proteins

Session 5: Protein structure and function

Lecture: Protein folding and action.

Lecture: Haemoglobin and myoglobin

Group Work: Protein structure and function

Session 6: Enzymes and enzyme regulation

Lecture: Enzyme activity.

Lecture: Regulatory strategies

Group Work: Enzymes and enzyme regulation

Session 7: Protein processing and targeting

Lecture: Protein processing in cells; the secretory pathway

Lecture: Proteolytic processing within the secretory pathway; collagen

Group Work: Control of cell growth

Session 8: Molecular diagnosis

Lecture: Molecular diagnosis 1

Lecture: Molecular diagnosis 2

Group Work and Lab demonstrations

Session 9: Mutations

Lecture: Mutagenesis and its effects

Lecture: Detecting mutations

Group Work: Mutations and their consequences

Session 10: Chromosomal abnormalities

Lecture: Chromosomal abnormalities 1

Lecture: Chromosomal abnormalities 2

Group Work: Chromosomal abnormalities

Session 11: Case studies

Lecture: Introduction to case studies

Group Work: Case studies

Session 12: Revision

Informal revision session

5 Secondary Learning Outcomes

In addition to meeting the outcomes described in Tomorrow's Doctors, at the completion of the unit students will be able to:

- Recognise the structural components of nucleic acids and distinguish between RNA and DNA.
- Explain the relationship between DNA, chromosome and genes.
- Outline the general features of DNA replication and compare and contrast mitosis and meiosis.
- Explain the principles of the genetic code and describe in general terms the processes of transcription, translation and post-translational modifications.
- Understand how basic cell structure relates to functional processes in the cell.
- Understand the chemistry of amino acids and how this relates to protein structure.
- Describe the action of enzymes and the major mechanisms for their regulation.
- Understand the link between the molecular structure and the physiological function of oxygen-transporting proteins.
- Outline the chromosomal basis of inheritance and understand the principles underlying pedigree analysis.
- Describe the different types of DNA mutation at the nucleotide and chromosomal levels.

- Understand the molecular methods used to analyse genes and proteins and appreciate the ways in which these methods are used in clinical situations and the ethical issues that may arise.
- Outline the molecular bases of a number of common inherited diseases.

6 Key Texts and/or Other Learning Materials

Essential

- *Marks' basic medical biochemistry: a clinical approach* - Michael Lieberman, Allan D. Marks, Alisa Peet 2013 1451100035 ,9781451100037
Lieberman M., Marks A.D. & Peet A., 2013. Marks' basic medical biochemistry: a clinical approach, Philadelphia, Pa: Wolters Kluwer Health/Lippincott Williams & Wilkins.
- *Biochemistry* - Denise R. Ferrier 2013 9781451187533
Ferrier D.R., 2013. Biochemistry, Philadelphia, Pennsylvania: Lippincott Williams & Wilkins.
- *Biochemistry* - Richard A. Harvey, Denise R. Ferrier c20119781609139988,9781608314126
Harvey R.A. & Ferrier D.R., 2011. Biochemistry, Philadelphia, Pa: Wolters Kluwer Health/Lippincott Williams & Wilkins.
- *New clinical genetics* - Andrew P. Read, D. Donnai 20111904842801,9781904842804
Read A.P. & Donnai D., 2011. New clinical genetics, Banbury: Scion.

Background

- *Human heredity: principles and issues* - Michael R. Cummings c2014 1133108636,9781133108634
Cummings M.R., 2014. Human heredity: principles and issues, Australia: Brooks/Cole Cengage Learning.
- *Essential cell biology* - Bruce Alberts, Dennis Bray 2013 9780815344551,9780815344544
Alberts B. & Bray D., 2013. Essential cell biology, New York: Garland Science.
- *Medical biochemistry* - John Baynes, Marek H. Dominiczak 2014
Baynes J. & Dominiczak M.H., 2014. Medical biochemistry, Philadelphia: Saunders. Available at: <http://ezproxy.lib.le.ac.uk/login?url=http://elsevierelibrary.co.uk/product/medical-biochemistry59110>.
- *Biochemistry* - Saeid Karandish 2010 1608311694,9781608311699
Karandish S., 2010. Biochemistry, Philadelphia, Pa: Lippincott Williams & Wilkins.
- *Cell and molecular biology* - Nalini Chandar, Susan Viselli 20101609133099,9781609133092
Chandar N. & Viselli S., 2010. Cell and molecular biology, Philadelphia, Pa: Lippincott Williams & Wilkins.

Document Version Information

Document Title: Unit Summary: Molecules, Genes and Diseases

Originator:

Date:

Replacing Document:

Approved:

Date: