



# MEDI219

## Genetics and Genomics in Medicine

S2 Day 2019

*Medicine and Health Sciences Faculty level units*

### Contents

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<a href="#"><u>General Information</u></a>	2
<a href="#"><u>Learning Outcomes</u></a>	2
<a href="#"><u>General Assessment Information</u></a>	3
<a href="#"><u>Assessment Tasks</u></a>	5
<a href="#"><u>Delivery and Resources</u></a>	6
<a href="#"><u>Unit Schedule</u></a>	7
<a href="#"><u>Policies and Procedures</u></a>	8
<a href="#"><u>Graduate Capabilities</u></a>	9

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#### **Disclaimer**

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## General Information

Unit convenor and teaching staff

Unit Convenor

Russell Diefenbach

[russell.diefenbach@mq.edu.au](mailto:russell.diefenbach@mq.edu.au)

Contact via 9850 2764

Level 1, 75 Talavera Road

By appointment

Credit points

3

Prerequisites

CBMS104

Corequisites

Co-badged status

Unit description

This unit introduces concepts which are core to biochemistry, cell and molecular biology. You will learn about the nature of chemical reactions that occur within the human body and how they are regulated. You will explore the structure and the relationships between the four main biomolecules (nucleic acids, proteins, carbohydrates and lipids) within the human body. You will gain an understanding of the fundamental structure of the cell and how this relates to function. Examples of how these processes are altered resulting in human diseases will also be provided. You will participate in practical classes which reinforce the lecture content and provide training in basic laboratory skills

## Important Academic Dates

Information about important academic dates including deadlines for withdrawing from units are available at <https://www.mq.edu.au/study/calendar-of-dates>

## Learning Outcomes

On successful completion of this unit, you will be able to:

Describe the major aspects of cell structure and organisation

Explain the role of compartmentalization and signalling in cells

Describe the properties and functions of the major groups of biomolecules

Discuss the role of glucose metabolism

Discuss the human genome structure and the properties of DNA

Describe the nature of variations in genetic material

Demonstrate development in practical scientific skills

Interpret and analyse experimental data

## General Assessment Information

**Assessment:** Your raw marks from assessments are combined into a weighted sum. The weighted sums for the whole class are ranked, and compared across other units for appropriate consistency. This process of comparison allows for the identification of unusual influences on class performance that might warrant the weighted sums of marks being scaled or otherwise altered. The numerical cut-off for each descriptive grade is then determined. The numerical value which you are issued with (i.e., Standardised Numerical Grade; SNG) is determined to match your descriptive grade by standardising weighted sums of raw marks to match standard scores out of 100. The SNG gives you an indication of how you have performed within the band for your descriptive grade. As the SNG is the result of scaling the weighted sum of your raw marks, you won't be able to:

- work out your exam mark based on the assignment marks you already know and the SNG;
- determine that you were "one mark away" from a different grade.

It is our professional responsibility as your mentors to assign you a grade that accurately reflects your performance. Our grading decisions are subject to scrutiny by academic colleagues at the Program, Faculty and University level.

Grades ranging from High Distinction to Fail are defined as follows:

Grade	SNG	Description
HD High Distinction	85-100	Work of outstanding quality. This may be demonstrated in areas such as criticism, logical argument, and interpretation of materials or use of methodology. This grade may also be awarded to recognise a high order of originality or creativity in student performance
D Distinction	75-84	Work of superior quality in the same areas of performance as above. This grade may also be awarded to recognise particular originality or creativity in student performance
Cr Credit	65-74	Work of predominantly good quality, demonstrating a sound grasp of content together with efficient organisation, selectivity and use of techniques
P Pass	50-64	Satisfactory achievement of unit objectives
F Fail	0-49	Failure to achieve unit objectives

Grade descriptors and other information concerning grading are contained in Schedule 1 of the Macquarie University Assessment Policy, which is available at: <https://staff.mq.edu.au/work/strategy-planning-and-governance/university-policies-and-procedures/policies/assessment>.

Further details for each assessment task will be available on iLearn.

All final grades in the Bachelor of Clinical Science are determined by a grading committee and are not the sole responsibility of the Unit Convenor.

Students will be awarded a final grade plus a Standardised Numerical Grade (SNG). The SNG is not necessarily a summation of the individual assessment components. The final grade and SNG that are awarded reflect the corresponding grade descriptor in the Grading Policy.

To pass this unit, students must demonstrate sufficient evidence of achievement of the learning outcomes, attempt all assessment tasks, meet any ungraded requirements including professionalism and achieve an SNG of 50 or better.

## Student Professionalism

In the Faculty of Medicine and Health Sciences, professionalism is a key capability embedded in all our courses. As part of developing professionalism, students are expected to attend all small group interactive sessions including tutorials, as well as clinical- and laboratory-based practical sessions.

Furthermore, lectures and seminars are key learning activities that you are expected to attend throughout completion of the Bachelor of Clinical Science. While audio recordings and lecture slides may be made available following these large group sessions, it is important to recognise that such resources are a study aid - and should not be considered an alternative to lecture or seminar attendance.

Students are required to attend a minimum of 80% of all small group interactive sessions. Students that do not meet this requirement may be deemed unable to meet expectations regarding professionalism and may be referred for disciplinary action (which may include exclusion from assessments and unit failure).

Similarly, as part of developing professionalism, students are expected to submit all work by the due date. Applications for assessment task extensions must be supported by appropriate evidence and submitted via [www.ask.mq.edu.au](http://www.ask.mq.edu.au). For further details please refer to the Special Consideration Policy available at <https://students.mq.edu.au/study/my-study-program/special-consideration>.

## Late Submission

All assignments which are officially received after the due date, and where no extension has been granted, will incur a deduction of 5% for the first day, and 5% for each subsequent day including the actual day on which the work is received. Weekends and public holidays are included. For example:

Due date	Received	Days late	Deduction	Raw mark	Final mark
Friday 14th	Monday 17th	3	15%	75%	60%

## Assessment Tasks

Name	Weighting	Hurdle	Due
<a href="#"><u>Online quiz</u></a>	0%	No	Week 3
<a href="#"><u>Mid-semester exam</u></a>	25%	No	Week 5
<a href="#"><u>Practical exam</u></a>	25%	No	Week 11
<a href="#"><u>Final Exam</u></a>	50%	No	University examination period

### Online quiz

Due: **Week 3**

Weighting: **0%**

Online quiz.

On successful completion you will be able to:

- Describe the properties and functions of the major groups of biomolecules
- Discuss the human genome structure and the properties of DNA

### Mid-semester exam

Due: **Week 5**

Weighting: **25%**

The mid-session exam will be composed of a mixture of multiple choice and short answer questions.

On successful completion you will be able to:

- Describe the major aspects of cell structure and organisation
- Explain the role of compartmentalization and signalling in cells
- Describe the properties and functions of the major groups of biomolecules
- Discuss the role of glucose metabolism
- Discuss the human genome structure and the properties of DNA
- Describe the nature of variations in genetic material
- Demonstrate development in practical scientific skills

- Interpret and analyse experimental data

## Practical exam

Due: **Week 11**

Weighting: **25%**

The practical exam will be composed of a mixture of multiple choice and short answer questions. All practicals will be assessed.

On successful completion you will be able to:

- Demonstrate development in practical scientific skills
- Interpret and analyse experimental data

## Final Exam

Due: **University examination period**

Weighting: **50%**

The final exam will be composed of a mixture of multiple choice and short answer questions. All content will be assessed.

On successful completion you will be able to:

- Describe the major aspects of cell structure and organisation
- Explain the role of compartmentalization and signalling in cells
- Describe the properties and functions of the major groups of biomolecules
- Discuss the role of glucose metabolism
- Discuss the human genome structure and the properties of DNA
- Describe the nature of variations in genetic material
- Demonstrate development in practical scientific skills
- Interpret and analyse experimental data

## Delivery and Resources

Lectures, 24hr; Tutorials, 12hr; Labs, 12hr; Total, 48hrs.

### Technology Used

Active participation in the learning activities throughout the unit will generally require students to have access to a tablet, laptop or similar device. Students who do not own their own laptop computer may borrow one from the university library.

### Required Unit Materials

All students are required to wear closed shoes and a lab coat/gown to attend practical classes.

### Recommended Readings

Unit readings for this unit are available via the iLearn and university library website.

The prescribed textbook for this unit is

**Alberts, B. (2014) *Molecular Biology of the Cell* (6th Edition). Garland Science**

Other recommended text (available at the library)

**Lodish, H. (2016) *Molecular Cell Biology* (8th Edition). MacMillan**

## Unit Schedule

Week	Lecture Topic	Delivered by	Tutorial	Practical	Assessment
1	Structure of the major biomolecules of cells - proteins, nucleic acids  Structure of the major biomolecules of cells - carbohydrates, lipids	Russell Diefenbach  Russell Diefenbach		Practical 1	
2	DNA replication and chromosome structure  Transcription and translation	Helen Rizos  Annika Van Hummel	Tutorial 1		
3	Structure and function of proteins  Enzymes	Russell Diefenbach  Russell Diefenbach		Practical 2	Online Quiz
4	Introduction to cells  Membrane structure and dynamics	Julie Atkin  Julie Atkin	Tutorial 2		
5	Biological membranes and transport  Vesicular trafficking	Sonam Parakh  Annika Van Hummel			Mid-semester exam
6	Membrane receptors  Biosignalling	Albert Lee  Sonam Parakh	Tutorial 3		
7	The cytoskeleton  Cellular adhesion	Russell Diefenbach  Annika Van Hummel		Practical 3	
	RECESS				
8	Cell cycle, mitosis and meiosis  Cell death	Lucinda McRobb  Shu Yang	Tutorial 4		

9	Principles of bioenergetics Glycolysis and gluconeogenesis	Russell Diefenbach Christine Chiu		Practical 4	
10	The citric acid cycle and oxidative phosphorylation Principles of metabolic regulation	Christine Chiu Christine Chiu	Tutorial 5		
11	Principles of gene regulation and epigenetics Principles of genetic variation	Ian Blair Ian Blair			Practical exam
12	Single-gene disorders Genetic variation producing disease-causing abnormalities in DNA	Ian Blair Helen Rizos	Tutorial 6		
13	Revision				

## Policies and Procedures

Macquarie University policies and procedures are accessible from [Policy Central \(https://staff.mq.edu.au/work/strategy-planning-and-governance/university-policies-and-procedures/policy-central\)](https://staff.mq.edu.au/work/strategy-planning-and-governance/university-policies-and-procedures/policy-central). Students should be aware of the following policies in particular with regard to Learning and Teaching:

- [Academic Appeals Policy](#)
- [Academic Integrity Policy](#)
- [Academic Progression Policy](#)
- [Assessment Policy](#)
- [Fitness to Practice Procedure](#)
- [Grade Appeal Policy](#)
- [Complaint Management Procedure for Students and Members of the Public](#)
- [Special Consideration Policy](#) (**Note:** *The Special Consideration Policy is effective from 4 December 2017 and replaces the Disruption to Studies Policy.*)

Undergraduate students seeking more policy resources can visit the [Student Policy Gateway \(https://students.mq.edu.au/support/study/student-policy-gateway\)](https://students.mq.edu.au/support/study/student-policy-gateway). It is your one-stop-shop for the key policies you need to know about throughout your undergraduate student journey.

If you would like to see all the policies relevant to Learning and Teaching visit [Policy Central \(https://staff.mq.edu.au/work/strategy-planning-and-governance/university-policies-and-procedures/policy-central\)](https://staff.mq.edu.au/work/strategy-planning-and-governance/university-policies-and-procedures/policy-central).

## Student Code of Conduct

Macquarie University students have a responsibility to be familiar with the Student Code of Conduct: <https://students.mq.edu.au/study/getting-started/student-conduct>



## Results

Results published on platform other than [eStudent](#), (eg. iLearn, Coursera etc.) or released directly by your Unit Convenor, are not confirmed as they are subject to final approval by the University. Once approved, final results will be sent to your student email address and will be made available in [eStudent](#). For more information visit [ask.mq.edu.au](#) or if you are a Global MBA student contact [globalmba.support@mq.edu.au](mailto:globalmba.support@mq.edu.au)

## Student Support

Macquarie University provides a range of support services for students. For details, visit <http://students.mq.edu.au/support/>

## Learning Skills

Learning Skills ([mq.edu.au/learningskills](http://mq.edu.au/learningskills)) provides academic writing resources and study strategies to improve your marks and take control of your study.

- [Workshops](#)
- [StudyWise](#)
- [Academic Integrity Module for Students](#)
- [Ask a Learning Adviser](#)

## Student Services and Support

Students with a disability are encouraged to contact the [Disability Service](#) who can provide appropriate help with any issues that arise during their studies.

## Student Enquiries

For all student enquiries, visit Student Connect at [ask.mq.edu.au](#)

If you are a Global MBA student contact [globalmba.support@mq.edu.au](mailto:globalmba.support@mq.edu.au)

## IT Help

For help with University computer systems and technology, visit [http://www.mq.edu.au/about\\_us/offices\\_and\\_units/information\\_technology/help/](http://www.mq.edu.au/about_us/offices_and_units/information_technology/help/).

When using the University's IT, you must adhere to the [Acceptable Use of IT Resources Policy](#). The policy applies to all who connect to the MQ network including students.

## Graduate Capabilities

### Commitment to Continuous Learning

Our graduates will have enquiring minds and a literate curiosity which will lead them to pursue knowledge for its own sake. They will continue to pursue learning in their careers and as they participate in the world. They will be capable of reflecting on their experiences and relationships with others and the environment, learning from them, and growing - personally, professionally and socially.

This graduate capability is supported by:

## **Learning outcomes**

- Describe the major aspects of cell structure and organisation
- Explain the role of compartmentalization and signalling in cells
- Describe the properties and functions of the major groups of biomolecules
- Discuss the role of glucose metabolism
- Discuss the human genome structure and the properties of DNA
- Describe the nature of variations in genetic material
- Demonstrate development in practical scientific skills
- Interpret and analyse experimental data

## **Assessment tasks**

- Online quiz
- Mid-semester exam
- Practical exam
- Final Exam

## **Discipline Specific Knowledge and Skills**

Our graduates will take with them the intellectual development, depth and breadth of knowledge, scholarly understanding, and specific subject content in their chosen fields to make them competent and confident in their subject or profession. They will be able to demonstrate, where relevant, professional technical competence and meet professional standards. They will be able to articulate the structure of knowledge of their discipline, be able to adapt discipline-specific knowledge to novel situations, and be able to contribute from their discipline to inter-disciplinary solutions to problems.

This graduate capability is supported by:

## **Learning outcomes**

- Describe the major aspects of cell structure and organisation
- Explain the role of compartmentalization and signalling in cells
- Describe the properties and functions of the major groups of biomolecules
- Discuss the role of glucose metabolism
- Discuss the human genome structure and the properties of DNA
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## Assessment tasks

- Online quiz
- Mid-semester exam
- Practical exam
- Final Exam

## Critical, Analytical and Integrative Thinking

We want our graduates to be capable of reasoning, questioning and analysing, and to integrate and synthesise learning and knowledge from a range of sources and environments; to be able to critique constraints, assumptions and limitations; to be able to think independently and systemically in relation to scholarly activity, in the workplace, and in the world. We want them to have a level of scientific and information technology literacy.

This graduate capability is supported by:

## Learning outcomes

- Describe the major aspects of cell structure and organisation
- Explain the role of compartmentalization and signalling in cells
- Describe the properties and functions of the major groups of biomolecules
- Discuss the role of glucose metabolism
- Discuss the human genome structure and the properties of DNA
- Describe the nature of variations in genetic material
- Demonstrate development in practical scientific skills
- Interpret and analyse experimental data

## Assessment tasks

- Online quiz
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- Practical exam
- Final Exam

## Problem Solving and Research Capability

Our graduates should be capable of researching; of analysing, and interpreting and assessing data and information in various forms; of drawing connections across fields of knowledge; and they should be able to relate their knowledge to complex situations at work or in the world, in order to diagnose and solve problems. We want them to have the confidence to take the initiative in doing so, within an awareness of their own limitations.

This graduate capability is supported by:

## **Learning outcomes**

- Describe the major aspects of cell structure and organisation
- Explain the role of compartmentalization and signalling in cells
- Describe the properties and functions of the major groups of biomolecules
- Discuss the role of glucose metabolism
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