MEDI209

Genetics and Genomics in Medicine

MED 0 2017

Medicine and Health Sciences Faculty level units

Contents

General Information

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Credit points 3
Prerequisites Admission to BClinSc and (12cp at 100 level) and (6cp at 200 level)
Corequisites
Co-badged status
Unit description During this unit you will explore the science and technologies underlying the use of genetics/ genomics and other "omics" and their application in personalised medicine. You will revise the molecular bases for inheritance, and the DNA/RNA technologies that are revolutionizing medical genetics and genomics. You will apply principles of classical genetics to understand the inheritance of defined traits and simple (monogenic) diseases, examine newer approaches for understanding the inheritance of common diseases, and learn about the behaviour of genes in populations. The emerging discipline of genomic medicine and the use of personal "omic" information for clinical care is examined. Cancer is emphasised as a leading example of the use of genomics and other "omics" data for the personalised diagnosis, prognosis, treatment and response to therapy of patients.

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:

Articulate a broad knowledge and understanding of the fundamental biological, chemical and physical sciences.

Demonstrate an understanding of the molecular basis of inheritance

Understand the principles of and interpret DNA/RNA (and other "omics") technologies and their applications (e.g., human genome & proteome projects) to personalised medicine

Apply the principle of Mendelian inheritance to examine transmission of defined traits and simple genetic diseases through pedigrees

Evaluate approaches to analyzing the inheritance of complex traits and common diseases

Apply simple population genetics tools to appreciate how genes behave in populations

Identify the online resources and tools that deal with the massive data sets released by

big data "omics" projects and evaluate the impact of genomics and the other "omics" on current and future medical practice.

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

Assessment Tasks

Name	Weighting	Hurdle	Due
Practical Session Write-Ups	30%	No	Tuesday 2pm, Weks 2,3,4
Oral Presentation	20%	No	Weeks 5-7
Essay	20%	No	Feb 17th 1pm
Final Exam	30%	No	end Week 6

Practical Session Write-Ups

Due: **Tuesday 2pm, Weks 2,3,4**
Weighting: **30%**

All laboratories will be conducted in groups. These have a highly investigative approach, where you will be conducting analysis to apply theoretical knowledge to understand genetics/genomics/omics data. You will be required to write reports for 3 practicals in MEDI209, each is only one week long (3 x 10% = 30% of your total assessment). The detailed requirements for each report will be given with notes available before or at the beginning of each practical class. Prac reports are due during the semester one week after the practical class is complete. Please check iLearn for due dates. All prac reports should be submitted to the MEDI209 coordinator directly.

On successful completion you will be able to:

- Articulate a broad knowledge and understanding of the fundamental biological, chemical and physical sciences.
- Apply simple population genetics tools to appreciate how genes behave in populations
- Identify the online resources and tools that deal with the massive data sets released by big data "omics" projects and evaluate the impact of genomics and the other "omics" on current and future medical practice.

Oral Presentation

Due: **Weeks 5-7**
Weighting: **20%**

You will be randomly (out-of-a-hat) assigned to either a Hot Topic team PPT oral presentation or to one side of a Debate (POSITIVE/NEGATIVE). These will occur during the practicals classes (hot topics first followed by debates) held in weeks 4-6 of the unit.

These will be chaired by the tutors assisting in the running of the practical classes.

Both Hot Topics and Debates will be video recorded for assessment and edited for future marketing of the BClinSci program and the MEDI209 unit.

- **Hot Topics** Student oral class presentations have been incorporated to assist you learn how to communicate science to your peers (peer-assisted learning) as well as to the public. The length of MEDI209 Hot Topics presentations will be 5min long plus 2min for questions whereas CBMS737/837 Hot Topics presentations will be 10min long plus 4min questions. Oral presentations will be given in the practical classes held in weeks 2 and the last week of semester (see below).
- **Debates:** Student debates (2 teams of three students) will contend an argument as a formal discussion before a public assembly. Speeches will be 5 minutes long with additional time for questions once completed. See Rules later in this guide

On successful completion you will be able to:

- Articulate a broad knowledge and understanding of the fundamental biological, chemical and physical sciences.
- Understand the principles of and interpret DNA/RNA (and other "omics") technologies and their applications (e.g., human genome & proteome projects) to personalised medicine
- Apply the principle of Mendelian inheritance to examine transmission of defined traits and simple genetic diseases through pedigrees

Essay

Due: **Feb 17th 1pm**
Weighting: **20%**

Maximum 2,000 word essay (not including tables, figures and references) on a topic of relevance to personalised/precision medicine.

On successful completion you will be able to:

- Articulate a broad knowledge and understanding of the fundamental biological, chemical and physical sciences.
- Demonstrate an understanding of the molecular basis of inheritance
- Understand the principles of and interpret DNA/RNA (and other "omics") technologies and their applications (e.g., human genome & proteome projects) to personalised medicine
- Evaluate approaches to analyzing the inheritance of complex traits and common diseases
- Identify the online resources and tools that deal with the massive data sets released by big data "omics" projects and evaluate the impact of genomics and the other "omics" on

current and future medical practice.

Final Exam

Due: **end Week 6**
Weighting: **30%**

The final exam (30% total assessment) will be composed of multiple short answer questions and is 2hr in length with 10min reading time. It is designed to address specific understanding of topics presented in lectures, practicals and peer-assisted oral presentation learning exercises. It also assesses that the knowledge you have obtained can be applied to new problems. It is Macquarie University policy to not set early examinations for individuals or groups of students.

On successful completion you will be able to:

- Articulate a broad knowledge and understanding of the fundamental biological, chemical and physical sciences.
- Demonstrate an understanding of the molecular basis of inheritance
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- Evaluate approaches to analyzing the inheritance of complex traits and common diseases
- Apply simple population genetics tools to appreciate how genes behave in populations

Delivery and Resources

Lectures, 12hr; Seminars/Tutorials, 12hr; Assessments, 60hr; Class Preparation, 50hr; Labs, 9hr; Other, 3hr; Total, 150hrs.

Unit Schedule

Lecture, **P**ractical, **H**ot **T**opic, **D**ebate	**MEDI209 Lecture, Practical, Hot Topic or Debate Topic** (*"Genetics & Genomics in Medicine"* Strachan et al., 2015 with MQ research examples)	**Academic Responsible**	**Date/ Due Date**	**Time**	**MQ Location**
Week 1 L1	DNA, Chromosomes & Cells (Chap 1; pp1-18)	MSB	Jan 9th	9-10	L1, 75 Talavera
L2	Gene Structure/Expression & the Human Genome (Chap 2; pp19-56)	MSB	Jan 10th	10-11	L1, 75 Talavera
Prac 1	Global Gene Expression Profiling (Transcriptomic Profiling)	HR	Jan 10th	13 -17	EMC2 Computer Lab

Week 2 L3	Underpinning DNA Technologies - PCR, Cloning (Chap 3; pp57-79)	HR	Jan 16th	9-10	L1, 75 Talavera
L4	Genome Sequencing Technologies Through the Ages	IB	Jan 17th	10-11	L1, 75 Talavera
Prac 2	Next-Gen Sequencing and the Integrated Genome Viewer for Disease Prediction and Susceptibility Analysis	IB/DB (CSIRO)	Jan 17th	13 -17	EMC2 Computer Lab
Week 3 L5	Single Gene Disorders, Inheritance, Allele Frequencies (Chap 5; pp 117-148)	IB	Jan 23rd	9-10	L1, 75 Talavera
L6	Identifying Disease Genes & Susceptibility (Chap 7; pp189-247)	IB	Jan24th	10-11	L1, 75 Talavera
Prac 3	Proteomics Big Data – HPP, Ingenuity, Human Protein Atlas & MissingProteinPedia	MSB	Jan 24th	13 -17	EMC2 Computer Lab
Week 4 L7	Epigenetics & Gene Regulation (Chap 6; pp149-188)	SG	Jan 30th	9-10	L1, 75 Talavera
L8	Genetic Counselling & Approaches to Treating Disease [Reading Task for Final Exam: Chaps 8 and 9; pp247-370]	AC	Jan 31st	10-11	L1, 75 Talavera
HT1	Nik-Zainal S et al., *Landscape of somatic mutations in 560 breast cancer whole-genome sequences.* Nature. 2016 May 2;534(7605):47-54. doi: 10.1038/nature17676.	MSB	Jan 31st	1 -1:30	L1, 75 Talavera
HT2	Cancer Genome Atlas Network. *Comprehensive molecular characterization of human colon and rectal cancer,* Nature, 487, 330-7. 2012. doi:10.1038/nature11252.	MSB	Jan 31st	1:30-2	L1, 75 Talavera
D1	*Next-gen human genome sequencing will allow the diagnosis and treatment of human cancers within a decade.*	MSB	Jan 31st	2-4	L1, 75 Talavera
Week 5 L9	Transcriptomics: Global Expression Analysis to Medicine	HR	Feb 6th	9-10	L1, 75 Talavera
L10	Big Data & the “Omics” Revolution	SR	Feb 7th	10-11	L1, 75 Talavera
HT3	*Single-cell genome sequencing: current state of the science*, Gawad, Koh & Quake. Nature Reviews Genetics 17, 175–188.2016.	MSB	Feb 7th	1 -1:30	L1, 75 Talavera

HT4	Kaiser J. *The gene editor CRISPR won't fully fix sick people anytime soon. Here's why.* May 3rd 2016. Science.	MSB	Feb 7th	1:30-2	L1, 75 Talavera
D2	*Personalised omics will solve the problem that currently "more than 90% of drugs only work in 30-50% of people".*	MSB	Feb 7th	2-4	L1, 75 Talavera
Week 6 L11	Cancer Genetics, Genomics & the TCGA (Chap 10; pp373-427)	MSB	Feb 13th	9-10	L1, 75 Talavera
Essay (1,500 word)	**Compare/Contrast the Human Genome & Human Proteome Projects as they Pertain to Personalised Medicine**	MSB	**Feb 13th**	**9am**	
L12	Personalised Cancer "Omics", Human Proteome Project & Human Protein Atlas	MSB	Feb 14th	10-11	L1, 75 Talavera
HT5	*Proteomic analysis of colon & rectal carcinoma using standard & customized databases.* Slebos et al., Scientific Data 2, 150022. 2015.	MSB	Feb 14th	1 -1:30	L1, 75 Talavera
HT6	*Facilitating a culture of responsible and effective sharing of cancer genome data.* Siu et al., Nature Medicine 22, 464–471. 2016.	MSB	Febr14th	1:30-2	L1, 75 Talavera
D3	*Healthy Lifestyle Is More Important than CVD Genetic Risk Factors.*	MSB	Feb 14th	2-4	L1, 75 Talavera
Exam (2hr)			**Feb 17th**	**2-4pm**	

Policies and Procedures

Macquarie University policies and procedures are accessible from Policy Central. Students should be aware of the following policies in particular with regard to Learning and Teaching:

Academic Honesty Policy http://mq.edu.au/policy/docs/academic_honesty/policy.html

Assessment Policy http://mq.edu.au/policy/docs/assessment/policy_2016.html

Grade Appeal Policy http://mq.edu.au/policy/docs/gradeappeal/policy.html

Complaint Management Procedure for Students and Members of the Public http://www.mq.edu.au/policy/docs/complaint_management/procedure.html

Disruption to Studies Policy (in effect until Dec 4th, 2017): http://www.mq.edu.au/policy/docs/disruption_studies/policy.html

Special Consideration Policy (in effect from Dec 4th, 2017): https://staff.mq.edu.au/work/strategy-planning-and-governance/university-policies-and-procedures/policies/special-consideration

In addition, a number of other policies can be found in the Learning and Teaching Category of 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/support/student_conduct/

Results

Results shown in *iLearn*, 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.

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) 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

IT Help

For help with University computer systems and technology, visit 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

Creative and Innovative

Our graduates will also be capable of creative thinking and of creating knowledge. They will be imaginative and open to experience and capable of innovation at work and in the community. We want them to be engaged in applying their critical, creative thinking.

This graduate capability is supported by:

Learning outcomes

- Articulate a broad knowledge and understanding of the fundamental biological, chemical and physical sciences.
- Demonstrate an understanding of the molecular basis of inheritance
- Apply simple population genetics tools to appreciate how genes behave in populations

Assessment tasks

- Practical Session Write-Ups
- Oral Presentation

Capable of Professional and Personal Judgement and Initiative

We want our graduates to have emotional intelligence and sound interpersonal skills and to demonstrate discernment and common sense in their professional and personal judgement. They will exercise initiative as needed. They will be capable of risk assessment, and be able to handle ambiguity and complexity, enabling them to be adaptable in diverse and changing environments.

This graduate capability is supported by:

Learning outcomes

- Demonstrate an understanding of the molecular basis of inheritance
- Understand the principles of and interpret DNA/RNA (and other "omics") technologies and their applications (e.g., human genome & proteome projects) to personalised medicine
- Identify the online resources and tools that deal with the massive data sets released by big data "omics" projects and evaluate the impact of genomics and the other "omics" on current and future medical practice.

Assessment tasks

- Oral Presentation
- Essay

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

- Articulate a broad knowledge and understanding of the fundamental biological, chemical and physical sciences.
- Understand the principles of and interpret DNA/RNA (and other "omics") technologies and their applications (e.g., human genome & proteome projects) to personalised medicine
- Apply simple population genetics tools to appreciate how genes behave in populations
- Identify the online resources and tools that deal with the massive data sets released by big data "omics" projects and evaluate the impact of genomics and the other "omics" on current and future medical practice.

Assessment tasks

- Practical Session Write-Ups
- Oral Presentation
- 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

- Articulate a broad knowledge and understanding of the fundamental biological, chemical and physical sciences.
- Demonstrate an understanding of the molecular basis of inheritance
- Understand the principles of and interpret DNA/RNA (and other "omics") technologies and their applications (e.g., human genome & proteome projects) to personalised medicine
- Apply the principle of Mendelian inheritance to examine transmission of defined traits and simple genetic diseases through pedigrees
- Evaluate approaches to analyzing the inheritance of complex traits and common diseases

- Apply simple population genetics tools to appreciate how genes behave in populations
- Identify the online resources and tools that deal with the massive data sets released by big data "omics" projects and evaluate the impact of genomics and the other "omics" on current and future medical practice.

Assessment tasks

- Practical Session Write-Ups
- Oral Presentation
- Essay
- 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

- Articulate a broad knowledge and understanding of the fundamental biological, chemical and physical sciences.
- Apply the principle of Mendelian inheritance to examine transmission of defined traits and simple genetic diseases through pedigrees
- Evaluate approaches to analyzing the inheritance of complex traits and common diseases
- Apply simple population genetics tools to appreciate how genes behave in populations
- Identify the online resources and tools that deal with the massive data sets released by big data "omics" projects and evaluate the impact of genomics and the other "omics" on current and future medical practice.

Assessment tasks

- Practical Session Write-Ups
- Oral Presentation
- Essay
- 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

- Demonstrate an understanding of the molecular basis of inheritance
- Understand the principles of and interpret DNA/RNA (and other "omics") technologies and their applications (e.g., human genome & proteome projects) to personalised medicine
- Apply the principle of Mendelian inheritance to examine transmission of defined traits and simple genetic diseases through pedigrees
- Evaluate approaches to analyzing the inheritance of complex traits and common diseases
- Identify the online resources and tools that deal with the massive data sets released by big data "omics" projects and evaluate the impact of genomics and the other "omics" on current and future medical practice.

Assessment tasks

- Practical Session Write-Ups
- Essay
- Final Exam

Effective Communication

We want to develop in our students the ability to communicate and convey their views in forms effective with different audiences. We want our graduates to take with them the capability to read, listen, question, gather and evaluate information resources in a variety of formats, assess, write clearly, speak effectively, and to use visual communication and communication technologies as appropriate.

This graduate capability is supported by:

Learning outcomes

- Articulate a broad knowledge and understanding of the fundamental biological, chemical and physical sciences.
- Understand the principles of and interpret DNA/RNA (and other "omics") technologies

and their applications (e.g., human genome & proteome projects) to personalised medicine
- Evaluate approaches to analyzing the inheritance of complex traits and common diseases

Assessment tasks

- Oral Presentation
- Essay

Engaged and Ethical Local and Global citizens

As local citizens our graduates will be aware of indigenous perspectives and of the nation's historical context. They will be engaged with the challenges of contemporary society and with knowledge and ideas. We want our graduates to have respect for diversity, to be open-minded, sensitive to others and inclusive, and to be open to other cultures and perspectives: they should have a level of cultural literacy. Our graduates should be aware of disadvantage and social justice, and be willing to participate to help create a wiser and better society.

This graduate capability is supported by:

Learning outcomes

- Understand the principles of and interpret DNA/RNA (and other "omics") technologies and their applications (e.g., human genome & proteome projects) to personalised medicine
- Apply simple population genetics tools to appreciate how genes behave in populations

Assessment task

- Oral Presentation

Socially and Environmentally Active and Responsible

We want our graduates to be aware of and have respect for self and others; to be able to work with others as a leader and a team player; to have a sense of connectedness with others and country; and to have a sense of mutual obligation. Our graduates should be informed and active participants in moving society towards sustainability.

This graduate capability is supported by:

Learning outcomes

- Understand the principles of and interpret DNA/RNA (and other "omics") technologies and their applications (e.g., human genome & proteome projects) to personalised medicine
- Apply simple population genetics tools to appreciate how genes behave in populations

Assessment task

- Oral Presentation

Changes from Previous Offering

Not offered previously