MEDI209

Genetics and Genomics in Medicine

MED 0 2018

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 revolutionising 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 broad knowledge and understanding of the fundamental biological, chemical and physical sciences that govern human genetics

Demonstrate an understanding of the molecular basis and biological implications of human inheritance

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

Apply the principle of Mendelian inheritance to analyse transmission of defined traits and simple genetic diseases through human 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.

Create an communal appreciation of ethical considerations around the application of modern "omics" technologies (whole genome and exome sequencing, metabolomics & proteomics) to modern evidence-based personalised medicine

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 (Weeks 2-4) will be conducted in computer data analysis labs. These labs will have a highly investigative approach, where you will be conducting analysis to apply theoretical knowledge to understand genetics, genomics, and other omics data. You will be required to write reports for each of the 3 MEDI209 practicals. Each is only one week long (i.e., 3 x 10% = 30% of your total assessment). The detailed requirements for each report will be given with notes available before or during each practical class. Prac reports are due during the semester two weeks after the practical class is complete. Please check iLearn for due dates. All prac reports should be submitted to the academic responsible for the labs directly.

On successful completion you will be able to:

- Articulate broad knowledge and understanding of the fundamental biological, chemical and physical sciences that govern human genetics
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Oral Presentation

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

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

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

Past Hot Topics and Debates have been video recorded for future marketing of the BClinSci program and MEDI209 unit and **may** occur if physically possible in 2018.

- **Hot Topics** (groups of 3 students) 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. These team presentations address contemporary issues in medicine.The length of each person's Hot Topic presentations will be 5min plus 2min for 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 (positive and negative) of 3 students) will contend an argument as a formal discussion before a public assembly of your peers. 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 broad knowledge and understanding of the fundamental biological, chemical and physical sciences that govern human genetics
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- Understand the principles of and interpret DNA/RNA (and other "omics") technologies, and evaluate their application (e.g., human genome and human proteome projects) to personalised medicine
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Essay

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

Maximum 1,500 word essay (not including any words used in tables, figures, legends or references) on a topic of relevance to personalised/precision medicine. This means that your synthesis of all the ideas you have read and accumulated into new figures, diagnrams, summaries and tables will be highly valued in this exercise.

On successful completion you will be able to:

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- 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.
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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 broad knowledge and understanding of the fundamental biological, chemical and physical sciences that govern human genetics
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- Evaluate approaches to analyzing the inheritance of complex traits and common diseases
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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	**MEDI 209 Lecture, Practical, Hot Topic or Debate Topic** (*"Genetics & Genomics in Medicine"* Strachan et al., 2015 with MQ research examples)	**Lead Academic**	**Date**	**Time**	**MQ Location**
Week 1 L1	DNA, Chromosomes & Cells (Chap 1; pp1-18)	MSB	Jan 8th	12-13	7 Wallys Walk E6B.149
L2	Gene Structure/Expression & the Human Genome (Chap 2; pp19-56)	MSB	Jan 9th	12-13	7 Wallys Walk E6B.149
Prac 1	Highlighting Protein Structure/Function in Rare Human Diseases (Serpinopathies)	MSB	Jan 10th	12-15	6 Eastern Rd E4B.214 PC Lab
Week 2 L3	Underpinning DNA Technologies - PCR, Cloning (Chap 3; pp57-79)	RD	Jan 15th	12-13	7 Wallys Walk E6B.149
L4	Genome Sequencing Technologies Through the Ages (Chap 3)	IB	Jan 16th	12-13	7 Wallys Walk E6B.149
Prac 2	Global Gene Expression Profiling (Transcriptomic Profiling)	HR	Jan 17th	12-15	6 Eastern Rd E4B.214 PC Lab
Week 3 L5	Single Gene Disorders, Inheritance, Allele Frequencies (Chap 5; pp 117-148)	KW	Jan 22nd	12-13	7 Wallys Walk E6B.149
L6	Identifying Disease Genes & Susceptibility (Chap 7; pp189-247)	KW	Jan 23rd	12-13	7 Wallys Walk E6B.149
Prac 3	Next-Gen Sequencing and Integrated Genome Viewer (Disease Prediction and Susceptibility Analysis)	DB (CSIRO)	Jan 24th	12-15	6 Eastern Rd E4B.214 PC Lab
Week 4 L7	Genetic Counselling & Approaches to Treating Disease [Reading Task for Final Exam: Chaps 8 and 9; pp247-370]	AC	Jan 29th	12-13	7 Wallys Walk E6B.149

L8	Epigenetics & Gene Regulation (Chap 6; pp149-188)	SG	Jan 30st	12-13	7 Wallys Walk E6B.149
HT1/2/3/4	1. Does Smoking Have a Long-Lasting Impact on Your Genome? Roby Joehanes et al. Epigenetic Signatures of Cigarette Smoking. Circulation: Cardiovascular Genetics, published online September 20, 2016; doi: 10.1161/circgenetics.116.001506 2. Green et al., Charting a course for genomic medicine from base pairs to bedside. Nature, 470, 204–213, 2011 3. Weisman, et al., Genetic alterations of TNBC by targeted Next-Gen sequencing & correlation with tumor morphology. Modern Path 29, 476–488. 2016. 4. What are the Consequences of Sequencing Healthy People? http://mobile.the-scientist.com/article/49739/the-consequences-of-sequencing-healthy-people/?utm_campaign=newsletter_ts_the-scientist-daily_2016&utm_source=hs_email&utm_medium=email&utm_content=53823557&_hsenc=p2anqtz-_wfuahizsoxngrfemr743sx4lgz2zllijbpj0-7fg8vy-enmfiua4stt5qlx163whmzyohog0nb-ki7fu2dntz1efmug&_hsmi=53823557	MSB	Jan 31st	12-15	4 Western Rd W5C.220 Tutorial Rm
D1	Healthy Lifestyle Is More Important than Known Genetic Risk Factors in Cardiovascular Disease.	MSB	Jan 31st	15-16	4 Western Rd W5C.220 Tutorial Rm
Week 5 L9	Transcriptomics: Global Expression Analysis to Medicine	HR	Feb 5th	12-13	7 Wallys Walk E6B.149
L10	Cancer Genetics, Genomics and the TCGA (Chap 10; pp373-427)	MSB	Feb 6th	12-13	7 Wallys Walk E6B.149
Short Essay (1,500 word)	Critically evaluate Dr Allen Roses (VP GlaxoSmithKline) statement that *"fewer than half of the patients prescribed some of the most expensive drugs actually derived any benefit from them".*	MSB	Feb 7th	10	75 Talavera, Level 1, Office 19

HT5/6/7/8	1. Kaiser J. The gene editor CRISPR won't fully fix sick people anytime soon. Here's why. May 3rd 2016. Science. 2. Ghajar CM &. Bissell MJ., Metastasis: Pathways of parallel progression. Nature 540, 528–529, December 2016. doi:10.1038/nature21104. 3. The Cancer Genome Atlas Network. Comprehensive molecular characterization of human colon and rectal cancer, Nature, 487, 330-7. 2012. doi:10.1038/nature 11252. 4. Can physicians personalize diets, matching specific dietary needs to individual genotypes/lifestyle? Piper MD, et al. Matching dietary amino acid balance to the in silico-translated exome optimizes growth and reproduction without cost to lifespan. Leitao-Goncalves, R et al. Commensal bacteria and essential amino acids control food choice behaviour and reproduction. PLoS Biology April 25, 2017.	MSB	Feb 7th	12-15	4 Western Rd W5C.220 Tutorial Rm
D2	Offspring generated through MRT (often termed as targeted mitochondrial replacement IVF) will (negative) or will not (affirmative) or be adversely affected from the deployment of what effectively is a three-parent technology http://www.biotechniques.com/news/biotechniquesNews/biotechniques-363775.html#.WH1IlH2Ct41	MSB	Feb 7th	15-16	4 Western Rd W5C.220 Tutorial Rm
Week 7 L11	Big Data: The "Omics" Revolution, HPP & Human Protein Atlas	SR	Feb 12th	12-13	7 Wallys Walk E6B.149
L12	Personalised Cancer "Omics", Human Proteome Project & Human Protein Atlas	MSB	Feb 13th	12-13	7 Wallys Walk E6B.149
HT9/10/11/12	1. Proteomic analysis of colon & rectal carcinoma using standard & customized databases. Slebos et al., Scientific Data 2, 150022. 2015. 2. Single-cell genome sequencing: current state of the science, Gawad, Koh & Quake. Nature Reviews Genetics 17, 175–188.2016. 3. Facilitating a culture of responsible and effective sharing of cancer genome data. Siu et al., Nature Medicine 22, 464–471. 2016. 4. Do personal genomics companies (e.g., Helix) provide accurate, reliable DNA-powered products offering insights on ancestry, entertainment, family, fitness, health and nutrition? http://www.businesswire.com/news/home/20170724005412/en/Helix-Launches-Online-Consumer-Marketplace-DNA-Powered-Products	MSB	Feb 14th	12-15	4 Western Rd W5C.220 Tutorial Rm
D3	Next-gen human genome sequencing will allow the diagnosis and treatment of all human cancers within the next decade.	MSB	Feb 14th	15-16	4 Western Rd W5C.220 Tutorial Rm
Final Exam **(2hr)**	An equal combination of Part A (multiple-choice) and Part B (short answer) questions	MSB	Feb 16th	9-11	7 Wallys Walk E6B.149

Lecturers: MSB (Mark Baker, Biomed Sciences FMHS); HR (Helen Rizos, Biomed Sciences FMHS); RD (Russell Diefenbach, Biomed Sciences FMHS); KW (Kelly Williams, Biomed Sciences); IB (Ian Blair, Biomed Sciences FMHS); AC (Ashley Crook, MUH); SG (Stuart Gallagher); SR (Shoba Ranganathan, CBMS MQ); DB (Denis Bauer, CSIRO eHealth transformational bioinformatics).

Spare Debates/Hot Topics (your team is able to propose one of your own BUT you will need coordinator pre-approval in advance to do so)

1. *It is ethical to generate and make available newborn's iPOPs (i.e., integrative personal omics profiles)?*
2. *Personalised omics evidenced-based medicine will solve the current problem that "more than 90% of drugs only work in 30-50% of people".*
3. *How Can We Measure and Eliminate CRISPR-Cas9 Off-Target Effects?* https://www.biotechniques.com/news/biotechniquesNews/biotechniques-362580.html#.WI_af_KCt40
4. *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.*

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). 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). 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).

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 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 broad knowledge and understanding of the fundamental biological, chemical and physical sciences that govern human genetics
- Demonstrate an understanding of the molecular basis and biological implications of human 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 and biological implications of human inheritance
- Understand the principles of and interpret DNA/RNA (and other "omics") technologies, and evaluate their application (e.g., human genome and human 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.
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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 broad knowledge and understanding of the fundamental biological, chemical and physical sciences that govern human genetics
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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 broad knowledge and understanding of the fundamental biological, chemical

and physical sciences that govern human genetics
- Demonstrate an understanding of the molecular basis and biological implications of human inheritance
- Understand the principles of and interpret DNA/RNA (and other "omics") technologies, and evaluate their application (e.g., human genome and human proteome projects) to personalised medicine
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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 broad knowledge and understanding of the fundamental biological, chemical and physical sciences that govern human genetics
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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 and biological implications of human inheritance
- Understand the principles of and interpret DNA/RNA (and other "omics") technologies, and evaluate their application (e.g., human genome and human proteome projects) to personalised medicine
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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 broad knowledge and understanding of the fundamental biological, chemical and physical sciences that govern human genetics
- Understand the principles of and interpret DNA/RNA (and other "omics") technologies, and evaluate their application (e.g., human genome and human proteome projects) to personalised medicine
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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 evaluate their application (e.g., human genome and human proteome projects) to personalised medicine
- Apply simple population genetics tools to appreciate how genes behave in populations

- Create an communal appreciation of ethical considerations around the application of modern "omics" technologies (whole genome and exome sequencing, metabolomics & proteomics) to modern evidence-based personalised medicine

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 evaluate their application (e.g., human genome and human proteome projects) to personalised medicine
- Apply simple population genetics tools to appreciate how genes behave in populations
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Assessment task

- Oral Presentation

Changes from Previous Offering

A review of the BClinSci program has been undertaken in late 2017. After consultation with the MEDI209 2017 class only minor changes have been incorporated into the 2018 unit, with significant changes anticipated at a programatic level in 2019. Following positive comments and constructive feedback, two of the MEDI209 practicals have been simplified/shortened with less emphasis on data construction and increased emphasis on data analysis/interpretation.