



COURSE OUTLINE

BIM202 Medical Genetics

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2021 | Semester 2

USC Sunshine Coast
USC Moreton Bay

ON CAMPUS

Most of your course is on campus but you may be able to do some components of this course online.

Please go to the USC website for up to date information on the teaching sessions and campuses where this course is usually offered.

1. What is this course about?

1.1. Description

Medical genetics will provide you with an overview of the role of genetic factors in human health and disease. Rapid advancements in technology, including human genome sequencing, are allowing genetics to play an increasingly important role in medicine. Medical genetics uses inquiry-based learning in lecture, laboratory and tutorial classes to explain concepts such as: personalised medicine, the genetics of cancer, epigenetics, genetic immunity, human microbiome, reproductive genetics, genetic counselling and ethics, and common molecular and cytogenetic diagnostic techniques.

1.2. How will this course be delivered?

ACTIVITY	HOURS	BEGINNING WEEK	FREQUENCY
ON CAMPUS			
Tutorial/Workshop 1 – Tutorials will be delivered fortnightly in odd weeks	1hr	Week 1	7 times
Laboratory 1 – Laboratory classes will be delivered fortnightly in even weeks.	3hrs	Week 2	6 times
Online – Online lecture modules	2hrs	Week 1	13 times
Online – Online pre-class tutorial activities	1hr	Week 1	7 times
Lecture – Weekly lectorial	1hr	Week 1	13 times

1.3. Course Topics

- Molecular tools for diagnosis and research
- Heritability of genetic diseases
- Ethical implications in medical genetics and genetic counselling
- Cancer genetics
- Biochemical genetics
- Immunogenetics
- Genetic screening
- Reproductive genetics
- Microbiome
- Epigenetics

2. What level is this course?

200 Level (Developing)

Building on and expanding the scope of introductory knowledge and skills, developing breadth or depth and applying knowledge and skills in a new context. May require pre-requisites where discipline specific introductory knowledge or skills is necessary. Normally, undertaken in the second or third full-time year of an undergraduate programs.

3. What is the unit value of this course?

12 units

4. How does this course contribute to my learning?

COURSE LEARNING OUTCOMES	GRADUATE QUALITIES
On successful completion of this course, you should be able to...	Completing these tasks successfully will contribute to you becoming...
1 Describe and explain the concepts and principles behind the genetic regulation of health and disease and how our understanding of these processes can lead to improved health outcomes for individuals and communities.	Knowledgeable Sustainability-focussed
2 Collect, accurately record, interpret and draw conclusions from scientific data.	Empowered
3 Describe and critically analyse ethical issues associated with genetic research, counselling and diagnostics.	Ethical
4 Gather, synthesise and critically evaluate information from a range of sources and communicate this information to a range of audiences.	Empowered

5. Am I eligible to enrol in this course?

Refer to the [USC Glossary of terms](#) for definitions of “pre-requisites, co-requisites and anti-requisites”.

5.1. Pre-requisites

LFS100 or LFS103

5.2. Co-requisites

Not applicable

5.3. Anti-requisites

Not applicable

5.4. Specific assumed prior knowledge and skills (where applicable)

It is recommended that students have some prerequisite knowledge of basic cell biology, and research methods

6. How am I going to be assessed?

6.1. Grading Scale

Standard Grading (GRD)

High Distinction (HD), Distinction (DN), Credit (CR), Pass (PS), Fail (FL).

6.2. Details of early feedback on progress

You will receive both formative and summative feedback on assessment items that are designed to allow you to apply your medical genetics knowledge in both written and practical contexts. BIM202 will include early formative feedback with the completion of weekly lecture, tutorial and laboratory class activities. A quiz in Week 3 will be the first summative low stakes assessment item.

6.3. Assessment tasks

DELIVERY MODE	TASK NO.	ASSESSMENT PRODUCT	INDIVIDUAL OR GROUP	WEIGHTING %	WHAT IS THE DURATION / LENGTH?	WHEN SHOULD I SUBMIT?	WHERE SHOULD I SUBMIT IT?
All	1	Quiz/zes	Individual	40%	2 hours	Throughout teaching period (refer to Format)	Online Test (Quiz)
All	2	Literature Review (or component)	Individual or Group	20%	500 words	Week 10	Online Assignment Submission
All	3	Examination - Centrally Scheduled	Individual	40%	2 hours	Exam Period	Online Test (Quiz)

All - Assessment Task 1: Tutorial and practical portfolio 40%

GOAL:	You will actively engage in and demonstrate learning of the theoretical and practical concepts of medical genetics that have been delivered in the online lectures, tutorial and laboratory classes.	
PRODUCT:	Quiz/zes	
FORMAT:	Four online quizzes, each worth 10%, based on the theoretical and practical concepts covered in lectures, tutorials and laboratory classes. The quizzes are to be completed individually in Weeks 3, 6, 9 and 12. The quizzes may consist of multiple-choice questions and written responses to short answer style questions.	
CRITERIA:	<p>No.</p> <p>1 You will be assessed on the following:</p> <ul style="list-style-type: none"> demonstration of depth and breadth of knowledge of the theoretical content ability to critically analyse and solve medical genetics problems ability to collect, accurately record, and interpret data 	<p>Learning Outcome assessed</p> <p>1 2 3</p>

All - Assessment Task 2: Medical Genetics assignment, 20%

GOAL:	You will explore and develop an understanding of the concepts associated with medical genetics, and will gain skills in the review and critical analysis of relevant literature, and the communication of information to a broader audience. If you opt to work in a group you will also gain skills in team work.	
PRODUCT:	Literature Review (or component)	
FORMAT:	You will work as either an individual or in a group to review scientific literature and produce a 500-word written assignment, with the incorporation of appropriate diagrams, to describe the molecular mechanisms associated with a genetic disorder. Detailed instructions along with other resources will be available on Blackboard. The assignment with in-text references and reference list will be submitted to Blackboard SafeAssign for review (text matching software for plagiarism checking) in Week 10.	
CRITERIA:	<p>No.</p> <p>1 critical analysis of the topic and synthesis of information,</p> <p>2 the scientific merit of the factual information presented,</p> <p>3 communication skills including writing style (grammar, vocabulary, spelling, logical order)</p>	<p>Learning Outcome assessed</p> <p>1 3 4</p> <p>4</p> <p>4</p>

All - Assessment Task 3: End of semester examination, 40%

GOAL:	Task 3 will assess your understanding of and ability to apply theoretical knowledge of the concepts relevant to medical genetics.	
PRODUCT:	Examination - Centrally Scheduled	
FORMAT:	An individual, two-hour final examination, consisting of multiple choice and short answer style questions.	
CRITERIA:	No.	Learning Outcome assessed
	1	You will be assessed on demonstration of knowledge, critical analysis and ability to provide correct answers to questions on the theoretical content covered in the course. 1

7. Directed study hours

A 12-unit course will have total of 150 learning hours which will include directed study hours (including online if required), self-directed learning and completion of assessable tasks. Directed study hours may vary by location. Student workload is calculated at 12.5 learning hours per one unit.

7.1. Schedule

PERIOD AND TOPIC	ACTIVITIES
Week 1, Topic 1: Introduction to Medical Genetics: Course overview. The place of genetics in medicine & the human genome project.	Online lecture modules and weekly lectorial. Tutorial 1: The biology & evolution of human skin colour.
Week 2, Topic 2: Molecular biology overview: DNA structure and replication, gene expression (RNA), protein synthesis, DNA mutation.	Online lecture modules and weekly lectorial. Practical 1: Practical skills development
Week 3, Topic 3: Molecular tools for diagnosis and research: Introduction to PCR, QPCR, karyotyping, microarrays, FISH and sequencing.	Online lecture modules and weekly lectorial. Tutorial 2: Alternative splicing.
Week 4, Topic 4: Genetic diseases – 3 main categories: Monogenic (single gene), chromosomal and multifactorial.	Online lecture modules and weekly lectorial. Practical 2: Fragile X diagnostic testing using PCR.
Week 5, Topic 5: Genetic counselling, ethical and social considerations.	Online lecture modules and weekly lectorial. Tutorial 3: Case study on genetic disease. Monogenic disorder – Thalassemia.
Week 6, Topic 6: Immunogenetics: Genetic immunity/resistance, organ transplant & HLA genetics, autoimmune diseases.	Online lecture modules and weekly lectorial. Practical 3: Haemochromatosis diagnostic testing - HFE gene mutation analysis.
Week 6, Topic 6: Immunogenetics: Genetic immunity/resistance, organ transplant & HLA genetics, autoimmune diseases.	Online lecture modules and weekly lectorial. Practical 3: Haemochromatosis diagnostic testing - HFE gene mutation analysis.
Week 7, Topic 7: Biochemical Genetics and Pharmacogenetics: Personalised medicine.	Online lecture modules and weekly lectorial. Tutorial 4: Early Onset Familial Alzheimer's Disease - Ethics case study.
Week 8, Topic 8: Cancer genetics	Online lecture modules and weekly lectorial. Practical 4: Detection of a Biochemical Disorder – Newborn screening of Phenylketonuria (PKU).
Week 9, Topic 9: Reproductive genetics	Online lecture modules and weekly lectorial. Tutorial 5: Case study on pharmacogenetics – acute lymphoblastic leukaemia.
Week 10, Topic 10: Epigenetics: Epigenetic regulation of gene expression (e.g. histone modification, DNA methylation, microRNAs).	Online lecture modules and weekly lectorial. Practical 5: Cancer Genetics - Li Fraumeni Syndrome (LFS) TP53 gene defect.
Week 11, Topic 11: Treatment of genetic disease	Online lecture modules and weekly lectorial. Tutorial 6: Identical Twins, Identical Fates? An Introduction to Epigenetics.
Week 12, Topic 12: Human Microbiome	Online lecture modules and weekly lectorial. Practical 6: Pathogen diagnostic testing.
Week 13: Semester revision	Weekly lectorial – semester review. Tutorial 7: Treatment of genetic disease.

8. What resources do I need to undertake this course?

Please note: Course information, including specific information of recommended readings, learning activities, resources, weekly readings, etc. are available on the course Blackboard site– Please log in as soon as possible.

8.1. Prescribed text(s) or course reader

Please note that you need to have regular access to the resource(s) listed below. Resources may be required or recommended.

REQUIRED?	AUTHOR	YEAR	TITLE	PUBLISHER
Required	Turnpenny, P. & Ellard, S.	2017	Emery's Elements of Medical Genetics	Elsevier
Recommended	Urry, L.A., Meyers, N., Cain, M.L., Wasserman, S.A., Minorsky, P.V., Reece, J.B.	2018	Campbell Biology	Pearson

8.2. Specific requirements

Laboratory coat, safety glasses, covered shoes and the BIM202 laboratory manual are required for practical classes.

9. How are risks managed in this course?

Risk assessments have been performed for all laboratory classes and a moderate level of health and safety risk exists. Moderate risks are those associated with laboratory work such as working with chemicals and hazardous substances. You will be required to undertake laboratory induction training and it is also your responsibility to review course material, search online, discuss with lecturers and peers and understand the health and safety risks associated with your specific course of study and to familiarise yourself with the University's general health and safety principles by reviewing the [online induction training for students](#), and following the instructions of the University staff

10. What administrative information is relevant to this course?

10.1. Assessment: Academic Integrity

Academic integrity is the ethical standard of university participation. It ensures that students graduate as a result of proving they are competent in their discipline. This is integral in maintaining the value of academic qualifications. Each industry has expectations and standards of the skills and knowledge within that discipline and these are reflected in assessment.

Academic integrity means that you do not engage in any activity that is considered to be academic fraud; including plagiarism, collusion or outsourcing any part of any assessment item to any other person. You are expected to be honest and ethical by completing all work yourself and indicating in your work which ideas and information were developed by you and which were taken from others. You cannot provide your assessment work to others. You are also expected to provide evidence of wide and critical reading, usually by using appropriate academic references.

In order to minimise incidents of academic fraud, this course may require that some of its assessment tasks, when submitted to Blackboard, are electronically checked through SafeAssign. This software allows for text comparisons to be made between your submitted assessment item and all other work that SafeAssign has access to.

10.2. Assessment: Additional Requirements

Eligibility for Supplementary Assessment

Your eligibility for supplementary assessment in a course is dependent of the following conditions applying:

- The final mark is in the percentage range 47% to 49.4%
- The course is graded using the Standard Grading scale
- You have not failed an assessment task in the course due to academic misconduct

10.3. Assessment: Submission penalties

Late submission of assessment tasks will be penalised at the following maximum rate:

- 5% (of the assessment task's identified value) per day for the first two days from the date identified as the due date for the assessment task.
- 10% (of the assessment task's identified value) for the third day
- 20% (of the assessment task's identified value) for the fourth day and subsequent days up to and including seven days from the date identified as the due date for the assessment task.
- A result of zero is awarded for an assessment task submitted after seven days from the date identified as the due date for the assessment task.

Weekdays and weekends are included in the calculation of days late.

To request an extension, you must contact your Course Coordinator and supply the required documentation to negotiate an outcome.

10.4. Study help

For help with course-specific advice, for example what information to include in your assessment, you should first contact your tutor, then your course coordinator, if needed.

If you require additional assistance, the Learning Advisers are trained professionals who are ready to help you develop a wide range of academic skills. Visit the [Learning Advisers](#) web page for more information, or contact Student Central for further assistance: +61 7 5430 2890 or studentcentral@usc.edu.au.

10.5. Wellbeing Services

Student Wellbeing provide free and confidential counselling on a wide range of personal, academic, social and psychological matters, to foster positive mental health and wellbeing for your academic success.

To book a confidential appointment go to [Student Hub](#), email studentwellbeing@usc.edu.au or call 07 5430 1226.

10.6. AccessAbility Services

Ability Advisers ensure equal access to all aspects of university life. If your studies are affected by a disability, learning disorder mental health issue, , injury or illness, or you are a primary carer for someone with a disability or who is considered frail and aged, [AccessAbility Services](#) can provide access to appropriate reasonable adjustments and practical advice about the support and facilities available to you throughout the University.

To book a confidential appointment go to [Student Hub](#), email AccessAbility@usc.edu.au or call 07 5430 2890.

10.7. Links to relevant University policy and procedures

For more information on Academic Learning & Teaching categories including:

- Assessment: Courses and Coursework Programs
- Review of Assessment and Final Grades
- Supplementary Assessment
- Administration of Central Examinations
- Deferred Examinations
- Student Academic Misconduct
- Students with a Disability

Visit the USC website: <http://www.usc.edu.au/explore/policies-and-procedures#academic-learning-and-teaching>

10.8. General Enquiries

In person:

- **USC Sunshine Coast** - Student Central, Ground Floor, Building C, 90 Sippy Downs Drive, Sippy Downs
- **USC Moreton Bay** - Service Centre, Ground Floor, Foundation Building, Gympie Road, Petrie
- **USC SouthBank** - Student Central, Building A4 (SW1), 52 Merivale Street, South Brisbane
- **USC Gympie** - Student Central, 71 Cartwright Road, Gympie
- **USC Fraser Coast** - Student Central, Student Central, Building A, 161 Old Maryborough Rd, Hervey Bay
- **USC Caboolture** - Student Central, Level 1 Building J, Cnr Manley and Tallon Street, Caboolture

Tel: +61 7 5430 2890

Email: studentcentral@usc.edu.au