Course Outline

Code: BIM202
Title: Medical Genetics

School: Health & Sport Sciences
Teaching Session: Semester 1
Year: 2019
Course Coordinator: Dr Anna Kuballa  Tel: (07) 5459 5582  Email: akuballa@usc.edu.au
Course Moderator: Dr Ann Parkinson

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
   BIM202 will provide you with an introduction to the principles and concepts integral to the field of medical genetics, using inquiry based learning in laboratory and tutorial classes. Rapid advancements in technology, including mapping of the human genome, are allowing genetics to play an increasingly important role in medicine. This course uses relevant examples to explain topics such as personalised medicine, the genetics of cancer, epigenetics, genetic immunity, human microbiome, reproductive genetics, and ethics.

   1.2 Course topics
   The topics covered include:
   - Gene inheritance, expression and regulation
   - Epigenetics
   - Heritable diseases
   - Cancer genetics
   - Biochemical genetics and pharmacogenetics
   - Immunogenetics
   - Genetic counselling and ethical issues in genetics
   - Reproductive genetics
   - Treatment of genetic disease
   - Human Microbiome

2. What level is this course?
   200 level Developing - Applying broad and/or deep knowledge and skills to new contexts. May require pre-requisites and introductory level knowledge/skills. Normally undertaken in the 2nd or 3rd year of an undergraduate program

3. What is the unit value of this course?
   12 units
4. **How does this course contribute to my learning?**

<table>
<thead>
<tr>
<th>Specific Learning Outcomes</th>
<th>Assessment Tasks</th>
<th>Graduate Qualities or Professional Standards mapping</th>
</tr>
</thead>
<tbody>
<tr>
<td>On successful completion of this course you should be able to:</td>
<td>You will be assessed on the learning outcome in task/s:</td>
<td>Completing these tasks successfully will contribute to you becoming:</td>
</tr>
</tbody>
</table>

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.

Task 1a, 1b, 2 & 3

Knowledgeable.

Sustainability-focussed.

Collect, accurately record, interpret and draw conclusions from scientific data.

Task 1a

Empowered.

Describe and critically analyse ethical issues associated with genetic research, counselling and diagnostics.

Task 1a & 1b

Ethical.

Gather, synthesise and critically evaluate information from a range of sources and communicate this information to a range of audiences using multi-media.

Task 2

Empowered.

5. **Am I eligible to enrol in this course?**

Refer to the [USC Glossary of terms](https://www.usc.edu.au/) for definitions of “pre-requisites, co-requisites and anti-requisites”.

5.1 **Enrolment restrictions**

Nil

5.2 **Pre-requisites**

LFS100 or (LFS103 and enrolled in Program SE303 or AE304) OR

5.3 **Co-requisites**

LFS100 and enrolled in SC357

5.4 **Anti-requisites**

Nil

5.5 **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 – High Distinction (HD), Distinction (DN), Credit (CR), Pass (PS), Fail (FL)

6.2 **Details of early feedback on progress**

BIM202 will include early assessment with the completion of weekly tutorial and laboratory class activities beginning in Week 3. You will receive both formative and summative in-class feedback on low stakes assessment items that are designed to allow you to apply your medical genetics knowledge in both written and practical contexts.
### 6.3 Assessment tasks

<table>
<thead>
<tr>
<th>Task No.</th>
<th>Assessment Tasks</th>
<th>Individual or Group</th>
<th>Weighting %</th>
<th>What is the duration / length?</th>
<th>When should I submit?</th>
<th>Where should I submit it?</th>
</tr>
</thead>
<tbody>
<tr>
<td>1a</td>
<td>Tutorial and practical portfolio</td>
<td>Individual</td>
<td>20</td>
<td>~1000 words cumulative</td>
<td>Weeks 3-12</td>
<td>In class</td>
</tr>
<tr>
<td>1b</td>
<td>Ethical implications in medical genetics: Case studies</td>
<td>Individual</td>
<td>10</td>
<td>~500 words</td>
<td>Week 7</td>
<td>Blackboard</td>
</tr>
<tr>
<td>2</td>
<td>Medical genetics assignment</td>
<td>Group</td>
<td>25</td>
<td>Scientific review of the literature ~1000 words</td>
<td>Week 10</td>
<td>Blackboard</td>
</tr>
<tr>
<td>3</td>
<td>End of Semester Examination</td>
<td>Individual</td>
<td>45</td>
<td>2 hours</td>
<td>Central Exam Period</td>
<td>Examination venue</td>
</tr>
<tr>
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</tbody>
</table>

|                           | 100%                                                                             |                     |             |                               |                       |                           |

**Assessment Task 1a: Tutorial and practical portfolio 20%**

**Goal:** This is designed to actively engage you in the theoretical and practical components of the role of genes in health and disease in tutorial and laboratory classes.

**Product:** Tutorial and laboratory pre-class and in class activities in association with 2 x closed book in class review quizzes.

**Format:** From weeks 3-12 you will complete both pre- and in class activities during your tutorial & laboratory classes. There will be 2 in-class review quizzes (weeks 6, and 12) that will be based on concepts from class. You will receive formative and summative feedback on the completion of tasks in tutorial & laboratory classes. Further details available on Blackboard.

**Criteria:**
- You will be assessed on the level of successful completion of the tutorial and laboratory activities (4%), and also on your understanding of the concepts with 2 in-class review quizzes each worth 8% in weeks 6 and 12.

**Assessment Task 1b: Ethical implications in genetics: Case studies, 10%**

**Goal:** This task has been designed to develop your critical thinking about ethical issues associated with medical genetics and genetic counselling.

**Product:** A discussion of ethical issues associated with medical genetics and genetic counselling.

**Format:** You will produce a written description of key ethical issues that relate to cases in medical genetics and genetic counselling. ~500 words (details for this task will be available on Blackboard)

**Criteria:**
- You will be assessed on
  - critical analysis
  - identification of key ethical issues
Assessment Task 2: Medical Genetics assignment, 25%

<table>
<thead>
<tr>
<th>Goal</th>
<th>You will explore and develop an understanding of the concepts associated with medical genetics, and will gain skills in group work, review and critical analysis of the relevant literature.</th>
</tr>
</thead>
<tbody>
<tr>
<td>Product</td>
<td>A scientific review of the literature (~1000 words).</td>
</tr>
<tr>
<td>Format</td>
<td>You will work either as an individual or in a group to produce a scientific review of the literature relevant to a medical genetics concept. Detailed instructions along with other resources will be available on Blackboard.</td>
</tr>
</tbody>
</table>
| Criteria | You will be assessed on  
• critical analysis of the topic and synthesis of information,  
• the scientific merit of the factual information presented,  
• communication skills including writing style (grammar, vocabulary, spelling, logical order) |

Assessment Task 3: End of semester examination, 45%

<table>
<thead>
<tr>
<th>Goal</th>
<th>This task has been designed to assess your understanding of, and ability to apply, theoretical knowledge of the principles behind medical genetics.</th>
</tr>
</thead>
<tbody>
<tr>
<td>Product</td>
<td>Written invigilated examination</td>
</tr>
<tr>
<td>Format</td>
<td>An individual, closed book, two hour final examination, consisting of multiple choice and short answer questions.</td>
</tr>
<tr>
<td>Criteria</td>
<td>A demonstration of knowledge, and ability to provide correct answers to questions on the theoretical content covered in the course.</td>
</tr>
</tbody>
</table>

7. What are the course activities?

7.1 Directed study hours

The directed study hours listed here are a portion of the workload for this course. 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.

<table>
<thead>
<tr>
<th>Location: Specific Campus(es) or online</th>
<th>Directed study hours for location:</th>
</tr>
</thead>
<tbody>
<tr>
<td>USC Sippy Downs</td>
<td>Lectures – two hours per week</td>
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<td></td>
<td>Tutorials – two hours per fortnight</td>
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<td></td>
<td>Practicals – two hour laboratory classes per fortnight</td>
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<tr>
<td></td>
<td>Tutorial and practical classes are delivered on alternating weeks</td>
</tr>
</tbody>
</table>

7.2 Course content

<table>
<thead>
<tr>
<th>Week # / Module #</th>
<th>What key concepts/content will I learn?</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>Introduction to Medical Genetics: Course overview. The place of genetics in medicine. The Human Genome Project</td>
</tr>
<tr>
<td>2</td>
<td>Molecular biology overview: DNA structure and replication, gene expression (RNA), protein synthesis, DNA mutation.</td>
</tr>
<tr>
<td>3</td>
<td>Molecular tools for diagnosis and research in medical genetics: Introduction to PCR, QPCR, karyotyping, microarrays, FISH, and next generation sequencing</td>
</tr>
<tr>
<td>4</td>
<td>Genetic diseases – 3 main categories: Monogenic (single gene), chromosomal and multifactorial</td>
</tr>
<tr>
<td>5</td>
<td>Genetic counselling, ethical and social considerations</td>
</tr>
<tr>
<td>6</td>
<td>Cancer genetics</td>
</tr>
<tr>
<td>7</td>
<td>Biochemical genetics and pharmacogenetics: Personalised medicine</td>
</tr>
</tbody>
</table>
8. What resources do I need to undertake this course?
Please note that 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)
Please note that you need to have regular access to the resource(s) listed below as they are required:

<table>
<thead>
<tr>
<th>Author</th>
<th>Year</th>
<th>Title</th>
<th>Publisher</th>
</tr>
</thead>
</table>

Recommended readings:

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

9. Risk management
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. It is your responsibility as a student 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. It is also your responsibility to familiarise yourself with the University’s general health and safety principles by reviewing the online Health Safety and Wellbeing training module 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:
a) The final mark is in the percentage range 47% to 49.4%
b) The course is graded using the Standard Grading scale
c) 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

In the first instance, you should contact your tutor, then the Course Coordinator. Additional assistance is provided to all students through Academic Skills Advisers. To book an appointment or find a drop-in session go to Student Hub.
Contact Student Central for further assistance: +61 7 5430 2890 or studentcentral@usc.edu.au

10.5 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
10.6 General Enquiries

In person:
- **USC Sunshine Coast** - Student Central, Ground Floor, Building C, 90 Sippy Downs Drive, Sippy Downs
- **USC South Bank** - 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