

# **BIOL3120**

## **Human Genetics and Evolutionary Medicine**

Session 1, In person-scheduled-weekday, North Ryde 2024

School of Natural Sciences

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#### Disclaimer

Macquarie University has taken all reasonable measures to ensure the information in this publication is accurate and up-to-date. However, the information may change or become out-dated as a result of change in University policies, procedures or rules. The University reserves the right to make changes to any information in this publication without notice. Users of this publication are advised to check the website version of this publication [or the relevant faculty or department] before acting on any information in this publication.

#### **General Information**

Unit convenor and teaching staff Oliver Griffith oliver.griffith@mq.edu.au

Credit points 10

Prerequisites 130cp at 1000 level or above including BIOL2110 or BIOL206(P)

Corequisites

Co-badged status

Unit description

This unit deals with the molecular, cellular and population basis of the genetics of human beings in relation to disease and evolutionary medicine. Topics include: genetics, genomics, disease gene discovery, evolutionary medicine and the social and ethical implications of studies in human genetics. Emphasis is placed upon the enormous impact which recent molecular advances have had upon the subject, as well as techniques of genetic analysis. Comparisons with the genetics of other vertebrates are made wherever appropriate.

#### Important Academic Dates

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

## **Learning Outcomes**

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

**ULO1:** Solve problems in human genetics using appropriate analytical methods and a variety of up to date resources

ULO2: Interpret and demonstrate understanding of the primary scientific literature

**ULO3:** Explain the importance of new techniques in human genetics for understanding human disease

**ULO4:** Explain the principles of evolutionary biology and their role in human health and disease

 $\textbf{ULO5:} \ \textbf{Utilise basic bioinformatic skills, including handling of genetic sequence data}$ 

**ULO6:** Understand genetics and its basis in human disease

## **General Assessment Information**

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#### **Requirements to Pass this Unit**

To pass this unit you must ahieve a total mark equal to or greater than 50%.

There are no hurdle assessments.

#### Late Assessment Submission and Penalties

Unless a Special Consideration request has been submitted and approved, a 5% penalty (of the total possible mark of the task) will be applied for each day a written report or presentation assessment is not submitted, up until the 7<sup>th</sup> day (including weekends). After the 7<sup>th</sup> day, a grade of '0' will be awarded even if the assessment is submitted. The submission time for all uploaded assessments is **11:55 pm**. A 1-hour grace period will be provided to students who experience a technical concern. For any late submission of time-sensitive tasks, such as scheduled tests/ exams, performance assessments/presentations, and/or scheduled practical assessments/labs, please apply for Special Consideration.

#### Assessments where Late Submissions will be accepted

- AT1 Problem Sets: NO special consideration required
- AT2 Literature Review: YES (standard late penalties apply)
- AT3 Final Exam: NO special consideration required

#### **Special Considerations**

The <u>Special Consideration Policy</u> aims to support students who have been impacted by shortterm circumstances or events that are serious, unavoidable and significantly disruptive, and which may affect their performance in assessment. If you experience circumstances or events that affect your ability to complete the assessments in this unit on time, please inform the convenor and submit a Special Consideration request through ask.mq.edu.au.

#### Assessment Tasks

Name	Weighting	Hurdle	Due
Examination	50%	No	Formal exam period
Literature review	25%	No	Week 9
Problem sets	25%	No	Week 3-11

#### Examination

Assessment Type 1: Examination Indicative Time on Task 2: 32 hours Due: **Formal exam period** Weighting: **50%** 

Formal examination covering all content of unit

On successful completion you will be able to:

- Solve problems in human genetics using appropriate analytical methods and a variety of up to date resources
- Explain the importance of new techniques in human genetics for understanding human disease
- · Explain the principles of evolutionary biology and their role in human health and disease
- Utilise basic bioinformatic skills, including handling of genetic sequence data
- · Understand genetics and its basis in human disease

#### Literature review

Assessment Type 1: Literature review Indicative Time on Task 2: 25 hours Due: **Week 9** Weighting: **25%** 

A written literature review of the methodology of a genomic technique

On successful completion you will be able to:

- · Interpret and demonstrate understanding of the primary scientific literature
- Explain the importance of new techniques in human genetics for understanding human disease
- Understand genetics and its basis in human disease

#### Problem sets

Assessment Type 1: Problem set Indicative Time on Task 2: 25 hours Due: Week 3-11 Weighting: 25%

Ongoing problem sets for practicals throughout the semester

On successful completion you will be able to:

- Solve problems in human genetics using appropriate analytical methods and a variety of up to date resources
- · Interpret and demonstrate understanding of the primary scientific literature
- Explain the principles of evolutionary biology and their role in human health and disease
- Utilise basic bioinformatic skills, including handling of genetic sequence data
- · Understand genetics and its basis in human disease

<sup>1</sup> If you need help with your assignment, please contact:

- the academic teaching staff in your unit for guidance in understanding or completing this type of assessment
- the Writing Centre for academic skills support.

<sup>2</sup> Indicative time-on-task is an estimate of the time required for completion of the assessment task and is subject to individual variation

#### **Delivery and Resources**

#### Some notes on unit delivery

#### Week 1 classes

In week 1 you will be required to engage and complete the lecture activities, available through iLearn. Please also attend the drop in session Thursday 2pm (zoom link on iLearn).

Practical classes will not run until week 3.

#### Methods of communication

We will communicate with you via your university email or through announcements on iLearn. Queries to convenors can either be placed on the iLearn discussion board or sent to oliver.griffith@mq.edu.au

#### **COVID** Information

For the latest information on the University's response to COVID-19, please refer to the Coronavirus infection page on the Macquarie website: <a href="https://www.mq.edu.au/about/coronavirus-">https://www.mq.edu.au/about/coronavirus-</a>

fags. Remember to check this page regularly in case the information and requirements change during semester. If there are any changes to this unit in relation to COVID, these will be communicated via iLearn.

Changes to the unit from last offering

We value student feedback to be able to continually improve the way we offer our units. As such we encourage students to provide constructive feedback via student surveys, to the teaching staff directly, or via the FSE Student Experience & Feedback link in the iLearn page.

Student feedback from the previous offering of this unit was very positive overall, with students pleased with the clarity around assessment requirements and the level of support from teaching staff. As such, no change to the delivery of the unit is planned, however we will continue to strive to improve the level of support and the level of student engagement.

## **Policies and Procedures**

Macquarie University policies and procedures are accessible from Policy Central (https://policie s.mq.edu.au). 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
- Assessment Procedure
- Complaints Resolution Procedure for Students and Members of the Public
- Special Consideration Policy

Students seeking more policy resources can visit <u>Student Policies</u> (<u>https://students.mq.edu.au/su</u> <u>pport/study/policies</u>). It is your one-stop-shop for the key policies you need to know about throughout your undergraduate student journey.

To find other policies relating to Teaching and Learning, visit <u>Policy Central</u> (<u>https://policies.mq.e</u> du.au) and use the <u>search tool</u>.

#### **Student Code of Conduct**

Macquarie University students have a responsibility to be familiar with the Student Code of Conduct: https://students.mq.edu.au/admin/other-resources/student-conduct

#### Results

Results published on platform other than <u>eStudent</u>, (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 <u>eStudent</u>. For more information visit <u>ask.mq.edu.au</u> or if you are a Global MBA student contact <u>globalmba.support@mq.edu.au</u>

### Academic Integrity

At Macquarie, we believe academic integrity – honesty, respect, trust, responsibility, fairness and courage – is at the core of learning, teaching and research. We recognise that meeting the expectations required to complete your assessments can be challenging. So, we offer you a range of resources and services to help you reach your potential, including free <u>online writing an</u> d maths support, academic skills development and wellbeing consultations.

#### Student Support

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

#### **The Writing Centre**

The Writing Centre provides resources to develop your English language proficiency, academic writing, and communication skills.

- Workshops
- Chat with a WriteWISE peer writing leader
- Access StudyWISE
- Upload an assignment to Studiosity
- Complete the Academic Integrity Module

The Library provides online and face to face support to help you find and use relevant information resources.

- Subject and Research Guides
- Ask a Librarian

#### Student Services and Support

Macquarie University offers a range of Student Support Services including:

- IT Support
- · Accessibility and disability support with study
- Mental health support
- Safety support to respond to bullying, harassment, sexual harassment and sexual assault
- Social support including information about finances, tenancy and legal issues
- <u>Student Advocacy</u> provides independent advice on MQ policies, procedures, and processes

#### **Student Enquiries**

Got a question? Ask us via AskMQ, or contact Service Connect.

## IT Help

For help with University computer systems and technology, visit <u>http://www.mq.edu.au/about\_us/</u>offices\_and\_units/information\_technology/help/.

When using the University's IT, you must adhere to the <u>Acceptable Use of IT Resources Policy</u>. The policy applies to all who connect to the MQ network including students.

Unit information based on version 2024.01R of the Handbook