



BIOL3120

Human Genetics and Evolutionary Medicine

Session 1, In person-scheduled-weekday, North Ryde 2022

School of Natural Sciences

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General Information

Unit convenor and teaching staff

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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 <https://www.mq.edu.au/study/calendar-of-dates>

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

ULO5: Utilise basic bioinformatic skills, including handling of genetic sequence data

ULO6: Understand genetics and its basis in human disease

General Assessment Information

General Faculty Policy on assessment submission deadlines and late submissions:

Online quizzes, in-class activities, or scheduled tests and exam must be undertaken at the time indicated in the unit guide. Should these activities be missed due to illness or misadventure, students may apply for Special Consideration.

All other assessments must be submitted by 5:00 pm on their due date. Should these assessments be missed due to illness or misadventure, students should apply for Special Consideration.

Late submissions will be accepted for all assessments in this unit with penalties.

Penalties will be applied for late submissions as follows:

A 12-hour grace period will be given after which the following deductions will be applied to the awarded assessment mark: 12 to 24 hours late = 10% deduction; for each day thereafter, an additional 10% per day or part thereof will be applied until five days beyond the due date. After this time, a mark of zero (0) will be given. For example, an assessment worth 20% is due 5 pm on 1 January. Student A submits the assessment at 1 pm, 3 January. The assessment received a mark of 15/20. A 20% deduction is then applied to the mark of 15, resulting in the loss of three (3) marks. Student A is then awarded a final mark of 12/20.

Assessment Tasks

| Name | Weighting | Hurdle | Due |
|--------------------------|-----------|--------|--------------------|
| <u>Examination</u> | 50% | No | Formal exam period |
| <u>Literature review</u> | 25% | No | Week 13 |
| <u>Problem sets</u> | 25% | No | Week 3-12 |

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 13**

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 3-12**

Weighting: **25%**

Ongoing problem sets for tutorials 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
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¹ 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.

² 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

Off-shore students

Off-shore students **must** email the convenor as soon as possible to discuss study options.

COVID Information and on-campus classes

On-campus teaching continues to be scheduled for Session 1, 2022. Masks are compulsory for all classes in indoor spaces and social distancing will be implemented wherever possible. Students will also be required to sanitise surfaces before and after use.

Students are requested to minimise the risk of spreading COVID to themselves and others in accordance with the university and NSW Health guidelines: <https://www.mq.edu.au/about/corona-virus-faqs> and <https://www.nsw.gov.au/covid-19/stay-safe>.

Any further requirements or changes to units in relation to COVID will be communicated to students via iLearn.

Policies and Procedures

Macquarie University policies and procedures are accessible from [Policy Central](https://policies.mq.edu.au) (<https://policies.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 [Student Policies](https://students.mq.edu.au/support/study/policies) (<https://students.mq.edu.au/support/study/policies>). 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 [Policy Central](https://policies.mq.edu.au) (<https://policies.mq.edu.au>) and use the [search tool](#).

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 [eStudent](#), (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 [eStudent](#). For more information visit ask.mq.edu.au or if you are a Global MBA student contact globalmba.support@mq.edu.au

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 [online writing and maths support](#), [academic skills development](#) and [wellbeing consultations](#).

Student Support

Macquarie University provides a range of support services for students. For details, visit <http://students.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](#)

Student Enquiries

Got a question? Ask us via [AskMQ](#), or contact [Service Connect](#).

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.