



BIOL3120

Human Genetics and Evolutionary Medicine

Session 1, Infrequent attendance, North Ryde 2020

Department of Biological Sciences

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General Information

Unit convenor and teaching staff

Michael Gibbs

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Kate Barry

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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. Topics include: pedigree analysis; population studies; inbreeding; cytogenetics; organisation of the human genome; identifying genes for single-gene and complex disorders; cancer genetics; developmental genetics; behavioural genetics; sex determination; immunogenetics; twin studies; genetic counselling; genetic screening; 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:** Use a variety of resources to find up to date information in human genetics
- ULO2:** Solve problems in human genetics using appropriate analytical methods
- ULO3:** Read and demonstrate understanding of the primary scientific literature
- ULO4:** Learn basic bioinformatic skills, including handling of genetic sequence data

Assessment Tasks

Coronavirus (COVID-19) Update

Assessment details are no longer provided here as a result of changes due to the Coronavirus (COVID-19) pandemic.

Students should consult [iLearn](#) for revised unit information.

[Find out more about the Coronavirus \(COVID-19\) and potential impacts on staff and students](#)

General Assessment Information

Problem sets are provided for weeks 2-3 and 8-12, and expected to take ~2 hours to complete. They will be released on Monday mornings and due at midnight Sunday night. Note that iLearn will release answers once the quizzes close, so late submissions will not be possible for problem sets.

The **bioinformatics report** is a short piece of work students will create using the skills they learn in bioinformatics practicals in weeks 4-6. Students will be provided a gene and patient sequences, and are expected to analyse and interpret the clinical significance of mutations found in these sequences. Note that a small part of the assignment (mutation identifications) needs to be submitted in week 5, and your bioinformatics report submitted in week 7.

The **topic essay** is an extended response to a selection of questions which will be provided in the second half of the semester. This essay should show a clear understanding of the genetics involved, as well as clinical and ethical considerations of the topic. Note that students are able to come up with their own question **in consultation with the unit convenor**, if they have a particular area of interest.

The **final exam** will have short answer and problem set style questions covering content from the entire unit.

Plagiarism: All written work must be in the student's own words. Assessments submitted via Turnitin will be subjected to plagiarism detection. Plagiarism will not be tolerated. Penalties for plagiarism range from loss of marks to awarding of a zero, depending upon the level of plagiarism, and reporting to the Faculty Disciplinary Committee.

Late submissions: Assessments submitted after the due date and without an approved Special Consideration will be penalised 10% of the total mark per day late. For example, if a student achieves a mark of 15/20 for an assignment submitted two days late, their penalty is -4 marks, and they will receive a final mark of 11/20.

Delivery and Resources

Coronavirus (COVID-19) Update

Any references to on-campus delivery below may no longer be relevant due to COVID-19.

Please check here for updated delivery information: https://ask.mq.edu.au/account/pub/display/unit_status

Online Unit: We are providing an online-only offering of BIOL3120 this year. There will be two lectures and a tutorial or practical provided online each week.

Note that support will be available from the unit convenor via Skype/Zoom, and in-person assistance may be possible depending on staff availability. There will be no difference for students enrolled as internal or external. For students who would prefer to take an internal unit, please consider taking [BMOL3402 - Molecular Biology and Genomics](#), which is open to students who achieved a credit or better in BIOL206.

Policies and Procedures

Macquarie University policies and procedures are accessible from [Policy Central \(https://staff.mq.edu.au/work/strategy-planning-and-governance/university-policies-and-procedures/policy-central\)](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.*)

Students seeking more policy resources can visit the [Student Policy Gateway \(https://students.mq.edu.au/support/study/student-policy-gateway\)](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 \(https://staff.mq.edu.au/work/strategy-planning-and-governance/university-policies-and-procedures/policy-central\)](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 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

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 help you improve your marks and take control of your study.

- [Getting help with your assignment](#)
- [Workshops](#)
- [StudyWise](#)
- [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

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

If you are a Global MBA student contact globalmba.support@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.

Changes since First Published

Date	Description
20/02/2020	Updated plagiarism and late submission policies.