



BIOL345

Human Genetics Theory

S1 Day 2018

Dept of Biological Sciences

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General Information

Unit convenor and teaching staff

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Credit points

3

Prerequisites

(39cp at 100 level or above) including 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:

- Use a variety of resources to find up to date information in human genetics
- Solve problems in human genetics using appropriate analytical methods
- Read and demonstrate understanding of the primary scientific literature
- Learn basic bioinformatic skills, including handling of genetic sequence data

General Assessment Information

If you receive special consideration for the final exam, a supplementary exam will be scheduled in the interval between the regular exam period and the start of the next session. By making a special consideration application for the final exam you are declaring yourself available for a resit during the supplementary examination period and will not be eligible for a second special consideration approval based on pre-existing commitments. Please ensure you are familiar with the policy prior to submitting an application. You can check the supplementary exam information page on FSE101 in iLearn (bit.ly/FSESupp) for dates, and approved applicants will receive an individual notification one week prior to the exam with the exact date and time of their supplementary examination.

Assessment Tasks

Name	Weighting	Hurdle	Due
Fortnightly online quiz	10%	No	Fortnightly
Database and literature search	5%	No	TBC
Human disease report	30%	No	TBC
Examination	50%	No	Exam Period
Participation in Practicals	5%	No	Ongoing

Fortnightly online quiz

Due: **Fortnightly**

Weighting: **10%**

The aim of this exercise is to become familiar with solving genetic problems.

On successful completion you will be able to:

- Solve problems in human genetics using appropriate analytical methods

Database and literature search

Due: **TBC**

Weighting: **5%**

The aim of this exercise is to encourage you to explore the resources in human genetics available on the internet, and to extract information from them to show a complete understanding.

On successful completion you will be able to:

- Use a variety of resources to find up to date information in human genetics
- Read and demonstrate understanding of the primary scientific literature

Human disease report

Due: **TBC**

Weighting: **30%**

The aim of this exercise is to apply knowledge from the computer practical classes and provide a diagnosis report of an unknown human disease.

On successful completion you will be able to:

- Use a variety of resources to find up to date information in human genetics
- Read and demonstrate understanding of the primary scientific literature
- Learn basic bioinformatic skills, including handling of genetic sequence data

Examination

Due: **Exam Period**

Weighting: **50%**

The final examination will require solving of analytical genetics problems, defining genetic terms and discussion questions.

On successful completion you will be able to:

- Solve problems in human genetics using appropriate analytical methods
- Read and demonstrate understanding of the primary scientific literature

Participation in Practicals

Due: **Ongoing**

Weighting: **5%**

Participation in computer practicals is mandatory and will count towards the final grade.

On successful completion you will be able to:

- Learn basic bioinformatic skills, including handling of genetic sequence data

Delivery and Resources

LECTURES

There are two one-hour lectures per week. Internal students will receive their lectures in the usual way. All lectures will be recorded live and will be available via the iLearn unit through Echo360.

COMPUTER PRACTICALS

There are fortnightly computer practicals that are compulsory, where students will learn basic bioinformatic skills to handle genetic data. External students will attend full-day on-campus session.

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.*)

Undergraduate 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 shown in *iLearn*, 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.

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 improve your marks and take control of your study.

- [Workshops](#)
- [StudyWise](#)
- [Academic Integrity Module for Students](#)
- [Ask a Learning Adviser](#)

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

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.

Graduate Capabilities

Creative and Innovative

Our graduates will also be capable of creative thinking and of creating knowledge. They will be imaginative and open to experience and capable of innovation at work and in the community. We want them to be engaged in applying their critical, creative thinking.

This graduate capability is supported by:

Assessment task

- Human disease report

Capable of Professional and Personal Judgement and Initiative

We want our graduates to have emotional intelligence and sound interpersonal skills and to demonstrate discernment and common sense in their professional and personal judgement. They will exercise initiative as needed. They will be capable of risk assessment, and be able to handle ambiguity and complexity, enabling them to be adaptable in diverse and changing environments.

This graduate capability is supported by:

Learning outcome

- Use a variety of resources to find up to date information in human genetics

Commitment to Continuous Learning

Our graduates will have enquiring minds and a literate curiosity which will lead them to pursue knowledge for its own sake. They will continue to pursue learning in their careers and as they participate in the world. They will be capable of reflecting on their experiences and relationships with others and the environment, learning from them, and growing - personally, professionally and socially.

This graduate capability is supported by:

Learning outcomes

- Use a variety of resources to find up to date information in human genetics
- Read and demonstrate understanding of the primary scientific literature
- Learn basic bioinformatic skills, including handling of genetic sequence data

Assessment tasks

- Database and literature search
- Participation in Practicals

Discipline Specific Knowledge and Skills

Our graduates will take with them the intellectual development, depth and breadth of knowledge, scholarly understanding, and specific subject content in their chosen fields to make them competent and confident in their subject or profession. They will be able to demonstrate, where relevant, professional technical competence and meet professional standards. They will be able to articulate the structure of knowledge of their discipline, be able to adapt discipline-specific knowledge to novel situations, and be able to contribute from their discipline to inter-disciplinary solutions to problems.

This graduate capability is supported by:

Learning outcomes

- Use a variety of resources to find up to date information in human genetics
- Read and demonstrate understanding of the primary scientific literature
- Learn basic bioinformatic skills, including handling of genetic sequence data

Assessment tasks

- Database and literature search
- Human disease report
- Examination
- Participation in Practicals

Critical, Analytical and Integrative Thinking

We want our graduates to be capable of reasoning, questioning and analysing, and to integrate

and synthesise learning and knowledge from a range of sources and environments; to be able to critique constraints, assumptions and limitations; to be able to think independently and systemically in relation to scholarly activity, in the workplace, and in the world. We want them to have a level of scientific and information technology literacy.

This graduate capability is supported by:

Learning outcomes

- Use a variety of resources to find up to date information in human genetics
- Solve problems in human genetics using appropriate analytical methods
- Read and demonstrate understanding of the primary scientific literature

Assessment tasks

- Fortnightly online quiz
- Human disease report
- Examination

Problem Solving and Research Capability

Our graduates should be capable of researching; of analysing, and interpreting and assessing data and information in various forms; of drawing connections across fields of knowledge; and they should be able to relate their knowledge to complex situations at work or in the world, in order to diagnose and solve problems. We want them to have the confidence to take the initiative in doing so, within an awareness of their own limitations.

This graduate capability is supported by:

Learning outcomes

- Solve problems in human genetics using appropriate analytical methods
- Read and demonstrate understanding of the primary scientific literature
- Learn basic bioinformatic skills, including handling of genetic sequence data

Assessment tasks

- Fortnightly online quiz
- Database and literature search
- Human disease report
- Examination

Effective Communication

We want to develop in our students the ability to communicate and convey their views in forms effective with different audiences. We want our graduates to take with them the capability to read, listen, question, gather and evaluate information resources in a variety of formats, assess, write clearly, speak effectively, and to use visual communication and communication

technologies as appropriate.

This graduate capability is supported by:

Assessment tasks

- Human disease report
- Examination