



BIOL345

Human Genetics Theory

S1 Day 2015

Dept of Biological Sciences

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General Information

Unit convenor and teaching staff

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Credit points

3

Prerequisites

39cp 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
- Explain the complexities of the relationship between genotype and phenotype
- Solve problems in human genetics using appropriate analytical methods
- Read and demonstrate understanding of the primary scientific literature
- Explain the process of scientific discovery in human genetics and the effects of recent advances.

General Assessment Information

Late assignments will be penalized at the rate of 5% of the value of the assignment per 24 hours.

Assessment Tasks

Name	Weighting	Hurdle	Due
Internet exercise	5%	No	16 March
Linkage assignment	10%	No	2 April
Essay	30%	No	15 May
Examination	55%	No	Exam Period

Internet exercise

Due: **16 March**

Weighting: **5%**

The aim of this exercise is to encourage you to explore the resources in human genetics available on the Web, and to extract information from them. Further instructions can be found on a page linked to the BIOL345 online unit. This exercise is to be submitted via the iLearn unit.

On successful completion you will be able to:

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

Linkage assignment

Due: **2 April**

Weighting: **10%**

This is an exercise in human gene mapping. It will require you to perform an analysis on an individual dataset and interpret your results. Information on marking standards can be found in

the iLearn unit.

On successful completion you will be able to:

- Solve problems in human genetics using appropriate analytical methods

Essay

Due: **15 May**

Weighting: **30%**

Your task is to analyse the science that led to the cloning of a gene for a human disease in the decade from 1990 to 2000, and compare it to modern approaches. The choice of disease gene is up to you. You will need to read the primary scientific papers describing the steps that led to the identification of this gene. Further information on the assessment and marking standards can be found in the iLearn unit. The essay is to be submitted to the Science Student Centre after online submission to Turnitin.

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
- Explain the process of scientific discovery in human genetics and the effects of recent advances.

Examination

Due: **Exam Period**

Weighting: **55%**

The final examination will require solving of analytical genetics problems [50% of the exam], defining genetic terms and discussion questions. Students need to gain at least 45% in the final examination to pass the unit.

On successful completion you will be able to:

- Explain the complexities of the relationship between genotype and phenotype
- Solve problems in human genetics using appropriate analytical methods
- Explain the process of scientific discovery in human genetics and the effects of recent advances.

Delivery and Resources

CLASSES

LECTURES

There are three 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.

TUTORIALS

There are weekly two-hour tutorials in the unit, starting from week 1. Internal students will be timetabled to a session. Please do not change sessions without consulting me. We will use the tutorials to work through the problem sets. You are expected to have attempted them before the tutorial. We will also discuss the assignments. **You will find the unit very difficult if you do not attend all the tutorials.**

REQUIRED AND RECOMMENDED TEXTS AND/OR MATERIALS

TEXT BOOK

All students will need access to the internet and a scientific hand calculator. It is recommended that you purchase the textbook.

Textbook: *Human Molecular Genetics*. (4th Ed) Tom Strachan & Andrew P. Read, *Garland Science*, 2011. ISBN: 978-0-815-34149-9 Distributed in Australia by McMillan

Suggestions for optional additional reading are available on the iLearn unit.

UNIT WEBPAGE AND TECHNOLOGY USED AND REQUIRED

BIOL345 iLearn unit

The BIOL345 online iLearn unit is the core resource in this unit. Updated unit information, links to lecture recordings, lecture notes, assignment details and discussion forums can be found there. All students must **make sure they check it regularly.**

Unit Schedule

Policies and Procedures

Macquarie University policies and procedures are accessible from [Policy Central](#). Students should be aware of the following policies in particular with regard to Learning and Teaching:

Academic Honesty Policy http://mq.edu.au/policy/docs/academic_honesty/policy.html

Assessment Policy <http://mq.edu.au/policy/docs/assessment/policy.html>

Grading Policy <http://mq.edu.au/policy/docs/grading/policy.html>

Grade Appeal Policy <http://mq.edu.au/policy/docs/gradeappeal/policy.html>

Grievance Management Policy http://mq.edu.au/policy/docs/grievance_management/policy.html

Disruption to Studies Policy http://www.mq.edu.au/policy/docs/disruption_studies/policy.html *The*

Disruption to Studies Policy is effective from March 3 2014 and replaces the Special Consideration Policy.

In addition, a number of other policies can be found in the [Learning and Teaching Category](#) of 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/support/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.

Late assignments

Late submissions for the Internet exercise will not be accepted. For the remaining assignments, late assignments will be penalized at the rate of 5% of the value of the assignment per 24 hours, except for cases in which an application for special consideration has been made via [Ask.mq.edu.au](http://ask.mq.edu.au) and approved.

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://informatics.mq.edu.au/hel>

p/.

When using the University's IT, you must adhere to the [Acceptable Use 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:

Learning outcomes

- Solve problems in human genetics using appropriate analytical methods
- Explain the process of scientific discovery in human genetics and the effects of recent advances.

Assessment task

- Essay

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:

Assessment task

- Essay

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
- Explain the complexities of the relationship between genotype and phenotype
- Read and demonstrate understanding of the primary scientific literature
- Explain the process of scientific discovery in human genetics and the effects of recent advances.

Assessment tasks

- Internet exercise
- Linkage assignment
- Essay
- Examination

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

- Explain the complexities of the relationship between genotype and phenotype
- Solve problems in human genetics using appropriate analytical methods
- Read and demonstrate understanding of the primary scientific literature
- Explain the process of scientific discovery in human genetics and the effects of recent advances.

Assessment tasks

- Linkage assignment
- Essay
- 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

- 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
- Explain the process of scientific discovery in human genetics and the effects of recent advances.

Assessment tasks

- Internet exercise
- Linkage assignment
- Essay
- 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:

Learning outcomes

- Explain the complexities of the relationship between genotype and phenotype
- Read and demonstrate understanding of the primary scientific literature

Assessment tasks

- Linkage assignment
- Essay

Unit-level Standards

The following standards will be used to determine your grade in BIOL345

Pass

- Demonstrates the ability to think analytically, and attempts to apply appropriate mathematical techniques to solve genetics problems.
- Demonstrates the ability to find and evaluate evidence from the primary scientific literature and to extract key messages from these sources.
- Demonstrates a basic understanding of the complexities of the relationship between

genotype and phenotype

- Demonstrates a basic understanding of the process of scientific discovery in human genetics and the effects of recent technological advances in the field.
- Demonstrates adequate communication skills, with the production of a piece of writing that addresses the task to a satisfactory level.

Credit

- Demonstrates the ability to think analytically and critically, and generally applies the appropriate mathematical techniques to solve genetics problems.
- Demonstrates the ability to critically evaluate evidence from the primary scientific literature and to extract key messages from these sources.
- Demonstrates a sound grasp of the complexities of the relationship between genotype and phenotype
- Demonstrates a sound understanding of the process of scientific discovery in human genetics and the effects of recent technological advances in the field.
- Demonstrates good communication skills, with the production of an effective piece of writing that fully addresses the task.

Distinction

- Demonstrates a superior ability to think analytically and critically, and consistently applies the appropriate mathematical techniques to solve genetics problems
- Demonstrates a sustained ability to critically evaluate evidence from the primary scientific literature and to extract key messages from these sources.
- Demonstrates a good grasp of the complexities of the relationship between genotype and phenotype.
- Demonstrates an excellent understanding of the process of scientific discovery in human genetics and the effects of recent technological advances in the field.
- Demonstrates excellent communication skills, with the production of an effective piece of writing that displays noticeable evidence of innovative thinking and originality.

High Distinction

- Demonstrates an outstanding ability to think analytically and critically, and consistently applies the appropriate mathematical techniques to solve genetics problems
- Demonstrates a sustained ability to critically evaluate evidence from the primary scientific literature and to extract key messages from these sources.
- Demonstrates a thorough grasp of the complexities of the relationship between genotype and phenotype.

- Demonstrates substantial depth and breadth of knowledge of the process of scientific discovery in human genetics and the effects of recent technological advances in the field.
- Demonstrates excellent communication skills, with the production of a sophisticated piece of writing that displays substantial innovative thinking and originality.