



# BIOL345

## Human Genetics Theory

S1 External 2014

*Dept of Biological Sciences*

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## General Information

### Unit convenor and teaching staff

Unit Convenor

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### Tutor

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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.

## Assessment Tasks

Name	Weighting	Due
<a href="#">Internet exercise</a>	5%	24 March
<a href="#">Linkage assignment</a>	10%	11 April
<a href="#">Essay</a>	30%	23 May
<a href="#">Examination</a>	55%	Exam Period

### Internet exercise

Due: **24 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: **11 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.

External students will be given an extension for this assignment, as we will be discussing relevant problems in the on-campus session on 13 April. The revised date will be communicated

via the iLearn unit.

On successful completion you will be able to:

- Solve problems in human genetics using appropriate analytical methods

## Essay

Due: **23 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. 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 cloning 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, and external students will listen to recorded lectures via the online unit. All lectures

will be recorded live and will be available via the iLearn unit through Echo360.

## TUTORIALS

There are weekly two-hour tutorials for internal students 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 or on-campus sessions.**

## ON-CAMPUS-SESSIONS FOR EXTERNAL STUDENTS

These will deal with the problem sheets, the essay and revision. There are two of these, both held on Sundays. There is no formal requirement to attend these sessions (they are voluntary as some students are interstate or overseas) but most students will derive a lot of benefit if they do so, and I **expect all local external students to attend**. Students will be expected to have attempted the relevant problems before the on-campus session and to be prepared to discuss their solutions. Each session will last the whole day. The dates are as follows:

1. Sunday 13 April end of week 6 problem sets 1- 5
2. Sunday 8 June end of week 12 problem sets 6 – 11

## ONLINE TUTORIALS FOR EXTERNAL STUDENTS

There will be some online tutorials in the evening for external students, using the online resource Scribblar. All arrangements, including timing and frequency, will be made with external students via the Discussion forum in the Online unit.

## 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](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](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](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/](https://students.mq.edu.au/support/student_conduct/)

## 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 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](http://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](http://ask.mq.edu.au)

## IT Help

For help with University computer systems and technology, visit <http://informatics.mq.edu.au/help/>.

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

### 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

## **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

## **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.