UNSW Course Outline

BABS3151 Human Molecular Genetics and Disease - 2024

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General Course Information

Course Code : BABS3151
Year : 2024
Term : Term 2
Teaching Period : T2
Is a multi-term course? : No
Faculty : Faculty of Science
Academic Unit : School of Biotechnology and Biomolecular Sciences
Delivery Mode : Multimodal
Delivery Format : Standard
Delivery Location : Kensington
Campus : Sydney
Study Level : Postgraduate, Undergraduate
Units of Credit : 6

Useful Links

Handbook Class Timetable

Course Details & Outcomes

Course Description

Personal genomics is revolutionising medicine. Soon, everyone in Australia is likely to have their genome sequenced at birth. What does this mean for how we see ourselves and treat disease? Is our future written in our genes, or can we influence and control our genetic destiny? This

course explores the central principles and applications of understanding our genetic makeup and how this is transforming biomedical science. Core concepts and methods in molecular genetics will be introduced and applied to a variety of topics on human evolution, development and disease. Students will examine the roles of “nature” versus “nurture”, including epigenetic regulation, and learn how developments in genome technology are opening new avenues for personalised medicine. Lectures and tutorials will be supported by laboratory and bioinformatics practicals using modern research methods and data. Skills developed during these practicals will provide excellent training for any students considering an Honours project in molecular bioscience.

Course Aims

This course will enable students to interpret human genomic data in relation to human evolution, development, and disease. It will explore core concepts and methods in molecular genetics and genomics, and how these relate to key societal issues such as genetic determination and personalized medicine. Emphasis will be placed on modern research approaches that use massively parallel sequencing technologies and genomics.

Course Learning Outcomes

Course Learning Outcomes
CLO1 : Discuss key concepts around current understanding of human genome structure, function and variation.
CLO2 : Discuss recent advances in genetic sequencing technologies and their impact on our understanding of human disease and human evolution.
CLO3 : Select and apply appropriate bioinformatics-based approaches to extract information on DNA sequence variation, gene expression and epigenetic regulation.
CLO4 : Explain the principles of fundamental methods in human genetic testing and gene discovery-based research and how these are used to further our understanding of the genetic basis of human diseases.
CLO5 : Critically interpret and evaluate experimental findings and communicate them in written and verbal formats.

Course Learning Outcomes	Assessment Item
CLO1 : Discuss key concepts around current understanding of human genome structure, function and variation.	• Presentation • Mid-term test • Final exam
CLO2 : Discuss recent advances in genetic sequencing technologies and their impact on our understanding of human disease and human evolution.	• Mid-term test • Final exam
CLO3 : Select and apply appropriate bioinformatics-based approaches to extract information on DNA sequence variation, gene expression and epigenetic regulation.	• Research Report • Mid-term test • Final exam
CLO4 : Explain the principles of fundamental methods in human genetic testing and gene discovery-based research and how these are used to further our understanding of the genetic basis of human diseases.	• Presentation • Mid-term test • Final exam
CLO5 : Critically interpret and evaluate experimental findings and communicate them in written and verbal formats.	• Research Report • Presentation

Learning and Teaching Technologies

Moodle - Learning Management System | Blackboard Collaborate

Assessments

Assessment Structure

Assessment Item	Weight	Relevant Dates
Presentation Assessment Format: Individual	20%	
Research Report Assessment Format: Individual Short Extension: Yes (2 days)	30%	
Mid-term test Assessment Format: Individual	25%	
Final exam Assessment Format: Individual	25%	

Assessment Details

Presentation

Assessment Overview

This task assesses your ability to critically evaluate published experimental findings and present them to a scientific audience.

You will need to deliver a 5 min. oral presentation on one of the research articles that discusses functional analysis of a gene of interest.

The presentations will take place during the practical sessions in Week 8 and 9.
A marking rubric will be used to mark the assessment and provide feedback.

Course Learning Outcomes

- CLO1 : Discuss key concepts around current understanding of human genome structure, function and variation.
- CLO4 : Explain the principles of fundamental methods in human genetic testing and gene discovery-based research and how these are used to further our understanding of the genetic basis of human diseases.
- CLO5 : Critically interpret and evaluate experimental findings and communicate them in written and verbal formats.

Detailed Assessment Description

Correction: The presentations will be 5 min in length followed by 5 minutes of Q&A, and will take place during Weeks 8 and 9.

Assignment submission Turnitin type

This is not a Turnitin assignment

Research Report

Assessment Overview

This task assesses your ability to select and use appropriate bioinformatic tools and databases to curate information about a gene of interest. You will be introduced to these resources during the practical sessions where you will also have the opportunity to receive feedback from the demonstrators.

You will need to submit a written scientific report summarising all your experimental findings collected throughout the term. The report should be no longer than 4 pages in length. Additional instructions will be provided during practical classes.

The assessment is due in Week 7 and should be submitted online. A marking rubric will be used to mark this assessment. Written feedback will also be provided where necessary.

Course Learning Outcomes

- CLO3 : Select and apply appropriate bioinformatics-based approaches to extract information on DNA sequence variation, gene expression and epigenetic regulation.
- CLO5 : Critically interpret and evaluate experimental findings and communicate them in written and verbal formats.

Detailed Assessment Description

Correction: This assessment is due at the end of Week 7. Additional information will be provided on the course Moodle site.

Assignment submission Turnitin type

This assignment is submitted through Turnitin and students do not see Turnitin similarity reports.

Mid-term test

Assessment Overview

This is a written test covering both lecture and practical material delivered in Weeks 1-4.

The Test is 2 hours in duration and is scheduled in Week 5. The test typically consists of essay type questions.

Mark/grade, along with general feedback will be provided 10 working days after the test.

Course Learning Outcomes

- CLO1 : Discuss key concepts around current understanding of human genome structure, function and variation.
- CLO2 : Discuss recent advances in genetic sequencing technologies and their impact on our understanding of human disease and human evolution.
- CLO3 : Select and apply appropriate bioinformatics-based approaches to extract information on DNA sequence variation, gene expression and epigenetic regulation.
- CLO4 : Explain the principles of fundamental methods in human genetic testing and gene discovery-based research and how these are used to further our understanding of the genetic basis of human diseases.

Detailed Assessment Description

Correction: The mid-term exam will be held during Week 5. Additional information will be provided on the course Moodle site.

Assignment submission Turnitin type

This is not a Turnitin assignment

Final exam

Assessment Overview

This is a written examination covering both lecture and practical material delivered during Weeks 7 - 10.

The exam is 2 hours in duration and is scheduled during the formal examination period. The exam typically consists of essay type questions.

Mark/grade will be released on official assessment results release date. Individual feedback will be provided upon request.

Course Learning Outcomes

- CLO1 : Discuss key concepts around current understanding of human genome structure, function and variation.
- CLO2 : Discuss recent advances in genetic sequencing technologies and their impact on our understanding of human disease and human evolution.
- CLO3 : Select and apply appropriate bioinformatics-based approaches to extract information on DNA sequence variation, gene expression and epigenetic regulation.
- CLO4 : Explain the principles of fundamental methods in human genetic testing and gene discovery-based research and how these are used to further our understanding of the genetic basis of human diseases.

Assessment Length

Please see the course Moodle site for additional information.

Assignment submission Turnitin type

This is not a Turnitin assignment

General Assessment Information

Grading Basis

Standard

Course Schedule

Attendance Requirements

Students are strongly encouraged to attend all lectures and review lecture recordings.

Students should maintain >80% attendance in practical classes to avoid potentially receiving an Unsatisfactory Fail grade.

Course Resources

Recommended Resources

Please the course Moodle site for detailed information.

Staff Details

Position	Name	Email	Location	Phone	Availability	Equitable Learning Services Contact	Primary Contact
Convenor	Joel Brame				By appointment	Yes	Yes

Other Useful Information

Academic Information

Upon your enrolment at UNSW, you share responsibility with us for maintaining a safe, harmonious and tolerant University environment.

You are required to:

- Comply with the University's conditions of enrolment.
- Act responsibly, ethically, safely and with integrity.
- Observe standards of equity and respect in dealing with every member of the UNSW community.
- Engage in lawful behaviour.
- Use and care for University resources in a responsible and appropriate manner.
- Maintain the University's reputation and good standing.

For more information, visit the UNSW Student Code of Conduct Website.

Academic Honesty and Plagarism

Referencing is a way of acknowledging the sources of information that you use to research your assignments. You need to provide a reference whenever you draw on someone else's words, ideas or research. Not referencing other people's work can constitute plagiarism.
Further information about referencing styles can be located at https://student.unsw.edu.au/referencing

Academic integrity is fundamental to success at university. Academic integrity can be defined as a commitment to six fundamental values in academic pursuits: honesty, trust, fairness, respect, responsibility and courage. At UNSW, this means that your work must be your own, and others' ideas should be appropriately acknowledged. If you don't follow these rules, plagiarism may be detected in your work.
Further information about academic integrity, plagiarism and the use of AI in assessments can be located at:

- The Current Students site,
- The ELISE training site, and
- The Use of AI for assessments site.

The Student Conduct and Integrity Unit provides further resources to assist you to understand your conduct obligations as a student: https://student.unsw.edu.au/conduct

Submission of Assessment Tasks

Penalty for Late Submissions

UNSW has a standard late submission penalty of:

- 5% per day,
- for all assessments where a penalty applies,
- capped at five days (120 hours) from the assessment deadline, after which a student cannot

submit an assessment, and
- no permitted variation.

Any variations to the above will be explicitly stated in the Course Outline for a given course or assessment task.

Students are expected to manage their time to meet deadlines and to request extensions as early as possible before the deadline.

Special Consideration

If circumstances prevent you from attending/completing an assessment task, you must officially apply for special consideration, usually within 3 days of the sitting date/due date. You can apply by logging onto myUNSW and following the link in the My Student Profile Tab. Medical documentation or other documentation explaining your absence must be submitted with your application. Once your application has been assessed, you will be contacted via your student email address to be advised of the official outcome and any actions that need to be taken from there. For more information about special consideration, please visit: https://student.unsw.edu.au/special-consideration

Important note: UNSW has a "fit to sit/submit" rule, which means that if you sit an exam or submit a piece of assessment, you are declaring yourself fit to do so and cannot later apply for Special Consideration. This is to ensure that if you feel unwell or are faced with significant circumstances beyond your control that affect your ability to study, you do not sit an examination or submit an assessment that does not reflect your best performance. Instead, you should apply for Special Consideration as soon as you realise you are not well enough or are otherwise unable to sit or submit an assessment.

Faculty-specific Information

Additional support for students

- The Current Students Gateway
- Student Support
- Academic Skills and Support
- Student Wellbeing, Health and Safety
- Equitable Learning Services
- UNSW IT Service Centre
- Science EDI Student Initiatives, Offerings and Guidelines