FACULTY OF SCIENCE
School of Biotechnology and Biomolecular Sciences

BABS3151

Human Genetics

Term 1, 2021

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Faculty of Science - Course Outline

1. Information about the Course

NB: Some of this information is available on the UNSW Handbook[1]

Year of Delivery	2021
Course Code	BABS3151
Course Name	Human Genetics
Academic Unit	School of Biotechnology and Biomolecular Sciences
Level of Course	3rd Year
Units of Credit	6UOC
Term(s) Offered	T1
Assumed Knowledge, Prerequisites or Co-requisites	BABS2204 or BABS2264
Contact Hours per Week	7
Number of Weeks	11 weeks + final exam period (starts Monday Feb 15th: final exam period 30th April-13th May)
Commencement Date	**Week 1**

Summary of Course Structure (for details see 'Course Schedule')

Component	HPW	Time	Day	Location
Lecture 1	1	14:00-15:00	Mon	Online via Moodle
Lecture 2	1	17:00-18:00	Tue	Online via Moodle
Lecture 3	1	13:00-14:00	Wed	Online via Moodle
Practical sessions	3-4	09:00-13:00	Wed (Weeks 1-5, 7-10)	BiolSci Teaching lab 10, Lvl1 E26 or online via Moodle
TOTAL	6-7			
Special Details	Students with clashes should consult with Dr. Emily Oates (e.oates@unsw.edu.au)			

[1]UNSW Online Handbook: http://www.handbook.unsw.edu.au

2. Staff Involved in the Course

Staff	Role	Name	Contact Details	Consultation Times
Course Convenor		Emily Oates (EO)	e.oates@unsw.edu.au	Email for appoint-ment
Course co-convenor		Dhanushi Abeygunawardena	dhanushi@unsw.edu.au	Email for appoint-ment
Additional Teaching Staff	**Lecturers & facilitators**	**Lecturers (BABS):** Richard Edwards (RE) Irina Voineagu (IV) Paul Waters (PW) Fatemeh Vafaee (FV) Frances Byrne (FB) Natalia Castaño Rodríguez (NCR) Nicole Green (NG) Marcel Dinger (MD) Kate Quinlan (KQ) :	**Email addresses:** richard.edwards@unsw.edu.au i.voineagu@unsw.edu.au p.waters@unsw.edu.au f.vafaee@unsw.edu.au frances.byrne@unsw.edu.au n.castanorodriguez@unsw.edu.au n.green@unsw.edu.au m.dinger@unsw.edu.au kate.quinlan@unsw.edu.au	Email for appoint-ment
	Guest Lecturers	Ozren Bogdanovic (OB), Garvan Robert Weatheritt (RW), Garvan Milita Zaheed (MZ), POWH Marc Cowley (MC), CCI Michelle Farrer (MF), SCH Daniel MacArthur (DM), Garvan Kristi Jones (KJ), CHW	Contact via Dr Oates	n/a
	Demon-strators:	Gavin Sutton (GS) Patricia Sullivan (PS) Andrei Smolnikov	gavin.sutton@unsw.edu.au PSullivan@ccia.org.au andrei.smolnikov@unsw.edu.au	n/a

3. Course schedule

	Lecture 1: Monday 1400-1500 Online	Lecture 2: Tue 1700-1800 Online	Lecture 3: Wed 1300-1400 Online	Tutorial/LAB Wed 0900-1300 BiolSci Teaching Lab 10, Lvl1 Rowntree Room wks 9 & 10
Week	Topic/Lecturer	Topic/Lecturer	Topic/Lecturer	Prac/Lab
1 Starts Mon 15/02	Introduction to human genetics/**EO** Date: 15/02	Human genome structure/**RE** 16/02	Human genome variation/**RE** 17/02	Getting to know your gene **EO, GS, PS, AS** 17/02
2 Starts Mon 22/02	Transcriptomics/**NG** 22/02	Alternative splicing & post-transcriptional regulation Garvan guest lecturer/**RW** 23/02	Sex determination/**PW** 24/02	Non-disease-causing variation within your gene **EO, GS, PS, AS** 24/02
3 Starts Mon 01/03	Massively parallel genomic sequencing technologies including WES & WGS/**AS** 01/03	Proteomics and its application to human genetics/**MW** 02/03	Epigenomics Garvan guest lecturer/**OB** 03/03	Disease-causing variation within your gene **EO, GS, PS, AS** 03/03
4 Starts Mon 08/03	Monogenic (Mendelian) disorders/**EO** 08/03	Mitochondrial, chromosomal & oligogenic disorders/**EO** 09/03	Multifactorial (complex) traits & disorders/**IV** 10/03	Interrogating RNA transcript data relevant to your gene **EO, GS, PS, AS** 10/03
5 Starts Mon 15/03	Different clinical genetic testing scenarios/**EO** 15/03	Pre-implantation genetic diagnosis Clinical (CHW) guest lecturer/**KJ** 16/03	Systems biology and its application to human genetics/**FV** 17/03	Midterm Exam (30%) Followed by "Tips for your research report" **EO, GS, PS, AS** 17/03
6 Starts Mon 22/03	Week 6 rest week			
7 Starts Mon 29/03	Personalized medicine & ethical considerations in human genetics/**MD** 29/03	Applications of human genome sequencing CCI guest lecturer/**MC** 30/03	Frontiers in human genetic research Garvan guest lecturer/**DM** 31/03	Experimental practical: Strategies to detect CYP2D6 & other genetic variants I 31/03 **EO, GS, PS, AS**
8 Starts Mon 05/04	EASTER MONDAY	Introduction to gene discovery and genetic filtering/**EO** 06/04	Behavioral genetics/**IV** 07/04	Experimental practical: Strategies to detect CYP2D6 & other genetic variants II 07/04 **EO, GS, PS, AS**
9 Starts Mon 12/04	Introduction to genetic treatment technologies/**EO** 12/04	Cancer genetics/**FB** 13/04	Familial cancer Clinical (POW) guest lecturer/**MZ** 14/04	Oral Presentations I **EO, GS, PS, AS** 14/04
10 Starts Mon 19/04	Immunogenetics/**NCR** 19/04	Development and implementation of SMA genetic treatment technologies Clinical (SCH) guest lecturer/**MF** 20/04	CRISPR and its role in human disease-related research and treatment /**KQ** 21/04	Oral Presentations II **EO, GS, PS, AS** 21/04
11 Starts 26/04	OFFICIAL STUDY PERIOD BEGINS SAT 24/04 FINAL EXAM (30%) DURING 30/04-13/05 EXAM PERIOD: Will be based on all lecture content delivered on/from 15/03 (Weeks 5-11)			

4. Course Details

Course Description[2] (Handbook Entry)	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? BABS3151 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 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 student considering an honours project in molecular bioscience or in genetic counselling.
Course Aims[3]	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 a genetic determination and personalised medicine. Emphasis will be placed on modern research approaches that use massively parallel sequencing and genomics.
Student Learning Outcomes[4]	Upon successful completion of this course, students should be able to: 1. Discuss recent advances in genomic technologies and how they have shaped our understanding of the human genome 2. Explain the core principles of molecular evolutionary theory and apply them to homology searching and genome annotation 3. Discuss the impact of population structure and genetic variation on human medicine 4. Assess claims made in the media about genetic causes or treatments for human disease 5. Compare and contrast the roles of "nature" and "nurture" in human development and disease 6. Critically evaluate the application of genomic technologies to "personalised medicine" 7. Apply bioinformatics sequence analysis tools to explore the function and evolutionaryhistory of DNA and protein sequences

Graduate Attributes Developed in this Course[5]

Science Graduate Attributes[5]	**FOCUS** *0 = NO FOCUS* *1 = MINIMAL* *2 = MINOR* *3 = MAJOR*	**Activities / Assessment**
Research, inquiry and analytical thinking abilities	3	Computer exercises & written reports
Capability and motivation for intellectual development	3	Lectures and computer practicals, group discussions & written reports
Ethical, social and professional understanding	2	Article discussions
Communication	3	Article discussions, written reports,
Teamwork, collaborative and management skills	2	Article discussions, computer practicals
Information literacy	3	Computer practicals, article discussions and written reports

[2] UNSW Handbook: http://www.handbook.unsw.edu.au
[3] Learning and Teaching Unit: Course Outlines
[4] Learning and Teaching Unit: Learning Outcomes
[5] Contextualised Science Graduate Attributes: http://www.science.unsw.edu.au/our-faculty/science-graduate-attributes

Major Topics (Syllabus Outline)	• Human Genome Structure, Variation and Function • Transcriptomics, Proteomics, Epigenomics • Alternative Splicing & post-transcriptional regulation • Sex determination. • Human Evolutionary Genetics • Genetic mechanisms that underlie mongenic, oligenic and multifactorial traits and diseases • Massively parallel sequencing technologies • CRISPR and its role in human disease-related research • Personalized medicine • Emerging genomic treatment technologies • Pre-implantation genetic diagnosis • Human genetics focused systems biology. • Genetic basis of specific traits/disorders (genetic muscle disorders, immunogenetics, cancer, behavioral genetics, Alzheimer's disease)
Relationship to Other Courses within the Program	• Builds on the second year course Genetics (BABS2204 or BABS2264) • Establishes foundation for Term 2 course Genes, Genomes & Evolution (BABS3291) Other courses related to BABS3151 Human Genetics: • Molecular Biology of Nucleic Acids (BABS3121 • Molecular Frontiers (BABS3281) • Microbial Genetics (BABS3021/MICR3621) • Bioinformatics Methods and Applications (BINF3010)

5. Rationale and Strategies Underpinning the Course

Teaching Strategies	The lectures, given by experts in the field, will introduce students to essential concepts and principles, as well as recent developments in the human genetics field. The practicals will further-explore many elements of the material introduced in lectures Additional relevant topics with be introduced during practicals to further expand each student's awareness of human genetic principals and the "real world" application of these principals. Computer-based exercises will provide hands-on exposure to methods, tools and concepts used in genetics. The oral presentation will further-develop each student's ability to critically evaluate and effectively communicate research results. Effective scientific communication is also developed and evaluated through written research reports.
Rationale for learning and teaching in this course[6]	This is a third year course that builds on ideas taught in second year genetics (BABS2204 or BABS2264). In developing these ideas, we aim to contextualise the material via the presentation of field-relevant examples. Emphasis is placed on critical thinking, analytical skills, information literacy and communication because these are qualities that will aid learning in the long term. The objectives and activities of this course are designed to develop UNSW and Science Faculty graduate attributes.

[6] Reflecting on your teachin6.

6. Lecture content

FUNDAMENTAL HUMAN GENETICS	• Human Genome Structure • Human Genome Variation • Proteomics • Transcriptomics • Sex determination • Human Evolutionary Genetics • Human genetics-related systems biology
HUMAN GENETIC VARIATION & DISEASE	• Human genetic mechanisms of disease • Oligogenic Traits • Multifactorial traits & heritability estimates • Whole exome and whole genome sequencing • Genome-wide association studies (GWAS) • CRISPR and it's role in human disease-related research • Personalized medicine and ethical considerations in human genetics
GENETIC BASIS OF SPECIFIC DISORDERS	• Gene discovery in muscle disorders • Immunogenetics • Cancer genetics • Behavioural genetics • Genetics of Alzheimer's Disease
SPECIALST TALKS BY INVITED SPEAKERS	• Alternative Splicing & post-transcriptional regulation • Epigenomics • Applications of Genome Sequencing • Frontiers in human genetic research • Emerging genomic treatment technologies • Familial cancer • Pre-implantation genetic diagnosis

7. Assessment Tasks and Feedback

Task	Knowledge & abilities assessed	Assessment Criteria	% of total mark	Date of		Feedback		
				Release	Sub-mission	WHO	WHEN	HOW
Pre-census formative assessment	Online multiple choice-based text based on lecture content during first two weeks of course	Formative test: result does NOT count towards final mark	N/A	Online from 0900 Tue 9th March	Must be completed online by 1700 Wed 10th March (prior to census date Sun 14th March 11.59pm)	E. Oates	Week 4	Students will receive a mark at end of test.
Mid-semester exam (Week 1-4 lecture content excluding Lecture 1)	Short answer questions based on pre-mid-semester exam lecture and practical content. (Essential disciplinary knowledge in genetics at 3rd year level)	Depth of understanding of examined material Clarity of explanation	30%	Online via Moodle during Week 5 practical session	At end of first hour of practical	E. Oates	Week 7	Via Moodle
Assignment 1: Report on the data analyses carried out during Week 2-4 practical sessions.	Ability to utilise bioinformatics tools to extract information about normal human/animal gene function, sequence variations (including likely-to-be-pathogenic mutations) and transcription. Ability to download gene, RNA and protein sequences. Ability to critically interpret extracted data.	Accurate completion of task Clear description of results Presentation of writing, including formatting, spelling/grammar references, structure. Detailed information will be provided on Moodle	20%	Week 2	Research report due by 1700 Thursday 6th April (Week 8)	E. Oates	Week 10	Via Moodle

Assignment 2: Presentation and critical assessment of a primary research article	Effective communication, critical thinking about research topics in genetics and genomics.	Appropriateness of evidence. Effectiveness of Arguments. Critical evaluation of evidence. Clarity of presentation style Clear understanding of statistical concepts. Detailed information will be provided on Moodle	20%	Week 7	Weeks 9,10	E. Oates	Week 10	Verbal feedback from academics and peers on day of presentation Results posted
Final Exam Week 5 - Week 11 content	Short answer questions based on post-mid-semester exam lecture and practical content. (Essential disciplinary knowledge in genetics at 3rd year level)	Depth of understanding of examined material. Clarity of explanation	30%	End of Term 1 exam period TBC	End of Term 1 exam period (2^{nd}-12^{th} May)	E. Oates	Check exam Timetable when available	Results posted

Approaches to assessment: http://teaching.unsw.edu.au/assessment

8. Additional Resources and Support

Text Books	There is no textbook set for this course because the topics covered are diverse and no single book covers all the material adequately. However, some useful texts are listed below, and lecturers will suggest additional reading material during the course.
Course Manual	**This document and the Moodle site for this course.**
Additional Readings	**Vogel and Motulsky's Human Genetics** **UNSW library e-Book:** https://link-springer-com.wwwproxy1.library.unsw.edu.au/book/10.1007%2F978-3-540-37654-5 **Oxford Desk Reference Clinical Genetics and Genomics 2nd edition** Helen V Firth and Jane A. Hurst **UNSW library e-Book:** http://oxfordmedicine.com.wwwproxy1.library.unsw.edu.au/view/10.1093/med/9780199557509.001.0001/med-9780199557509 **A Primer of Human Genetics** Greg Gibson **Textbook available from UNSW library** **No e-Book available in Australia** UNSW library will scan and make available several of the most important sections of this textbook for our BABS3151 students
Recommended Internet Sites	**Moodle site for this course:** http://moodle.telt.unsw.edu.au/course/view.php?id=3151 Library website and resources: http://info.library.unsw.edu.au/ NCBI: http://www.ncbi.nlm.nih.gov/ PubMed: http://www.ncbi.nlm.nih.gov/sites/entrez?db=PubMed
Societies	Human Genetics Society of Australasia https://www.hgsa.org.au/ Genetics Society of Australasia www.genetics.org.au/ Australasian Evolution Society ausevo.com/ Australian Genomics Health Alliance https://www.australiangenomics.org.au

9. Administration Matters

Expectations of Students	Attendance at all practicals is compulsory unless you have a valid and documented medical (with medical certificate) or other reason. Be punctual. Do not talk or eat during lectures. Mobile phones must be switched off during lectures and practicals. Attendance at practicals will be recorded.
Assignment Submissions	**Late submissions will incur a penalty of 10% of the total value per day.**

10. UNSW Academic Honesty and Plagiarism

What is Plagiarism?

Plagiarism is the presentation of the thoughts or work of another as one's own.
*Examples include:

- direct duplication of the thoughts or work of another, including by copying material, ideas or concepts from a book, article, report or other written document (whether published or unpublished), composition, artwork, design, drawing, circuitry, computer program or software, web site, Internet, other electronic resource, or another person's assignment without appropriate acknowledgement;
- paraphrasing another person's work with very minor changes keeping the meaning, form and/or progression of ideas of the original;
- piecing together sections of the work of others into a new whole;
- presenting an assessment item as independent work when it has been produced in whole or part in collusion with other people, for example, another student or a tutor; and
- claiming credit for a proportion a work contributed to a group assessment item that is greater than that actually contributed†.

For the purposes of this policy, submitting an assessment item that has already been submitted for academic credit elsewhere may be considered plagiarism.

Knowingly permitting your work to be copied by another student may also be considered to be plagiarism.

Note that an assessment item produced in oral, not written, form, or involving live presentation, may similarly contain plagiarised material.

The inclusion of the thoughts or work of another with attribution appropriate to the academic discipline does *not* amount to plagiarism.

The Learning Centre website is main repository for resources for staff and students on plagiarism and academic honesty. These resources can be located via:

https://student.unsw.edu.au/plagiarism

The Learning Centre also provides substantial educational written materials, workshops, and tutorials to aid students, for example, in:

- correct referencing practices;
- paraphrasing, summarising, essay writing, and time management;
- appropriate use of, and attribution for, a range of materials including text, images, formulae and concepts.

Individual assistance is available on request from The Learning Centre.

Students are also reminded that careful time management is an important part of study and one of the identified causes of plagiarism is poor time management. Students should allow sufficient time for research, drafting, and the proper referencing of sources in preparing all assessment items.

* Based on that proposed to the University of Newcastle by the St James Ethics Centre. Used with kind permission from the University of Newcastle
† Adapted with kind permission from the University of Melbourne

11. Special Consideration and Further Assessment

Students who believe that their performance, either during the session or in the end of session exams, may have been affected by illness or other circumstances may apply for "special consideration".

Applications can be made for compulsory class absences (laboratories), assessment tasks, mid-session examination and final examination.

*UNSW operates under a Fit to Sit/ Submit rule for all assessments. If a student wishes to submit an application for special consideration for an exam or assessment, the application must be submitted **prior to the** start of the exam or before an assessment is submitted. If a student sits the exam/ submits an assignment, they are declaring themselves well enough to do so.*

The Assessment Implementation Procedure can be found here:
https://www.gs.unsw.edu.au/policy/documents/assessmentimplementationprocedure.pdf

Lab/Tutorial Attendance

Lab/tutorials are **compulsory** and attendance will be marked. If you are absent from a lab class you must apply for special consideration and provide a medical certificate or other professional documentation that supports the reason for your absence.

Assignments

If you are unable to submit an assignment by the due submission day and time, you must apply for special consideration and provide a medical certificate or other professional documentation that supports the reason for your absence. Note that a **10% penalty per day** will ensue if you do not provide a satisfactory explanation.

In-session and Final Exams

If you are absent from an in-session or final exam/test, you must also apply for special consideration and provide a medical certificate or other professional documentation that supports the reason for your absence. Further assessment granted to students who miss an exam due to valid unavoidable circumstances will be conducted on the designated

Term 1 2021 supplementary exam period: 24 –28 May 2021.

Further assessment exams will be offered on this day ONLY and failure to sit for the appropriate exam on this day may result in an overall failure for the course. Further assessment will NOT be offered on any alternative dates.

NOTES

- *Applying for special consideration does NOT guarantee approval!!!* Students who do not provide valid reasons and/or valid supporting documentation will be rejected and appropriate penalties for late submission or absences will be applied.

- Students should also consult the information provided on “Special Consideration” via UNSW website for further information about general rules covering examinations, assessment, special consideration and other related matters. This is information is available on the web at: https://student.unsw.edu.au/special-consideration
- Please send enquires via unsw/webforms.