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Faculty of Science - Course Outline

1. Information about the Course
   NB: Some of this information is available on the UNSW Handbook¹

<table>
<thead>
<tr>
<th>Component</th>
<th>HPW</th>
<th>Time</th>
<th>Day</th>
<th>Location</th>
</tr>
</thead>
<tbody>
<tr>
<td>Lecture 1</td>
<td>1</td>
<td>13:00-14:00</td>
<td>Mon</td>
<td>Mathews Theatre D</td>
</tr>
<tr>
<td>Lecture 2</td>
<td>1</td>
<td>15:00-16:00</td>
<td>Thu</td>
<td>Mathews Theatre C</td>
</tr>
<tr>
<td>Tutorial</td>
<td>1</td>
<td>09:00-10:00</td>
<td>Fri</td>
<td>Bioscience (D26) G08</td>
</tr>
<tr>
<td>Computer Labs</td>
<td>3</td>
<td>1000-1300</td>
<td>Fri</td>
<td>Bioscience (D26) G08</td>
</tr>
<tr>
<td><strong>TOTAL</strong></td>
<td>6</td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

Special Details: Students with clashes should consult with Dr Irina Voineagu

2. Staff Involved in the Course

<table>
<thead>
<tr>
<th>Staff</th>
<th>Role</th>
<th>Name</th>
<th>Contact Details</th>
<th>Consultation Times</th>
</tr>
</thead>
<tbody>
<tr>
<td>Course Convenors</td>
<td></td>
<td>Irina Voineagu (IV)</td>
<td><a href="mailto:i.voineagu@unsw.edu.au">i.voineagu@unsw.edu.au</a></td>
<td>Email for appointment;</td>
</tr>
<tr>
<td></td>
<td></td>
<td>Richard Edwards (RJE)</td>
<td><a href="mailto:richard.edwards@unsw.edu.au">richard.edwards@unsw.edu.au</a></td>
<td></td>
</tr>
<tr>
<td>Additional Teaching Staff</td>
<td>Lecturers &amp; Facilitators</td>
<td>Bill Ballard (BB)</td>
<td><a href="mailto:b.ballard@unsw.edu.au">b.ballard@unsw.edu.au</a></td>
<td>Email for appointment;</td>
</tr>
<tr>
<td></td>
<td></td>
<td>Bill Sherwin (WS)</td>
<td><a href="mailto:w.sherwin@unsw.edu.au">w.sherwin@unsw.edu.au</a></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td>Paul Waters (PW)</td>
<td><a href="mailto:p.waters@unsw.edu.au">p.waters@unsw.edu.au</a></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td>Akira Gokoolparsadh</td>
<td><a href="mailto:a.gokoolparsadh@unsw.edu.au">a.gokoolparsadh@unsw.edu.au</a></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td>Brigid Betz Stablein</td>
<td><a href="mailto:b.betz-stablein@unsw.edu.au">b.betz-stablein@unsw.edu.au</a></td>
<td></td>
</tr>
<tr>
<td>Guest Lecturers</td>
<td></td>
<td>Dr Ozren Bogdanovic, Garvan (OB)</td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td>Dr Marc Cowley, Garvan (MC)</td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td></td>
<td>Contact via Dr Voineagu</td>
<td></td>
<td>n/a</td>
</tr>
</tbody>
</table>

¹ UNSW Online Handbook: http://www.handbook.unsw.edu.au
### 3. Course Schedule (Lectures, Practicals and Tutorials)

<table>
<thead>
<tr>
<th>Week</th>
<th>Date</th>
<th>Topic/ Instructor</th>
<th>Date</th>
<th>Topic/ Instructor</th>
<th>Date</th>
<th>Topic/ Instructor</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>27.02</td>
<td>Introduction to Human Genetics; modes of inheritance /IV</td>
<td>02.03</td>
<td>Human Genome Structure/RE</td>
<td>03.03</td>
<td>Introduction to R / BBS</td>
</tr>
<tr>
<td>2</td>
<td>06.03</td>
<td>Human Genome Variation/RE</td>
<td>09.03</td>
<td>Sex determination/PW</td>
<td>10.03</td>
<td>Statistics for Human Genetics/ BBS</td>
</tr>
<tr>
<td>3</td>
<td>13.03</td>
<td>Transcriptomics/IV</td>
<td>16.03</td>
<td>Proteomics/ RE</td>
<td>17.03</td>
<td>Writing and presentation workshop/AG</td>
</tr>
<tr>
<td>4</td>
<td>20.03</td>
<td>Epigenomics/OB</td>
<td>23.03</td>
<td>Linkage Analysis and Monogenic Traits/IV</td>
<td>24.03</td>
<td>Human Genome Variation and Databases/ IV, AG</td>
</tr>
<tr>
<td>5</td>
<td>27.03</td>
<td>Oligogenic Traits /IV</td>
<td>30.03</td>
<td>Multifactorial Inheritance /IV</td>
<td>31.03</td>
<td>Gene Expression Variation (GTEX)/IV, AG</td>
</tr>
<tr>
<td>6</td>
<td>03.04</td>
<td>Genome Wide Association Studies/IV</td>
<td>06.04</td>
<td>Lessons from Genome Wide Association Studies/IV</td>
<td>07.04</td>
<td>ENCODE, Epigenomics Roadmap/IV, AG</td>
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<tr>
<td>7</td>
<td>10.04</td>
<td>Heritability Estimates/ IV</td>
<td>13.04</td>
<td>Midsemester Exam</td>
<td>14.04</td>
<td></td>
</tr>
<tr>
<td>Break</td>
<td>17.04</td>
<td>20.04</td>
<td>21.04</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>8</td>
<td>24.04</td>
<td>Whole Exome and Whole Genome Sequencing/IV</td>
<td>27.04</td>
<td>Applications of Genome Sequencing/MC</td>
<td>28.04</td>
<td>Experimental practical: Detecting CYP2D6 polymorphisms I/IV, AG</td>
</tr>
<tr>
<td>9</td>
<td>01.05</td>
<td>Human Evolutionary Genetics I/BB</td>
<td>04.05</td>
<td>Human Evolutionary Genetics II/BB</td>
<td>05.05</td>
<td>Experimental practical: Detecting CYP2D6 polymorphisms II/Iv, AG</td>
</tr>
<tr>
<td>10</td>
<td>08.05</td>
<td>Animal models of disease/ IV</td>
<td>11.05</td>
<td>Personalized Medicine/AG</td>
<td>12.05</td>
<td>Review of Quantitative Genetics; Fingerprints Practical/WS</td>
</tr>
<tr>
<td>11</td>
<td>15.05</td>
<td>Genotoxicity/WS</td>
<td>18.05</td>
<td>Cancer genetics/FB</td>
<td>19.05</td>
<td>Next generation sequencing</td>
</tr>
<tr>
<td>12</td>
<td>22.05</td>
<td>Genetic Basis of Intelligence/WS</td>
<td>25.05</td>
<td>Genetics of Alzheimer’s Disease/MJ</td>
<td>26.05</td>
<td>Presentations</td>
</tr>
<tr>
<td>13</td>
<td>29.05</td>
<td>No lecture</td>
<td>01.06</td>
<td>No lecture</td>
<td>02.06</td>
<td>Presentations</td>
</tr>
</tbody>
</table>
**4. Course Details**

### Course Description

(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 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 personalised medicine. Emphasis will be placed on modern research approaches that use next generation sequencing and genomics.

### Student Learning Outcomes

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. Run bioinformatics tools in both Windows and Linux, using graphical user interfaces and command line prompts
8. Apply bioinformatics sequence analysis tools to explore the function and evolutionary history of DNA and protein sequences

### Graduate Attributes Developed in this Course

<table>
<thead>
<tr>
<th>Science Graduate Attributes</th>
<th>FOCUS</th>
<th>Activities / Assessment</th>
</tr>
</thead>
<tbody>
<tr>
<td>Research, inquiry and analytical thinking abilities</td>
<td>3</td>
<td>Computer exercises and written reports</td>
</tr>
<tr>
<td>Capability and motivation for intellectual development</td>
<td>3</td>
<td>Lectures and computer practicals, group discussions and written reports</td>
</tr>
<tr>
<td>Ethical, social and professional understanding</td>
<td>2</td>
<td>Article discussions</td>
</tr>
<tr>
<td>Communication</td>
<td>3</td>
<td>Article discussions, written reports,</td>
</tr>
<tr>
<td>Teamwork, collaborative and management skills</td>
<td>2</td>
<td>Article discussions, computer</td>
</tr>
</tbody>
</table>

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3 Learning and Teaching Unit: [Course Outlines](http://www.handbook.unsw.edu.au)
4 Learning and Teaching Unit: [Learning Outcomes](http://www.handbook.unsw.edu.au)
| Information literacy | 3 | Computer practicals, article discussions and written reports |

**Major Topics (Syllabus Outline)**

- Human Genome Structure and Function
- Modes of inheritance and genetics methods
- Next generation sequencing and its applications to human genetics
- Human evolutionary genetics
- Research topics in human genetics

**Relationship to Other Courses within the Program**

- Builds on the second year course Genetics (BIOS2021/BIOS2621)
- Establishes foundation for session 2 course Genes, Genomes & Evolution (BABS3291)
- Other courses related to Genes in Time and Space:
  - Molecular Biology of Nucleic Acids (BABS3121)
  - Molecular Genomics (BABS3281)
  - Microbial Genetics (BABS3021/MICR3621)
  - Bioinformatics Methods and Applications (BINF3010)
  - Conservation Biology and Biodiversity (BIOS3071)
  - Population and Community Ecology (BIOS3111)
  - Evolution (BIOS3171).

**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 in genetics as well as recent developments. The practicals explore some aspects of the material introduced in lectures and extend the discussion to other relevant topics and skills. Computer-based exercises will provide hands-on exposure to methods, tools and concepts used in genetics. The presentation emphasises developing the ability to communicate and evaluate research results. Effective communication of science is also evaluated through written reports.

**Rationale for learning and teaching in this course**

This is a third year course that builds on ideas taught in second year genetics (BIOS2021/BIOS2621). In developing these ideas, we aim to contextualise the material by using examples of current relevance in the discipline and in society. 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.

*Reflecting on your teaching*
6. Lecture content

**Topic 1: Human Genome Structure and Function**
1. Introduction to Human Genetics /IV
2. Human Genome Structure/RE
3. Human Genome Variation/RE
4. Sex determination/PW
5. Transcriptomics/ IV
6. Proteomics/ RE
7. Epigenomics/OB

**Topic 2: Modes of inheritance and genetics methods**
8. Linkage Analysis and Monogenic Traits/IV
9. Oligogenic Traits /IV
10. Multifactorial Inheritance /IV
11. Genome Wide Association Studies/IV
12. Lessons from Genome Wide Association Studies/IV
13. Heritability Estimates/ IV

**Topic 3: Next generation sequencing and its applications to human genetics**
14. Whole Exome and Whole Genome Sequencing/IV
15. Applications of Genome Sequencing/MC

**Topic 4: Human evolutionary genetics**
16. Human Evolutionary Genetics I/BB
17. Human Evolutionary Genetics II/BB

**Topic 5: Research topics in human genetics**
18. Animal models of disease/ IV
19. Personalized Medicine/AG
20. Genotoxicity/WS
21. Cancer genetics/FB
22. Genetic Basis of Intelligence/WS
23. Genetics of Alzheimer’s Disease/MJ
24. Forensics/PW
### 7. Assessment Tasks and Feedback

<table>
<thead>
<tr>
<th>Task</th>
<th>Knowledge &amp; abilities assessed</th>
<th>Assessment Criteria</th>
<th>% of total mark</th>
<th>Date of Feedback</th>
<th>Feedback</th>
</tr>
</thead>
</table>
| Assignment 1: Report paper on the data analysis carried out in weeks 4-6 | Ability to utilise bioinformatics tools to extract information on DNA sequence variation, gene expression and epigenetic modifications. Ability to critically interpret the data. | - Accurate completion of task  
- Clear exposition of results  
- Presentation of writing, including formatting, spelling/grammar, references, structure. | 20 | Week 6 | Week 9 | I. Voineagu | Week 11 | Marks, written comments on report |
| Mid-term Exam | Essential disciplinary knowledge in genetics at 3rd year level; emphasis on genomics. | - Depth of understanding of examined material  
- Clarity of explanation | 30 | Week 7 | Week 7 | Lecturers in the first 6 weeks | Week 9 | Results posted |
| 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 | 20 | Week 5 | Weeks 12,13 | I. Voineagu | Weeks 12,13 | Verbal feedback from academics and peers Results posted |
| Final Exam | Essential disciplinary knowledge in genetics at 3rd year level; emphasis on evolutionary genetics. | - Depth of understanding of examined material  
- Clarity of explanation | 30 | End of year exam period | End of year exam period | Lecturers from weeks 7 to 12 | Check exam Timetable when available | Results posted |

8. Additional Resources and Support

<table>
<thead>
<tr>
<th>Text Books</th>
<th>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.</th>
</tr>
</thead>
<tbody>
<tr>
<td>Course Manual</td>
<td>This document and the Moodle site for this course.</td>
</tr>
</tbody>
</table>
Library website and resources: [http://info.library.unsw.edu.au/](http://info.library.unsw.edu.au/)  
| Computer Laboratories or Study Spaces | Room G08 - but only when no classes are being held in this room |
9. Administration Matters

<table>
<thead>
<tr>
<th>Expectations of Students</th>
<th>Attendance at all practicals and lectures is compulsory unless you have a valid and documented medical or other reason. Be punctual. Do not talk or eat during lectures. Mobile phones must be switched off during lectures and practicals. Attendance and arrival times at practicals will be marked.</th>
</tr>
</thead>
<tbody>
<tr>
<td>Assignment Submissions</td>
<td>All assignments must have a signed cover sheet unless submitted via Turn-It-In online. Submissions without cover sheets will not be marked. All assignments must be typed. Hand-written submissions will not be marked. Assignment must be submitted by 4.30 pm on the due date. A late submission will incur a penalty of 10% of the total value per day.</td>
</tr>
</tbody>
</table>

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
SEMIESTER 1 2017

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 such as (laboratories and tutorials), in-session assessments tasks, and final examinations. **Students must make a formal application for Special Consideration** for the course/s affected as soon as practicable after the problem occurs and **within three working days of the assessment to which it refers.**

Students should consult the “Special Consideration” section of Moodle for specific instructions related to each BABS course they are studying. Further general information on special consideration can also be found at https://student.unsw.edu.au/special-consideration.

**HOW TO APPLY FOR SPECIAL CONSIDERATION**

Applications must be made via Online Services in myUNSW. **You must obtain and attach Third Party documentation before submitting the application. Failure to do so will result in the application being rejected.** Log into myUNSW and go to My Student Profile tab > My Student Services channel > Online Services > Special Consideration. After applying online, students must also verify supporting their documentation by submitting to **UNSW Student Central:**

- Originals or certified copies of your supporting documentation (Student Central can certify your original documents), and
- A completed Professional Authority form (pdf - download here).

The supporting documentation must be submitted to Student Central for verification **within three working days** of the assessment or the period covered by the supporting documentation. Applications which are not verified will be rejected.

**Students will be contacted via the online special consideration system as to the outcome of their application. Students will be notified via their official university email once an outcome has been recorded.**

**SUPPLEMENTARY EXAMINATIONS:**

The University does not give deferred examinations. However, further assessment exams may be given to those students who were absent from the final exams through illness or misadventure. Special Consideration applications for final examinations and in-session tests will only be considered after the final examination period when lists of students sitting supplementary exams/tests for each course are determined at School Assessment Review Group Meetings. Students will be notified via the online special consideration system as to the outcome of their application. **It is the responsibility of all students to regularly consult their official student email accounts and myUNSW in order to ascertain whether or not they have been granted further assessment.**

For Semester 1 2017, BABS Supplementary Exams will be scheduled on:

- All BABS coded courses (except BABS1201): Wednesday 12th of July
- All BIOC coded courses: Thursday 13th of July
- BABS1201 and MICR-coded courses: Friday 14th of July
Further assessment exams will be offered on this day ONLY and failure to sit for the appropriate exam may result in an overall failure for the course. Further assessment will NOT be offered on any alternative dates.