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Course Summary

The field of human genetics has been transformed over the past few years by the wealth of whole genome sequence information that has been obtained from human populations and individuals throughout the world. This has allowed us fundamental insights into the genetic basis of the human condition, from the phenotypic differences that distinguish us as individuals through to disease susceptibility. The aim of this course is to give you an overview of current research findings in the field of human genetics and the importance played by the ready availability of genome sequence data to this enterprise.

Course Aims & Learning Outcomes

The subject-specific learning outcomes are such that, at the end of the course, students should be able to:

- understand the importance played by the ready availability of genome sequence data from human populations and individuals throughout the world in the field of human genetics. Examples will include an analysis of genome databases.
- understand the processes, both directed and random, that shape the formation and reformation of the human genome and how these processes, if mis-directed, can lead to disease.
- Interrogate genome databases and identify genes, gene regulatory elements as well as features of variation within genomes.
- Understand the importance of gene regulation in health and how gene mis-regulation may contribute to disease.

Course Teaching Staff

Course Co-ordinator(s):
Dr Alasdair Mackenzie (alasdair.mackenzie@abdn.ac.uk)

Other Staff:
Dr. Mary–Joan, Macleod (mjmacleod@abdn.ac.uk)
Dr Elizabeth Hay.

Assessments & Examinations

This course is assessed via a written examination (worth 70% of the overall course grade) in the May exam diet and one piece of continuous assessment (worth 30% of the overall course grade).

The continuous assessment associated with this course is detailed below, with two other pieces of work being associated with your other “Option” course and the “Core” course.

It is vital that the deadlines for your continuous assessments are adhered to. Submit an incomplete piece of work rather than miss a deadline. Work not submitted on time will not be accepted unless accompanied by either a medical certificate or a written explanation justifying this.

A complete submission of your work consists of:

- uploading an electronic copy of the work via MyAberdeen before 12 NOON on the deadline date.

The deadlines for all three pieces of work are:

- Core course Research Perspective: 12 NOON, Monday 30th September.
- Option 1 course Research Tutorial Spotlight: 12 NOON, Monday 4th November.
- Option 2 course Essay: 12 NOON, Monday 6th December.
Research Essay

The essay topics are given below. You should select one title.

1. What evidence is there that DNA-methylation impacts health, plays a role in disease and represents a possible therapeutic target?
2. Review the evidence that histone methylation plays a role in health and disease.
3. Discuss the genomic mechanisms that control multicellularity and how they support human health and disease.
4. Discuss the success of GWAS analysis in our understanding of disease and what technologies might replace it.

You are welcome to use subheadings to structure the essay as you see fit, but the following should be included on a title page.

- Title (do not modify the title from above)
- Name
- Student ID
- Word Count

Word limit for your Research Essay is **2,000 words** and you can use as many appropriate figures/tables as you wish. The word limit does not include text in tables, figure legends, or references.

Scientific Writing

Writing is an important scientific skill. Its function in the Honours courses is to provide you with training in finding, reading, analysing and communicating scientific ideas. Although it is usually necessary to start your reading from reviews that provide an expert overview of a topic, it is critical to your development that you read a significant number of original papers that describe the experiments underpinning key scientific advances. Central to these skills is the development of the ability to judge the important points made in a paper and what are the central pieces of evidence that support those points. Finally, it is important for all graduates to have a working knowledge of the key experimental procedures and techniques that generate the data that we use to test hypotheses.

**Word Limit:** Adhering to a word limit (excluding figure legends, tables and the reference list) requires you to be disciplined in the preparation of the piece of work; being able to write to a required length is a very useful skill, so we expect you to stay within the limit set. Your computer will give you a word count; this must be included at the end of the work submitted. **We reserve the right to return work exceeding the word count for shortening. Submissions returned for shortening must be re-submitted within 24 h.** Having to resubmit your work again will delay marking and subsequent feedback.

**Assessment:** The continuous assessment for Honours will be assessed by two members of staff, using criteria that will be published in MyAberdeen alongside the submission links for each piece of work. This assessment is not open to negotiation, although if asked, the markers will clarify any points of constructive criticism. Please use the assessment criteria as a guide and read them with care; the notes on scientific writing also give you guidance on what we judge to be important in a well-written piece of work. If you have particular doubts about your ability to write scientifically, either in terms of organising material or in the mechanics of good scientific writing, seek help from a member of staff or the Honours Coordinators during the first term. Do not wait until your first assignment is causing you anxiety.

All submissions should make reference to the latest literature on the subject you have chosen. While you may be guided through an unfamiliar subject area by reference to a review, your work should specifically not paraphrase the review article, but should be a synthesis of your own views of the subject, written in your own words arrived at by reading of the original research papers from resources such as Web of Science/Medline/PubMed/Google Scholar. This will give insight into how information is derived (one criteria assessed) as well as helping in preparation for the Data Analysis exam at the end of the year, where understanding of a research paper is tested.
Avoiding Plagiarism

The definition of Plagiarism is the use, without adequate acknowledgement, of the intellectual work of another person in work submitted for assessment. A student cannot be found to have committed plagiarism where it can be shown that the student has taken all reasonable care to avoid representing the work of others as his or her own.

The instruction given above to write assignments in your own words and not to copy whole sentences from articles is crucially important to avoid plagiarism.

The University views this offence extremely seriously; indeed, it can have dire consequences, including the awarding of no higher than a pass degree.

Continuous assessment assignments and your thesis are all submitted as electronic copies via MyAberdeen so they can be checked for originality. The programme will detect passages of text copied from other sources, and also if sentences from various text sources were used throughout the text, both indicators of plagiarism. MyAberdeen accepts most common formats, but it is advised that you submit your work as PDF files to avoid problems with reformatting of figures and/or text during the submission process. Any evidence of copying from other sources that is detected in your final submissions will be brought to the attention of the Head of School, who will investigate and determine whether cheating has occurred and take the appropriate action.

Feedback

As for all elements of continuous assessment, you will be given feedback on the Honours classification your work has attained, with the grading on the University Common Grading Scale (CGS). Feedback is normally given within 3 weeks of submission.

Guide to Writing

Students should refer to "A Guide to Scientific Writing" by David Lindsay (Longman Cheshire) for more general guidance on writing. What follows is not a substitute for reading this book, but gives general guidance on writing and on how we assess your work.

PLANNING YOUR WRITING

Think

- What do I know already?
- Where will I find the information needed to develop my views on this issue?
- Where can I find more information?
- What are the best examples to illustrate the points that I want to make?
- How many words do I devote to each example?

Prepare

- Read a mix of reviews and use these to identify the major original scientific papers that have resulted in our current understanding of the topic.
• Read these papers and make notes on: research strategy use to analyse the problem, key experimental procedures that generate the data and critical controls that validate the data.
• Devise a set of themes and ideas for your work using the core information from above.
• Organise evidence under the theme headings: remember that arguments pro and contra are equally important.
• Select illustrations (diagrams/schemes) that reflect the themes and ideas.

Plan
• Place themes in a logical order, and have a clear, and planned, introduction and conclusion.
• Start simply and develop towards more complex arguments.
• Do not hop from one theme to another and then back again.
• Identify the links between themes as a mechanism of ensuring continuity.

Execute
• Write short sentences and keep clauses simple.
• Use appropriate tenses.
• Be consistent in the organisation of sections.
• Have diagrams in front of you when writing about them.
• Support statements with evidence, usually a citation; ensure your citation style is consistent.

Complete
• Read over what you have written - can you read it out loud without stumbling?
• Have you answered the question?
• Have you done what you said you would do at the start of the assignment?
• Have you checked it carefully for typographical errors?

Assessment of Written Work

Every piece of work in your Honours year will be assessed using a standardised assessment form. The assessment forms ensure that you get useful feedback on your written work. The Continuous Assessment form covers the following criteria.

Content and Presentation

Each piece of work will be judged on content and also on style of presentation. More marks are given for the content of the work than are given for the presentation. Look at the structure of the feedback form to see what the priorities are in giving marks. However, remember also that a written piece of work must always be more than a collection of facts and ideas. Good presentation is central to clear communication.

Knowledge: It is expected that any piece of work will contain a substantial body of facts gleaned from appropriate original literature, which should be cited within the text (Citations). The length of the work and its intended audience will dictate how many facts can be given in support of a given statement.

Analysis: Students are expected to develop their analytical skills. This is most readily demonstrated by use of carefully selected examples, which should show a good understanding of the material. Remember that examples may either support or undermine an argument.

Understanding: Students are expected to display a clear grasp of fundamental concepts in the context of the work and their discipline. This is sometimes illustrated by the lack of mistakes about fundamentals of the cell and cellular processes, but it is also expected a student will develop, through their reading, an understanding of the subject area and display this by writing logically about it.
**Techniques**: Scientific information is derived from experimentation. It is important to understand how information is derived. For example what technique was used, how was the experiment conducted etc.

**Figures**: An argument can often be supported by Figures or Tables that present information more effectively than text alone. Figures and Tables should not be an add-on, but must be an integral feature of the text and must be described and discussed. A poor or inappropriate figure or table will usually detract from the work. Appropriate figures prepared by hand or using a drawing programme are preferred to reproductions of complex diagrams from other people's work (if used, make sure you acknowledge the source).

**Citations**: Papers and reviews used as source material should be cited in the text. Direct quotes should be indicated by quotation marks, although their use should be kept to a minimum, and they must be referenced (see University Web page on plagiarism). Use of the Harvard style of citation is essential and a list of citations should be presented at the end of the work (referencing of EMBO Journal articles is a good example). The reference list does not have to be included in your word count.

In the text a reference should be cited by author and date; e.g. 'Water is known to boil at 100°C (Jones and Brown, 1872; Brown et al., 1873) and freeze at...'. Not more than two authors may be cited per reference; if there are more than two authors use et al. References should be listed alphabetically according to the initial letter of the surname of the first author. Where the same authors have published more than one paper, list them in the order in which their papers appeared. If necessary use a and b, e.g. 1990a., with the authors' surnames and initials inverted.

References should include, in the following order:

- authors' names; year; article or chapter title; editors (books only); journal or book title; name and address of publisher (books only); volume number and inclusive page numbers. The name of each journal should be abbreviated according to the World List of Scientific Periodicals (see an EMBO J. paper for reference) and italicised. References should therefore be listed as follows:


**Structure**: A good piece of writing will be clearly structured by division into appropriate sections, including an introduction, which provides a clear and concise statement of the issue to be discussed, and a conclusion, which briefly sums up the issues discussed.

**Introduction**: a clear and brief introduction of the topic of the work that describes the specific areas questions or issues that the reader should focus on.

**Viewpoint**: Students should form a view on the subject about which they are writing and should be able to support their views with balanced use of appropriate examples. A balanced piece of work will consider the relative strengths of the arguments for and against a particular point of view.

**Conclusions**: this section is used to pull the main themes of the work together and to briefly state the principal outcome of the analysis that you have performed. It should leave the reader with a clear impression of what you think about the subject matter presented.
**Sentence construction, spelling, grammar:** Students are expected to spell correctly and to follow the basic rules of grammar. Short, clear sentences are preferable to complex, tortuous, rambling constructions. You should be able to pick up the eight clear grammatical, punctuation and spelling errors in the sentence that follows. If you can’t, then revise your grammar/spelling rules. “It’s clear to the company that their commercial targeted young people of the same age as John’s friends who were clearly able to receive it’s message.” Where possible reduce your use of TLAs (three letter abbreviations). As a guide, only use TLAs if the words you are abbreviating are used more than 5 times.

**Organisation:** A written assignment is easier to read if it is attractively set out on the page (wide margins, double spaced, font size ≥12) with a logical progression and structure.

**Specific comments:** This section is provided for the staff to make comments that amplify the box assessments in the top half of the form.

**Note that computer failure is not accepted as a reason for late submission - it is good practice to maintain at least two copies of computer files.**
Sample Assessment/Feedback Form

<table>
<thead>
<tr>
<th>Criteria</th>
<th>1st Class</th>
<th>2.1 Class</th>
<th>2.2 Class</th>
<th>3rd Class</th>
<th>Bare Pass</th>
</tr>
</thead>
<tbody>
<tr>
<td>CONTENT</td>
<td>Excellent demonstration of knowledge and understanding, grasp of fundamental concepts, selective use of arguments.</td>
<td></td>
<td></td>
<td></td>
<td>Little or no relevant content, superficial knowledge, lack of grasp of fundamentals, arguments not relevant.</td>
</tr>
<tr>
<td>TECHNICAL INSIGHT</td>
<td>Clear recognition of how information was derived.</td>
<td></td>
<td></td>
<td></td>
<td>Lacking insight and a demonstration of how information was derived.</td>
</tr>
<tr>
<td>STRUCTURE</td>
<td>Clear logical structure with and meaningful introduction, main text and conclusion sections, clearly argued.</td>
<td></td>
<td></td>
<td></td>
<td>Poorly structured, confused order of topics, poorly focused.</td>
</tr>
<tr>
<td>FIGURES</td>
<td>Well integrated with text, with appropriate legend, clearly illustrated.</td>
<td></td>
<td></td>
<td></td>
<td>Not appropriate, poorly integrated, legends irrelevant or missing, untidy, poorly labelled.</td>
</tr>
<tr>
<td>REFERENCING</td>
<td>Good use of a range of references.</td>
<td></td>
<td></td>
<td></td>
<td>Citations lacking or erroneous, format inconsistent.</td>
</tr>
<tr>
<td>PRESENTATION</td>
<td>Visually attractive, well-organised, legible.</td>
<td></td>
<td></td>
<td></td>
<td>Untidy, badly organised, illegible.</td>
</tr>
<tr>
<td>SENTENCE CONSTRUCTION, SPELLING AND GRAMMAR</td>
<td>Sentence construction good, readability high, spelling and grammar correct.</td>
<td></td>
<td></td>
<td></td>
<td>Sentence construction poor, incoherent, many errors.</td>
</tr>
</tbody>
</table>
**Class Representatives**

**We value students’ opinions in regard to enhancing the quality of teaching and its delivery; therefore, in conjunction with the Students’ Association we support the Class Representative system.**

In the School of Medicine, Medical Sciences & Nutrition we operate a system of course representatives, who are elected from within each course. Any student registered within a course that wishes to represent a given group of students can stand for election as a class representative. You will be informed when the elections for class representative will take place.

**What will it involve?**

It will involve speaking to your fellow students about the course you represent. This can include any comments that they may have. You will attend a Staff-Student Liaison Committee and you should represent the views and concerns of the students within this meeting. As a representative, you will also be able to contribute to the agenda. You will then feedback to the students after this meeting with any actions that are being taken.

**Training**

Training for class representatives will be run by the Students Association. Training will take place within each half-session. For more information about the Class representative system visit www.ausa.org.uk or email the VP Education & Employability vped@abdn.ac.uk. Class representatives are also eligible to undertake the STAR (Students Taking Active Roles) Award with further information about this co-curricular award being available at: www.abdn.ac.uk/careers.

**Problems with Coursework**

If students have difficulties with any part of the course that they cannot cope with, alone they should notify the course coordinator immediately. If the problem relates to the subject matter general, advice would be to contact the member of staff who is teaching that part of the course. Students with registered disabilities should contact the medical sciences office, (medsci@abdn.ac.uk) (based in the Polwarth Building, Foresterhill) to ensure that the appropriate facilities have been made available. Otherwise, you are strongly encouraged to contact any of the following as you see appropriate:

- Course student representatives
- Course co-ordinator
- Convenor of the Medical Sciences Staff/Student Liaison Committee (Professor Gordon McEwan)
- Personal Tutor
All staff are based at Foresterhill and we strongly encourage the use of email or telephone the Medical Sciences Office. You may have a wasted journey travelling to Foresterhill only to find staff unavailable.

If a course has been completed and students are no longer on campus (i.e. work from second half session during the summer vacation), coursework will be kept until the end of Fresher’s Week, during the new academic year. After that point, unclaimed student work will be securely destroyed.

**Course Reading List**

This course does not have recommended textbooks as you are expected to read the primary research literature and the most recent reviews (most of which will be recommended during lectures) so no specific course texts will be recommended. That said, your previous text books from third year would be useful for basic and fundamental knowledge.

**Lecture Synopsis**

The lectures on this course will critically assess the benefits that have accrued from the sequencing of the human genome and the development of genomic technologies. The lectures will provide you with a basis for further reading to understanding of how modern molecular genomic techniques can help us to understand the biological and genomic systems that have evolved to keep us alive and healthy and how genome variation and environment can influence health to cause disease. You will be able to understand the most up-to-date developments in genomics and molecular biology and to access and understand the huge wealth of information available relating to the biology of the human genome and how we are able to predict the effects of genetic and epigenetic variation on the ability of the human population to age healthily. Lecture content: the post genomic age, genomics, evolution and disease, ENCODE!, the non-coding genome, enhancers, chromosomes and TADs, introduction to genome data mining, functional analysis of regulatory SNPS, mammalian disease models, genome editing, the future of genetics, molecular epigenetics, clinical epigenetics.

**Practical/Lab/Tutorial Work**

This course contains two research tutorial sessions.

1. **Data mining:** The initial tutorial session will allow students to explore the bioinformatics used in a manuscript by Oldridge et al (2015) “Genetic predisposition to neuroblastoma mediated by a LMO1 super-enhancer polymorphism”, Nature 528, 418-421. **Due to Covid restrictions this class will take place on-line through Collaborate on MyAberdeen.**

2. **Video presentation:** The second tutorial will involve the sharing of a pre-recorded 5-minute online video presentation by each of the students summarising subjects discussed in essay 2. The content of the video need not reproduce the entire content of the essay but reflect an aspect of the essay which the student found particularly interesting. **These tutorials will be presented on-line due to Covid restrictions** and will serve to broaden the field of knowledge of the audience whilst developing the oral presentation skills of each student. Feedback on presentation skills, accessibility, content and understanding will be provided by both members of staff and students and a prize will be awarded for the best presentation. These can be recorded in Powerpoint using...
the “Slide Show” option and submitted to MyAberdeen before the date displayed in the timetable. These presentations will be presented on Collaborate on MyAberdeen as per the timetable. These formative assessments are compulsory.

University Policies

Students are asked to make themselves familiar with the information on key education policies, available here. These policies are relevant to all students and will be useful to you throughout your studies. They contain important information and address issues such as what to do if you are absent, how to raise an appeal or a complaint and how the University will calculate your degree outcome.

These University wide education policies should be read in conjunction with this programme and/or course handbook, in which School specific policies are detailed. These policies are effective immediately, for the 2022/23 academic year. Further information can be found on the University’s Infohub webpage or by visiting the Infohub.

The information included in the institutional area for 2022-23 includes the following:

- Assessment
- Feedback
- Academic Integrity
- Absence
- Student Monitoring/ Class Certificates
- Late Submission of Work
- Student Discipline
- The co-curriculum
- Student Learning Service (SLS)
- Professional and Academic Development
- Graduate Attributes
- Email Use
- MyAberdeen
- Appeals and Complaints

Where to Find the Following Information:

C6/C7- University of Aberdeen Homepage > Students > Academic Life > Monitoring and Progress > Student Monitoring (C6 & C7)
https://www.abdn.ac.uk/students/academic-life/student-monitoring.php#panel5179

Absences- To report absences you should use the absence reporting system tool on Student Hub. Once you have successfully completed and sent the absence form you will get an email that your absence request
has been accepted. The link below can be used to log onto the Student Hub Website and from there you can record any absences you may have.

Log In - Student Hub (ahttps://www.abdn.ac.uk/studenthub/loginbdn.ac.uk)

Submitting an Appeal- University of Aberdeen Homepage > Students > Academic Life > Appeals and Complaints

https://www.abdn.ac.uk/students/academic-life/appeals-complaints-3380.php#panel2109

Academic Language & Skills support
For students whose first language is not English, the Language Centre offers support with Academic Writing and Communication Skills.

Academic Writing
- Responding to a writing task: Focusing on the question
- Organising your writing: within & between paragraphs
- Using sources to support your writing (including writing in your own words, and citing & referencing conventions)
- Using academic language
- Critical Thinking
- Proofreading & Editing

Academic Communication Skills
- Developing skills for effective communication in an academic context
- Promoting critical thinking and evaluation
- Giving opportunities to develop confidence in communicating in English
- Developing interactive competence: contributing and responding to seminar discussions
- Useful vocabulary and expressions for taking part in discussions

More information and how to book a place can be found here
<table>
<thead>
<tr>
<th>Grade</th>
<th>Grade Point</th>
<th>% Mark</th>
<th>Category</th>
<th>Honours Class</th>
<th>Description</th>
</tr>
</thead>
</table>
| A1    | 22          | 90-100 | Excellent  | First         | • Outstanding ability and critical thought  
• Evidence of extensive reading  
• Superior understanding  
• The best performance that can be expected from a student at this level |
| A2    | 21          | 85-89  |            |               |                                                                            |
| A3    | 20          | 80-84  |            |               |                                                                            |
| A4    | 19          | 75-79  |            |               |                                                                            |
| A5    | 18          | 70-74  |            |               |                                                                            |
| B1    | 17          | 67-69  | Very Good  | Upper Second  | • Able to argue logically and organise answers well  
• Shows a thorough grasp of concepts  
• Good use of examples to illustrate points and justify arguments  
• Evidence of reading and wide appreciation of subject |
| B2    | 16          | 64-66  |            |               |                                                                            |
| B3    | 15          | 60-63  |            |               |                                                                            |
| C1    | 14          | 57-59  | Good       | Lower Second  | • Repetition of lecture notes without evidence of further appreciation of subject  
• Lacking illustrative examples and originality  
• Basic level of understanding |
| C2    | 13          | 54-56  |            |               |                                                                            |
| C3    | 12          | 50-53  |            |               |                                                                            |
| D1    | 11          | 47-49  | Pass       | Third         | • Limited ability to argue logically and organise answers  
• Failure to develop or illustrate points  
• The minimum level of performance required for a student to be awarded a pass |
| D2    | 10          | 44-46  |            |               |                                                                            |
| D3    | 9           | 40-43  |            |               |                                                                            |
| E1    | 8           | 37-39  | Fail       | Fail          | • Weak presentation  
• Tendency to irrelevance  
• Some attempt at an answer but seriously lacking in content and/or ability to organise thoughts |
| E2    | 7           | 34-36  |            |               |                                                                            |
| E3    | 6           | 30-33  |            |               |                                                                            |
| F1    | 5           | 26-29  | Clear Fail | Not used for Honours | • Contains major errors or misconceptions  
• Poor presentation |
<p>| F2    | 4           | 21-25  |            |               |                                                                            |</p>
<table>
<thead>
<tr>
<th>Date</th>
<th>Time</th>
<th>Place</th>
<th>Subject</th>
<th>Session</th>
<th>Staff</th>
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<tbody>
<tr>
<td><strong>Week 13</strong></td>
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<tr>
<td>Mon 24 Oct</td>
<td>14.00-16.00</td>
<td>1M:001</td>
<td>Introduction and The Post Genomic Age.</td>
<td>Lecture</td>
<td>AM</td>
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<td>Genomics, Evolution and Disease</td>
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<td>Tue 25 Oct</td>
<td>14:00-16:00</td>
<td>1M:001</td>
<td>ENCODE!</td>
<td>Lecture</td>
<td>AM</td>
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<td>The Non-coding Genome</td>
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<td>Wed 26 Oct</td>
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<td>Thu 27 Oct</td>
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<td>Fri 28 Oct</td>
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<td><strong>Week 14</strong></td>
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<tr>
<td>Mon 31 Oct</td>
<td>15.00-16.00</td>
<td>1M:001</td>
<td>Enhancers, Chromosomes and TADs</td>
<td>Lectures</td>
<td>AM</td>
</tr>
<tr>
<td>Tue 1 Nov</td>
<td>10:00-12:00</td>
<td>1M:001</td>
<td>Introduction to genome data mining</td>
<td>Lectures</td>
<td>AM</td>
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<td>Functional analysis of regulatory SNPs</td>
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<td>Wed 2 Nov</td>
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<td>Thu 3 Nov</td>
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<td>Fri 4 Nov</td>
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<td><strong>Week 15</strong></td>
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<tr>
<td>Mon 7 Nov</td>
<td>14.00-16.00</td>
<td>1M:001</td>
<td>Mammalian Disease Models</td>
<td>Lectures</td>
<td>AM</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td>Genome editing</td>
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<td></td>
</tr>
<tr>
<td>Tue 8 Nov</td>
<td>11.00-13.00</td>
<td>1M:001</td>
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Staff Code. AM; Alasdair MacKenzie, MJMcL; Mary Joan MacLeod, EH; Elizabeth Hay.

Campus Maps - Foresterhill