



GN4010

Honours Genetics (Option 1)

Course Handbook
2019-20

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Cover image:

Confocal micrograph of fluorescently labelled HeLa cells.

Nuclei are labelled in blue, tubulin in green and actin fibres in red.

Courtesy of:

Kevin Mackenzie

Microscopy and Histology Core Facility

Institute of Medical Sciences

University of Aberdeen

<http://www.abdn.ac.uk/ims/microscopy-histology>

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 disease susceptibility through to the features that distinguish us from our closest relatives, the great apes. The aim of this module 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 tools and techniques used to analyse and extract biological information from human genome sequence data;
- understand the forces, both directed and random, that shape the formation and reformation of genomes, including selective pressure, genetic drift and recombination;
- understand the approaches used to show that selection has been operating on the human genome in the recent and distant past, and the limitations and caveats that apply.

Course Teaching Staff

Course Co-ordinator(s):

Dr Jonathan Pettitt (j.pettitt@abdn.ac.uk)

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 2nd December.

Research Tutorial Spotlight

This exercise aims to create a lay summary of the topic covered in the research tutorials contained within this course. A lay summary is often used in grant applications and is also appearing more frequently when submitting research articles for publication. It is a brief summary of a research project or a research proposal that has been written for members of the public, rather than researchers or professionals. Something similar to the style encountered in a New Scientist article would be a good term of reference. It should be written in plain English, avoid the use of jargon and explain any technical terms that have to be included. Overall, this piece of work should have a more journalistic style and be engaging.

Your submission should be short, should use a simple figure to summarise the work (a visual abstract), and have a concise list (3-5 articles) of relevant references. As guidance, you should take the reader through the following:

The background: what did we know before; why were the studies conducted? In particular, place the subject matter in a broader context for a lay reader – explain why they should pay attention to the topic – what is important and why?

The subject/topic itself: what did the researchers do and find; some strengths and limitations; what are the implications?

The future: what are the next steps; are there unanswered questions?

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

- *Title* (8 words maximum)
- *Name*
- *Student ID*
- *Word Count*

Word count for your Research Spotlight is **800 words** and you can use a maximum of one figure. The word limit does not include text in the figure or references.

As an excellent resource and some guidance on writing for a lay audience, see the link below to the “Make it Clear” campaign, aimed at making scientific writing and research as clear and understandable as possible.

<http://www.invo.org.uk/makeitclear/>

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 re-formatting 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:

Tugendreich, S., Bassett, D.E., Jr, McKusick, V.A., Boguski, M.S. and Hieter, P. (1994) Genes conserved in yeast and humans. *Hum. Mol. Genet.*, 3, 1509-1517.

Gehring, W. (1994) A history of the homeobox. In Duboule, D. (ed.), Guidebook to the Homeobox Genes. Oxford University Press, Oxford, UK, pp. 1-10.

Lewin, B. (1994) Genes V. Oxford University Press, Oxford, UK.

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 there commercial targeted young people of the same age as Johns friends who were clearly able to receive its message."

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 MyAberdeen Rubric

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

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 Mrs Jenna Reynolds (medsci@abdn.ac.uk) in the School Office (based in the IMS, Foresterhill), or Mrs Sheila Jones (s.jones@abdn.ac.uk) in the Old Aberdeen office associated with the teaching laboratories, 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 (Prof Gordon McEwan)
- Adviser of studies
- Medical Sciences Disabilities Co-ordinator (Dr Derryck Shewan)

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 semester during the summer vacation), coursework will be kept until the end of Freshers' 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, so no specific course texts will be recommended. That said, your previous texts from third year would be useful for basic and fundamental knowledge.

For students that would like background reading, perhaps because they feel they have gaps in their population genetic backgrounds, the following textbook will be useful. There are copies of it in the library.

Mark Jobling, Edward Hollox, Toomas Kivisild, Chris Tyler-Smith. *Human Evolutionary Genetics* 2nd Edition (2014). ISBN 9780815341482

Lecture Synopsis

The aim of this course is to provide an in-depth, up-to-date understanding of genetics, with a special emphasis on recent findings from studies in the fields of genome, evolutionary and population biology. The course provides you with insights into human genetics through the availability of multiple human genome sequences and how these are shedding light on the evolution of important human traits. You will 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 range from disease susceptibility through to the features that distinguish us from apes. You will be able to understand the forces, both directed and random, that shape the formation and reformation of human genomes.

Lecture content: The application of new sequencing techniques to human genetics, disease susceptibility, human evolution, genetic diseases, pedigree analysis and genetic risk assessment.

Practical/Lab/Tutorial Work

This course contains one set of research tutorial sessions; small-group teaching exercises in which, supported by a staff member, you will discuss a specialist subject, based upon self-directed reading of the literature. Five papers have been chosen that reflect the development of a particular topic, **and you must have read the five papers before attending the first research tutorial**. The papers will be posted on MyAberdeen.

You will be required to develop an understanding of what constitutes a key and important paper, how the information is derived (techniques and their application), the design of those experiments, an understanding of the crucial data and an appreciation of what in the field is controversial. You are also expected to read outside the prescribed five papers and such 'use of extensive original literature' (CAS marking scheme) will be taken as an indicator of a first-class student.

You will thus build up a set of your own notes on the particular subject area within the research tutorial. For each tutorial, you will be divided into small groups and attend two tutorials, each of 1-2 hours. You will subsequently be examined on your knowledge and understanding of the subjects, in particular *how* they are studied and how given practical techniques function, *what* are the merits of different approaches, and in what situation their application is appropriate. In addition, your understanding of the biology described in the five papers will be tested.

Preparation:

The time spent with the tutors for each tutorial is very limited. For this reason, it is important to 'hit the ground running' and go into the first of your two sessions fully prepared. This reading of the papers should be thorough, making sure in particular you understand the rationale of the results sections, how all the techniques work that are described and how the results lead to the main conclusions of the paper. If anything is not clear, bring those questions along to the tutorial with you; if you are critical of anything in the papers, bring these discussion points along as well. Remember, the tutors are there to support your learning, not to spoon-feed you with the answers. You should therefore go to the tutorial prepared to ask questions, to think, and to contribute to the group discussion subjects suggested in the tutorial notes (below). The tutorials are spaced so as to allow time to prepare for each of the two 2-hour sessions, each of which will have a different focus and address a different set of questions.

University Policies

Students are asked to make themselves familiar with the information on key institutional policies which have been made available within MyAberdeen (<https://abdn.blackboard.com/bbcswebdav/institution/Policies>). 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 indicate how seriously the University takes your feedback.

These institutional policies should be read in conjunction with this programme and/or course handbook, in which School and College specific policies are detailed. Further information can be found on the [University's Infohub webpage](#) or by visiting the Infohub.

The information included in the institutional area for 2019/20 includes the following:

- Absence
- Appeals & Complaints
- Student Discipline
- Class Certificates
- MyAberdeen
- Originality Checking
- Feedback
- Communication
- Graduate Attributes
- The Co-Curriculum

Medical Sciences Common Grading Scale

Grade	Grade Point	Category	Honours Class	Description
A1	22	Excellent	First	<ul style="list-style-type: none"> 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			
A3	20			
A4	19			
A5	18			
B1	17	Very Good	Upper Second	<ul style="list-style-type: none"> 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			
B3	15			
C1	14	Good	Lower Second	<ul style="list-style-type: none"> Repetition of lecture notes without evidence of further appreciation of subject Lacking illustrative examples and originality Basic level of understanding
C2	13			
C3	12			
D1	11	Pass	Third	<ul style="list-style-type: none"> 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			
D3	9			
E1	8	Fail	Fail	<ul style="list-style-type: none"> Weak presentation Tendency to irrelevance Some attempt at an answer but seriously lacking in content and/or ability to organise thoughts
E2	7			
E3	6			
F1	5	Clear Fail	Not used for Honours	<ul style="list-style-type: none"> Contains major errors or misconceptions Poor presentation
F2	4			
F3	3			
G1	2	Clear Fail/ Abysmal	-	<ul style="list-style-type: none"> Token or no submission
G2	1			
G3	0			

Course Timetable GN4010: 2019-2020

Date	Time	Venue	Subject	Session	Staff
Week 7					
Mon 9 Sep	14:00-15:00	1:032/033	Darwin and Demography: Forces that shape human genetic variation	Lecture	Dr J Pettitt
	15:00-16:00	1:032/033	Dawn of the Planet of the Apes: Genetic origins of modern humans 1	Lecture	Dr J Pettitt
Tue 10 Sep	13:00-14:00	1:147	Dawn of the Planet of the Apes: Genetic origins of modern humans 2	Lecture	Dr J Pettitt
	14:00-15:00	1:147	Rise of the Planet of the Apes: admixtures and introgressions	Lecture	Dr J Pettitt
Week 8					
Mon 16 Sep	14:00-15:00	1:032/033	Rise of the Planet of the Apes: the impact of migrations	Lecture	Dr J Pettitt
	15:00-16:00	1:032/033	Human phenotypic variation 1 – dietary adaptation	Lecture	Dr J Pettitt
Tue 17 Sep	13:00-14:00	1:147	Human phenotypic variation 2 – lifestyle adaptation	Lecture	Dr J Pettitt
	14:00-15:00	1:147	Human phenotypic variation 3 – pigmentation	Lecture	Dr J Pettitt
Week 9					
Tue 24 Sep	11:00-12:00	1:143/144	Human phenotypic variation 4 – adaptation to infectious disease	Lecture	Dr J Pettitt
Week 10					
Mon 30 Sep	14:00-15:00	1:143/144	Paternal Effect Mutations	Lecture	Dr J Pettitt
Tue 1 Oct	13:00-14:00	1:147	<i>De novo</i> and rare mutations	Lecture	Dr J Pettitt
Week 11					
Mon 7 Oct	14:00-15:00	1:154	Human Exome Aggregation Consortium Tutorial 1 (Group 1)	Tutorial	Dr J Pettitt
	15:00-16:00	1:154	Human Exome Aggregation Consortium Tutorial 1 (Group 2)	Tutorial	Dr J Pettitt
Tue 8 Oct	13:00-14:00	1:039/044	Human Exome Aggregation Consortium Tutorial 2 (Group 1)	Tutorial	Dr J Pettitt
	14:00-15:00	1:039/044	Human Exome Aggregation Consortium Tutorial 2 (Group 2)	Tutorial	Dr J Pettitt