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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

With the sequencing of the human genome an understanding of how the one-dimensional information of the genome results in a healthy human has never been closer. Understanding these processes will lead to a better understanding of how they can go wrong, producing malformation and disease. This course introduces critical aspects of this process and the first of four modules describes what DNA is, how it replicates and how it is packaged into the genomes of bacteria and higher organisms. This first module will also describe how we can manipulate DNA in order to explore its secrets. The second module will explore the processes that control the expression of genes in both bacteria and higher organisms. This critical aspect of gene function controls where, when and the extent to which genes are turned on; this is the key to multi-cellular complexity and environmental response. The third module will describe the properties of amino acids, peptides and proteins, which contribute to the structure and function of our bodies. This module will also describe how we can analyse the structure and function of proteins. The fourth and final module will be devoted to the study of genes in families and populations. This final module will also discuss how genes can go wrong and lead to disease.

Course Aims & Learning Outcomes

The aims of the course are to enable students:

- To establish an understanding of the structures and functions of nucleic acids and to examine their importance in packaging, replicating and maintaining genetic information;
- To understand the main principles of protein biochemistry, structure and function;
- To understand the basic mechanisms that controls the expression of genes at the levels of transcription and translation in both prokaryotes and eukaryotes
- To understand the principles of how genetic processes can go wrong as a result of mutation and genome rearrangements, thus leading to disease.

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

- To compare and contrast the structure of genomes in eukaryotes and prokaryotes and how they are replicated and packaged;
- To describe the molecular mechanisms involved in gene transcription and translation with specific examples;
- To appreciate the role of protein chemistry and structure in function;
- To describe how genetic mechanisms can go wrong to produce malformation and disease.

Practical skills advanced comprise:

- Ability to follow set protocols and develop competence in measuring volumes and calculating concentration; and the ability to obtain, record, collate and analyse information in the laboratory.

Numerical and Communication skills are encouraged by opportunities:
• To analyse laboratory-acquired information and approach some tutorial problems; and produce written laboratory reports and verbally address topics during tutorials.

Interpersonal and Teamwork skills are encouraged by opportunities:

• To work productively with others in the laboratory; and recognise and respect the views and opinions of others during tutorials.

Self-Management skills are needed in:

• Balancing the various demands of this and other courses you are studying.
• Achieving learning outcomes through a series of lectures, tutorials and practical classes. These three elements complement each other.

The function of the **lectures** is to enable information to be transmitted to a large group. Lectures provide an indication of the quality and quantity of knowledge and understanding expected to be gained by the end of the course. In the majority of cases attendance at lectures is directly linked to exam success. Lectures serve to introduce the student to the information required to pass the exam in a measured and easily digestible format. Because of the need to social distance as a result of the Covid-19 pandemic all lecture materials will be made available as pre-recorded and captioned recordings on the MyAberdeen website. **It is essential that all lecture materials be studied well in advance of attempting the on-line tests!**

**Tutorials.** Every week from week 8 between 12noon and 1pm on Monday one of the lectures and the course coordinator will lead a tutorial to allow students to ask questions or to discuss any aspect of the course material. Please take maximum advantage of these sessions.

**Practical classes** enable the class to use a few of the techniques mentioned in the lectures, and to gain experience in the acquisition, recording, evaluation, interpretation and presentation of experimental results. Subject-specific, intellectual and written communication skills are assessed in the written degree examination and in practical reports. Practical and numerical skills are assessed in practical reports. Communication, interpersonal and teamwork skills are encouraged in tutorials and practicals; these are not formally assessed. Students will be allotted one of four practical groups per practical class. Changes in the group allocation will only be made only if the student has a good reason to change group or is able to swap with a student in another group. Both practical sessions are compulsory. Each practical is assessed using a 60 minute on-line test that will contribute 20% of the final mark.

**The continuous assessment** sessions that follow each of the course modules will be used to assess engagement of students with lecture materials. Each of these 4 X 60 minute assessments can be attempted by the students on any University networked computer at any time over a period of a week after the finish of the relevant block of lectures. Each assessment will contribute 15% to the final marks.
Course Teaching Staff

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

Other Staff:
Prof Iain McEwan (iain.mcewan@abdn.ac.uk)
Dr John Barrow (j.barrow@abdn.ac.uk)
Prof Martin Collinson (m.collinson@abdn.ac.uk)
Dr Alexander Lorenz (a.lorenz@abdn.ac.uk)

Assessments & Examinations

Assessment in BI20M3 consists of:

- Assessment of practical reports (40% of final mark);
- 4 X one hour multiple choice exam (60% of final mark).

Computer based continuous tests ~ online computer based tests will be available online for one week after the last lectures on each module. These tests will not be invigilated and only one hour will be allowed to complete the test once the session is started by the students so it is advisable that the students know their lecture material before the start of the test to make best use of their time. It is also advisable that students have a calculator available as many questions may include simple calculations. Only one attempt will be allowed so it is advisable to prepare well before the test. These tests will contribute 60% of the student’s final marks. For further enquiries, please contact the course co-ordinator Dr MacKenzie, or E-learning (Dr Andrew Yule /Dr Sara Preston).

Computer based assessment of practicals. Practical sessions will be followed immediately by the opportunity to test your knowledge of the practical subject matter and results in an on-line test available via MyAberdeen. These tests will be available for over one week and, once activated, the tests will remain open for one hour and will permit the student to answer 20-30 question. Only one opportunity will be provided. Each practical test will contribute 20% of the final marks together contributing to 40% of the final mark.

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 Science Office (medsci@abdn.ac.uk) in the Medical Sciences Office (based in the Polwarth Building, Foresterhill), or Kelly Reid (k.l.reid@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

For students intending to study Biochemistry and or Molecular Biology as a final degree option the following is advised. Also note that this is the textbook for the BI25M7 Energy for Life course that runs next term –


Or


For Dr Collinson’s Lectures;

Recommended texts include Human Molecular Genetics by Strachan and Read (3rd edition) and Emery’s Elements of Medical Genetics (any edition).

Students who are studying other degrees may prefer the following instead:


Lecture Synopsis

Lecture 1
Title: Introduction to BI20M3 course
Lecturer: Dr Alasdair MacKenzie
Content: A welcome and introduction to the course. This lecture will include a summary of course requirements and student responsibilities.

Module 1; Nucleic Acids

This module will provide an overview of nucleic acid biochemistry with emphasis on the dynamic structure of DNA and the way in which it is replicated and packaged into chromosomes. The basic principles of modern recombinant DNA technologies will also be introduced.

Lectures 1 and 2
Title: Chemistry and structure of DNA and RNA
Lecturer: Dr John Barrow
Content: The genetic code is held within a complex biological polymer, DNA. Moreover, to convert this genetic information into molecules that do work (proteins), a second nucleic acid, RNA, is needed. This lecture describes the structure of DNA and RNA, how their structures were elucidated and the relationship between structure and function in nucleic acids. The concept of chemical instabilities within DNA giving rise to mutations is also introduced.
Lecture 3
Title: Chromosome structure
Lecturer: Dr John Barrow
Content: Chromosomes must not only contain genes but must also encode sequences that regulate gene transcription and the replication and segregation of chromosomes. Thus these huge DNA molecules, especially when considered in relation to the size of a cell, must be packaged into a more compact form. This is a particular problem for eukaryotes with their multiple very large chromosomes. DNA supercoiling is essential to compact DNA but is not sufficient on its own. Histones and other proteins bound to the DNA are needed to pack the DNA into highly ordered structures. The same principles apply to the compaction of prokaryotic chromosomes but these nucleoprotein structures are less ordered in prokaryotes.

Lecture 4
Title: DNA replication
Lecturer: Dr John Barrow
Content: The complex structure of DNA requires complex systems to replicate it. This lecture will describe the fundamental rules of DNA replication and experiments which helped to uncover these rules. The basic reaction of DNA replication will be described followed by a detailed description of how this reaction is catalysed in E. coli. The essential requirements of accuracy and processivity will be related to the structure and function of the E. coli replication machinery.

Lecture 5
Title: DNA repair and genome evolution
Lecturer: Dr John Barrow
Content: The consequences of the chemical instability of DNA (mentioned in the first two lectures) will be described in relation to the generation of mutations. How cells try and minimise the chances of alterations in the genetic code occurring will be described. No biological process is 100% accurate and some of these changes to the DNA sequence arise as a result of errors made by DNA polymerases. How these errors are corrected by mismatch repair will be discussed. Other changes to the genetic code arising from non-enzymatic transformations (DNA damage) must also be minimised and the many repair options that exist to deal with the different types of DNA damage will be described. The effects of DNA damage on DNA replication and genome evolution will also be introduced.

Lectures 6 and 7
Title: DNA technologies
Lecturer: Dr John Barrow
Content: The late 20th century explosion in molecular biological techniques has facilitated major leaps in our understanding of fundamental biological processes. It has also allowed the genetic engineering of organisms. This lecture will describe the basic techniques of DNA cloning. It will also describe how the chemical properties of DNA can be exploited to analyse recombinant DNA molecules by hybridisation, by DNA sequencing, and by the polymerase chain reaction. The use of DNA microarrays to analyse not just single genes but whole genomes will also be discussed.
Lecture 8
Title: Genome alterations – genetically modified organisms
Lecturer: Dr John Barrow

Content: The technologies of molecular biology discussed in lectures six and seven can be exploited to alter the genomes of a wide range of organisms. This lecture will describe how recombination can be harnessed to introduce stretches of engineered DNA into the genomes of bacteria, plants and animals.

Module 2; Gene expression and regulation

One of the most important questions within modern biology centres on how one-dimensional information held within the DNA is turned into healthy living 3-dimensional organisms that are able to interact with their environments. This module will describe how this information is decoded by transcription and translation to form proteins and how organisms control these processes to ensure that the correct proteins are produced in the correct cells at the correct times and in the correct amounts. Textbook references, Lehninger Principles of Biochemistry (4th ed), Chapter 26, 27 and 28. This module will be followed by a one hour on-line assessment available on MyAberdeen covering the contents of modules 1 and 2.

Lecture 1
Title: Transcription in prokaryotes
Lecturer: Dr Alasdair MacKenzie

Content: The first lecture in this series will deal with basal transcription in prokaryotes and will describe how the RNA polymerase enzyme "transcribes" the genetic information in DNA to the messenger RNA that will encode a protein. We will also describe how rates of prokaryotic transcription are controlled using specific examples of metabolic enzymes whose activity is controlled at the level of transcription (e.g. β-galactosidase).

Key words: Transcription, mRNA, tRNA, rRNA, DNA-directed RNA polymerase, σ-factor, Template strand, termination, Operator, Operon, lac operon

Lecture 2
Title: Transcription in Eukaryotes
Lecturer: Dr Alasdair MacKenzie

Content: This lecture will describe the major differences between prokaryotic and eukaryotic transcription. Although the basic principles are the same, eukaryotic transcription is a much more complex process than prokaryotic transcription and allows for the greater degrees of control required to produce complex multicellular organisms. This lecture will introduce basic concepts in eukaryotic transcription such as the assembly of the basal transcriptional machinery to form the preinitiation complex (PIC).
**Key words:** RNA polymerase II, TATA binding protein, Pre-initiation complex (PIC), Initiator sequence, promoter sequence, Transcription factors. Initiation complex, elongation, termination

**Lectures** 3 and 4  
**Title:** Gene regulation in Eukaryotes  
**Lecturer:** Dr Alasdair MacKenzie  
**Content:** In order for complex processes such as embryonic development and human health to occur the activity of a large number of genes within the genome must be coordinated with an enormous degree of precision. This precision is controlled by a class of DNA binding proteins called DNA-binding transactivators that, when activated by the cellular machinery, displace histones to bind DNA sequences called enhancers to form enhancesome complexes. Enhancesome complexes may form at some distance from the gene being activated and it is now known that they interact with the pre-initiation complex through long distance looping of intervening DNA. This lecture will introduce these processes many of which have only recently been discovered.  

**Key Words:** Enhancer, promoter, DNA-binding transactivators, Histones, co-activators, TFIID.

**Lecture 5**  
**Title:** RNA processing  
**Lecturer:** Dr Alasdair MacKenzie  
**Content:** This lecture will introduce the concepts of post transcriptional processing of eukaryotic mRNA and will describe how the primary transcript is spliced, "capped" and "tailed" before being transported from the nucleus to the cytoplasm to take part in the next stage of gene expression; translation.  

**Key words:** Intron, Exon, 5’ cap, Poly(A) tail, Splicing, alternative splicing

**Lecture 6**  
**Title:** The genetic code  
**Lecturer:** Dr Alasdair MacKenzie  
**Content:** This set of lectures deals with the way in which genetic information carried in the structure of DNA is translated into the structure of proteins encoded by DNA. Three bases read in sequence along a DNA template form a ‘triplet’ code-word. Sequences of triplets are transcribed into sequences of complementary three-base ‘codons’ in messenger RNA (mRNA). Codon sequences are translated into sequences of amino-acid residues in polypeptides. Each of the twenty coded amino-acids has at least one codon. The genetic code (that is, which 3-base code-words specify which amino-acids) was solved using synthetic RNA molecules as mRNAs in a test-tube protein-synthesising system. The code turns out to be degenerate (several amino-acids are specified by more than one code-word), non-overlapping (individual code-words are discrete), comma-less (individual code-words are not separated by non-coding bases) and near-universal (with few exceptions, the same code-words are assigned to the same amino-acids throughout nature). Code-words that are similar to one another tend to be assigned to the same amino-acid or to amino-acids with similar structures. This non-random assignment and degeneracy in general minimise deleterious effects of mutation.
Key Words: Translation, Messenger RNA (mRNA), Central Dogma of Molecular Biology, Genetic Code, Triplet, Codon, Degeneracy, Non-overlapping, comma-less, near-universal nature of code, Frame-shift mutation, Point (single-base) mutation, Silent mutation, Conservative mutation, Open reading frame, Overlapping genes

Lecture 7
Title: Transcription
Lecturer: Dr Alasdair MacKenzie
Content: The genetic code contained within the mRNA must be “translated” into proteins. This lecture will describe the fascinating choreography of the ribosome subunits, tRNA, mRNA and Amino-acyl tRNA synthetases required for the production of a complete polypeptide.

Key Words: Transfer RNA (tRNA), Adaptor function, Clover-leaf structure, Anticodon, Amino-acyl tRNA synthetase, Wobble hypothesis, Proof-reading during translation.

Lecture 8
Title: Post transcriptional modification and degradation.
Lecturer: Dr Alasdair MacKenzie
Content: Once translated many proteins in eukaryotes need to be further altered by a number of processes that include proteolytic cleavage, the formation of disulfide bonds, glycosylation and phosphorylation. Collectively these processes are called post-translational modifications and they are essential for the normal functioning of most human proteins. In eukaryotes any of these processes occur in a part of the cell called the endoplasmic reticulum (ER). Once translated and modified proteins are then sorted and distributed by another cellular structure called the Golgi apparatus. Another critical aspect of protein metabolism is protein degradation where proteins are “tagged” for destruction. This avoids the build up of excessive un-needed protein in the cell.

Key words: Post-translational modification, disulfide bonds, glycosylation, phosphorylation endoplasmic reticulum, Golgi apparatus, protein degradation.

Module 3; Proteins

This module will provide a comprehensive introduction to protein biochemistry, building on the basic chemistry of amino acids and peptides. The properties of proteins will be described, using a number of specific examples. The final lectures in the module will consider the methods used to study proteins. These provide the information that underlies our current understanding of protein structure and function.

Lecture 1
Title: Amino acid biochemistry; amino acids as buffers; amino acid diversity
Lecturer: Prof Iain McEwan
Content: Proteins fulfil a diversity of functions, for example as enzymes, as structural elements of cells and tissues, as carriers of gases and nutrients, as contractile elements in muscle, as antibodies, and as
hormones. All this diversity comes from relatively simple building-blocks, L-amino-acid residues. Amino-acids act as zwitterions, and may therefore be used as buffers for biological studies. Buffering ability is an important property of proteins, the charge of which alters as the pH changes. The pH at which a particular protein has no net charge is called its isoelectric point.

**Keywords:** Amino acid, buffering

**Lecture 2**  
**Title:** Protein structure  
**Lecturer:** Prof Iain McEwan  
**Content:** Four levels of structure in a protein molecule may be distinguished. The primary structure is the sequence of amino-acid residues, which is always written with the N-terminus on the left and the C-terminus on the right. The terms secondary and tertiary structure describe features of the three-dimensional folding of the polypeptide chain; they determine the final shape of the molecule and the juxtaposition of individual amino-acid residues within the folded structure. Secondary structural features such as the α-helix and the β-sheet occur in varying proportions in different proteins. Tertiary structure relies on a number of different types of force, including hydrogen bonds, ionic bonds, hydrophobic interactions and disulphide bonds. Quaternary structure describes the aggregation of several polypeptide chains, with specific interactions between the polypeptide sub-units (also called monomers); the sub-units are held together mainly by hydrophobic interactions.

**Keywords:** three-dimensional structure, α-helix, β-sheet

**Lectures 3 and 4**  
**Title:** Globular proteins (1 and 2)  
**Lecturer:** Prof Iain McEwan  
**Content:** Different types of protein structure are required for different functions. All proteins fall into two broad classes: globular and fibrous proteins. Globular proteins include insulin, which is important in glucose homeostasis, and immunoglobulins, which are one of the body’s responses to infection. Other globular proteins include myoglobin, which acts as an oxygen carrier and contains a haem prosthetic group. Haemoglobin is a member of the same family, but is more complex in its structure. It contains four subunits, held together by hydrophobic forces. It shows co-operative binding of oxygen and allosteric regulation by carbon dioxide and protons. The mode of action of transcription factor proteins is reliant on their modular structure. We will examine how transcription factors can generally be divided into 2 components; a DNA binding domain and an RNA polymerase activation domain. This lecture will describe the different DNA binding domains or "motifs" and how these allow DNA binding. In addition, the modes of action of various activation domains will be described.

**Keywords:** globular protein, insulin, immunoglobulin, myoglobin, haemoglobin, transcription factors

**Lecture 5**  
**Title:** Fibrous proteins – keratin, elastin and collagen  
**Lecturer:** Prof Iain McEwan
In contrast to most enzymes, circulating and intracellular proteins, which are globular, fibrous proteins have structural roles. An example is keratin, which is made up of $\alpha$-helices. Collagens contain an unusual triple helix that is quite distinct from the $\alpha$-helix. These helices form only when there are repeat structures in the polymer, in which glycine occurs at every third monomer position. Collagen is also rich in proline and lysine residues, both of which may be hydroxylated; this is an example of a post-translational modification. Elastin achieves the necessary flexibility by means of unique cross-links between lysine residues.

**Keywords:** fibrous protein, $\alpha$-keratin, collagen, elastin

**Lecture 6**
**Title:** Membrane protein, transmembrane proteins
**Lecturer:** Prof Iain McEwan
**Content:** Membranes represent a barrier but also contain important activities, reflecting their protein components. The structures of important membrane proteins will be explained, with emphasis on how an $\alpha$-helix can form the transmembrane part of a protein. Proteins are a major component of cells and are present in all cellular compartments.

**Key words:** Membrane, phospholipid, GPI-anchor, hydrophobic, hydrophilic, cell-cell interactions, membrane fusion, membrane transport, receptors

**Lecture 7**
**Title:** How we study proteins (1)
**Lecturer:** Prof Iain McEwan
**Content:** Proteins are analysed by techniques like electrophoresis, which can give information on size and charge. The important technique of SDS-PAGE will be described in detail, as will the analysis of data to allow us to estimate the molecular mass of proteins and their component chains.

**Keywords:** Electrophoresis, SDS-PAGE

**Lecture 8**
**Title:** How we study proteins (2)
**Lecturer:** Prof Iain McEwan
**Content:** Proteins may be identified by determining their amino-acid composition and, especially, their N-terminal sequence. Many analyses of proteins require them to be cut into smaller pieces by specific proteases.

**Keywords:** amino-acid composition, amino-acid sequence, proteases

**Lecture 9**
**Title:** How we study proteins (3)
**Lecturer:** Prof Iain McEwan
**Content:** The specificity of antibodies is used to provide fast and sensitive assays in many different applications. Among these are enzyme-linked immunosorbent assays (ELISA) and immunoblotting (or western blotting).
Module 4; Genetic disease

This module will explain how genomes can be compromised by mutation and chromosomal rearrangements leading to disorders such as Downs syndrome, cystic fibrosis, fragile-X syndrome and cancer. Recommended text *Human Molecular Genetics* by Strachan and Read (3rd edition) and Emery’s Elements of medical genetics (any edition). This module will be followed by a one hour on-line assessment available on MyAberdeen covering the contents of modules 3 and 4.

Lecture 1

**Title:** Genetic disease

**Lecturer:** Prof Martin Collinson

**Content:** This first lecture will revise and extend 1st year lectures on genetic inheritance. The autosomal recessive condition, cystic fibrosis, is carried by around 1 in 25 of the Caucasian population. The gene for the condition, the CFTR gene, produces a protein which forms a chloride iron channel. Mutations within the gene can have different effects on the production of the protein and on its function within the cell. The spectrum of different mutations that give rise to the disease will be described.

**Keywords:** Mendelian, inheritance, Cystic fibrosis, autosomal, recessive, mutation, ion channel

Lecture 2

**Title:** Cancer and autosomal dominant inheritance

**Lecturer:** Prof Martin Collinson

**Content:** Cancer affects 1 in 3 individuals in their lifetime but only a very small percentage of them are associated with the genetic predisposition. Most cancer is multifactorial but there are several types that show autosomal dominant inheritance. In this lecture we will consider one of these, colorectal cancer, which can be associated with several genetic conditions for which the genes and their mutations have been identified. As a result of this individuals found to be predisposed may be offered appropriate screening to decrease their risk.

**Keywords:** Cancer, autosomal, dominant, multifactorial

Lecture 3

**Title:** Genome rearrangements and disease

**Keywords:** antibodies, enzyme-linked immunosorbent assay (ELISA), immunoblotting.
Lecturer: Prof Martin Collinson

Content: Triplet repeat diseases – Huntington’s disease, fragile X mental retardation, myotonic dystrophy and others – are caused by expansions of unstable trinucleotide sequences such as (CTG)n. Mechanisms of DNA instability and triplet repeat expansion will be described. We will also look at chromosomal deletions and translocations, their effects on gene expression, and their importance in genetic disease and cancer. X-linked and autosomal dominant patterns of inheritance will be revised.

Keywords: Huntington, myotonic dystrophy, fragile X, trinucleotide repeat, DNA instability, deletion, translocation, cancer

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Lecture 4

Title: Genetic diagnosis and gene therapy

Lecturer: Prof Martin Collinson

Content: The implications of genetics research for prenatal diagnosis, predictive testing and surveillance will be discussed and put in context. Methods for diagnosis based on DNA testing of samples obtained by amniocentesis and chorionic villus sampling will be described. The role of the genetic counsellor and other professionals, and the ethical and practical issues that arise, will be discussed. Recent advances in gene therapy will also be covered. Mitochondrial and Y-linked inheritance will be mentioned.

Keywords: Diagnosis, prenatal, gene therapy, genetic counselling

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Lecture 5 Genes, Nutrition, and the Environment

Content: The action of genes can be modified by pharmacological and nutritional factors. This lecture explores why piglets born to vitamin A-deficient mothers have no eyes, and goes on to explain the role of retinoids derived from the diet in embryonic development, through a direct control of gene expression.

Keywords: Vitamin A, retinoic acid, control of gene expression.

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Lecture 6

Title: The Human genome and genome projects

Lecturer: Prof Martin Collinson

Content: Humans and other species, such as experimental animals and agriculturally important animals and plants, are having their genomes sequenced. This lecture gives an overview of technology – the factory-scale application of molecular genetics with computers and automation; the basic dideoxy sequencing method and its automation; top-down and bottom-up strategies, and contigs. We
will look at web-based databases of genome project information, and how these can help scientists quickly find genome information. The power of sequencing technology has opened up ancient genomes, and we will explore how hybridisation with Neanderthals affected modern humans.

**Keywords:** Genome, human, homologue, DNA sequence, contig

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**Practical Lab Work**

See BI20M3 practical manuals which are available online. It is strongly advised that students take time to familiarise themselves with the practical content prior to the start of class.

Practical reports are to be completed by the end of the practical and retained by the student to act as a reference to answering a series of on-line questions that will be available for two weeks after the final practical. You will be given one hour to complete 20-30 questions relating to the practical. These assessments will comprise 40% of your final mark.

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**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 2021/22 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 2021-22 includes the following:

- Absence
- Appeals & Complaints
- Avoiding Plagiarism
- Assessment
- Email Use
- Feedback
- Graduate Attributes
- Late Submission of Work
- MyAberdeen
- Professional and Academic Development
- Student Learning Service (SLS)
- Student Monitoring/Class Certificates
- Student Discipline
- The Co-curriculum
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| A1    | 22          | 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          |                |               |                                                                                                       |
| A3    | 20          |                |               |                                                                                                       |
| A4    | 19          |                |               |                                                                                                       |
| A5    | 18          |                |               |                                                                                                       |
| B1    | 17          | 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          |                |               |                                                                                                       |
| B3    | 15          |                |               |                                                                                                       |
| C1    | 14          | 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          |                |               |                                                                                                       |
| C3    | 12          |                |               |                                                                                                       |
| D1    | 11          | 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          |                |               |                                                                                                       |
| D3    | 9           |                |               |                                                                                                       |
| E1    | 8           | 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           |                |               |                                                                                                       |
| E3    | 6           |                |               |                                                                                                       |
| F1    | 5           | Clear Fail     | Not used for Honours | • Contains major errors or misconceptions  
• Poor presentation |
| F2    | 4           |                |               |                                                                                                       |
| F3    | 3           |                |               |                                                                                                       |
| G1    | 2           | Clear Fail/Abysmal | -              | • Token or no submission                                                                 |
| G2    | 1           |                |               |                                                                                                       |
| G3    | 0           |                |               |                                                                                                       |
Course Timetable BI20M3: 2021-2022

All lecture recordings will be available at the start of the course on MyAberdeen. Lecture dates provided below are recommended to allow time to study for on-line tests. Students are strongly encouraged to watch each lecture and to become familiar with the lecture material prior to on-line tests. Synchronised learning sessions on Collaborate (on-line face-to-face) sessions are highlighted in yellow.

<table>
<thead>
<tr>
<th>Date</th>
<th>Time</th>
<th>Place</th>
<th>Subject</th>
<th>Online tests</th>
<th>Session</th>
<th>Staff</th>
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<tbody>
<tr>
<td><strong>Week 9</strong></td>
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<tr>
<td>Mon 27 Sep</td>
<td>12:00</td>
<td>MyAbdn</td>
<td>Introduction to the course on Collaborate</td>
<td>Lecture</td>
<td>AM</td>
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<td>13:00</td>
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<tr>
<td></td>
<td>Own time</td>
<td>MyAbdn</td>
<td>1. Chemistry and Structure of DNA and RNA 1.</td>
<td>Lecture</td>
<td>JB</td>
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<tr>
<td>Fri 1 Oct</td>
<td>Own time</td>
<td>MyAbdn</td>
<td>2. Chemistry and Structure of DNA and RNA 2.</td>
<td>Lecture</td>
<td>JB</td>
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</tbody>
</table>

| **Week 10**|        |         |                                                  |                                                  |         |       |
| Mon 4 Oct  | Own time| MyAbdn  | 3. Chromosome Structure                          | Lecture                                          | JB      |       |
|            | 12:00   | Collaborate |Tutorial                                        | Tutorial                                         | JB +    | AM    |
|            | 13:00   |         |                                                  |                                                  |         |       |
|            | Own time| MyAbdn  | 4. DNA replication   | Lecture                                          | JB      |       |
|            |         |         |                                                  |                                                  |         |       |
| Fri 8 Oct  | Own time| MyAbdn  | DNA technologies 1                                 | Lecture                                          | JB      |       |

| **Week 11**|        |         |                                                  |                                                  |         |       |
| Mon 11 Oct | Own time| MyAbdn  | DNA technologies 2                                 | Lecture                                          | JB      |       |
|            | 12:00   | Collaborate |Tutorial                                        | Tutorial                                         | JB+AM   |       |
|            | 13:00   |         |                                                  |                                                  |         |       |
|            | Own time| MyAbdn  | Genetically modified organisms                    | Lecture                                          | JB      |       |
|            |         |         |                                                  |                                                  |         |       |
| Fri 15 Oct | Own time| MyAbdn  | Transcription 1                                   | Lecture                                          | AM      |       |

| **Week 12**|        |         |                                                  |                                                  |         |       |
| Mon 18 Oct | Own time| MyAbdn  | Transcription 2                                   | Lecture                                          | AM      |       |
|            | 12:00   | Collaborate |Tutorial                                        | Tutorial                                         | JB+AM   |       |
|            | 13:00   |         |                                                  |                                                  |         |       |
|            | Own time| MyAbdn  | Gene Regulation 1                                 | Lecture                                          | AM      |       |
|            |         |         |                                                  |                                                  |         |       |
| Fri 21 Oct | Own time| MyAbdn  | Gene Regulation 2                                 | Lecture                                          | AM      |       |

| **Week 13**|        |         |                                                  |                                                  |         |       |
| Mon 25 Oct | Own time| MyAbdn  | RNA processing and the genetic code              | Lecture                                          | AM      |       |
|            | 12:00   | Collaborate |Tutorial                                        | Tutorial                                         | JB+AM   |       |
|            | 13:00   |         |                                                  |                                                  |         |       |
|            | Own time| MyAbdn  | Translation                                      | Lecture                                          | AM      |       |
|            |         |         |                                                  |                                                  |         |       |
| Fri 29 Oct | Own time| MyAbdn  | Translation and the ribosome                     | Lecture                                          | AM      |       |

| **Week 14**|        |         |                                                  |                                                  |         |       |
| Mon 1 Nov  | Own time| MyAbdn  | Post translational processing                    | Lecture                                          | AM      |       |
|            | 12:00   | Collaborate |Tutorial                                        | Tutorial                                         | JB+AM   |       |
|            | 13:00   |         |                                                  |                                                  |         |       |
|            | Own time| MyAbdn  | Transcription 3                                  | Lecture                                          | AM      |       |

<p>| <strong>Week 15</strong>|        |         |                                                  |                                                  |         |       |
| Mon 8 Nov  | Own time| MyAbdn  | Protein Structure 1                              | Lecture                                          | IM      |       |
|            | 12:00   | Collaborate |Tutorial (tips on completing DNA Practical)     | Tutorial                                         | IM+AM   |       |</p>
<table>
<thead>
<tr>
<th>Date</th>
<th>Time</th>
<th>Location</th>
<th>Activity</th>
<th>Staff</th>
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<tbody>
<tr>
<td>Fri 12 Nov</td>
<td>Own time</td>
<td>MyAbdn</td>
<td>Protein Structure 2</td>
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<td>Mon 15 Nov</td>
<td>Own time</td>
<td>MyAbdn</td>
<td>4. Globular Proteins (2)</td>
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<td>Tutorial IM+AM</td>
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<td>Lecture IM</td>
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<td>Wed 16 Nov</td>
<td>A 09:00-11:00</td>
<td>Collaborate</td>
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<td>B 12:00-14:00</td>
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<td>C 15:00-17:00</td>
<td>Zoology Basement Labs</td>
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<tr>
<td>Fri 19 Nov</td>
<td>D 09:00-11:00</td>
<td>E 12:00-14:00</td>
<td>F 15:00-17:00</td>
<td>Protein Practical (ELISA)</td>
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<td>Fri 19 Nov</td>
<td>Own time</td>
<td>MyAbdn</td>
<td>6. Transmembrane Proteins</td>
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<td>Mon 22 Nov</td>
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<td>MyAbdn</td>
<td>7. Studying proteins 1</td>
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<td>Tutorial IM +AM</td>
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<td>Fri 26 Nov</td>
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<td>9. Studying proteins 3</td>
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<td>Mon 29 Nov</td>
<td>Own time</td>
<td>MyAbdn</td>
<td>1. Genetic Disease and autosomal recessive inheritance</td>
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<td>3. Genome rearrangements and disease</td>
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<td>MyAbdn</td>
<td>4. Genetic Diagnosis and Gene Therapy</td>
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<td>Lecture MC</td>
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**Weeks 16-19**

**Staff**

- Alasdair MacKenzie (Course co-ordinator)
- Martin Collinson
- Iain McEwan
- John Barrow
- Alexander Lorenz