UNIVERSITY of BRADFORD

Module Descriptor

Medical Genetics

Module Code:	BIS6011-B
Academic Year:	2018-19
Credit Rating:	20
School:	School of Chemistry and Biosciences
Subject Area:	Biomedical Science
FHEQ Level:	FHEQ Level 6
Module Leader:	Dr Mark Sutherland

Additional Tutors:
Dr Steven Picksley, Dr Michael Fessing, Dr Krzysztof Poterlowicz

Pre-requisites:	Molecular Genetics 2017-18
Co-requisites:	

Contact Hours

Type	Hours
Lectures	24
Tutorials	9
Directed Study	167

Availability Periods

Occurrence	Location/Period
BDA	University of Bradford / Semester 1 (Sep - Jan)

Module Aims

Develop a comprehensive understanding and detailed knowledge of how variation in the human genome contributes to disease. Deepen students' knowledge of molecular biology of the gene, linking errors in the molecular mechanisms responsible for gene expression to specific diseases. Inform student's how genetic variability can either directly lead to specific diseases, such as cystic fibrosis, or predispose individuals to complex disorders, including diabetes, heart disease and cancer.

Make students aware of how genetic diseases are inherited and how inheritance is predicted. Promote an understanding of how advances in genetics are driving forward research into novel treatments for many of the diseases described throughout the module.

Outline Syllabus

Human genome structure, next generation sequencing, ENCODE project, bioinformatics, epigenetics, gene expression, mRNA processing, non-coding RNA and disease. The role of genetic variability either as a direct cause of, or a predisposing factor to human diseases. Single gene disorders, including case studies of Cystic Fibrosis and Huntington's disease. Multifactorial disease, genetic linkage, case studies including Type I diabetes. Overview of cancer genetics, hereditary cancers and a case study on p53 mutations. Gene therapy, both existing and emerging, novel treatments for genetic disease. Stem cell technology and mutation detection techniques.

Module Learning Outcomes

On successful completion of this module, students will be able to...

1 Discuss the detailed structure of the eukaryotic (human) genome (HCPC standard 13).

2 Review how the expression of eukaryotic genes is regulated at the level of transcription and post-transcriptionally (HCPC standard 13)

3 Critically appraise the effects of genes and genetic factors (including epigenetics) in health and disease; discuss in detail, specific examples of human disease with genetic aetiologies (HCPC standards 2, 13)

4 Evaluate the methods used to investigate genes (their function and their expression), genetic mutations and polymorphisms (HCPC standards 2, 13).

5 Observe and interpret clinical and laboratory data relating to genetic studies, engage with problem solving aspects of mutation detection and determining the relative risk of genetic inheritance via pedigree analysis (HPC standards 8, 14)

6 Demonstrate personal responsibility for self-directed learning and time management (HCPC standards 1, 3).

7 Reflect and review your practice (HCPC standard 11/12).

Learning, Teaching and Assessment Strategy

The curriculum to develop the knowledge and understanding required for this module is delivered in lectures. The core knowledge for this module is supplemented by reference to current published scientific literature which requires extensive further reading and autonomous learning by the students. Development of knowledge and understanding and other skills, such as problem solving, is also achieved using workshop sessions. In the workshops you will work individually to research information, interpret data, solve problems and develop your understanding of medical genetics. The workshop exercises will require you to work under pressure and are first formatively and then summatively assessed. During

directed study hours, students are expected to undertake reading to consolidate and expand on the content of formal taught sessions; research and prepare for assessments; revise material from formal taught sessions; and undertake specific elements of reading as directed.
Reassessment of failed elements will be as per the initial method of assessment.

Mode of Assessment

Type	Method	Description	Length	Weighting	Final Assess'
Summative	Examination - closed book	One 2-hour examination comprising two from a choice of five essays (LO 1-6)	2 hours	60%	Yes
Summative	Coursework	Data interpretation and problem-solving and knowledge-based assessment (LO 1-7)	1 hour	20%	No
Summative	Coursework	Written report (LO 1-7)	0-1000 words	20%	No

Legacy Code (if applicable)

BM-3123D

Reading List

To view Reading List, please go to **rebus:list**.