Liverpool John Moores University

Title:	Medical Genetics
Status:	Definitive
Code:	**7102BSBMOL** (126683)
Version Start Date:	01-08-2021
Owning School/Faculty:	Pharmacy & Biomolecular Sciences
Teaching School/Faculty:	Pharmacy & Biomolecular Sciences

Team	Leader
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Academic Level:	FHEQ7	**Credit Value:**	20	**Total Delivered Hours:**	40
Total Learning Hours:	200	**Private Study:**	160		

Delivery Options
Course typically offered: Semester 1

Component	Contact Hours
Lecture	20
Practical	10
Tutorial	2
Workshop	6

Grading Basis: 50 %

Assessment Details

Category	Short Description	Description	Weighting (%)	Exam Duration
Exam	exam	The exam is a combination of problem solving and factual recall	50	2
Report	report	This is the final report based on the linked practical sessions within the module. Typical word length 2500.	50	

Aims

To provide an overview of the roles and investigations of genes and disease.

Learning Outcomes

After completing the module the student should be able to:

1	Demonstrate a reflective understanding of the genetic basis of disease and its investigation.
2	Apply principles of interpretation to genetic data.
3	Critically evaluate the literature on genetic disease and research techniques.

Learning Outcomes of Assessments

The assessment item list is assessed via the learning outcomes listed:

Examination	1	2	3
Lab report	1	3	

Outline Syllabus

Lectures:
The module provides a critical appreciation of the human genome, its regulation, functional significance of gene mutations and current approaches of identification of human genetic disorders. Topics covered include:
The human genome, epigenetics, genomic medicine, high throughput genomic techniques, bioinformatics, inborn errors of metabolism, molecular medicine, stem cells, molecular basis of host-pathogen interaction, immunogenetics and disease.

Workshops:
Gene mapping: pedigree analysis and gene mapping using recombination frequencies.

Practical:
Analysis of single nucleotide polymorphisms (SNPs) by qPCR.
The practical sessions will involve the use of recombinant DNA technologies (restriction endonucleases, PCR, and electrophoretic DNA analyses) to identify and characterise selected SNPs within the human genome. Students will be expected to research the literature and design experimental regimes based on information relating to the methodology. Some method development will be required, and the students will be expected to have carried out DNA sequence analyses and literature search on the SNPs and DNA sequence before starting the practical work.

Tutorials
Two pastoral tutorials will be provided.

Learning Activities

Lectures, practicals, workshops, tutorials and student-centred activities.

Notes

This module aims to develop understanding of the contribution of chromosomes and genes to health and disease. A range of disorders will be covered with an introduction to relevant techniques and therapeutic approaches.