

Liverpool John Moores University

Title: MEDICAL AND CLINICAL GENETICS A
Status: Definitive
Code: **6008BMBMOL** (101496)
Version Start Date: 01-08-2011

Owning School/Faculty: Pharmacy & Biomolecular Sciences
Teaching School/Faculty: Pharmacy & Biomolecular Sciences

Team	Leader
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Academic Level: FHEQ6 **Credit Value:** 12.00 **Total Delivered Hours:** 26.00

Total Learning Hours: 120 **Private Study:** 94

Delivery Options

Course typically offered: Semester 1

Component	Contact Hours
Lecture	16.000
Online	1.000
Tutorial	3.000
Workshop	4.000

Grading Basis: 40 %

Assessment Details

Category	Short Description	Description	Weighting (%)	Exam Duration
Test	AS1	Coursework: In class analysis of a published paper	40.0	
Exam	AS2	Exam	60.0	2.00

Aims

To develop understanding of the contribution of chromosomes and genes to health and disease

Learning Outcomes

After completing the module the student should be able to:

- 1 discuss the genetic underpinning of health and the role of genetic factors in disease
- 2 discuss the link between mutation and phenotype in a range of named single gene defects and chromosomal abnormalities
- 3 discuss approaches to therapy involving genes and gene products
- 4 locate and use sources of genetic information

Learning Outcomes of Assessments

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

CW	1	2	3	4
EXAM	1	2	3	4

Outline Syllabus

Overview of the roles of genes and chromosomes in disease
Chromosome rearrangements and their effects, associated disorders. Non-disjunction, changes in ploidy, associated disorders
Biochemical and physiological effects of mutations within genes. Examples may include haemoglobinopathies, thalassaemias, disorders of carbohydrate metabolism, haemophilia, cystic fibrosis, Duchenne muscular dystrophy, Huntington's disease, Fragile X
Recombinant proteins, approaches to gene therapy
Introduction to diagnostic techniques.
Introduction to bioinformatics.

Learning Activities

Lectures, On line workshops, paper analysis, tutorials

References

Course Material	Book
Author	Turnpenney, P and Ellard, S
Publishing Year	2007
Title	Emery's Elements of medical genetics
Subtitle	
Edition	13th
Publisher	Saunders

ISBN	9780702029172
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Course Material	Book
Author	Russell, PJ
Publishing Year	2006
Title	iGenetics
Subtitle	A Mendelian Approach
Edition	
Publisher	Pearson Benjamin Cummings
ISBN	080534666X

Course Material	Book
Author	Strachan, T and Read, AP
Publishing Year	2004
Title	Human Molecular genetics
Subtitle	
Edition	3rd edition
Publisher	Garland Science
ISBN	0815341849

Course Material	Book
Author	Trends in Genetics
Publishing Year	0
Title	
Subtitle	
Edition	
Publisher	
ISBN	

Course Material	Book
Author	Trends in Molecular Medicine
Publishing Year	0
Title	
Subtitle	
Edition	
Publisher	
ISBN	

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. Some of these concepts will be further developed in BMLBM3043