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Title: Medical Genetics
Status: Definitive
Code: **6014BMBMOL** (117400)
Version Start Date: 01-08-2018

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

Team	Leader
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Jari Louhelainen	
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Academic Level: FHEQ6 **Credit Value:** 12 **Total Delivered Hours:** 24
Total Learning Hours: 120 **Private Study:** 96

Delivery Options

Course typically offered: Semester 1

Component	Contact Hours
Lecture	14
Workshop	10

Grading Basis: 40 %

Assessment Details

Category	Short Description	Description	Weighting (%)	Exam Duration
Portfolio	Wksh reps	workshop reports	50	
Reflection	AB	Annotated bibliography	50	

Aims

To provide an overview of the roles and investigation of genes in disease

Learning Outcomes

After completing the module the student should be able to:

- 1 demonstrate a reflective understanding of the basis of genetic disease and its investigation
- 2 apply principles of interpretation of genetic data
- 3 critically evaluate literature on genetic topics

Learning Outcomes of Assessments

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

Workshop reports	1	2
Annotated bibliography	3	

Outline Syllabus

Gain and loss of function mutations, single gene defects, multifactorial conditions, epigenetics, familial and sporadic disease. Typical examples: sickle cell disease, cystic fibrosis, Duchenne Muscular dystrophy, haemophilia, Fragile X, Huntington's disease, Alzheimer's disease, Parkinson's disease.

Developmental genetics and abnormalities.

Phenotype to genotype and genotype to phenotype. human genome project.

Bioinformatics.

Overview of diagnostic and research techniques including PCR, multiplex assays, blotting techniques, microarrays, FISH.

Approaches to gene therapy

Learning Activities

Lectures, workshops including on line activities, paper analysis,

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.