

## Module Specification

Module Title  Module Code   
Credit Value  Level  Mode of Delivery  Semester

Pre-requisite modules	Co-requisite modules	Overlapping modules
Molecular Genetics (BIO163)	None	None
Chromosomes & gene functions (BMD111)	None	None

### 1) Content Description

Provide a description of the module, as it will appear in the Module Directory and on the Student Information System (approx. 70-80 words).

This module explores human hereditary disease in terms of genetics, pathogenesis, clinical features and clinical management. We will look at key examples of chromosomal abnormalities (e.g. Trisomy 21), monogenic disease (e.g. cystic fibrosis, Huntington's disease and Duchenne Muscular Dystrophy) and common disease (e.g. cancer). Students will be introduced to methods and techniques for identifying genetic loci associated with disease (e.g. homozygosity mapping, genome-wide association studies, DNA sequencing). Finally, we will discuss issues around genetic screening, testing and counselling.

### 2) Module Aims

Specify the aims of the module, i.e. the broad educational purposes for offering this module.

The aims of this module are to:

- Develop broad knowledge and understanding of human medical genetics.
- Describe the genetics, clinical features and clinical management of key examples of inherited disease.
- Discuss approaches to identifying genetic loci associated with disease and their relevance to specific types of genetic disease.
- Understand patterns of inheritance and relate this to scenarios in genetic testing, screening and counselling.

### 3) Learning Outcomes

Identify the learning outcomes for this module, i.e. knowledge, skills and attributes to be developed through completion of this module. Outcomes should be referenced to the relevant [QAA benchmark statements](#) and the [Framework for Higher Education Qualifications in England, Wales and Northern Ireland \(2008\)](#). The [SEEC Credit Level Descriptors for Further and Higher Education 2003](#) and [Queen Mary Statement of Graduate Attributes](#) should also be used as a guiding framework for curriculum design.

Academic Content:	
A1	Basic principles of medical genetics
A2	Key examples of inherited disease
A3	identification of genetic loci associated with inherited disease
A4	Issues in genetic testing, screening and counselling
Disciplinary Skills - able to:	
B1	Apply medical genetics knowledge and principles in combination with problem-solving skills in a range of theoretical situations
B2	Reason critically
B3	Plan, undertake and report a bibliographically-based piece of research
B4	Evaluate scientific literature and synthesise a coherent precis of research within medical genetics
Attributes:	
C1	Communicate effectively via spoken and written English
C2	Acquire new learning in a range of ways, both individually and collaboratively
C3	Develop respect for the opinions of others
C4	grasp the principles and practices of the field of medical genetics
C5	Explain and argue clearly and concisely

#### 4) Reading List

Provide an indicative reading list for the module. This should include key texts and/or journals but **should not** be an exhaustive list of materials.

The primary textbooks for the module will be "Emery's Elements of Medical Genetics" (15th Edition) by Turnpenny & Ellard and "Genetics & Genomics in medicine" by Strachan, Goodship & Chinnery. In addition, students will be pointed towards key review papers relevant to individual topics.

## 5) Teaching and Learning Profile

Provide details of the method of delivery (lectures, seminars, fieldwork, practical classes, etc.) used to enable the achievement of learning outcomes and an indicative number of hours for each activity to give an overall picture of the workload a student taking the module would be expected to undertake. This information will form the Key Information Set for each undergraduate programme and will be used to populate the KIS widget found on the QMUL programme information pages. More information can be found [online](#) about KIS. You may also wish to refer to the [QAA guidance on contact hours](#) when completing this section.

Activity Type	KIS Category	Time Spent (in hours)
Lecture	Scheduled	22
Practical Classes and workshops	Scheduled	8
Total		30

Specify the total module notional study hours. This should be a total of the hours given for each activity. The notional study hours for each academic credit point is 10. A 15 credit point module therefore represents 150 notional study hours.

Activity Type	Total Time Spent (in hours)	Percentage of Time Spent
Scheduled learning and teaching	30	20
Placement	0	0
Independent Study	120	80
Total	150	100

Use the information provided in the box above to specify the total time spent and the percentage time spent in each category of teaching and learning activity.

## 6) Assessment Profile

Provide details of the assessment methods used to assess the achievement of learning outcomes.

Description of Assessment	Assessment Type	KIS Category	Duration / Length	% Weighting	Final element of assessment?	Qualifying Mark
Coursework	Written assignments	Coursework		25	No	
Examination	Written Exam	Written	2.5 hours	75	Yes	

**Final element of assessment:** The assessment that takes place last. **There should normally be only one element of assessment marked as final unless two assessment or submission dates occur on the same day.**

**Qualifying mark:** A specified minimum mark that must be obtained in one or more elements of assessment in order to pass a module. **This is in addition to, and distinct from, the requirement to achieve a pass in the module mark to pass the module.**

## Reassessment

Provide details of the reassessment methods used, specifying whether reassessment is either standard reassessment or synoptic reassessment.

Standard Reassessment

Synoptic Reassessment

Synoptic reassessment details (if you have indicated synoptic reassessment above, please give details)		
Brief Description of Assessment	Assessment Type	Duration / Length of Examination / Coursework
Resit examination	Written Exam	2.5 hours

### Section 3 - Alternative Assessment Arrangements for Associate Students

This section **must only** be completed if the module will be made available to associate students in Semester A and where the credit value of the "associate" version is the same as for the main version, and the main version is assessed by exam in May which is not available to the associate students. All other aspects of the module specification remain the same as indicated in Section 2 above. To add alternative assessment arrangements please click 'Add Alternative Assessment'.

### Section 4a - Half Module for Associate Students (for a half module to be taught in Semester A)

This section must be completed if the proposed module will take place over 2 semesters but will be made available to single-semester associate students in a half-credit format in **Semester A**. Modules worth less than 30 credits taken over 2 semesters may not be made available in a half-credit format. To add details for the half module please click 'Add Half Module (Semester A)'.

### Section 4b - Half Module for Associate Students (for a half module to be taught in Semester B)

This section must be completed if the proposed module will take place over 2 semesters but will be made available to single-semester associate students in a half-credit format in **Semester B**. Modules worth less than 30 credits taken over 2 semesters may not be made available in a half-credit format. To add details for the half module please click 'Add Half Module (Semester B)'.