

2007/08 Taught Postgraduate Module Catalogue

BIOL5241M

Chromosomal and Molecular Basis of Genetic Disorders

10 credits

Module manager Dr Martin Richards

Email: M.B.Richards@leeds.ac.uk

Taught Semester 1 [View Timetable](#)

Year running 2007/08

Pre-requisite qualifications

BSc or equivalent.

Module replaces BIOL5234M

This module is not approved as an Elective

Objectives

On completion of this module, students should be able to:

Understand some of the basic methodologies of DNA diagnostics

Appreciate factors determining the incidence of genetic diseases, discriminating between single gene, polygenic and multifactorial diseases.

understand the reasons why certain methodologies are used to study chromosomes and have knowledge of the different types of samples analysed in cytogenetic laboratories

understand the processes of cell division, gametogenesis and sex determination

appreciate the different types of structural and numerical chromosome rearrangements and the effects these have on human survival and disease.

have knowledge of the facts to be considered for the prenatal diagnosis of disease

understand the skills needed in the identification of normal and abnormal karyotypes

be able to critically assess a range of scientific literature, using their knowledge and understanding of basic human genetics and cytogenetics

Syllabus

Molecular genetics of inherited conditions, genetic heterogeneity and pharmacogenetics and genetic complexity. Chromosome structure, composition and nomenclature. Chromosome banding and FISH. Mitosis and meiosis. Cell cycle and gametogenesis. Sex determination and X-inactivation. Structural chromosome rearrangements. Sex chromosome rearrangements. Segregation of chromosome rearrangements. Types and origins of numerical chromosome abnormalities. Phenotype/karyotype correlations. Genomic

imprinting and uniparental disomy. Chromosome mosaicism. Cytogenetics of leukaemia. Cytogenetics of solid tumours. Techniques and considerations for prenatal diagnosis. Physical mapping. Future developments in cytogenetics.

Teaching methods

Delivery type	Number	Length hours	Student hours
Lecture	21	1.00	21.00
Practical	1	2.00	2.00
Private study hours			77.00
Total Contact hours			23.00
Total hours (100hr per 10 credits)			100.00

Private study

2 hours reading per lecture; $23 \times 2 = 46$ hours

14 hours practical write-up

17 hours data analysis

Progress monitoring

Data Analysis: 55%

Write-up of 'dry' practical: 45%

Methods of assessment

Coursework

Assessment type	Notes	% of formal assessment
Written Work	Write-up of the 'dry' practical	45.00
Source Analysis	Completion of a data analysis exercise	55.00
Total percentage (Assessment Coursework)		100.00

Reading list

The [reading list](#) is available from the Library website