2006/07 Taught Postgraduate Module Catalogue

BIOL5234M

Chromosomal and molecular basis of genetic disorders
15 credits

Module manager Dr Martin Richards
Email: m.b.richards@leeds.ac.uk

Taught Semester 1 View Timetable

Year running 2006/07

Pre-requisite qualifications
BSc or equivalent.

Module replaces BIOL5222M and BIOL5219M

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
Lectures: 21 x 1 hour;
Tutorials: 4 x 1 hour
'Dry' practical: 1 x 2 hours

Private study
18 hours preparing for oral presentations
3 hours reading per lecture
18 hours practical write-up
24 hours data analysis

Progress monitoring
Oral presentation

Methods of assessment
Oral presentation (30%)
Data analysis (40%),
write-up of `dry practical (30%)

Reading list
The reading list is available from the Library website