Module Title:
Genetic Testing

Module Code:
AM0310

Maximum Number of Students:
20

Total ECTS Credits
2

Notional Learning Hours
(a) Contact Time - 15 h
(b) Private Study – 35 h

Format of Teaching:
Lectures 10h
Laboratories or Practicals 5h

Teaching Strategy:
Formal lectures in 60/90 min timetable. One 5h laboratory practical.

Convener:
Vera Ribeiro

University / Department:
University of Algarve/Department of Chemistry, Biochemistry and Pharmacy

Language of Tuition:
English

Module Description - The Purpose or Aims:

1. To provide the basis and practical aspects of the methodologies available for genetic testing
2. To present specific examples of application, focusing on clinical correlations, significance of results as well as ethical aspects.

Specific Learning Outcomes for this module: (contributing to general learning outcomes GLO 1 – GLO 10)

At the end of the module the learner is expected to be able to:
1. correctly describe the principles underlying the most commonly used methods in molecular diagnostics
2. propose strategies for the characterization of specific genetic traits in individuals and the detection/quantification of gene expression levels in organisms, tissues or cells.
3. correctly perform genotyping tests and interpret the results

Summary of Course Content:

Nature of the hereditary material and the flow of information from DNA to protein. Inter-individual genetic variability and its impact in the predisposition to disease and in the response to therapeutics. The Human Genome Project. Genetic polymorphisms. Restriction digestion and electrophoresis. Southern and Northern blotting. DNA sequencing (chemical, chain termination, pyrosequencing). Recombinant DNA. Polymerase chain reaction (PCR) and PCR-based techniques (PCR-RFLP, PCR-ARMS, PCR-ASO, OLA, SSCP). Real-time PCR. Microarrays. Applications: microbiology, GMOs, forensics, prenatal/newborn screening, disease risk testing, pharmacogenetics. Benefits and risks of gene testing.

Transferable Skills Taught:

Communication:
Writing literature-based reports and practical reports
Information Technology:
Use of Web resources for database search (literature, DNA sequence)

Assessment Methods:

1. LO1 and LO2 – Written Examination, 75% (format to be decided - either a test or an essay on one specific topic)
2. LO3 – lab report, 25%

Assessment Criteria:

Threshold
LO1 – to correctly describe the principles of a given technique used in genetic testing
LO2 – to identify the information that can be obtained from each technique
LO3 – to adequately perform a genotyping protocol

Good
LO1 – to describe the advantages and limitations of each method
LO2 – to be able to choose the most adequate technique to answer a specific biological/clinical question
LO3 – to correctly assign the individual genotype for a specific gene polymorphism

Excellent
LO1 – to understand the most recent and emergent technological developments in genotyping strategies
LO2 – to be able to discuss the implications of the results that can be obtained as well as the ethical issues involved
LO3 - to critically analyse the lab results obtained in terms of clinical outcome

Resource Implications of Proposal and Proposed Solutions:

Lecture notes and selected papers will be available for students.

Recommended reading:

Hartl, DL, Jones, EW Genetics / analysis of genes and genomes, 2000, ISBN 0-7637-0913-1