



CORPORATE AND ACADEMIC SERVICES

MODULE SPECIFICATION

Part 1: Basic Data					
Module Title	Medical Genetics (MSc)				
Module Code	USSKP3-30-M	Level	M	Version	1
Owning Faculty	Health and Life Sciences	Field	Applied Sciences		
Contributes towards	MSc Biomedical Science				
UWE Credit Rating	30	ECTS Credit Rating	15	Module Type	Standard
Pre-requisites			Co- requisites		
Excluded Combinations			Module Entry requirements		
Valid From	September 2012		Valid to	September 2018	
CAP Approval Date	30 th May 2012				

Part 2: Learning and Teaching	
Learning Outcomes	<p>On successful completion of this module students will be able to:</p> <ul style="list-style-type: none"> • Critically discuss Chromosome morphology and classification. Discuss the future potential of human genetics and its ethical dilemmas. (exam - A1 and/or coursework - B) • Discuss and detail the consequences of aberrations of chromosome number and structure on the human phenotype, including medical syndromes and cancer. (exam – A1 and/or coursework - B) • Demonstrate the ability to identify the modes of inheritance of a wide range of specific autosomal and sex-linked genetic disorders together with phenotypic findings. (exam – A1 and/or coursework - B) • Critically review the current molecular approaches to gene cloning, characterisation and mapping, and the mechanisms involved in disease pathogenesis; understand the standard molecular techniques and their adaptation in the utility of genetic disease diagnosis (exam – A1 and/or coursework - B) • Evaluate the various prenatal diagnostic tests in terms of the procedural approaches and types of abnormality that might be detected. (exam – A1 and/or coursework - B) • Demonstrate an in depth appreciation of the application of medical genetics to diagnosis and therapy of genetic disease. (exam – A1 and/or coursework - B) • Integrate principles of Mendelian genetics, cytogenetics, and molecular genetics with their clinical application in modern medicine. (exam – A1 and/or coursework - B) <p>Non-assessed outcomes will include:</p> <ul style="list-style-type: none"> • Group working on presentations • Communication with peers in understanding the impact of genetic diseases for the patient, the family and the healthcare worker
Syllabus Outline	Overview: scientific basis of medical genetics – human genome- structure and function; human genome mapping; modes of inheritance of genetic disorders; clinical

	<p>applications – genetic assessment, prenatal diagnosis, treatment and prevention of disease</p> <p><u>Mechanisms of genetic modification</u>; DNA damage and repair mechanisms, cell cycle, epigenetics, imprinting, clinical conditions related to genetic modification.</p> <p><u>DNA analysis</u> – indirect & direct mutant gene tracking; techniques for demonstration of DNA mutation/polymorphisms including PCR, MLPA, Sequencing etc. Chromosome analysis – karyotyping, FISH, etc; heteromorphisms; mitochondrial chromosomes; chromosome aberrations.</p> <p><u>Gametogenesis</u> – meiosis; spermatogenesis; oogenesis; fertilisation ; Lyonisation; sex determination and differentiation; genomic imprinting</p> <p><u>Inheritance modes of genetic disorders</u> – autosomal and sex-linked; non-Mendelian inheritance – multifactorial – continuous and discontinuous; twin concordance, family correlation studies. Somatic cell disorders; mitochondrial disorders.</p> <p><u>Clinical applications</u>- genetic assessment, communication of advice, medical ethics; Prenatal diagnosis; population screening; prevention and treatment of genetic disease; gene therapy; Genetics of common diseases; Immunogenetics, cancer genetics, inborn errors of metabolism, RNA biology and alternative splicing, disorders of development</p>
<p>Contact Hours/Scheduled Hours</p>	<ul style="list-style-type: none"> • Students will meet staff weekly for a 2 hour lecture on the scheduled topic. Extra tutorial sessions will be scheduled to more deeply explore the subject areas. Presentation sessions will allow students to engage with the applications of various techniques in the diagnosis and prognosis of genetic diseases and these will be explored in a class setting with the input of staff members. • Research material from group presentations will be loaded onto Blackboard for knowledge exchange between students and to contribute to learning. <p>Students on the module will also be required to attend a conference week at an appropriate time in the year (dependent on changes to the academic calendar). During this week a range of visiting lecturers will be brought in to give keynote lectures (for example based on their clinical practice) or research focused lectures that map to the syllabus content. The conference week will also give students an experience of what it is like to attend a scientific conference, with an intensive schedule of talks across the week to be attended.</p>
<p>Teaching and Learning Methods</p>	<ul style="list-style-type: none"> • The module will be delivered as mix of lectures and integrated tutorial sessions – with computer-learning support together with a student centred case study and coupled to an assessed presentation on genetic techniques. Tutor led seminars of advanced topics of current issues in medical genetics will act to engage student with leading edge research. • Students will be expected to be independently engaged in further research indicated by the subject matter covered in the lectures and indicated by specific reading and reference lists; students will be expected to develop the content with self-directed learning. Students will also engage in group work to prepare an assessed presentation and in the preparation of key topics for an assessed timed essay as exam practice. • M level students will study the subject matter alongside level 3 students, performing the basic principles described above. M level students will additionally have tutorial sessions to further develop the critical analysis of the lecture material and to extend the knowledge base of the material covered.
<p>Reading Strategy</p>	<p>At Masters level students are expected to demonstrate the ability to find information, assess its relevance and utilise it in their studies in an independent manner; however the programme team recognise that students entering the programme may be at different levels of the development of the skills required to undertake this successfully. Therefore module leaders will provide you with a</p>

	<p>starting point in terms of core readings and the lecture material will also give you a strong starting point. However it is in the area of further reading that you need to show the independence of skills and of knowledge development, so you will need to find the Further Readings yourself. However, the skills required to do this are covered during the early stages of the course, during induction week you will have a library induction session, in the Research Methods and Practical Skills module that you take during the first semester we will cover how to undertake a literature search and how to assess and use the material you find appropriately. The programme tutorials will provide opportunities for you to further develop these skills and to ask any questions that you have. Further support and guidance is available through the library which runs workshops that you can sign up to, and also has advice in its website.</p> <p>Module leaders will give you a clear indication of any essential reading, and point you towards the appropriate textbooks and journals for their discipline. This will usually be in the form of a reading list in the module guide; the indicative list on this module specification is as it states indicative as the relevant available books and journals can change regularly – and the module specification is a document written only once when a module is modified and can last for many years. So it is important that you refer to the reading list for your specific year group as the definitive document.</p> <p>All students will be encouraged to make full use of the print and electronic resources available to them through membership of the University. These include a range of electronic journals and a wide variety of resources available through web sites and information gateways. The University Library's web pages provide access to subject relevant resources and services, and to the library catalogue. Many resources can be accessed remotely.</p>
Indicative Reading List	<p>Textbooks – current editions of</p> <ul style="list-style-type: none"> • “Essential Medical Genetics.” Connor M. & Ferguson-Smith M. F. (Wiley-Blackwell). • “Human Genetics.” Gardner A. & Davies T. (Scion). • “Human Genetics: concepts and applications.” Lewis R. McGraw-Hill. • “Colour Atlas of Genetics.” Passarge E. (Thieme). • “Genetic Analysis: an integrated approach.” Sanders M.F. & Bowman J.L. (Pearson). • “Ethical Issues in Modern Medicine.” Steinbock B., Arras J. D., London A. J. (McGraw-Hill). • “Human Molecular Genetics.” Sudbury P. (Pearson Education). • “Emery’s Elements of Medical Genetics” Turnpenny P & Ellard S. (Elsevier). • “Medical Genetics.” Young I. D. (Oxford University Press). <p>Appropriate current journals, relevant to the course content, as indicated by the academic staff</p>

Part 3: Assessment	
Assessment Strategy	<p>All specialist subject modules on the MSc BMS programme have a 50:50 weighting of course work to final exam. Coursework as decided by the module leader in line with the programme assessment strategy.</p> <ul style="list-style-type: none"> • The module will be assessed by a 3hour examination under controlled conditions on the lecture material, together with 2 pieces of coursework designed to encourage extra reading beyond the lecture notes, encompassing enhanced concepts of the lecture material as well as wider reading of the topic itself. • Feedback will be provided on all coursework and there will also be opportunity in tutorials to discuss student progress and understanding. • Details of the requirements for each component will be provided in the module handbook together with a marking criteria and mark sheet

	by which students can guide their performance.	
Identify final assessment component and element	3 hour examination	
% weighting between components A and B (Standard modules only)	A:	B:
	50	50
First Sit		
Component A (controlled conditions) Description of each element	Element weighting (as % of component)	
1. 3 hour examination – final assessment	100	
Component B Description of each element	Element weighting (as % of component)	
1. Practical portfolio	60	
2. Poster presentation of technique based case study	40	
Resit (further attendance at taught classes is not required)		
Component A (controlled conditions) Description of each element	Element weighting (as % of component)	
1. 3 hour examination	100	
Component B Description of each element	Element weighting (as % of component)	
1. Extended case study	100	
If a student is permitted an EXCEPTIONAL RETAKE of the module the assessment will be that indicated by the Module Description at the time that retake commences.		