



CORPORATE AND ACADEMIC SERVICES

MODULE SPECIFICATION


Part 1: Basic Data					
Module Title	Medical Genetics				
Module Code	USSKBH-30-3	Level	3	Version	1
Owning Faculty	Health and Applied Sciences	Field	Biological, Biomedical and Health Sciences		
Contributes towards	BSc (hons) Biomedical Science BSc (hons) Healthcare Science (Life Science) BSc (Hons.) Forensic Science BSc (Hons.) Forensic Science (Biology)				
UWE Credit Rating	30	ECTS Credit Rating	15	Module Type	Standard
Pre-requisites	Molecular Genetics (USSKB7-15-2)	Co- requisites	none		
Excluded Combinations	none	Module Entry requirements	N/A		
Valid From	September 2014	Valid to	September 2020		

CAP Approval Date	28/03/2014
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Part 2: Learning and Teaching	
Learning Outcomes	<p>On successful completion of this module students will be able to:</p> <p>All L.O assessed in both A&B</p> <ul style="list-style-type: none"> • Discuss Chromosome morphology and classification. • Discuss the future potential of human genetics and its ethical dilemmas; • Identify the modes of inheritance of specific autosomal and sex-linked genetic disorders together with phenotypic findings; • Review the current molecular approaches to gene cloning, characterisation and mapping, and the mechanisms involved in disease pathogenesis; • Evaluate the various prenatal diagnostic tests in terms of the procedural approaches and types of abnormality that might be detected; • Outline the application of medical genetics to diagnosis, counselling and therapy of genetic disease; • Integrate principles of Mendelian genetics, cytogenetics, and molecular genetics with their clinical application in modern medicine.
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>Integral to the module will be a series of workshops on ethical aspects of genetic testing and manipulation, including consideration of cloning, preimplantation genetic diagnosis and “saviour siblings”.</p>
Contact Hours	<ul style="list-style-type: none"> • Students will meet staff weekly for a 2 hour lecture on the scheduled topic. Extra workshops will be provided to cover the ethical aspects and these will cover the underpinning genetic techniques and limitations that contribute to the ethical dilemmas. Presentation sessions will allow students to engage with the issues surrounding genetic testing and 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 will be able to attend MSc tutorials and conference week by choice if they wish to expand their knowledge of genetics applications.</p>
Teaching and Learning Methods	<p>The module will be delivered as mix of lectures and integrated tutorial sessions – with computer-learning support together with a student centred research exercise on genetic testing</p> <ul style="list-style-type: none"> • 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. <p>Scheduled learning includes lectures, seminars, tutorials.</p> <p>Independent learning includes hours engaged with essential reading, case study preparation, assignment preparation and completion etc. These sessions constitute an average time per level as indicated in the table below. Scheduled sessions may vary slightly depending on the module choices you make.</p>
Key Information Sets Information	<p>Key Information Sets (KIS) are produced at programme level for all programmes that</p>

this module contributes to, which is a requirement set by HESA/HEFCE. KIS are comparable sets of standardised information about undergraduate courses allowing prospective students to compare and contrast between programmes they are interested in applying for.

<i>Number of credits for this module</i>				30	
Hours to be allocated	Scheduled learning and teaching study hours	Independent study hours	Placement study hours	Allocated Hours	
300	72	228	0	300	

The table below indicates as a percentage the total assessment of the module which constitutes a -

Written Exam: Unseen written exam

Coursework: Written assignment or essay, presentation

Please note that this is the total of various types of assessment and will not necessarily reflect the component and module weightings in the Assessment section of this module description:

Total assessment of the module:		
Written exam assessment percentage		60%
Coursework assessment percentage		40%
Practical exam assessment percentage		0%
		100%

Reading Strategy

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. Students will be presented with opportunities within the curriculum to develop their information retrieval and evaluation skills in order to identify such resources effectively.

Any **essential reading** will be indicated clearly, along with the method for accessing it, e.g. students may be expected to purchase a set text, be given or sold a print study pack or be referred to texts that are available electronically, etc. This guidance will be available either in the module handbook, via the module information on Blackboard or through any other vehicle deemed appropriate by the module/programme leaders. If **further reading** is expected, this will be indicated clearly. If specific texts are listed, a clear indication will be given regarding how to access them and, if appropriate, students will be given guidance on how to identify relevant sources for themselves, e.g. through use of bibliographical databases.

Indicative Reading List

Textbooks – current editions of

- Connor, M. & Ferguson-Smith, M. F. *Essential Medical Genetics*. Oxford: Wiley-Blackwell.

	<ul style="list-style-type: none"> • Gardner, A. & Davies, T. <i>Human Genetics</i>. Bloxham: Scion. • Lewis, R. <i>Human Genetics: concepts and applications</i>. McGraw-Hill. • Passarge, E. <i>Colour Atlas of Genetics</i>.. Stuttgart: Thieme. • Sanders, M.F. & Bowman, J.L <i>Genetic Analysis: an integrated approach</i>. Harlow: Pearson. • Steinbock B., Arras J. D., London A. J. <i>Ethical Issues in Modern Medicine</i>. McGraw-Hill. • Sudbury P. <i>Human Molecular Genetics</i>. Harlow: Pearson Education. • Turnpenny P & Ellard S. <i>Emery's Elements of Medical Genetics</i> Philadelphia, PA: ElsevierChurchill Livingstone.. • Young I. D. <i>Medical Genetics</i>.Oxford: Oxford University Press. <p>Appropriate current journals, relevant to the course content, as indicated by the academic staff</p>
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Part 3: Assessment	
Assessment Strategy	<p>All specialist subject modules on the BSc BMS programme have a 40:60 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 two pieces of coursework designed to encourage extra reading beyond the lecture notes provided at the ethical workshops. • Feedback will be provided on all coursework and there will also be opportunity in tutorials to discuss student progress and understanding. <p>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.</p>

Identify final assessment component and element		
% weighting between components A and B (Standard modules only)	A: 60	B: 40
First Sit		
Component A (controlled conditions) Description of each element	Element weighting (as % of component)	
1. Examination (3 hours)	100	
2.(etc)		
Component B Description of each element	Element weighting (as % of component)	
1. Research essay (1500 words)	50	
2. Ethical debate	50	

Resit (further attendance at taught classes is not required)

Component A (controlled conditions) Description of each element	Element weighting (as % of component)
1. Examination (3 hours)	100
2.(etc)	
Component B Description of each element	Element weighting (as % of component)
1.Extended research exercise (3000 words)	100
2.(etc)	
<p>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.</p>	