University of the West of England
BRISTOL

CORPORATE AND ACADEMIC SERVICES

MODULE SPECIFICATION

Part 1: Basic Data						
Module Title	Medical Genetics					
Module Code	USSKP3-30-M		Level	M	Version	2
UWE Credit Rating	30	ECTS Credit Rating	15	WBL module?	No	
Owning Faculty	Health and Applied Sciences		Field	Applied Sciences		
Department	Biological Biomedical and Analytical Sciences		Module Type	Standard		
Contributes towards	MSc Biomedical Science					
Pre-requisites	Study of genetics at undergraduate level		Co- requisites	None		
Excluded Combinations			Module Entry requirements	Study of genetics at undergraduate level		
First CAP Approval Date	30th May 2012		Valid from	September 2012		
Revision CAP Approval Date	2nd February 2016		Revised with effect from	September 2016		

Review Date	~ 5 years post approval for PSRB requirements

Part 2: Learning and Teaching	
Learning Outcomes	On successful completion of this module students will be able to: • Critically discuss Chromosome morphology and classification. Discuss the future potential of human genetics and its ethical dilemmas. (exam and/or coursework) • Discuss and detail the consequences of aberrations of chromosome number and structure on the human phenotype, including medical syndromes and cancer. (exam and/or coursework) • 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 and/or coursework) • 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 and/or coursework) • Evaluate the various prenatal diagnostic tests in terms of the procedural approaches and types of abnormality that might be detected. (exam and/or coursework) • Demonstrate an in depth appreciation of the application of medical genetics to diagnosis and therapy of genetic disease. (exam and/or coursework) • Integrate principles of Mendelian genetics, cytogenetics, and molecular genetics

	with their clinical application in modern medicine. (exam and/or coursework) Non-assessed outcomes will include: • Group working and critical analysis in tutorials • 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 applications – genetic assessment, prenatal diagnosis, treatment and prevention of disease <u>Mechanisms of genetic modification</u>; DNA damage and repair mechanisms, cell cycle, epigenetics, imprinting, clinical conditions related to genetic modification. <u>DNA analysis</u> – indirect & direct mutant gene tracking; techniques for demonstration of DNA mutation/polymorphisms including PCR, MLPA, Sequencing, Southern blotting, microarray. Chromosome analysis – karyotyping, FISH, etc; heteromorphisms; mitochondrial chromosomes; chromosome aberrations. <u>Gametogenesis</u> – meiosis; spermatogenesis; oogenesis; fertilisation ; Lyonisation; sex determination and differentiation; genomic imprinting <u>Inheritance modes of genetic disorders</u> – autosomal and sex-linked; non-Mendelian inheritance – mutilfactorial – continuous and discontinuous; twin concordance, family correlation studies. Somatic cell disorders; mitochondrial disorders. <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
Contact Hours/Scheduled Hours	• 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 key inherited case studies, and discuss the applications of various techniques in the diagnosis and prognosis of genetic diseases; these will be explored in a class setting with the input of staff members. • 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.
Teaching and Learning Methods	• 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 poster presentation on genetic disease and diagnostic 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, in particular to appreciate the applications and limitations of various molecular diagnostic techniques and the implications in their use for termination decisions.
Reading Strategy	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 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. 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. 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.
Indicative Reading List	Textbooks – current editions of • Tobias, E., Connor, M. and Ferguson-Smith, M. F. (2011) *Essential Medical Genetics*. 6th ed. Chichester: Wiley-Blackwell. • Gardner, A. and Davies, T. (2009) *Human Genetics*. 2nd ed. Bloxham: Scion. • Lewis, R. (2008) *Human Genetics: concepts and applications*. London: McGraw-Hill. • Sanders, M.F. and Bowman, J.L. (2014) *Genetic Analysis: an integrated approach.* Online: Pearson. • Steinbock, B., Arras, J. D. and London, A. J. (2009) *Ethical Issues in Modern Medicine.* 7th ed. London: McGraw-Hill. • Turnpenny, P. and Ellard, S. (2012) *Emery's Elements of Medical Genetics.* 14th ed. Philadelphia: Elsevier Churchill Livingstone. • Young, I. D. (2010) *Medical Genetics*. Oxford: Oxford University Press. • Strachan, T. and Read, A. (2011) Human Molecular Genetics. 4th ed. Abingdon: Garland Science Appropriate current journals, relevant to the course content, as indicated by the academic staff

Part 3: Assessment	
Assessment Strategy	The MSc BMS Programme has a programme level assessment strategy (see Programme Specification appendix 1), and all modules have their assessments designed to relate to that document. For parity across all routes the specialist subject modules on the MSc BMS programme have a 50:50 weighting of course work to final exam – this module is one of the specialist modules. Therefore the coursework has been designed in line with the programme assessment strategy. Specialist module coursework is designed to test the ability of students to express their chosen specialist discipline in both written form and in oral form. The coursework essay is similar in style to a review article in a journal, and the presentation is designed to replicate those given at conferences. Both are highly relevant assessments for higher level science graduates to have undertaken, preparing them for future academic style writing and presentation in their professional lives. The assessments are marked to the BBAS standard PG marking criteria, and students are fully briefed on the assessment both in writing and through a tutorial session. Students also develop several transferable skills during this assessment including negotiation (they are allowed to pick their own title and refine it), critiquing of published literature, scientific writing etiquette, and editing documents to a high editorial standard. The exam enables students to demonstrate a breadth of knowledge that it would be reasonable for future employers to see in a Masters graduate in relation to their chosen specialism.

Identify final assessment component and element	**A1**	
% 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 –open book exam	100
Component B **Description of each element**	**Element weighting** **(as % of component)**
1. Essay (3000 words)	60
2. Poster presentation of genetic case study – 20 minutes including oral defence	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; open book	100
Component B **Description of each element**	**Element weighting** **(as % of component)**
1. Extended case study (5000 words maximum)	100

If a student is permitted a **RETAKE** of the module the assessment will be that indicated by the Module Description at the time that retake commences.