MODULE SPECIFICATION

Part 1: Information			
Module Title	Medical Genetics		
Module Code	USSKP3-30-M	Level	Level 7
For implementation from	2020-21		
UWE Credit Rating	30	ECTS Credit Rating	15
Faculty	Faculty of Health & Applied Sciences	Field	Applied Sciences
Department	HAS Dept of Applied Sciences		
Module type:	Standard		
Pre-requisites	None		
Excluded Combinations	None		
Co- requisites	None		
Module Entry requirements	None		

Part 2: Description

Features: Module Entry Requirements: Study of genetics at undergraduate level

Educational Aims: See learning outcomes.

In addition to the learning outcomes, 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

Outline Syllabus: 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.

Mechanisms of genetic modification; DNA damage and repair mechanisms, cell cycle, epigenetics, imprinting, clinical conditions related to genetic modification.

DNA analysis – indirect and 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.

Gametogenesis – meiosis; spermatogenesis; oogenesis; fertilisation ; Lyonisation; sex determination and differentiation; genomic imprinting.

Inheritance modes of genetic disorders – autosomal and sex-linked; non-Mendelian inheritance – mutilfactorial – continuous and discontinuous; twin concordance, family correlation studies. Somatic cell disorders; mitochondrial disorders.

Clinical applications- 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.

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.

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.

Part 3: Assessment

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. This will be an online exam with a 24 hour submission window,

First Sit Components	**Final Assessment**	**Element weighting**	**Description**
Written Assignment - Component B		30 %	3000 word essay
Presentation - Component B		20 %	Poster presentation of genetic case study - 20 minutes including oral defence
Examination (Online) - Component A	✓	50 %	Online examination (24 hours)
Resit Components	**Final Assessment**	**Element weighting**	**Description**
Examination (Online) - Component A	✓	50 %	Online examination (24 hours)
Case Study - Component B		50 %	Extended case study (5000 words maximum)

Part 4: Teaching and Learning Methods

Learning Outcomes

On successful completion of this module students will achieve the following learning outcomes:

Module Learning Outcomes	**Reference**
Critically discuss Chromosome morphology and classification. Discuss the future potential of human genetics and its ethical dilemmas	MO1
Discuss and detail the consequences of aberrations of chromosome number and structure on the human phenotype, including medical syndromes and cancer	MO2
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	MO3
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	MO4
Evaluate the various prenatal diagnostic tests in terms of the procedural approaches and types of abnormality that might be detected	MO5
Demonstrate an in depth appreciation of the application of medical genetics to diagnosis and therapy of genetic disease	MO6
Integrate principles of Mendelian genetics, cytogenetics, and molecular genetics with their clinical application in modern medicine	MO7

Contact Hours

Independent Study Hours:

Independent study/self-guided study	234

	Total Independent Study Hours:	234
	Scheduled Learning and Teaching Hours:	
	Face-to-face learning	66
	Total Scheduled Learning and Teaching Hours:	66
	Hours to be allocated	300
	Allocated Hours	300
Reading List	*The reading list for this module can be accessed via the following link:* https://uwe.rl.talis.com/modules/usskp3-30-m.html	

Part 5: Contributes Towards
This module contributes towards the following programmes of study: Biomedical Science (Medical Genetics) [Sep][FT][Frenchay][1yr] MSc 2020-21 Biomedical Science [Sep][FT][Frenchay][1yr] MSc 2020-21