MODULE SPECIFICATION

Part 1: Information			
Module Title	Medical Genetics		
Module Code	USSKBH-30-3	Level	Level 6
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	Molecular Genetics 2020-21		
Excluded Combinations	None		
Co- requisites	None		
Module Entry requirements	None		

Part 2: Description

Educational Aims: See Learning Outcomes

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 etc. 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.

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".

Teaching and Learning Methods: Students will interact weekly (3 hr timeslot) with staff for lectures/lectorials/tutorials 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. Workshops tutorials will also cover a range of genetic testing techniques to underpin knowledge for assessments and lectures. Presentation sessions will allow students to engage with the issues surrounding genetic testing and will be explored 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.

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.

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.

Scheduled learning includes lectures, lectorials, seminars, tutorials.

Independent learning includes hours engaged with essential reading, case study preparation, assignment preparation and completion etc. These sessions constitute an average time per level. Scheduled sessions may vary slightly depending on the module choices you make.

Part 3: Assessment

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.

The module will be assessed by an online examination (with a 24 hour window for submission) on the lecture material, together with two pieces of coursework designed to encourage extra reading beyond the lecture notes provided at the ethical and techniques workshops. The first piece of coursework involves a detailed assessment of a genetic disease including: symptoms, epidemiology, underpinning genetics and molecular pathways, current and novel approaches to treatment - presented as a 1500 word essay. The second piece of coursework will be a group debate on ethical issues in genetic testing.

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.

First Sit Components	**Final Assessment**	**Element weighting**	**Description**
Written Assignment - Component B		20 %	Research essay (1500 words)
Set Exercise - Component B		20 %	Ethical debate
Examination (Online) - Component A	✓	60 %	Online open book examination (24 hours)

Resit Components	Final Assessment	Element weighting	Description
Written Assignment - Component B		40 %	Extended research exercise (3000 words)
Examination (Online) - Component A	✓	60 %	Online open book examination (24 hours)

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
Discuss Chromosome morphology and classification	MO1
Discuss the future potential of human genetics and its ethical dilemmas	MO2
Identify the modes of inheritance of specific autosomal and sex-linked genetic disorders together with phenotypic findings	MO3
Review the current molecular approaches to gene cloning, characterisation and mapping, and the mechanisms involved in disease pathogenesis	MO4
Evaluate the various prenatal diagnostic tests in terms of the procedural approaches and types of abnormality that might be detected	MO5
Outline the application of medical genetics to diagnosis, counselling 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/usskbh-30-3.html

Part 5: Contributes Towards
This module contributes towards the following programmes of study: Biomedical Science [Sep][FT][Frenchay][4yrs] MSci 2018-19 Healthcare Science (Genetic Science) [Sep][FT][Frenchay][3yrs] BSc (Hons) 2018-19 Biomedical Science [Sep][FT][Frenchay][3yrs] BSc (Hons) 2018-19 Biological Sciences [Sep][FT][Frenchay][3yrs] BSc (Hons) 2018-19 Biological Sciences [Sep][FT][Frenchay][4yrs] MSci 2018-19