



Module Specification

Medical Genetics

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Part 1: Information

Module title: Medical Genetics

Module code: USSKBH-30-3

Level: Level 6

For implementation from: 2021-22

UWE credit rating: 30

ECTS credit rating: 15

Faculty: Faculty of Health & Applied Sciences

Department: HAS Dept of Applied Sciences

Partner institutions: None

Delivery locations: Frenchay Campus

Field: Applied Sciences

Module type: Standard

Pre-requisites: Genes and Biotechnology 2020-21, Human Health and Disease 2020-21, Molecular Cell Biology 2019-20, Studies in the Biology of Disease 2021-22

Excluded combinations: None

Co-requisites: None

Continuing professional development: Yes

Professional, statutory or regulatory body requirements: None

Part 2: Description

Overview: Pre-requisites: students must take (USSKAT-30-3 Studies in the Biology of Disease AND USSJXR-15-2 Molecular Cell Biology) OR (USSKAN-30-2 Human Health and Disease OR USSKAM-30-2 Genes and Biotechnology)

Features: Not applicable

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 – multifactorial – 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”.

Part 3: Teaching and learning methods

Teaching and learning methods: 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.

Students will be able to attend MSc tutorials and conference week by choice if they wish to expand their knowledge of genetics applications.

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, 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.

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

MO1 Discuss Chromosome morphology and classification

MO2 Discuss the future potential of human genetics and its ethical dilemmas

MO3 Identify the modes of inheritance of specific autosomal and sex-linked genetic disorders together with phenotypic findings

MO4 Review the current molecular approaches to gene cloning, characterisation and mapping, and the mechanisms involved in disease pathogenesis

MO5 Evaluate the various prenatal diagnostic tests in terms of the procedural approaches and types of abnormality that might be detected

MO6 Outline the application of medical genetics to diagnosis, counselling and therapy of genetic disease

MO7 Integrate principles of Mendelian genetics, cytogenetics, and molecular genetics with their clinical application in modern medicine

Hours to be allocated: 300

Contact hours:

Independent study/self-guided study = 228 hours

Face-to-face learning = 72 hours

Total = 300

Reading list: The reading list for this module can be accessed at [readinglists.uwe.ac.uk](https://uwe.rl.talis.com/modules/usskbh-30-3.html) via the following link <https://uwe.rl.talis.com/modules/usskbh-30-3.html>

Part 4: Assessment

Assessment strategy: All specialist subject modules on the BSc 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.

The module will be assessed by an online examination (with a 24hr 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 workshops.

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.

Assessment components:

Examination (Online) - Component A (First Sit)

Description: Online Examination (24 hours)

Weighting: 50 %

Final assessment: Yes

Group work: No

Learning outcomes tested: MO1, MO2, MO3, MO4, MO5, MO6, MO7

Written Assignment - Component B (First Sit)

Description: Case study (1500 words)

Weighting: 25 %

Final assessment: No

Group work: No

Learning outcomes tested: MO1, MO2, MO3, MO4, MO5, MO6, MO7

Set Exercise - Component B (First Sit)

Description: Ethical debate

Weighting: 25 %

Final assessment: No

Group work: No

Learning outcomes tested: MO1, MO2, MO3, MO4, MO5, MO6, MO7

Examination (Online) - Component A (Resit)

Description: Online Examination (24 hours)

Weighting: 50 %

Final assessment: Yes

Group work: No

Learning outcomes tested:

Written Assignment - Component B (Resit)

Description: Extended research exercise (3000 words)

Weighting: 50 %

Final assessment: No

Group work: No

Learning outcomes tested:

Part 5: Contributes towards

This module contributes towards the following programmes of study:

Healthcare Science (Genetic Science) {Foundation} [Sep][FT][Frenchay][4yrs] BSc (Hons) 2018-19

Biomedical Science [Sep][SW][Frenchay][5yrs] MSci 2018-19

Biomedical Science {Foundation} [Sep][FT][Frenchay][5yrs] MSci 2018-19

Applied Biomedical Science {Top-Up}[Sep][FT][INTUNI][1yr] BSc (Hons) 2019-20

Biological Sciences [Sep][FT][Frenchay][3yrs] BSc (Hons) 2019-20

Biological Sciences [Sep][FT][Frenchay][4yrs] MSci 2019-20

Biomedical Science [Sep][FT][Frenchay][3yrs] BSc (Hons) 2019-20

Biomedical Science [Sep][FT][Frenchay][4yrs] MSci 2019-20

Applied Biomedical Science [Sep][FT][Frenchay][3yrs] BSc (Hons) 2019-20

Biological Sciences [Sep][SW][Frenchay][4yrs] BSc (Hons) 2018-19

Biological Sciences [Sep][SW][Frenchay][5yrs] MSci 2018-19

Biomedical Science [Sep][SW][Frenchay][4yrs] BSc (Hons) 2018-19

Biological Sciences {Foundation} [Sep][FT][Frenchay][5yrs] MSci 2018-19

Biomedical Science {Foundation} [Sep][FT][Frenchay][4yrs] BSc (Hons) 2018-19

Biological Sciences {Foundation} [Sep][FT][Frenchay][4yrs] BSc (Hons) 2018-19