



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: 2022-23

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: Genetics 2022-23, Human Health and Disease 2022-23, Molecular Biotechnology 2022-23, Molecular Cell Biology 2022-23, Studies in the Biology of Disease 2022-23

Excluded combinations: None

Co-requisites: None

Continuing professional development: Yes

Professional, statutory or regulatory body requirements: None

Part 2: Description

Overview: In this module students will gain an in-depth knowledge of human inheritance and genetic disease.

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 OR USSKFQ-15-2 Genetics).

Features: Not applicable

Educational aims: This module aims to teach students to think holistically about factors influencing mutation and how this leads to genetic conditions. Further, students will consider the applications, limitations and ethical issues associated with the use of genetic technologies and their impact on society.

Outline syllabus: Overview: scientific basis of medical genetics – human genome-structure and function; 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 – techniques for demonstration of DNA mutation/polymorphisms including PCR, Multiplex Ligation Dependent Probe Amplification (MLPA), Sequencing etc. Chromosome analysis – karyotyping, Fluorescence In-situ Hybridisation (FISH), etc; mitochondrial chromosomes; chromosome aberrations.

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

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; medical ethics; Prenatal diagnosis; population screening; prevention and treatment of genetic disease; 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: This module is delivered as a mixture of lectures and consolidating interactive tutorial classes.

The debate 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 and students, who are encouraged to contribute their own views and to consider the societal factors which have shaped these.

The module also includes monthly check-in quizzes, which enable students to assess their knowledge on the previous months lecture material. Students receive immediate feedback on these quizzes and staff use the results to inform student support and follow on teaching.

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

MO1 Critically discuss chromosome morphology and classification by reviewing current molecular approaches and the mechanisms involved in disease pathogenesis.

MO2 Critically 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 Critically 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 and therapy of genetic disease

MO6 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: Component A:

Component A is a 3 hour examination. An examination has been selected to assess the breadth and depth of student knowledge across the syllabus and the ability of students to synthesise and critically evaluate current research in the field.

Students are supported in this assessment by tutorials on examination structure, preparation and strategies. Exemplar questions are also completed and reviewed in class.

Component B:

Component B is a debate on controversial issues relating to genetic research and technology. Students will work in groups to explore pros, cons, anti-pros and anti-cons of a self selected topic. This topic can be selected from a suggested list, or students are invited to propose their own topic of interest. Each student presents a different viewpoint relating to the issue for ten minutes and answers questions relating to their presentation for five minutes.

This assessment encourages in-depth research and critical analysis pertaining to a

particular genetic dilemma. Moreover, this assessment develops valuable graduate skills in debate, empathy and engenders inclusivity and respect for alternative viewpoints and societal norms, to their own.

Students are supported in this assessment by tutorial classes which explore the assessment. These tutorial classes also use example debates and students are guided regarding ambiguous cases, which are appropriate for debate.

Assessment components:

Examination - Component A (First Sit)

Description: Three Hour Examination

Weighting: 50 %

Final assessment: Yes

Group work: No

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

Presentation - Component B (First Sit)

Description: Ethical debate. Students will work in small groups, presenting for 10 minutes each and answering questions for 5 minutes each.

Weighting: 50 %

Final assessment: No

Group work: Yes

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

Examination - Component A (Resit)

Description: Three Hour Examination

Weighting: 50 %

Final assessment: Yes

Group work: No

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

Presentation - Component B (Resit)

Description: Ethical Debate. Students will present for 10 minutes each and answer questions for 5 minutes.

Weighting: 50 %

Final assessment: No

Group work: No

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

Part 5: Contributes towards

This module contributes towards the following programmes of study:

Healthcare Science (Genetic Science) [Sep][FT][Frenchay][3yrs] BSc (Hons) 2020-21

Biomedical Science [Sep][SW][Frenchay][5yrs] MSci 2019-20

Biomedical Science {Foundation} [Sep][FT][Frenchay][5yrs] MSci 2019-20

Healthcare Science (Genetic Science) {Foundation} [Sep][FT][Frenchay][4yrs] BSc (Hons) 2019-20

Biomedical Science {Foundation} [Sep][SW][Frenchay][6yrs] MSci 2018-19

Biological Sciences [Sep][FT][Frenchay][3yrs] BSc (Hons) 2020-21

Biological Sciences [Sep][FT][Frenchay][4yrs] MSci 2020-21

Applied Biomedical Science [Sep][FT][Frenchay][3yrs] BSc (Hons) 2020-21

Biomedical Science [Sep][FT][Frenchay][4yrs] MSci 2020-21

Biomedical Science [Sep][FT][Frenchay][3yrs] BSc (Hons) 2020-21

Biological Sciences {Foundation} [Sep][FT][Frenchay][4yrs] BSc (Hons) 2019-20

Biological Sciences [Sep][SW][Frenchay][4yrs] BSc (Hons) 2019-20

Biological Sciences [Sep][SW][Frenchay][5yrs] MSci 2019-20

Biological Sciences {Foundation} [Sep][FT][Frenchay][5yrs] MSci 2019-20

Biomedical Science [Sep][SW][Frenchay][4yrs] BSc (Hons) 2019-20

Biomedical Science {Foundation} [Sep][FT][Frenchay][4yrs] BSc (Hons) 2019-20

Biomedical Science {Foundation} [Sep][SW][Frenchay][5yrs] BSc (Hons) 2018-19

Biological Sciences {Foundation} [Sep][SW][Frenchay][5yrs] BSc (Hons) 2018-19

Biological Sciences {Foundation} [Sep][SW][Frenchay][6yrs] MSci 2018-19