Health Education England, National Health Service England and Genomics England Ltd.

Masters (MSc) Programme in Genomic Medicine Curriculum Document

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Prepared by: HEE Genomics Education Programme in conjunction with Genomics England Ltd.

Contents

Purpose

This document details the development of an exciting and innovative new Masters programme in Genomic Medicine designed to be applicable and accessible to all healthcare professions. It has been developed through collaboration between Health Education England (HEE), NHS England (NHSE) and Genomics England Ltd. (GeL). The purpose of the programme is to provide genomic education for many different healthcare professionals to support:

- an understanding of the 100,000 Genomes Project and the research and clinical opportunities it provides
- wider education in genomic medicine and the transformation of both the specialist and general workforce in the NHS
- building capacity and capability in the NHS workforce in genomic medicine both clinical and research.

Background

Health Education England

As part of our mandate from the Department of Health (DH), HEE is tasked with delivering recruitment, education, training and development of the healthcare workforce.

HEE exists to improve the quality of health and healthcare for the people and patients of England. Each year HEE has the responsibility for investing nearly £5 billion of public funding in training and education to ensure that we provide staff in the right numbers, with the right values, skills and behaviours to meet current and future patient needs. HEE has developed an Education Outcomes Framework that has adopted five high level domains outlined below:

1. **Excellent education:** education and training is commissioned and provided to the highest standards, ensuring learners have an excellent experience and that all elements of education and training are delivered in a safe environment for patients, staff and learners.
2. **Competent and capable staff:** there are sufficient healthcare staff educated and trained, aligned to service and changing care needs, to ensure that people are cared for by staff who are properly inducted, trained and qualified, who have the required knowledge and skills to do the jobs the service needs, whilst working effectively in a team.

3. **Adaptable and flexible workforce:** the workforce is educated to be responsive to innovation and new technologies with knowledge about best practice, research and innovation that promotes adoption and dissemination of better quality service delivery to reduce variability and poor practice.
4. **NHS values and behaviours:** healthcare staff have the necessary compassion, values and behaviours to provide person centred care and enhance the quality of the patient experience through education, training and regular Continuing Personal and Professional Development (CPPD) that instils respect for patients.
5. **Widening participation:** talent and leadership flourishes, free from discrimination, with fair opportunities to progress, enabling everyone to participate and fulfil their potential, recognising individual as well as group differences, treating people as individuals, and placing positive value on diversity in the workforce; in addition, there are opportunities to progress across the five leadership framework domains.

The HEE Genomics Education Programme

A key objective for HEE is the establishment of a Genomics Education Programme that will both support and enhance the 100,000 Genomes Project and provide education and training in genomics at the appropriate level for all staff in the NHS. This will prepare for the legacy of the 100,000 Genomes Project in healthcare.

The importance of education and training, across the NHS and the wider research field, as a central pillar in ensuring the effective delivery of genomic technologies has been recognised from the outset. The core aims of the HEE Genomics Education Programme are to:

- respond to the immediate task of supporting the 100,000 Genomes Project
- provide the foundations for long term education and training to underpin workforce development in bioinformatics and the use of genomic data for specialist staff
- a wider awareness programme on genomic technologies for non-specialist healthcare workers and allied healthcare professionals
- support workforce planning, in collaboration with its partners in the NHS, Public Health England, research and academia to ensure that there is a holistic approach to genomics education and training from bench to bedside
- ensure that the education and training programmes put in place can support the translational aspects of the 100,000 Genomes Project, building upon its legacy in a way that is sustainable through the development of the knowledge and skills of the workforce and its transformation.

HEE will provide an education and training programme that will train existing NHS staff and develop the next generation of clinicians, scientists and multidisciplinary healthcare teams to ensure that NHS capacity to harness genomic medicine is the most advanced in the world.

Professor Ian Cumming (Chief Executive Officer of HEE) has established a Genomic Advisory Board with a number of key functions, including:

- determining the education and training implications of the 100,000 Genomes Project and the timescales for appropriately developing the workforce;
- ensuring that the views of patients and the public inform all workforce and education training and developments; and
- appointing Task and Finish working groups to undertake specific projects.

Professor Sue Hill (Chief Scientific Officer for NHS England) has engaged with members across the healthcare delivery sector who, as members of specific Task and Finish groups, will input into work-streams derived from a high level work plan which will include the following:

- Governance and Strategy
- Workforce Communications and Awareness Raising
- Education and Training Resources
- Workforce Planning and Commissioning
- Implementation and Workforce Transformation
- Research and Innovation.

Genomics England and the 100,000 Genomes Project, and its implications within the NHS

As part of the UK Government's Life Sciences Strategy, the Prime Minister announced the 100,000 Genomes Project in December 2012. In July 2013 the Department of Health formed a wholly owned limited company called Genomics England to oversee and deliver the 100,000 Genomes Project. As well as working closely with the Department of Health, Genomics England will develop its programme in partnership with the DH and other key delivery partners, including NHS England (NHSE), Public Health England (PHE) and Health Education England (HEE).

This programme aims to transform the capability and capacity of the NHS to apply genomic medicine for patient benefit. The 100,000 Genomes Project has the following main aims:

- sequence up to 100,000 genomes by the end of 2017 focusing upon rare inherited disorders, cancers and infectious disease
- create an ethical and transparent NHS transformation programme based on informed consent
- enable new scientific discovery and medical insights

- enable the NHS to become world-leaders in the application of genomic medicine by creating state-of the art genomic medicine education and training programmes that build both capability and capacity
- bring benefit to patients by improved diagnosis and treatment regimes
- stimulate the development of a vibrant UK genomics industry.

This unrivalled knowledge will facilitate clinicians and multidisciplinary healthcare teams in their understanding and application of genomic medicine, leading to better and earlier diagnosis and personalised care.

The value of this programme will be the alignment of the highest fidelity and most comprehensive whole genome DNA sequence produced from patients to date with detailed clinical information stored within a safe haven data infrastructure. This will allow ongoing updates and refreshment of the clinical data from primary, secondary and tertiary NHS care to offer a picture of life-course health and disease progression. After curation and quality control GeL will return findings to NHS healthcare teams caring for patients with these conditions. In addition, there will be access to anonymised builds of the DNA sequence and clinical data within the GeL data infrastructure, as these accrue for clinical and academic research, training and capacity building and for use by industry.

This project is the most ambitious and most advanced of its kind. Partnership with the NHS is essential to ensure success because of its capacity to deliver high-quality initial phenotypic data; a flow of electronic health data from primary care, hospital and social care records; and an opportunity to work with clinicians and patients to acquire further information on conditions, co-morbidities and outcomes. For patients with rare inherited disease, cancer and infectious diseases we will work to construct, maintain and annotate a rich, life course refreshable dataset to enable and accelerate clinical genomics research.

In recognition of its key role as a delivery partner in the 100,000 Genomes Project, NHS England has recently embarked on a process of selecting NHS Genomic Medicine Centres to enable sample acquisition and preparation from patients with rare inherited diseases and cancer to start in January 2015 when the main phase of the project rolls out. Selected centres will enter into an agreement with NHS England on the number of patients to be enrolled into the project and will need to demonstrate strict adherence to standard operating procedures. The implementation of a comprehensive HEE Genomic Education Training programme is therefore central to the success of the NHS to fully engage with the 100,000 Genomes Project.

The Genomics England Clinical Interpretation Partnership

Genomics England is working with key funders to establish the Genomics England Clinical Interpretation Partnership (GeCIP) as a well-defined mechanism for public, charitable or

philanthropic funders, clinicians and researchers and those in training to engage with the project and add value in terms of clinical interpretation and collaborative research.

For further information on the Genomics England 100,000 Genomes Project and Clinical Interpretation Partnership visit; http://www.genomicsengland.co.uk/.

Transforming Genomic Medicine Training for the NHS

Health Education England and Genomics England recognise that there are substantial opportunities for national training programmes to be developed that harness the potential of the 100,000 Genomes Project. We believe these must offer the quality and quantity of engagement with the 100,000 Genomes Project that ensures they will be equipped to operate at the leading-edge of genomic medicine within the NHS. A key element of this is the new Masters in Genomic Medicine which will facilitate access to the data and interpretation pathway as part of a recognised training function with GeCIP.

Masters Programme in Genomic Medicine

Structure and Curriculum

Aim

To establish a new and innovative flexible full- or part-time Health Education England/Genomics England Master's (MSc) programme in Genomic Medicine for training healthcare professionals from different professional backgrounds and groups (e.g. medicine, nursing, scientists and technologists) for whom knowledge of genomics will impact on their service delivery to patients. The aim of the degree is to provide a multi-disciplinary and multi-professional perspective in genomics applied to clinical practice and medical research to enhance knowledge and skills in this rapidly evolving field.

Individual course fees will be funded by HEE for NHS staff that enroll on either the full MSc or any part of the programme. The modular design of the programme will allow stand-alone modules that can be accessible for all staff as part of CPPD.

Commissioning of the Masters Programme in Genomic Medicine

HEE will look to commission HEIs with a geographical spread across England, potentially 6 to 8 institutions which are recognised for research and teaching expertise in this area. They should have a proven track record of teaching and training at postgraduate level and research excellence. HEIs should be able to demonstrate partnerships/potential partnerships with the NHS Genomic Medicine Centres and excellent working relationships with relevant

clinical departments. HEIs will also be required to be part of a national network to ensure a standard approach of HEIs delivering the MSc in Genomic Medicine.

Programme Structure

The proposed programme should have a modular structure and use elements of blended learning as appropriate, which can be delivered flexibly to include the following:

- a full-time option, delivered over one year
- a part-time option delivered over 2 years
- access to individual modules or combination of modules for CPPD, with flexibility of learning offered via core modules and a range of optional modules
- combinations of module credits that can lead to a Postgraduate Certificate (PG Cert 60 credits at FHEQ Level 7) or Postgraduate Diploma (120 credits at FHEQ Level 7)
- a significant research component that can either access the GeL training embassy and involve whole genome sequencing analytics, or a clinically related research dissertation that can carry either 60 or 30 credits. The dissertation could be undertaken in the students hosting NHS department or Genome sequencing facility and could be co-supervised by the host department and HEI.

This MSc in Genomic Medicine is structured such that each student is required to complete 7 core 15 credit modules plus a Research Module which may be 60 or 30 credits. In addition, they will be required to complete a number of optional modules to enable students to tailor the course to their own interests/speciality. In addition, where students elect or are advised by their supervisor to undertake a 30-credit research module instead of the 60 credit research module then they will need to choose additional optional modules in order to achieve 180 credits for the award of an MSc.

Core Modules: 15 credits per module unless otherwise stated

Module 1	An Introduction to Human Genetics and Genomics
Module 2	Omics Techniques and their Application to Genomic Medicine
Module 3	Genomics of Common and Rare Inherited Diseases
Module 4	Molecular Pathology of Cancer and Application in Cancer Diagnosis, Screening, and Treatment
Module 5	Pharmacogenomics and Stratified Healthcare
Module 6	Application of Genomics in Infectious Disease
Module 7	Bioinformatics, Interpretation, Statistics and Data Quality Assurance
Module 8	Research Project (60 credit or 30 credit options, see page 29 for details)

Potential Optional Modules

HEIs can suggest other additional optional modules to reflect the expertise available within the Institution in addition to the core modules.

Module 9	Ethical, legal and social issues in applied genomics
Module 10	Counselling skills for genomics
Module11	Economic models and human genomics
Module 12	Expanding the content of the MSc in Genomic Medicine with workplace-based modules
Module 13	Professional and Research skills
Module 14	Advanced Bioinformatics module

Example of potential module combinations

	Combination 1	**Combination 2**
Core Modules 1-7	7 x 15 credits = 105 credits	7 x 15 credits =105 credits
Research Module	1 x 60 credits = 60 credits	1 x 30 credits = 30 credits
Optional Modules	1 x 15 credits = 15 credits	3 x 15 credits = 45 credits
Total Credits	180 credits	180 credits

To achieve an MSc award, all the modules (180 credits with a minimum of 150 credits at FHEQ Level 7) will have to be successfully completed. HEIs may choose to offer different optional modules, but must put a programme together that includes the core modules.

Entry Requirements

Applicants will be recruited from a range of professional backgrounds, including those with first degrees or equivalent experience, and those with higher degrees. HEIs offering the Masters programme must ensure that their entry criteria will permit applicants from a wide range of health related backgrounds and ensure that candidates satisfy HEI regulations for admission and fitness to pursue and complete the course of study. Entry level guidelines for English Language; a minimum IELTS score of 7.0 is required for the programme. We acknowledge some HEIs may require higher IELTS scores. Where this is the case, the HEI will be expected to indicate what support is available to bring students up to the required level.

Entry Routes

For HEE funded places all candidates must have the support of their employer and the agreement of their Local Education and Training Board.

Award Titles

The title of the degree awarded will be *MSc in Genomic Medicine*.

Relevant Quality Assurance Agency (QAA) Code(s) of Practice

Provider HEIs will adhere to the current QAA Code of Practice for the Assurance of Academic Quality and Standards in Higher Education. Further details can be found on the QAA website: www.qaa.ac.uk/Pages/default.aspx.

Awarding Body

While the full programme may be delivered and awarded by a single HEI provider, collaborative partnerships between HEIs will also be considered and encouraged. It is expected that where collaborative provision is proposed a memorandum of agreement or understanding and contracts would be necessary, with delivery arrangements clearly defined, including the academic and logistical responsibilities of each partner as well as clarification of the financial arrangements between the HEI and its partner. The awarding HEI must be satisfied that the partner institution is able to meet their responsibilities to a satisfactory standard and will be responsible for the overall quality assurance of the programme.

Accreditation of Prior Learning

A process for Accreditation of Prior Learning (APL) that conforms to the guidelines below must be defined by each HEI provider. This must clearly define the minimum and maximum level of APL that will be awarded, the timing, costs and process, and align to statutory requirements for healthcare science. Good practice supports the view that such prior learning should only be used once since double counting is not recommended.

QAA 'Higher education credit framework for England: guidance on academic credit arrangements in higher education in England', August 2008
www.qaa.ac.uk/Publications/InformationAndGuidance/Pages/Highereducation-credit-framework-for-England-guidance-on-academic-creditarrangements-in-higher-education-in-England-Augu.aspx

QAA 'Guidelines on the accreditation of prior learning', September 2004
www.qaa.ac.uk/Publications/InformationAndGuidance/Pages/Guidelineson-the-accreditation-of-prior-learning-September-2004.aspx

Programme Delivery and Monitoring

The tender and subsequent accreditation process will require an HEI to provide a detailed description of the content and educational level of each module and the teaching, learning and assessment strategy to demonstrate how the programme and module aims/learning outcomes will be met.

Programme Delivery

HEIs are expected to ensure that all teaching, learning and assessment is up to date and informed by research to ensure that at graduation, students meet the Framework for Higher Education Qualifications (FHEQ) descriptor at level 7 (www.qaa.ac.uk). By undertaking a research project, students should become aware of the major contribution the workforce makes to research and innovation to benefit patients and the delivery of healthcare.

Academic Induction

It is expected that there will be a period of academic induction at the start of each MSc programme.

Teaching and Learning

It is expected that a blended learning approach will be adopted, based on a model of student-centred adult learning. This should balance and integrate face-to-face teaching, e-learning, etc., and consider the requirements of each student, with learning activities utilised that are appropriate to achieve the desired learning outcomes. Students should be enabled to gain the study skills necessary to manage their own learning, and to exercise initiative, and personal and professional responsibility.

Assessment – Accreditation

The assessment programme should be designed to enable the student to obtain regular constructive feedback on progress and achievement. It should encourage critical reflection and action planning, identifying both strengths and areas for development and improvement. The overall schedule of assessments should be documented in a clear and accessible manner with lines of accountabilities clearly allocated. The assessment programme should also demonstrate how the approach is based on a sound understanding of the evidence base, academic literature and good practice in assessment.

Key areas that must be covered by the assessment programme include:

- A clear statement of accountabilities, including the governance structure for assessment
- The balance between formative and summative assessments

- The assessment of each module, including the weighting of the contribution of individual elements of assessments within the module
- Progression criteria
- The range of valid, reliable and appropriate assessment techniques that will be utilised across the programme and for each module
- The process for providing clear and timely information for students
- How examiners will be trained (including refresher training) and the guidelines that will be given
- How criterion for referencing and standard setting are undertaken
- The nature and timelines for student feedback will be given
- The arrangements for assessment of students with a declared disability
- An assessment blueprint demonstrating the relationship between each assessment and the learning outcomes of the programme
- Exemplar criteria and marking scheme, including critical reflective writing
- The systems used to ensure equivalence of standards and areas of good practice, in addition to the acknowledged external examiner process
- The process of appointing external examiners
- A defined role for external examiners that includes contributing to the review and development of assessment strategies and providing advice from an overarching perspective.

Curriculum

Throughout the degree course students will be expected to gain a range of generic scientific and professional skills including:

- Originality in the application of knowledge, and understanding of how the boundaries of knowledge are advanced through research.
- Apply analytical and synthetic skills to investigate and test new hypotheses
- Integrate information from a variety of sources to construct a coherent thesis on a scientific topic
- Critically appraise and analyse the scientific literature on relevant subject and the ability to judge and interpret findings
- Construct hypotheses pertinent to the experimental exploration of topical questions in the field of medical genomics
- Evaluate the significance of experimental results in the context of previous work
- Engage and communicate effectively with diverse communities including the lay public and professionals involved in research and clinical practice

- Summarise and disseminate information including test results in oral and written forms to colleagues, patients and the public
- Critically evaluate the published literature with respect to the patient and carer perspective of genomic medicine
- Exercise initiative and personal responsibility
- Make decisions in complex and unpredictable situations
- Learn independently as part of a commitment to continuing professional development.

Core Modules

Module 1: An Introduction to Human Genetics and Genomics (15 credits)

Aim

This is the introductory module for the MSc programme, it aims to provide the student with an introduction to the key areas of genomics, human genetics and genetic variation. It will prepare participants to understand disease genetics and how genomic medicine can be utilised to elucidate disease mechanisms and biology. In addition, this module will also cover the fundamentals of Information Governance in the context of genomic medicine and its applications providing underpinning knowledge for later modules in bioinformatics and statistics. This module will serve as a foundation for those wishing to advance their careers within the NHS in genomic medicine.

Content Description

This module will provide clear understanding of the structure and variations in genetic material. Covering basic genetics and genomics, it will prepare participants to understand the role of genetics in disease and how genomic information can be utilised to elucidate disease mechanisms and biology.

The first section 'Genome Structure & Sequence variation' will review the architecture of the human genome and the functional units embedded in it, for example enhancers, promoters, coding exons, untranslated regions, etc. It will then cover DNA sequence variation (e.g. single nucleotide variants (SNVs), insertions and deletions, copy number variants (CNVs) and how variation is structured across the genome, explaining the principles of linkage disequilibrium and its extent in human populations (HapMap project).

The next part 'Biology of Genomes' will cover in more detail aspects of gene regulation (enhancers, promoters, transcription factors, silencers) and chromatin structure (histone modifications; DNase-I hypersensitive sites, open chromatin). It will then discuss genetic control of functional elements introducing the basic principles of quantitative trait loci (QTL) analyses.

Learning Outcomes

By the end of this module the student will be able to:

1. Discuss the human genome structure and the properties of DNA
2. Critique genome architecture and its variation across human populations
3. Critically evaluate the regulation of gene expression, transcription and translation
4. Appraise and interpret variation in genome structure and sequence in the context of physiological function and disease
5. Discuss and analyse epigenetic modifications and imprinting and its role in disease
6. Correlate genetic markers to phenotype and interpret output of association studies both for dichotomous and quantitative traits
7. Discuss and justify the ethical and governance frameworks in place within the NHS and how they apply to medical genomics including patient safety, data sharing and confidentiality
8. Identify the range, purposes, benefits and potential risks of sharing, integrating and aggregating clinical data and information. Describe and evaluate the purpose, structures, use and storage of health records.

Indicative Content

- Architecture of the human genome and genetic variation within it
- DNA sequence variation, type and frequency e.g. single nucleotide variants, small insertions and deletions, copy number variation, rearrangements and tandem repeats
- How variation arises and its extent in populations (e.g. HapMap)
- Gene regulation: enhancers, promoters, transcription factors, silencers
- Epigenetics and imprinting
- Mutational mechanisms: how different types of DNA variants affect gene function or expression to cause disease; correlation of genotype with phenotype
- Concepts of heterogeneity and pleiotropy
- Modes of inheritance for clinical manifestation of human variation
- Legislation, Codes of Practise, Caldicott Guardian and Information Commissioner
- Patient identifiable data and information, relationship between data and information

- Information system risks to patient safety, electronic and paper copies, safe havens, encryption, secondary uses of data, audit and research
- Secure information exchange between professionals
- Sharing and communication with patients and careers, consent
- Handling requests for information about patients /clients.

Module 2: Omics Techniques and their Application to Genomic Medicine (15 credits)

Aim

This module will cover areas of 'omics' technologies, their interpretation and application in key areas of healthcare such as cancer, rare inherited diseases and infectious diseases, as well as research. A specific focus will be on the approaches supporting the 100,000 Genomes Project. This core module will provide the underpinning knowledge to enable students to understand the remaining taught modules and to support those undertaking their research project utilising the 100,000 Genomes Project data sets.

Content Description

This module explores the state of the art genomics techniques used for DNA sequencing (e.g. targeted approaches, whole exome and whole genome sequencing) and RNA sequencing, using highly parallel techniques, together with current technologies routinely used to investigate genomic variation in the clinical setting. This module will introduce the bioinformatics approaches required for the analysis of genomic data, which together with data governance covered in Module 1 will provide a solid foundation for the Bioinformatics and Statistics module. The module will also cover the use of array based methodologies and RNA sequencing in estimating levels of protein expression, micro RNAs and long non–coding RNAs. A comprehensive introduction to metabolomics and proteomics, which are important for the functional interpretation of genomic data and discovery of disease biomarkers will also be included. Students will also learn about the strategies employed to evaluate pathogenicity of variants for clinical reporting.

Learning Outcomes

By the end of this module the student will be able to:

1. Describe and critically evaluate a range of up-to-date genomic technologies and platforms used to sequence targeted parts of the genome or whole genomes
2. Discuss the application of other techniques (for example array comparative genome hybridisation, MLPA, qPCR) commonly used to interrogate genomic variation in the

clinical setting using examples in cancer and rare inherited diseases and infectious diseases

3. Acquire the knowledge of selecting appropriate technology platforms for applications in medical genomics either for research or medical diagnostic purposes
4. Critique how these techniques and their applications in RNA expression can be applied to metabolomics and proteomic analysis
5. Discuss and critically appraise approaches to the bioinformatics analysis and interpretation of 'omics' data
6. Critically evaluate the different 'omics' technologies and platforms and their application to genomic medicine and the impact of personalised medicine
7. Discuss the approaches required to evaluate the pathogenicity of variants identified in whole genome sequencing and other genomic technologies.

Indicative Content

- Basis of genotyping and detection of genetic variation:
  - Whole exome and whole genome sequencing, including library preparation methods, sequencing chemistries and platforms
- Brief overview of methodologies for detecting base substitutions (SNV), small insertions and deletions (indels), copy number variants (CNV) or rearrangements, to include Sanger sequencing, pyrosequencing, ARMS, MLPA, qFPCR, microarray
- Genomic testing strategies as: gene focused, multiple genes, or whole genome or exome, and for detection of sequence, copy number or rearrangements
- Additional techniques: RNA expression profiling (expression array) and RNA sequencing, metabolomics; proteomics techniques
- Overview of bioinformatic approaches to the analysis of genomic data
- Approaches to the evaluation of pathogenicity of variants in the context of an NHS clinical report.

Module 3: Genomics of Common and Rare Inherited Diseases (15 credits)

Aim

The number of rare monogenic disorders is estimated to be greater than 7,000, but only in approximately half of these are the underlying genes known. Common diseases such as intellectual disability, diabetes, schizophrenia and autism are thought to arise from a complex interplay of genetic and environmental factors but deeper understanding of the genetic and mechanistic basis of these diseases is necessary for clinical translation.

The aim of this module is to provide a brief introduction to the clinical presentation and manifestations of rare inherited and common diseases and consider the patient and family perspective with respect to the role and impact of genomics. The module will also offer a comprehensive overview of the traditional and current strategies and techniques used to identify genes responsible for both common multifactorial and rare inherited diseases, focusing mainly on the latter. Building further on the techniques covered in module 2, students will learn how to identify disease phenotypes and how to select cases with unmet diagnostic need that will benefit from either exome or whole genome sequencing.

Indicative Content

This module will initially explore the clinical presentation and course of a range of common and rare inherited diseases. The principles and practise of medical genetics, and the management and treatment of probands and their families will be discussed. In addition, the role of genomics in a care pathway will be examined including the patient and family perspective.

The students will then learn about the Genomics England 100,000 Genomes Project and data infrastructure and through practical examples learn how to select cases with unmet diagnostic need that will benefit from exome or whole genome sequencing. Building on knowledge gained in Module 2, students will further explore the analytical challenges in genomics as applied to rare inherited diseases.

This MSc module will explore the traditional and current approaches used to identify genes responsible for common and rare inherited diseases, focusing on the latter.

Learning Outcomes

By the end of this module the student will be able to:

1. Examine the landscape of common and rare inherited diseases
2. Explain the genetic architecture of common and rare inherited diseases
3. Critically evaluate traditional and current approaches used to identify genes for common and rare inherited diseases
4. Synthesise information gained from exome / whole genome analysis with patient information / medical records to determine diagnosis, penetrance or prognosis for a number of examples of common and rare inherited conditions
5. Discuss and evaluate the Genomics England Programme and the Data Infrastructure
6. Identify phenotype, select cases and relevant family information for whole exome or whole genome based approaches for hypothesis free whole exome or whole genome sequencing

7. Discuss and critically evaluate the implications of patient access to their medical records and clinical information for medical genomics, inter-professional practice and multidisciplinary care.

Indicative Content

- Clinical presentation and course of a range of rare inherited and common diseases
- Principles and practise of medical genetics; risk stratification and management of patients and their families
- Approaches and techniques used to identify genes responsible for common and rare inherited diseases (e.g. candidate gene, positional mapping, genome wide association studies, exome / whole genome sequencing, use of population data sets)
- Basic statistics to aid interpretation of GWAS and analysis of populations
- The Genomics England 100,000 Genomes Project and data infrastructure
- Selection of tractable cases with unmet diagnostic need suitable for whole genome analysis
- Analytical challenges in genomics as applied to rare inherited diseases including:
    - the potential of electronic health records to enrich patient data
    - importance of phenotyping and use of databases such as ClinVar
    - use of large population datasets
    - sharing information e.g. Human Variome Project
- Impact of patient on-line access to their health records, test results etc. on medical genomics.

Module 4: Molecular Pathology of Cancer and Application in Cancer Diagnosis, Screening, and Treatment (15 credits)

Aim

This module will equip the student with detailed knowledge and understanding of the molecular mechanisms involved in cancer development. This will include the ways in which interrogation of a person's own genome and the genome of tumour cells can facilitate the diagnosis and treatment of cancer.

Content Description

This module covers the molecular mechanisms that underlie cancer development, growth and metastasis, and the differences between different cancers. It will explore the different molecular and cellular actions of anti-cancer treatments, the genomic factors affecting response and resistance to treatment, and the research approaches to anti-cancer drug design and development. Broad situations which confer a high cancer risk to a person

and/or to other members of the same family will be discussed in the context of how genomic information may be integrated into cancer screening programmes. This module will prepare the students to interrogate the cancer data sets from the 100,000 Genomes Project.

Learning Outcomes

By the end of this module the student will be able to:

1. Apply the principles of cancer development and emerging changes in classification
2. Compare and contrast the genomic basis of cancer predisposition, and how this is used to identify people and families at higher risk of cancer
3. Critically evaluate how genomic information is currently applied in the diagnosis, classification, treatment selection and monitoring of cancer (e.g. leukaemia, breast, melanoma, lung cancers)
4. Analyse how information from exome and whole genome analysis of tumour tissue can be used to investigate the molecular and cellular processes leading to cancer and inform strategies for drug development.

Indicative Content

- Tumour classification systems
- Cellular properties of tumours: growth, division, invasion, aberrant hormone or toxin production, immunogenicity
- Factors in tumour formation: molecular mechanisms and role of microenvironment, molecular signatures & changing classification
- Diagnosis, molecular sub-classification, aggressiveness (prognosis) characterisation of metastases
- Monitoring disease following treatment (medical, surgical or bone marrow transplant)
- Genomic testing of cell free tumour DNA in blood, for diagnosis and monitoring of solid cancers
- Importance of sample quality for tumour genomic analysis
- Molecular basis of single gene subsets; research evidence (co-segregation studies) identifying sequence alterations (single gene Sanger sequencing and NGS panel tests); how to interpret molecular results for pathogenicity – literature, databases, & *in silico* tools
- Other molecular predisposition; GWAS studies; other predisposition biomarkers
- Environmental factor and lifestyle predisposition and protection; molecular action; genomic interaction; epigenetic factors
- Genomic and cellular markers and optimal treatment regimes:
    - in haematological cancer

- in solid tumours
- Companion diagnostics in cancer
- Breakthrough tumour /metastases and molecular mechanisms.

Module 5: Pharmacogenomics and Stratified Healthcare (15 credits)

Aim

Pharmacogenomics and stratified health care ensure that healthcare professionals offer the 'right treatment, for the right person, at the right time' is a fast developing area. This module will provide a comprehensive overview of the analytical strategies and techniques used in pharmacogenomics and explore some of the challenges and limitations in this field (e.g. availability of patient material for studies of adverse drug reactions which tend to be rare, allelic heterogeneity between different ethnic groups, patient compliance etc.). Biomarkers are the predictive tools for optimising drug response and preventing adverse drug reactions thus this module will also provide an overview of the different type of genomic biomarkers currently in use or emerging.

Content Description

Pharmacogenomics is playing a key role in our health care system. This module will describe the complexity of pharmacogenomics and the effect of medication on individuals based on their genetic make-up i.e. tailoring drug treatment to improve patient response and techniques to stratify patients at risk of adverse drug reactions. The module will use examples of known validated pharmacogenomic tests relevant to the use of drug treatments.

Learning Outcomes

By the end of this module the student will be able to:

1. Discuss and evaluate the mechanism of several examples of genomically-determined differential drug response, and drug reaction
2. Appraise the strategies and analytical approaches for stratifying patients for optimal drug response or adverse drug reactions including ethnic differences, and how these translate into 'companion diagnostics'
3. Identify and analyse the challenges and limitations of pharmacogenetic studies
4. Identify and evaluate the different types of current and emerging biomarkers used in personalised medicine
5. Discuss and critically evaluate how genomic information can enable development of drugs targeted for particular genotypes

6. Identify the ethical, legal and social issues (ELSI) that could accompany patient stratification for healthcare advice or intervention and defend the use of patient stratification to improve the diagnosis and treatment of disease.

Indicative Content

- Genomic basis of: drug reaction, drug efficacy, ethnic differences in both these; and how these are applied in prescribing practice
- Use of genomic information, for targeted drug development
- Companion diagnostics and options for NHS service delivery models
- Different types and examples of genomic-targeted intervention (examples of genomically-targeted clinical, therapeutic or lifestyle choices)
- Genomic biomarkers: SNPs, variability of short sequence repeats, haplotypes, DNA modifications, e.g. methylation, deletions or insertions, copy number variants, RNA expression levels, RNA splicing, microRNA levels
- Use of biomarkers in treatments other than cancer.

Module 6: Application of Genomics in Infectious Disease (15 credits)

Aim

From this module the student will understand how genomics can be used to provide more accurate diagnosis, predict which drugs are likely to be more effective and monitor treatment and control of infectious disease in individuals and populations.

Content Description

The student will learn about the genomic structure of infectious agents, implication of acquisition or loss of nucleotides, genes and plasmids on pathogenicity, sensitivity of a pathogen to drug treatment and response to the host.

Learning Outcomes

1. Demonstrate an in-depth knowledge of and explain the differences between prokaryote and eukaryote genomes
2. Discuss and appraise how the genome sequence of pathogens can be used to track cross infection and outbreaks of infections among the population
3. Review the emerging action of drugs in controlling infection e.g. HIV, TB
4. Critique the molecular basis of organism drug resistance in some infections and how this directs drug research

5. Evaluate how sequencing of the genome of infective organisms can be used in infectious disease for assessing: diagnosis, sub-classification & strain identity, pathogenicity, drug resistance and drug selection; and for epidemic control.

Indicative Content

- Infection as a cause of national and global morbidity and mortality
- Transmission of human infections: person to person, food and waterborne, sexually transmitted, vector-borne
- Prokaryotes, their genome, replication and population genetics
- Genomic characterisation of viruses: DNA and RNA genomes, single-stranded, double stranded, segmented
- Genomic comparisons of microbial strains in the context of outbreaks and transmissions in hospitals and the community
- Anti-infective drug action
- Mutation rate & drug resistance
- Genomic evidence of individual susceptibility to specific infection
- Role of genomics in: infectious disease diagnosis, prognosis, drug selection, resistance, monitoring, epidemic control, drug research.

Module 7: Bioinformatics, Interpretation, and Data Quality Assurance in Genome Analysis (15 credits)

Aim

The main challenge for application of genomic data is in its analysis and interpretation. The aim of this module is to enable students to gain the knowledge and understanding required to critically interpret existing genomic research, and develop the skills to formulate their own research questions as well as to collect, analyse and interpret their own NHS data using a basic range of statistical and bioinformatics techniques.

Content Description

The module will cover the fundamental principles of informatics and bioinformatics applied to clinical genomics, find and use major genomic and genetic data resources; use software packages, *in silico* tools, databases and literature searches to align sequence data to the reference genome, critically assess, annotate and interpret findings from genetic and genomic analyses. Theoretical sessions will be coupled with practical assignments of analysing and annotating predefined data sets.

This is a core module that is central to the MSc programme in Genomic Medicine as it will provide the student with the skills to analyse genomic data. Upon completion of this module students will be eligible to base their MSc research project on data from the 100,000 Genomes Project data set.

Learning Outcomes

By the end of this module the student will be able to:

1. Analyse the principles applied to quality control of sequencing data, alignment of sequence to the reference genome, calling and annotating sequence variants, and filtering strategies to identify pathogenic mutations in sequencing data
2. Interrogate major data sources, e.g. of genomic sequence, protein sequences, variation, pathways, (e.g. EVS, dbSNP, ClinVar, etc.) and be able to integrate with clinical data, to assess the pathogenic and clinical significance of the genome result
3. Acquire relevant basic computational skills and understanding of statistical methods for handling and analysing sequencing data for application in both diagnostic and research settings
4. Gain practical experience of the bioinformatics pipeline through the Genomics England programme.
5. Justify and defend the place of Professional Best Practice Guidelines in the diagnostic setting for the reporting of genomic variation.

Indicative Content

- Aligning genome data to reference sequence using up to date alignment programmes (e.g. BWA)
- Assessment of data quality through application of quality control measures
- How to determine the analytical sensitivity and specificity of genomic tests
- Use of tools to call sequence variants e.g. GATK, annotation of variant-call files using established databases
- Filtering strategies of variants, in context of clinical data, and using publically-available control data sets
- Use of multiple database sources, *in silico* tools and literature for pathogenicity evaluation, and familiarity with the statistical programmes to support this
- Principles of integration of laboratory and clinical information, and place of best-practice guidelines for indicating the clinical significance of results
- Principles of downstream functional analysis e.g. knock-outs, and other cellular model
- How to analyse genomic data to identify epigenetic and other variation that modifies phenotype

- Practice in examples of analysis of genomic data in the Training Embassy within the Genomics England Data Centr**e.**

Module 8: Research Project (60 or 30 credit options)

The research module can be approached by two different routes depending on student preference: a full research project of 60 credits or a literature review dissertation of 30 credits. If the latter is chosen, additional optional modules must be undertaken to fulfil the requirements of 180 credits for a Masters degree.

Research Project (60 credit option)

Aim

The aim of this module is for students to build on their previous knowledge, skills and experience of undertaking research by undertaking a medical genomics research project that shows originality in the application of knowledge, together with a practical understanding of how established techniques of research and enquiry are used to create and interpret knowledge in a specialism of healthcare science. Research projects should be designed to take into account the research training required by individual students and the needs of the department in which the research is to be conducted. The research dissertation should be presented in the form of a scientific report to be considered for publication in a scientific journal, i.e. use of sub-headings, tables, figures, references, etc. This can be based on a workplace project or the Genomics England dataset.

Content Description

Students will use both the theoretical knowledge they will acquire throughout the taught part of the course and the analytical skills they will develop in order to tackle a research question by themselves. Undertaking of the research project will involve formulating the question, acquiring and analysing the data and finally present and discuss results. The project could be carried out in the hosting NHS laboratory, research department and industry under joint supervision i.e. tutors from both the hosting department and the programme. Research projects should be presented in the format of a paper for publication (additional figures and tables can be presented as supplementary material).

Learning Outcomes

By the end of this module the student will be able to:

1. Justify the rationale for research governance and ethical frameworks when undertaking research or innovation in the NHS

2. Identify a research question and critically evaluate the novelty and importance of the research question
3. Design, plan and undertake a research project to test a hypothesis from conception to completion/archiving in accordance with ethical and research governance regulations drawing on expert advice where necessary and involving patients and service users
4. Analyse the data using appropriate methods and statistical techniques, and interpret, critically discuss and draw conclusions from the data
5. Prepare a written report that describes and critically evaluates the research project, clearly identifying the strengths and weaknesses
6. Present a summary of the research project and outcome that conforms to the format of a typical scientific presentation at a national or international scientific meeting, responding to questions appropriately
7. Prepare and present a summary of the research project to specialist, non-specialist and lay audiences seeking feedback on the presentation(s) and critically reflecting on the experience and feedback.

Literature Based Project (30 credit option)

Aim

The aim of this module is to allow students to carry out an in depth literature based project on specific subjects e.g. cardiovascular genomics or epigenetics. This module could potentially provide a 30 credit module but then must be done in conjunction with additional optional modules to attain the total of 180 credits.

Optional Modules

These are suggestions for optional modules and HEIs can submit additional modules.

Module 9: Ethical, Legal and Social Issues in Applied Genomics (15 credits)

Aim

The module aims to provide a framework for ethical understanding of medical genomics. Students will be provided with a platform of ethical understanding from which to consider issues of human confidentiality, autonomy, disclosure, informed consent and natural justice. Upon this platform, students will consider the impact of genomic technologies on individual lives and those of demographic and ethnic groupings. The social implications of the availability of genetic testing and screening will be considered, especially in the context of reproductive technologies. Finally students will be provided with a legal framework for

patenting of genetic information as well as the use of genetic data for research, diagnostic and therapeutic purposes.

Content Description

Many genomic tests have wider implications for the patient and their family, particularly where these may have a predictive aspect, provide incidental information, have potential for being misleading or increase uncertainty. The student will explore the ethical, legal and social issues (ELSI) involved in genomic testing and in specific integrated pathways.

Learning Outcomes

By the end of this module the student should be able to:

1. Explain and defend the ethical principles of confidentiality, autonomy, disclosure, informed consent and natural justice to scenarios within genomic medicine in a global context
2. Critically evaluate the challenges of emerging genomic technologies on society and societal values
3. Critically appraise the current legal framework within which genomic medicine in practised in the UK, Europe, USA and globally
4. Identify and critically evaluate the ethical, legal and social impact of genomic medical advances in a clinical pathway
5. Critically evaluate and defend the principles of the Helsinki Declaration under which all medical research is performed with specific reference to genetic research
6. Critically appraise the global impact of genetic databases on human autonomy, healthcare provision and discrimination
7. Identify those clinical pathways in their own specialty or field of practice where the use of genomic investigation/techniques may be beneficial to patients, and be able to argue for those benefits.

Indicative Content

- ELSI issues for genomic medicine – confidentiality, autonomy, disclosure, informed consent, natural justice
- Effects of opportunistic predictive, incidental or uncertain-significance results
- Impact of genomic testing on individuals, and demographic and ethnic differences
- Social implications of genetic testing and screening, particularly in the context of human reproduction
- Legal aspects of genomic information in relation to diagnostic, therapeutic, and research use and patenting of genomic information and tests
- Obtaining ethical approval for research projects

- Specific ELSI issues as applied to clinical pathways.

Module 10: Counselling Skills for Genomics

Aim

The aim of this module is to equip students with the knowledge, communication and counselling skills and appropriate attitudes and behaviours towards the diagnosis and management of patients whose care will be influenced by genomic investigations.

Content Description

This is an introduction to counselling skills for genomics. Students undertaking this module will be taught how to communicate and provide appropriate support to individuals and their families. Development of counselling skills will be achieved via theoretical and practical sessions. Students will understand the importance of a family history and communication of pathogenic and/or uncertain results.

Learning Outcomes

By the end of this module the student will be able to:

1. Explain and justify the importance of and application of informed consent in the field of genomic medicine generally and as applied to the 100,000 Genomes Project
2. Understand the different purposes of genomic testing in patients with rare inherited diseases, cancer and infectious diseases
3. Explain genomic results in terms of diagnosis prediction and uncertainty
4. Acquire an understanding of the skills necessary to support individuals who have genomic results that affect their care
5. Critique the concepts of genetic and genomic predispositions to illnesses
6. Critique the consequences of genomic test results on the patient and the wider family including incidental findings
7. Analyse and discuss the communication and counselling skills needed to engage and communicate effectively in a compassionate manner with patients
8. Demonstrate a critical understanding of screening pathways used to test for inherited and acquired disorders
9. Demonstrate an in depth understanding of the ethical, legal and social issues around genome testing and whole genome sequencing.

Indicative Content

- Consent and what it means in relation to the 100,000 Genomes Project
- Ethical and social implications of genomic testing

- How to record and interpret a family history, recognising what is or may be relevant
- How to verify personal and family history information; consent, confidentiality, access to records
- Different purposes of genomic testing
- Approaches to prenatal testing, pre-implantation testing (PGD) and pre-conception carrier screening in relation to new technologies
- Strategies of approach to lifelong patient management of whole genome information
- Managing and explaining complex genome results
- Sources for patient support: patient support groups, on-line resources, other resources
- Communication and counselling skills
- How to access and use patient databases e.g. Decipher.

Module 11: Economic Models and Human Genomics

Aim

With the UK government's effort to develop national clinical guidelines to secure consistent, high quality, evidence-based medicine predicated on outcomes and cost, it is important that students understand the role of economic modelling in the decision-making process. The module will explore the potential impact of genomic technologies on the healthcare system. Economic models will be used to demonstrate the anticipated costs and benefits of new technological approaches.

Content Description

Technological advances in the area of genomic medicine have led to new tests with major impact on improving disease diagnosis and effectiveness of treatments. However, the continuous growth in the use of genomic technologies has cost implications. Using established economic models it is possible to predict the costs of new treatments and assess benefits to patients in the context of the available resources for health care. In addition, this module will explore the factors that determine the effects of the rapid development of genomics on health care systems covering the role and relative influence that government, doctors and the public exert in this process. These will be analysed to assess whether clients/patients are best served by current arrangements and whether people's health outcomes match reasonable expectations. Students will be encouraged to propose ways of tackling perceived shortcomings.

Learning Outcomes

Understand the importance of economic models in the field of medical genomics. By the end of this module the student should be able to:

1. Appreciate the role of 'quality of life' studies in outcome evaluation
2. Make informed choices between treatment options based on measures of disease burden such as quality-adjusted life years and disability-adjusted life years
3. Critique cost-minimization analysis (CMA) in relation to genomic medicine
4. Critique cost-effectiveness analysis (CEA) in relation to genomic medicine
5. Critique cost-utility analysis (CUA) in relation to genomic medicine
6. Critique cost-benefit analysis (CBA) in relation to genomic medicine
7. Acquire and apply knowledge in economic models and human genomics
8. Evaluate issues around the cost of health interventions
9. Critique issues around the measurement benefits in healthcare.

Indicative Content

- Evaluate the significance of economic models in genomic medicine
- Display skill in selection of appropriate economic models and understanding of their effect on the public health
- Summarise, critically analyse and disseminate results of appropriate tests
- Ability to justify economic based decisions in the field of genomics.

Module 12: Expanding the Content of the MSc in Genomic Medicine with Workplace-based Modules

There is a potential to consider a work-based module which could explore genomic practice in an individual's work base. This could be in the form of a short case based portfolio of study.

Suggested generic learning outcomes:

- Critically evaluate existing practice in genomic medicine
- Devise and critique methods and approaches in genomic medicine
- Apply technical expertise to a range of cases, then evaluate and reflect on the utility of these technologies to the practise of genomic medicine techniques
- Apply clinical expertise to a range of cases and reflect on the utility of the genomic testing.

Module 13: Professional and Research Skills

Aim

The overall aim of this module is to ensure that the trainee has the underpinning knowledge of the importance of research, development and innovation across the NHS and in healthcare science in particular and to provide the underpinning knowledge for the research project.

Content Description

This module is designed to give the student a thorough background to the principles of scientific research to equip them to undertake relevant research within the NHS, research and industry departments, as an individual and as part of a research team. Using this knowledge the student will be able to undertake relevant scientific research within the NHS, students will learn about the importance of ethics for a project and understand the legal framework in which research with patients and/or patient samples must be undertaken. Students will develop the skills to present their research both to fellow healthcare professionals and to non-scientific audiences. The process of developing clinical guidelines from research results will be explored.

Learning Outcomes

By the end of this module the student should be able to:

1. Critique the context within which research and audit are undertaken within the NHS and examine the contribution of the healthcare workforce to undertaking cutting-edge translational research for patient benefit and promoting innovation within the NHS
2. Evaluate the difference between audit and research and know different types of research approaches including qualitative, quantitative and systematic review
3. Appreciate the issues regarding current ethical approval processes for research and audit, the requirements for continuous monitoring, progress reporting, adverse event monitoring, study closure and archiving
4. Appraise how clinical guidelines are produced and the concept of evidence based practice including the role of current statutory and advisory regulatory bodies.

Indicative Content

- Prepare a project proposal including the ethical approval mechanism in the NHS
- Present research findings in oral and written forms including scientific presentations
- Critically review the literature to establish current knowledge with respect to a research question and summarise the findings

- Identify and discuss an audit project that has resulted in change specific to their specialism
- Identify and discuss a research study that has resulted in an improvement in patient care relevant to their specialism.

Other Potential Optional Modules:

- Advanced Bioinformatics
- Epigenetics
- Clinical based projects / modules e.g. cardiovascular disease