Masters (MSc)

Postgraduate Certificate (PG Cert)

Postgraduate Diploma (PG Dip)

in

Genomic Medicine

COURSE INFORMATION

(2015/2016 INTAKE)

National Heart and Lung Institute
Faculty of Medicine
Imperial College, London

The Graduate School

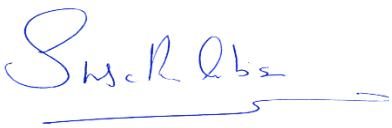
Welcome from Professor Sue Gibson, Director of the Graduate School

The Graduate School has several roles but our main functions are to provide a broad, effective and innovative range of professional skills development courses and to facilitate interdisciplinary interactions by providing opportunity for students to meet at academic and social events. Whether you wish to pursue a career in academia, industry or something else, professional skills development training will improve your personal impact and will help you to become a productive and successful researcher.

Professional skills courses for Master's students are called "Masterclasses" and they cover a range of themes, for example, presentation skills, academic writing and leadership skills (http://www3.imperial.ac.uk/graduateschool/currentstudents/professionalskillsmasters/masterclassprogramme). All Masterclasses are free of charge to Imperial Master's students and I would encourage you to take as many as you can to supplement your academic training. The Graduate School works closely with the Graduate Students' Union (GSU) and is keen to respond to student needs so if there is an area of skills training, or an activity that you would like us to offer, but which is not currently provided, please do get in touch (graduate.school@imperial.ac.uk).

The Graduate School also runs a number of exciting social events throughout the year which are an opportunity to broaden your knowledge as well as to meet other students and have fun. Particular highlights include the Ig Nobel Awards Tour Show, the Chemistry Show and the 3 minute thesis competition. You should regularly check the Graduate School's website and e-Newsletters to keep up to date with all the events and training courses available to you.

Finally, I hope that you enjoy your studies here at Imperial, and I wish you well.



Sue Gibson

Welcome from Dr Janet De Wilde, Head of Postgraduate Professional Development

I would like to welcome you to the graduate school courses for postgraduate professional development. The team of tutors here come from a wide variety of experiences and we understand just how important it is to develop professional skills whilst undertaking postgraduate studies and research. Not only will this development improve success during your time at Imperial College, but it will also prepare you for your future careers. We are continually working to develop and innovate the courses we offer and over this year you will see many new offerings both face to face and online. I encourage you to explore and engage with the diverse range of opportunities on offer from the team at the graduate school and I wish you well in your studies.

Janet De Wilde

Welcome from the MSc Genomic Medicine Course Director: Professor Michael Lovett

m.lovett@imperial.ac.uk

Welcome to the new MSc programme in Genomic Medicine at Imperial College London. Completion of this course can lead to the Masters in Science (MSc) degree, a Postgraduate Diploma (PG-DIP) or a Postgraduate Certificate (PG-CERT) in Genomic Medicine.

Genomic approaches are increasingly impacting upon health care. In the near future we will have multiple types of genomic data from hundreds of thousands, if not millions, of individuals world-wide. We are in a new era of personalised and stratified health care for many human diseases, based upon genomic data. We aim to educate students from a wide range of backgrounds in how to apply genomic approaches and how to interpret the outputs from those analyses. This is a rapidly evolving field, so we also want to provide students with the skills to continue to learn and understand new approaches as they become relevant to clinical practice. We provide the highest level of training in all aspects of genomic science and we provide it in one of the most cutting-edge scientific environments in the world. I hope you enjoy the innovative and flexible training programme that we have devised.

Michael Lovett

Genomic Medicine

Background to the course

This Master's level programme educates students from a wide range of backgrounds (e.g. medicine, nursing, healthcare scientists, basic scientists and technologists) to interpret and understand genomic data that increasingly impacts on service delivery to patients. The programme is flexible and modular and includes full- and part-time MSc options, delivered over one or two years respectively. There are also full-time and part-time Postgraduate Certificate (PG Cert) or Postgraduate Diploma (PG Dip) options. If choosing the full-time PG Cert or PG Dip mode of study students should be aware that module choices will be more limited. Students will be made aware of their options during the admissions process.

The aim of the programme is to enhance knowledge and skills in this rapidly evolving field by providing a flexible, multi-disciplinary and multi-professional perspective in genomics applied to clinical practice and medical research. In so doing, it fulfills the requirements of Genomics England, Health Education England and Public Health England for MSc, PG Dip and PG Cert programmes to transform the NHS workforce in readiness for the100,000 Genomes Project, set out in a tender in 2014. Imperial College was successful in its bid to run the programmes and has been designated a preferred provider by Health Education England and our course values and upholds

the NHS Constitution.

The programme comprises core and optional taught modules of 7.5 ECTS each that will be taught using a blended approach (direct teaching and online distance learning) to provide flexibility for health professionals to combine their study with NHS and Public Health service duties. The MSc programme also includes a core research module (30 ECTS) with opportunities to access the emerging data from the 100,000 Genomes Project through the Genomics England Clinical Interpretation Partnership (GeCIP) training domains.

Most modules consist of one week of face-to face teaching and up to three weeks of eLearning and independent study (exceptions are noted below). Teaching will be delivered across various College Campuses, including the Royal Brompton Campus, the St Mary's Campus, Hammersmith Campus and South Kensington Campus.

The modules are offered on a cycle of 12 months, so that all modules become available at least once every 12 months. There are two student intakes, once in October and once in March. However, the full-time MSc option is only available to students who start in October. Before students start on either intake date, they are required to take a short on-line pre-sessional refresher course (not for credit) that covers the basic molecular genetics necessary to begin the programme. The first Core module in the programme is "Core Concepts in Human Genetics & Genomics" that all students must take. It is offered twice in each 12 month cycle corresponding to the two intake dates.

The programme includes collaborations with the Institute for Cancer Research (providing the Core Cancer Genomics Module), Buckinghamshire New University (collaborating on the Optional Workplace-Based Module), and Brunel University London (providing the Optional Economic Evaluation in Human Genomics Module). The Institute of Cancer Research and Brunel may also co- supervise research projects.

Introduction to the Programme Leaders

Professor Michael Lovett (m.lovett@imperial.ac.uk**)** is Chair in Systems Biology within NHLI at Imperial College, London. He is based at the Royal Brompton Campus and is Course

Director for the Programme in Genomics Medicine and module lead in the Core Bioinformatics module, the Optional Laboratory Skills module and co-lead of the MSc Research Project.

Dr Claire Shovlin (c.shovlin@imperial.ac.uk) is Reader in Clinical and Molecular Medicine within NHLI at Imperial College London. She is based at the Hammersmith Campus and is module lead in the Core Concepts module, Core Genomics and the Patient module, and co-module lead on the Optional Workplace-Based module.

Professor Rosalind Eeles (Ros.Eeles@icr.ac.uk) is Head of Oncogenetics at The Institute of Cancer Research (ICR) and Honorary Consultant at the Royal Marsden. She is module lead of the Core Molecular Pathology of Cancer module.

Dr Fiona Culley (f.culley@imperial.ac.uk) is Lecturer in Respiratory Infectious Diseases within NHLI at Imperial College London. She is based at the St Mary's Campus and is module lead for the Core Genomics of Infectious Diseases module.

Dr. Inga Prokopenko (i.prokopenko@imperial.ac.uk) is Senior Lecturer in Human Genomics within the School of Public Health at Imperial College London. She is based at the Hammersmith Campus and is module lead for the Core Omics Technologies module.

Professor Geraldine Thomas (geraldine.thomas@imperial.ac.uk) is Chair in Molecular Pathology in the Department of Surgery & Cancer at Imperial College London. She is based at the Charing Cross Campus and is lead for the Core Ethical, Legal and Social Issues module and co-lead of the MSc Research Project.

Dr Deborah Morris-Rosendahl (d.morris-rosendahl@imperial.ac.uk) (left) is Head of Clinical Genetics and Genomics at the Royal Brompton campus at Imperial College London. **Dr Anna Need** (a.need@imperial.ac.uk) (right) is Lecturer in Neuropsychiatric Genetics within the Department of Medicine, at Imperial College London. She is based at the Hammersmith Campus. Together they co-lead the Genomics of Common and Rare Inherited Diseases module.

Professor Uta Greisenbach (u.griesenbach@imperial.ac.uk) is Professor of Molecular Medicine within NHLI at Imperial College, London. She is based at the Royal Brompton Campus and is lead for the Optional Genome-Based Therapeutics module.

Dr Letizia Foroni (l.foroni@imperial.ac.uk) is Principal Teaching Fellow within the Department of Medicine, at Imperial College London. She is based at the Hammersmith Campus and is lead for the Optional Pharmacogenomics and Stratified Medicine module.

Mr Steven Pearce (Steven.Pearce@bucks.ac.uk) is Principal Lecturer within the Department of Advanced & Continuing Professional Development at Bucks New University. He is co-lead on the Optional Workplace-Based module.

Dr Subhash Pokhrel (subhash.pokhrel@brunel.ac.uk) is Senior Lecturer in the Institute of Environment, Health and Societies at Brunel University London. He is lead for the Optional Economic Evaluation in Human Genomics module.

Dr. Amir Hakim (a.hakim08@imperial.ac.uk) is a Research Associate within NHLI, Imperial College London. He is lead for the Professional and Research Skills E-learning module.

Other Contacts within the Programme

NHLI Education Administrator: Miss Ellie Wilde

NHLI Education Manager: Ms Eleanor Tucker

NHLI Director of Education: Professor Sue Smith

NHLI Deputy Director of Education: Dr Duncan Rogers

Head of Institute: Professor Kim Fox

Institute Manager: Dr Jane Evers

Institute Lead for Women: Professor Sian Harding

Institute Lead for Outreach: Professor Sara Rankin

Other key individuals are listed here: http://www.imperial.ac.uk/nhli/contacts/

Course overview

What courses are offered?

Master's Programme (90 ECTS)

All students will participate in 7 compulsory Core modules (see below for details) plus one Optional module of their choice (from a total of six options, see below). **Each Module is four weeks in length and in most cases this consists of one week of face-to-face teaching and three weeks of distance e-learning**. The students will then conduct a 4 month ***research project***, which includes preparation of a report and a oral presentation of their research findings. In some cases students may be able to conduct their research projects at their place of work, if the project meets the required standards, has an Imperial College co-supervisor and is approved by Professors Lovett and Thomas. All modules carry 7.5 ECTS and the project carries 30 ECTS.

Core Modules

Core Concepts in Human Genetics and Genomics
Molecular Pathology of Cancer and Application in Cancer Diagnosis, Screening and Treatment (delivered by the Institute of Cancer Research)
Application of Genomics in Infectious Disease
Omics Technologies and their Application to Genomic Medicine
Ethical, Legal and Social Issues in Applied Genomics
Genomics of Common and Rare Inherited Diseases
Either Bioinformatics, Quality Control, Analysis & Interpretation of Genome Sequencing Data
OR Genomics and the Patient

Elective/Optional Modules
Pharmacogenomics and Stratified Medicine
Economic Evaluation in Human Genomics (delivered by Brunel University London)
Laboratory Skills for Genomics
Genome-Based Therapeutics
Professional and Research Skills (eLearning module)
Workplace-Based Module (all distance learning) (co-delivered with Buckingham New University)

Postgraduate Diploma (60 ECTS)

The PG-Dip degree consists of successful completion of the 7 Core modules plus one Optional module as shown above. It does not require completion of a research project.

Postgraduate Certificate (30 ECTS)

The PG-Cert degree consists of successful completion of four modules, of which one must be the Core Concepts module and 3 further modules, 1 of which can be from the optional list above.

Short Courses

Modules will also be offered as Short Courses. Modules run by Imperial College will be part of the Short Course portfolio with the Short Course Quality Committee (SCQC)'s accreditation (http://www3.imperial.ac.uk/cpd/qualityassurance).

Registration

Students will register for the Master's programme or the Postgraduate Diploma (PG Dip) or the Postgraduate Certificate (PG Cert). Students who register for the PG Dip or PG Cert in the first instance can transfer to the part-time (2-year) MSc, subject to satisfactory performance in the coursework assessments. Alternatively, they may take the Diploma or Certificate and return to the MSc programme in a subsequent academic year.

What are the entry criteria?

An upper second class (2.1.) honours degree in a relevant biological subject from a UK university equivalent.

A candidate with a degree below the 2.2 entry requirement, but who has at least three years relevant work experience after graduation and has two supportive references on file may be considered under the College's special circumstances policy.

The programme will not be suitable for entrants without degree level knowledge of a relevant biological subject

Applicants will be required to meet the College's standard English proficiency requirements for postgraduate students.

Information for students who already hold the PG Cert or PG Diploma:

For the direct entry route to the PG Diploma students wishing to attend on a full-time or part-time basis will have to have successfully completed the PG Cert.

For the direct entry route to the MSc, students wishing to attend on a part-time basis will have to have successfully completed the PG Diploma.

For the direct entry route to the MSc, students wishing to attend on a full-time basis will have to have completed either the PG Cert or PG Diploma

How long does course take to complete?

This is a modular, flexible programme.

For MSc: 1 year full-time (12 months), 2 years part-time (24 months)

PG Diploma (PG Dip): 8 months full-time, 2 years part-time (minimum 9-maximum 24 months continuous enrolment)

For PG Certificate (PG Cert): 4 months full-time 1 year part-time (minimum 5-maximum 12 months continuous enrolment)

Cohort Entry Points

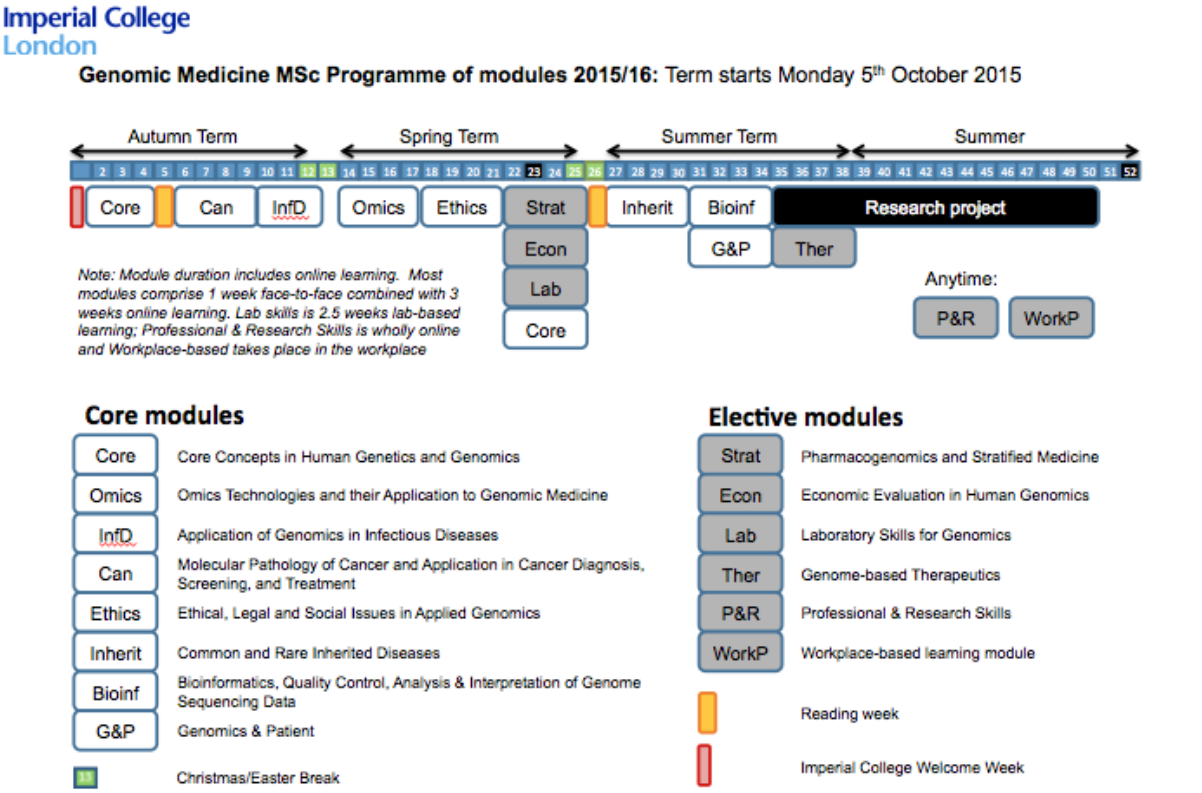
Annually in October for full-time MSc. Annually in October and March for part-time MSc.

Annually in October for full-time PG Dip. Annually in October and March for part-time PG Dip.

Annually in October and March for full-time and part-time PG Cert.

Students who have already completed the PG Cert can register for direct entry on to either the part-time or full-time PG Dip or either the full-time or part-time MSc at an appropriate time to be agreed by the Programme Director.

An overview of the timetable is shown in Figure 1 below.



Can the course be completed via distance-learning?

As noted above, the programme includes a large e-learning component, usually three weeks in every module, but it is not available as a complete online course. Two of the optional modules (Professional and Research Skills and the Workplace-Based module) are entirely distance learning. However, all of the Core modules include at least one week of face-to-face teaching. All E-learning materials will be handled through Blackboard to provide an online hub for all of the resources and communication. Most assessments will also be conducted through Blackboard and e-learning exams/assessments.

Why study with us?

We believe our programme offers a unique and fulfilling student experience as:

- The programme will be delivered by Imperial academic scientists and clinicians and invited outside speakers who are experts in their field.
- We offer a truly comprehensive programme that includes all aspects of Genomics within the field of human health and disease.
- The face-to-face teaching experiences are designed to impart complex concepts that may require longer explanations and to foster discussion and in-depth understanding of the material.

- The e-learning materials are designed to allow students to progress at their own pace by distance learning with available on-line help and support.
- Our programme encompasses both theory and practice, thereby developing skills that can be applied to scientific experimentation and not just the assimilation of theoretical knowledge.
- The course will be delivered by researchers at the cutting edge of genetics and genomics, many of whom are physician scientists who employ genomics in a clinical setting. The course involves leading researchers from every department within the Faculty of Medicine at Imperial College who will provide state-of-the-art education in this exciting and rapidly evolving field.
- Our programme will give students experience of working in four of the largest campuses of Imperial College: at South Kensington, St Mary's, Hammersmith and the Royal Brompton sites. This will give access to an unparalleled range of scientific resources, as well as the lively social cultures in each area.
- Imperial College has a world renowned reputation and is currently rated in the top ten universities worldwide. In the most recent Times survey of the best Universities to study in the UK it was rated 3rd, with high scores for research, teaching and the student experience.

Programme philosophy

We aim to place our students at the cutting edge of today's developments in Genomic Medicine and Genome Technologies. We want them to understand how these approaches are applied, there scope, potential and limitations. Students will study with, and then work alongside, world-leading scientists who have been deeply involved with the development and application of these approaches. Finally, in the MSc, they can potentially contribute, through laboratory projects, to real advances in understanding human genetics and genomics .

Overall Aims of the Course

- Independent learning
- Working in groups of varying sizes as team leader and member
- Information retrieval, appraisal and assimilation
- Understanding and applying core health and safety requirements
- Understanding success, failure and the uncertainty inherent in research
- Communication of scientific information in both written and oral forms
- Communication with both scientifically-literate and lay audiences
- Time management
- Project management in the context of the four-month research project
- Problem-solving strategies in experimental and translational design
- Awareness of the wider societal, ethical and commercial context which drives the fields, including the construction of business models for genomics
- Ability to use a number of resources to research
- Demonstrate the ability to self-critique by reflecting on coursework evaluation, project reports, critical reviews of scientific papers and have the ability to impartially review peers work.

- Manage information effectively by compiling results into a presentable and simple format, compiling reviews and discussion essays.
- Show autonomy by acting as an independent self-critical learner with minimum guidance for module tasks
- Effective communications skills that have been developed through group tasks, written and oral presentations

Upon graduation, all students will be able to:

For the PG Cert

- Critically appraise and synthesise genomic medicine data from a range of sources
- Communicate core concepts in genomic medicine clearly and effectively with both scientifically-literate and lay audiences
- Evaluate the potential of large-scale patient genome analysis to revolutionise healthcare in at least one domain
- Self-critique by reflecting on coursework evaluation, project reports, critical reviews of scientific papers.

For the PG Dip
Learning Objectives for the PG Cert plus

- Evaluate the potential of large-scale patient genome analysis to revolutionise healthcare across inherited disease, cancer and infectious disease and its implications in the healthcare setting
- Demonstrate awareness of the societal and ethical context of genomic medicine, including the complexities of protecting patient information
- Critically evaluate strengths and limitations of techniques suitable for assessing genomic variation relating to different clinical problems and disease states.
- Demonstrate competency in analysing and interpreting patient genomic analysis results and communicating their implications effectively to the patient.

For the MSc
Learning Objectives for PG Cert and PG Dip plus

- Demonstrate synoptic knowledge and deep understanding of medical genomics
- Deploy effective problem-solving strategies in data analysis and experimental design
- Appreciate the success, failure and the uncertainty inherent in research
- Effectively communicate scientific information in both written and oral forms
- Use project management skills in the context of the research project
- Select and deploy suitable research resources and strategies
- Synthesise complex research findings into a clear dissertation and oral presentation

Assessments

Assessment is designed to align with Learning Outcomes for each module and to be balanced equally across all taught modules with respect to student effort and timing of assessments. Taught modules will be assessed by coursework alone (9 modules), examination and coursework (3 modules), practical assessment and coursework (1 module) or examination and practical assessment (1 module). Students will therefore be assessed via a range of assessment methods. Choice of modules will be agreed with the Programme Director. There are specific College regulations that students should be aware of and must adhere to. These are summarized at the end of this document.

Coursework assessment includes :

- Critical analysis of complex clinical data sets/case studies
- Completion of ethical approval form
- Critical analysis of clinical diagnostic reports
- Report on analysis of bioinformatics data
- Lay summary of the 100,000 Genomes Project
- Protocol suitable for delivering DNA sequence findings to patients
- Critical appraisal of published evaluation
- Report of experimental findings written as a research paper
- Research proposal
- Report on clinical practice development
- Review article written for “Bioessays” format
- Examination format:
- 1-hour: Single Best Answer & Short Answer Question sections
- 2-hour: Short Answer Question section and long essay
- Practical assessment:
- Deliver analysed genomic data to a simulated patient (actor)
- Critical analysis of a research paper, presented orally to a group

The MSc research project is largely assessed by a written report. Assessment also includes a presentation to a mixed student & lay audience, an oral assessment with two internal examiners and supervisor assessment of research and analytical skills.

Exam Boards

There will be two exam boards per year, a mid year board in March and a final exam board in September.

Course Fees

MSc 1 year full time	Home/EU £12,000 Overseas £29,100
MSc 2 years part-time	Home/EU £6,000 per year

	Overseas £14,550 per year
PG-Dip 8 months full-time	Home/EU £8,000 Overseas £19,400
PG-Dip 2 years part-time	Home/EU £4,000 per year Overseas £9,700 per year
Pg-Cert 1 year part-time	Home/EU £4,000 Overseas £14,500

For the most up to date information please refer to:
http://www3.imperial.ac.uk/studentfinance/2015-16tuitionfees#PGT

Scholarships and Grants

Full time NHS employees may be eligible for full payment of fees through Health Education England (HEE). See
https://www.genomicseducation.hee.nhs.uk/genomicsmsc/programmes-and-events

There are also opportunities available to home and overseas students. For more information visit: http://www3.imperial.ac.uk/studentfinance

How to apply

All applications are coordinated on-line via the EMBARK registry system. Please visit
http://www3.imperial.ac.uk/pgprospectus/applicationforms

Course Induction

All students will be required to attend a course induction week which will include: registration, general orientation, introduction to various staff, short presentations of their work by key staff (involved in project supervision), lead lectures, library tour, training in search engines and reference manager systems and essential briefings on health and safety.

Brief Descriptions of Modules

Core Modules

Core Concepts in Human Genetics and Genomics

This module aims to build on the Pre- sessional Course to provide the student with vocabulary and understanding, either to pursue later aspects of the course, or as a standalone module to support specific researcher's development. Effectively, this module will serve as a foundation for those wishing to advance their careers within the NHS in genomic medicine. Students will commence by revisiting genomic architecture (particularly functional genomic units in eukaryotes and prokaryotes), principles of gene regulation, chromatin structure, and DNA variation. Mendelian and

non Mendelian patterns of inheritance will be used to emphasise the potential functional consequences of genomic variation, epigenetic modifications, and imprinting. Commonly used methodologies to annotate variation in human populations will be developed, with reference to human disease states. This section will emphasise how genomic medicine can be utilised to elucidate disease mechanisms and biology, and develop the concept of stratified medicine. Students will also learn how to use specific investigative approaches to correlate genetic markers to simple (dichotomous) and continuous (quantitative trait) phenotypic distributions. To more specifically prepare students for the wider goals of the 100,000 genomes project, DNA sequence variants will be explored in potential contexts spanning silent variation, physiological variation, disease, and uncertainty. This theme will be developed throughout the course. Additionally, the NHS ethical and governance frameworks that apply to medical genomics will be explored. Particular emphasis will be placed on underpinning knowledge for later modules in bioinformatics and statistics, and potential compromise of patient safety and confidentiality. The purposes and benefits of data sharing, across health records to genomic annotations will be summarised, and optimal forms of NHS data storage discussed. There will be one week (30 hours) of face- to- face teaching, in the form of lectures, seminars, workshops etc. Students will also complete 30 hours of directed e- learning. Student will also complete 127.5 hours of independent self-study and preparation for assessment.

Molecular Pathology of Cancer and Application in Cancer Diagnosis, Screening, and Treatment

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.

Specific content:
Tumour classification systems; cellular properties of tumours; factors in tumour formation; diagnosis, molecular subclassification, aggressiveness (prognosis) characterisation of metastases; breakthrough tumour/metastases and molecular Mechanisms. Models of genetic predisposition; GWAS studies; rarer higher penetrance genetic mutations and risk modelling; environmental factor and lifestyle predisposition and protection; molecular action; genomic interaction; epigenetic factors. Monitoring disease following treatment. 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 (cosegregation studies) identifying sequence alterations (single gene Sanger sequencing and NGS panel tests); how to interpret molecular results for pathogenicity – literature, databases, & in silico tools; genomic cellular markers and optimal treatment regimes; companion diagnostics in cancer. Principles of counselling strategies and ethical issues in cancer genomics including the patient perspective; consenting issues surrounding cancer genomics; consideration of the issues surrounding incidental findings in cancer genomics.

There will be one week (30 hours) of face- to- face teaching, in the form of lectures, seminars, workshops etc. Students will also complete 30 hours of directed e- learning. Student will also complete 127.5 hours of independent self- study and preparation for assessment.

Application of Genomics in Infectious Diseases

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. 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. The course will illustrate the above points by examples such as TB, HCV, HIV and Flu. It will include a discussion of the impact of metagenomics and studies of the microbiome on disease. It will also include a description of host genetic factors that may influence susceptibility and prognosis. There will be one week (30 hours) of face- to- face teaching, in the form of lectures, seminars, workshops etc. Students will also complete 30 hours of directed e- learning. Student will also complete 127.5 hours of independent self- study and preparation for assessment.

Omics Technologies and their Application to Genomic Medicine

This module will explain the underlying principles of the techniques that are the basis of the majority of clinical and research applications in genomic medicine. The techniques covered will focus on the analysis of genomic variation at the DNA level. DNA sequencing techniques willspecifically be explored in the context of the 100,000 Genomes project. The whole range of techniques available will be explored, including genomic, epigenomic, transcriptomic, metabolomic and proteomic techniques. Their clinical utility will be illustrated by the use of examples from rare monogenic and common complex genetic disease, including cancer and infectious diseases. The main aim of the module will be to allow the student to identify appropriate genomic medicine techniques to reach specific research or clinical goals. On completion of this module, students should be able to outline an investigation of a clinical case using standard genomic techniques, from the initial sample to a description of identified genomic features of possible clinical interest.

There will be one week (30 hours) of face- to- face teaching, in the form of lectures, seminars, workshops etc. Students will also complete 30 hours of directed e- learning. Student will also complete 127.5 hours of independent self- study and preparation for assessment.

Ethical, Legal and Social Issues in Applied Genomics

This module will focus on the ethical principles that govern the implementation of genomic medicine in clinical practice, and how these will be used to inform the societal changes that will naturally ensue with regard to the acceptance of stratification for treatment in medicine. The key historical events that underlie the development of ethics in medicine will be discussed in order for the students to understand how this has been framed. The various laws and guidance that is provided to healthcare professionals in the UK, Europe and the USA will be identified so that students are

aware of national variations that may need to be addressed when international studies are planned. The many challenges that will need to be overcome with respect to the use of new technologies will be discussed. These will include storage of data from genomics studies and rights of access to it and how this affects patient privacy, and strategies to ensure that access to these technologies is made available to minority populations within society to avoid discrimination in healthcare access. The first week of the module will consist of a mixture of lectures, debates, roll play and interaction with patients who have genetically determined disease. This will be supplemented by online resources to provide a deeper understanding of ethical issues around genomic research and its likely societal impact. Students will be provided with a scenario for a research study and will be asked to produce a document that addresses any ethical and legal issues raised. They will be asked to complete a version of the IRAS form for a REC application and be asked to defend this in an interview.

There will be one week (30 hours) of face- to- face teaching, in the form of lectures, seminars, workshops etc. Students will also complete 30 hours of directed e- learning. Student will also complete 127.5 hours of independent self- study and preparation for assessment.

Genomics of Common and Rare Inherited Diseases

The goal of this module is to provide an understanding of the different way that genetic variation can cause disease, including common and rare genetic variants, and multifactorial, polygenic and Mendelian modes of inheritance. Using examples of specific genetic disorders, the students will learn when and why genetic analysis can be useful in the clinic and will be taught traditional and current approaches to genetic diagnosis. The module will be provide an overview of the status of genetic testing in the UK and internationally and learn about the different tools currently used for genetic testing in rare and complex disorders. We will discuss when it may be appropriate to perform clinical investigation of common diseases, but the predominant focus will be on the identification and interpretation of rare, highly penetrant pathogenic genetic variants. The module will aim to provide students with a rounded view of the use of genetics in clinical medicine today and in the future, considering the experience from the point of view of patients, their families, doctors and other health care professionals. Students will consider ethical issues such as implications for other family members and incidental findings, as well as problematic subjects such as variants of unknown significance, genetic heterogeneity, pleiotropy and phenotypic variability.

There will be one week (30 hours) of face- to- face teaching, in the form of lectures, seminars, workshops etc. Students will also complete 30 hours of directed e- learning. Student will also complete 127.5 hours of independent self- study and preparation for assessment.

As their seventh Core Module MSc students will be required to choose between two alternative Core Modules; either Bioinformatics, or Genomics and the Patient, both summarized below:

Bioinformatics, Quality Control, Analysis & Interpretation of Genome Sequencing Data

The aim of this module is to enable students to gain a basic knowledge and understanding of the concepts and methods required to analyse and interpret genome sequencing data. Students will develop the skills to formulate their own research

questions as well as to collect, analyse and interpret their own NHS data using a range of statistical and bioinformatics techniques. A basic understanding of R and UNIX shell scripting will be taught to allow students to use and understand tools for the mapping and manipulation of sequencing and variant data such as samtools, BWA, Genome Analysis Toolkit (GATK), vcftools, etc. The module will also cover: Variant calling software tools such as Genome Analysis Toolkit (GATK), samtools and Platypus. Variant annotation tools such as Annovar and Variant Effect Predictor and pathogenicity prediction tools such as Polyphen, SIFT and Mutationtaster. Multiple databases and tools for associating variant calls with possible phenotypes such as HGMD, ClinVar, OMIM, Dapple and Genemania. Probability theory, estimation theory, hypothesis testing, linear models and experimental design. Subtleties, ethics and concepts of relative risk and how complex genomic data is communicated to patients.

There will be one week (30 hours) of face- to- face teaching, in the form of lectures, seminars, workshops etc. Students will also complete 30 hours of directed e- learning. Student will also complete 127.5 hours of independent self- study and preparation for assessment.

Genomics and the Patient

This module will focus on development of tools to assist the HealthCare professional in their future roles delivering NGS/genomic results, including “looked for, additional clinically important findings”, i.e. unanticipated genomic findings of potential clinical significance. Using patient faculty with a range of expertise including scientific/professionals (offers received), and a group of experienced Imperial- based Faculty, this module will build the necessary skills stepwise. Participants will identify and interpret key features of NGS and other genomics reports, and specifically develop interpretation skills that allow them to place results in context for the patient. This will require practical application of genetic risk calculations covered in the Core Concepts and Inherited Diseases modules; appreciation of predicted and plausible molecular results of particular patterns of sequence change (from Core Concepts); and a practical application of relevant ethical issues. In the first two weeks of the Module, they will use supported online resources to develop a draft personal template for provision of results to a non- technical person, and indicate the language in which they would wish to deliver this information verbally. During the face- to face week, in cycles of continuing improvement, they will use their template with different NGS scenarios to peers; academic faculty, and patient faculty. The finalised protocol template will be handed in as course work for the module. Practical communication skills will be assessed using a role play of an example set of NGS results designed to require understanding of key elements of genomic risk and ethical implications of rephenotyping.

There will be one week (30 hours) of face- to- face teaching, in the form of lectures, seminars, workshops etc. Students will also complete 30 hours of directed e- learning. Student will also complete 127.5 hours of independent self- study and preparation for assessment.

Research project (compulsory module for MSc)

The objective of this module is to provide the student with an understanding of how to identify a research question in genomic medicine; to identify whether sources of data already exist to answer that question, or whether additional samples need to be analysed. If appropriate, to design a study using new samples, and to understand how these samples can be used ethically and legally, to choose an appropriate method to execute the study, including appropriate statistical methodology. The module will also provide the opportunity to obtain laboratory skills in an omic technology and to understand the correct use of appropriate controls for procedures used in the study. The student will be expected to present the results of the study in the form of a written article for a scientific journal and orally to a lay audience. By the end of the project, the student should be able to assess whether the original hypothesis of the study was correct, whether additional hypotheses should be investigated, and to suggest additional studies that might strengthen or disprove the original hypothesis.

Types of projects that will be available to students will include:

- Laboratory projects
- Workplace based projects in a healthcare science setting
- Computer based projects
- Library based projects

The project will run over 14 weeks full- time (it will also be available to part- time students, normally to be spread out over a period of up to a year). Students will be expected to work a 35 hour week in their project Part- time students will be advised on registration, and again when choosing their project, on the types of project best suited to part- time study. Some of the laboratory based projects may not be available part time.

Full time students must complete projects by September. >50 indicative project titles from prospective clinical and non-clinical supervisors have already been submitted**.** All MSc students will be required to take their project under the supervision or co-supervision of a member of College staff (who may hold an honorary contract with the College). In some cases students may be able to conduct their research projects at their place of work, if the project meets the required standards, has an Imperial College co-supervisor and is approved by Professors Lovett and Thomas. Both project content and supervisory arrangements must be approved by the course director.

For the MSc and the PG-Dip degrees the student must also complete one optional module

Optional Modules

Pharmacogenomics and Stratified Medicine

Pharmacogenomics is the study of how the genetic profiles of different individuals have been found to influence and modulate drug response and drug reaction. This module will provide an understanding of the different genotypic profiles, and how these are identified molecularly to guide the choice of drugs depending on the individual genetic profile. The student will learn the technology underlying this field, how this is

used in the context of personalised medicine and pharmacogenomics and its scope. Specific content: Teaching will concentrate on drug interactions in HIV, acute and chronic lymphoid and myeloid leukaemia; autoimmune disorders and inflammation and drug interaction in neurological disorders. The course will concentrate on the description of molecular techniques for the identifications of SNPs using PCR, Sanger sequencing and /or high throughput sequencing. The student will be trained to critically review the applications of the technologies and the impact of assigning specific genotypes to individual in the context of specific therapies and how to balance drug efficacy and toxicity. The course will review papers that have led to the discovery of drugs driven by different genetic profiles and review future applications for the development of new drugs in the future.

There will be one week (30 hours) of face- to- face teaching, in the form of lectures, seminars, workshops etc. Students will also complete 30 hours of directed e- learning. Student will also complete 127.5 hours of independent self- study and preparation for assessment.

Genome- Based Therapeutics

The students will learn how genetic information contributes to drug development.
The module will comprehensively cover the current know- how on gene and nucleic acid based therapies typically including a review of pertinent pre- clinical and clinical research related to cystic fibrosis, haemophilia, SCID and other diseases for which gene therapy and genome- based therapies have advanced over the last 10 years. Students will be introduced to basic tools related to gene and nucleic acid- based therapies such as commonly used viral and non- viral vectors and vector production methods. In addition the use of anti- sense technology, genome editing and applications involving small non- coding RNAs will be covered. Students will also learn how the identification and characterisation of mutations and polymorphisms has led to development of personalised medicines and mutation- specific treatments for a range diseases. Students will be exposed to basic concepts related to intellectual property, commercialisation strategies and health economics

There will be two weeks (50 hours) of face- to- face teaching, in the form of lectures, seminars, workshops etc. Students will also complete 10 hours of directed e- learning Student will also complete 127.5 hours of independent self- study and preparation for assessment.

Laboratory Skills for Genomic Medicine

The purpose of this module is to give students experience of experimental design and laboratory skills relevant to genomic medicine research. It will take place over 3 weeks, with three distinct phases. Phase 1: Experimental design phase in groups, with the support of guided, interactive workshops. Phase 2: Laboratory execution of experiments. Phase 3: Data analysis and discussion of how to present the results in the form of a research paper. Within the guiding framework, students will have experimental design freedom and a stake in their research. This will prepare students for participating in a laboratory- based research project later in the course and give them skills in evaluating and presenting research in authentic ways. At the end of this module, students will have their case history, an experimental design and sequencing results. They will have all the necessary parts for writing up a research paper reporting

a novel finding of genomic variation related to a clinical case.

There will be three weeks (60 hours) of face- to- face teaching, in the form of lectures and laboratory sessions. Students will also complete 10 hours of directed e- learning. Student will also complete 117.5 hours of independent self- study and preparation for assessment.

Research and Professional Skills

This optional module is wholly online. The module comprises twelve online chapters, with an automatically scored test at the end of each chapter, which the students will have to pass before they can progress to the next one. The chapters will include teaching material and diagrams with audio voiceover, podcasts and vodcasts, and worked examples. The module leader will provide online feedback during the development of the research proposal assignment and assist with the identification of a project supervisor should the proposal become the basis of the research Dissertation. Content will include: Searching the literature: using medical databases, Reference Manager, Cochrane Library, NHS evidence, Boolean search, Quiz/Likert scale Basic statistics: percentages, mean/median, standard deviation, bias, odds, confounding, ratios, probability (p- values) Extended statistics: data collection, data handling, data analysis, specific statistical tests Critical appraisal: understanding learning outcomes and aims, systematic reviews, randomised controlled trials, qualitative research studies, epidemiology/cohort studies, diagnostic test studies, economic evaluation studies, case control Study design: creating a hypothesis, research question, study design methodology, power calculations, research ethics thesis writing and viva skills

Students will complete 60 hours of directed e- learning. Student will also complete 127.5 hours of independent self- study and preparation for assessment.

Economic Evaluation in Human Genomics

Technological advances in the area of genomic medicine are leading to new tests with major potential to improve disease diagnosis and the effective targeting of treatments. However, growth in the use of genomic technologies also has the potential to increase costs and to divert scarce healthcare resources from other uses. Using established methods of economic evaluation and decision modelling, it is possible to estimate and compare the impact of new technologies on costs and health outcomes. This module will introduce the main approaches to economic evaluation, including cost- utility analysis and cost- benefit analysis. Methods for quantifying costs or savings associated with alternative healthcare interventions and methods for measuring and valuing health outcomes will be described. The role of decision models in synthesising and extrapolating epidemiological and effectiveness evidence will be discussed, and the most common types of model (decision trees and Markov models) will be described. Throughout the module, concepts and methods will be illustrated using real examples and case studies drawn from genomic medicine. Students will be encouraged to consider whether existing methods and processes of economic evaluation are appropriate for this rapidly developing field, and to discuss alternative approaches. For the assessment, students will critically appraisal a published economic evaluation of a genomic technology.

There will be one week (30 hours) of face- to- face teaching, in the form of lectures, seminars, workshops etc. Students will also complete 30 hours of directed e- learning. Student will also complete 127.5 hours of independent self- study and preparation for assessment.

Workplace- based module

This module provides the healthcare practitioner with the opportunity to focus on, develop and extend personal, academic and professional ability in relation to an area of clinical practice in the genomic medicine setting. We anticipate students will start preparation for this module during earlier parts of the programme. At the outset of this module, they will (in collaboration with a clinical mentor and an academic supervisor) produce a learning contract to guide their individualised study and assessment topic. The topic will focus on a genomics related innovation in their own personal practice areas. Contact time will consist of face to face and electronic supervision sessions with an academic supervisor (normally 6 hours of tutorials), in line with individual students' learning needs and clinical objectives. In addition there will be 4 hours of online learning. The exact delivery pattern will be dependent on student numbers: face to face sessions will predominantly be one to one with the supervisor, with additional structured sessions that will utilise proven, well- received peer support feedback mechanisms. This module is not available on a stand- alone basis, and must be undertaken as part of the PG Cert/PG Dip/MSc programme. This module does not need to be taken at a fixed time in the year. Students taking this module on a part- time basis will agree with their supervisor an appropriate timeframe for completing the module. As the module must be completed at a student's place of work it is not expected that full- time students will take this module. However, if a full- time student who has taken study leave to complete the programme is able to arrange a placement to complete this module at their place of work, they may be permitted to take this module during the optional module slot in their programme (March for October starters, June for March starters). These students will agree with their supervisor an appropriate deadline These students will agree with their supervisor an appropriate deadline for submission of their coursework, with counselling from the module co-leads on how to balance their assessment load across the year.

Rules of Progression

Full-time students register for the qualification they wish to exit with: PG Cert, PG Dip or the MSc programme. Part-time students should register for the PG Cert in the first instance and progress to the other awards should they so wish. Students registered for the PG Cert who wish to continue on the programme without a break can transfer their registration to the PG Dip after completing Core Concepts and 2 other modules, provided they have demonstrated satisfactory academic progress in their first 3 modules. Similarly, students registered for the PG Dip can transfer their registration to the MSc after completing two further modules, provided they have demonstrated satisfactory academic progress.

Satisfactory progress will normally be judged achieving at least 50% for module assessments completed. PG Cert students who have failed a single module with a mark between 45-49% may still transfer their registration to PG Dip, subject to confirmation on passing the failed coursework and/or exam on the second and final

attempt. The same applies to PG Dip students who have failed one of their two post-PG Cert modules.

Students who register for and successfully complete the PG Cert may use the credit gained towards registration for the PG Dip or the MSc at a later point, provided that the PG Dip is completed within 4 years and the MSc is completed within 5 years, of initial registration for the Postgraduate Cert. These students may be asked to surrender the associated PG Cert and/or PG Dip on registration for the higher award. Likewise, students who register for and successfully complete the PG Dip may use the credit gained towards the MSc, provided that the MSc is completed within 5 years of their initial registration of the lowest award. These students may be asked to surrender the associated PG Dip on registration for the MSc.

Students who register for the MSc in the first instance will have a “virtual” PG Cert confirmed if the requirements for the PG Cert have been met. The pass mark for the PG Cert will be carried forward and the credits will accumulate towards the next level. On successful completion of eight modules (seven Core and one Optional), students will have a “virtual” PG Dip. The pass mark will be carried forward and the credits will accumulate towards the MSc. Should these students subsequently fail to achieve the requirements for the MSc they will be awarded the PG Dip.

The pass mark for each module is 50%. Within a module, no assessment less than 40% will be considered as a condoned pass regardless of the aggregate mark for the module.

PG Cert:

Students may be permitted one module marginal fail (45%-49%), but must pass the other three modules and achieve an overall aggregate score of 50% in order to pass the PG Cert.

PG Dip:

Students may be permitted two module marginal fail (45%-49%) but, in total, must pass six modules.

MSc:

Students who have already completed the taught pathway must pass the research project module (with a minimum aggregate mark of 50%) in order to be awarded the MSc. Students may be permitted a marginal fail (45-49%) in one of the oral presentation elements and or the supervisor assessment as long as they achieve a mark of 50% or above.

Pastoral Care

Once accepted on the course, each student will be allocated a personal tutor who will be an NHLI staff member independent of the course. They will guide the student throughout the course, dealing with both academic and personal aspects. We recognise that students may join the programme with very different backgrounds. One of the key roles of the tutors will be to work with the students to identify those areas where they would benefit from further instruction and experience, and to design a programme that will best fit those needs. The Programme Director oversees the tutoring system and will also provide independent advice for students if required.

Postgraduate teaching and social facilities

There are extensive facilities for postgraduate students at the Imperial College Royal Brompton/South Kensington, St Mary's and Hammersmith Campuses. The Imperial library provides first class provision for learning and research materials in the relevant disciplines and also houses extensive computing facilities for students. Students also have access to the services at the other Imperial College university campuses.

Further enquiries

For any further information please contact:

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Some additional notes on Coursework and Assessment Policies

Coursework should be submitted by 4pm on the day specified in the module details, in *full* through Turnitin or Blackboard Learn using the links in the assessment folder for that module. A failure to do this by the submission deadline will be classified as a 'non-submission'.

If the word count is exceeded normally a 1% penalty for every 1% excess will be made at the discretion of the examination board (eg. if a piece of work is 4450 words for a 4000 word piece of work, you will be penalised for 50 words, as you are allowed only 10% over the stated word count.)

If the word count is under, normally a 1% penalty for every 1% under will be made at the discretion of the examination board (eg if a piece of work is 3550 words for a 4000 word piece of work, you will be penalised for 50 words, as you are allowed only 10% under the stated word count)

Unauthorised late submissions will be awarded 0%.

Students are advised to keep a copy of the email receipt from turnitin as proof of submission.

Requests for an extension to the submission date should be submitted in writing to the module leader, at least 7 days before the submission deadline.

Written submissions ***must*** be anonymised. The front page of the submission should include the student's CID number and the total word count.

The submission should also include the student's CID number on every page and include page numbers within the footer of each page.

Words within titles, tables, figures and appendices are not counted in the word count. References bracketed within the text are not counted in the word count i.e. (Jones et al., 2010) but are counted if they are within a sentence structure e.g. *A study by Jones et al., (2010).* The reference list does not contribute to the word count.

Tables, Figures and Appendices should be used appropriately (only to substantiate material in the main text) and not as a means of "free word" material. Every table and figure used should be numbered and labelled and should be referred to in the main text. If appropriate reference is not made to them in the main text, then they will not be taken into account during marking.

Submissions should be word-processed with a minimum of a 2cm margin, at least 1.5 spacing for text and be fully referenced (Harvard style).

All authors listed on a publication should be included in the reference list.

Students will be permitted to re-enter a failed examination on a single occasion. Resits will be scheduled in August of each year. At the discretion of the Programme Director, students may be counselled to wait until the following year to re-sit.

A failed piece of coursework can be resubmitted on one occasion, with a deadline to be agreed with the relevant module leader and Programme Director, normally between 1 week and 2 months after the original submission deadline.

If the MSc research project is failed, students may resubmit on one occasion in the following academic year, with a deadline to be agreed with their project supervisor and the Programme Director.

Candidates with mitigating circumstances will be dealt with according to the relevant College policy
https://workspace.imperial.ac.uk/registry/Public/Exams/MitigatingCircumstancesPolicyProcedures