100,000 Genomes Project

General Practice Information
100,000 Genomes Project:
General Practice

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Directorate of Genetics
The 100,000 Genomes Project
Information for General Practitioners

Introduction
Genomics England Limited (GEL), a company owned by the Department of Health, is creating a lasting legacy for patients, the NHS and the UK economy through the sequencing of 100,000 genomes, all with the consent of participants in the 100,000 Genomes Project. The Project will transform the application of genetics in healthcare and contribute to the development of a personalised medicine service in the NHS.

The North Thames NHS Genomic Medicine Centre (NT GMC) is one of 13 newly-founded GMC sites located across England that are working in partnership with NHS England and GEL to realise the goals of the 100,000 Genomes Project. The NT GMC is a partnership between Great Ormond Street Hospital for Children, Barts Health, London North West Healthcare, Moorfields Eye Hospital, the Royal Free London, Royal National Orthopaedic Hospital and University College London Hospitals NHS Trusts. All staff working in the NHS across these sites are considered to be part of the NT GMC and fall within its transformation remit. The NT GMC is led by Great Ormond Street, located in Holborn.

What is the 100,000 Genomes Project planning to do?
The Project will sequence 100,000 whole genomes from around 70,000 people as part of the largest national genome sequencing initiative in the world. Participants are patients with a rare genetic disease and their families and patients with cancer. The results will be linked with patients’ medical records and stored securely. By combining these two sets of information and allowing authorised researchers to access it, the Project aims to:

- Provide a diagnosis for some patients with rare diseases
- Learn how to adapt cancer treatment by looking at the genome of the tumour
- Develop a genomic medicine service for the NHS
- Support clinicians and researchers in hospitals, universities and companies of all sizes to develop new medicines, therapies and diagnostic tests

An additional and critically important spin off is the importance of this huge amount of data to researchers. This includes those wanting to understand more about the genomes but also to those wanting to develop new treatments, diagnostics, devices and medicines including academics and those in life science industries such as well-known pharmaceutical and biotechnology companies.

Who can participate?

- Patients with an eligible rare disease or cancer are usually identified by their clinicians and invited to learn more about the 100,000 Genomes Project.
- After the Project has been explained by a trained practitioner and they have had as much time as they need to consider whether to take part and ask questions, patients will be asked to:
  - Discuss and complete a consent form with the practitioner.
- Give blood samples (approximately 30ml); occasionally other samples such as saliva might also be requested.
- Give a piece of their tumour (for cancer patients), collected during their routine treatment.
- Give their personal details, contact details and health information to the Project.

**What results may participants receive?**

- After a participant’s whole genome is sequenced and analysed, the results will be sent to their clinical team who will discuss them with the participant.
- At first, findings will take some time to be returned – current timescales in 2017 are 12-18 months. This is due to the demands of designing and implementing the necessary infrastructure and embedding the new technology within the NHS, the likes of which has not been carried out on this scale previously. In time, results should be returned within a couple of weeks as the technology and our understanding of the results develops.
- The results may provide information that helps guide a participant’s treatment, but alternatively it is also possible that there will be no information to report at this time. All participants’ genomic information will remain within the data system, where research carried out may still lead to a diagnosis or the return of other results in the future.
- If something is found which could be important for the health of other members of the participant’s family, their clinical team will advise them about what information would be helpful to pass on, and will support them in doing this.
- Participants can also request that their genomes are tested for the presence of genomic changes that are known to be linked with the development of certain diseases such as bowel, breast and ovarian cancers and familial hypercholesterolaemia. These diseases can often be prevented or their effects reduced by early diagnosis through screening programmes and NHS treatment.
- In addition to participants choosing to be informed of any secondary findings, they can also ‘opt in’ to have their genomes tested to find out if they are carriers of genetic diseases such as cystic fibrosis as this may have an impact on the health of the participant’s children.

**The Future**

The aim of the 100,000 Genomes Project is to create a new genomic medicine service for the NHS, transforming the way people are cared for. Patients may be offered a diagnosis where there wasn’t one before. In time, there is the potential of new and more effective treatments. The project will enable new medical research. Combining genomic sequence data with medical records is a ground-breaking resource, allowing researchers to study how best to use genomics in healthcare and how best to interpret the data to help patients. The causes, diagnosis and treatment of disease will also be investigated.

We also aim to kick-start a UK genomics industry as part of what is currently the largest national sequencing project of its kind in the world. Genomics England’s legacy will be a genomics service ready for adoption by the NHS, new medicines, treatments and diagnostics and a country which hosts the world’s leading genomic companies.

*It is a bold ambition with benefits for all!*
The GP’s role

Clinicians have always personalised patient management. There is a now growing momentum to improve this further through the integration of genomic information into clinical care. This will incorporate powerful new tools through which clinicians can further tailor healthcare, improving disease prevention, prediction, diagnosis and treatment. Advances in genetic technology and understanding, coupled with an increasing patient demand for genetic and genomic investigation, is driving this. The healthcare workforce, including General Practitioners, needs to be empowered to identify the opportunities for genomic medicine and feel confident in their ability to deliver personalised care effectively and compassionately. They will need to have sufficient understanding of genomics to communicate effectively, support their patients and institute appropriate management.

To produce a whole genome sequence, the 100,000 Genomes Project requires blood samples from eligible patients who have given their informed consent to take part. The patient’s DNA is extracted from their blood sample at the regional lab at Great Ormond Street Hospital and is then sent to be processed at the national sequencing facility in Cambridge.

If it is not possible to take blood samples from patients during their consent appointment, we supply them with a Blood Sample Pack so they can arrange to have their samples taken at a later date – patients often choose the familiarity and convenience of their GP surgery for this. The packs contain blood tubes, and a genetic test request form including patient identification. The pack also contains secure, pre-paid packaging in which patients can return their samples and forms to the Project Team at the Genomic Medicine Centre in the regular post.

The 100,000 Genomes Project would not be possible without the combined efforts of NHS staff across multiple disciplines and we thank you for your cooperation in the collection of blood samples required for patients to participate if they choose to book phlebotomy appointments at your facility.

If you have questions or concerns about this or any other aspect of the project please call us on 0207 762 6701.

Rare Disease: The GP’s Role
Video by the Genomics Education Programme
https://youtu.be/1CNGk5lovv4
How can the Genomics Education Programme help you?

The Genomics Education Programme (GEP) is a £20 million, three-year project supported by Health Education England (HEE) to ensure staff in the NHS have the knowledge, skills and experience to keep this country a world leader in genomic and precision medicine.

To achieve its goals, the Genomics Education Programme is:

- Providing online resources and self-directed education for NHS staff, including general courses, such as Introduction to Genomics and Introduction to Bioinformatics.
- Developing curricula and assuring delivery of formal academic-based training programmes for the entire workforce, such as the MSc in Genomic Medicine and Higher Specialist Scientist Training in genomics.
- Co-ordinating workforce planning for the specialist staff required to deliver genomic technologies.
- Commissioning NHS-funded places for staff on training programmes.
- Supporting multi-professional clinical research fellowships and doctoral posts in genomics and bioinformatics and genomics.
- Through HEE, funding at least 550 places for NHS staff on a Master's Degree in Genomic Medicine delivered by partner Universities. Funding is also available to study individual, standalone modules for Continuing Personal and Professional Development (CPPD) across all NHS professions.

Funded Postgraduate courses at Queen Mary University of London for NHS staff

The Master’s Degree in Genomic Medicine is being delivered by Queen Mary University of London in the North Thames region. Funding for this initiative is due to end in 2018, although HEE are working to extend this. This means that enrolment for the MSc and PG Dip are now suspended but staff can still apply to study individual modules of the programme. Modules can be studied on an assessed or non-assessed basis with staff either choosing to take assessments and gain accreditation or alternatively only attend the face-to-face lectures delivered by the university without the requirement to carry out self-directed study outside of the sessions, complete assignments or sit exams. More information can be found on the Genomics Education website: www.genomicseducation.co.uk

Free-to-access education resources

The GEP have also produced a number of free-to-access, online resources designed to be used by NHS staff to increase their knowledge in genomics. The resources range from short, one-hour training programmes to more in-depth, Massive Open Online Courses (MOOCs) available through the FutureLearn platform which include around nine hours of study spanning a number of weeks.

Online educational resources include the short courses ‘Introduction to Genomics’ and ‘Introduction to Bioinformatics’ and the Massive Open Online Course ‘Whole Genome Sequencing: Decoding the Language of Life and Health’. All of these resources are freely available for anyone to use.

A number of free-to-access, online study modules have also been developed by a team of healthcare staff and geneticists as part of the Gen-Equip project. These modules are aimed at all healthcare professionals working in Primary Care and cover topics such as ‘Inherited Cancer Syndromes’, ‘Before and During Pregnancy’ and ‘Inherited Cardiovascular Conditions’. All of the modules are accredited by the RCGP for CPD. To find out more and register, visit Gen-Equip’s Primary Care Genetics website: http://bit.ly/NYGMCPcg
What can the NT GMC Project Team do for you?

Developments in genomics within cancer diagnosis and treatment, Familial Cancer, Pharmacogenomics, Rare Diseases, Infectious Diseases and Complex Diseases all have the potential to impact primary care. HEE have released a strategy document identifying the educational need and their existing resources and outlining their action plan to raise awareness of genomics in primary care. You can view and download this document from http://bit.ly/HEEstatement

The NT GMC 100,000 Genomes Project Team are available to support NHS staff from across the region with regards to the Project, the mainstreaming of genomic medicine and opportunities for education and training in genomics.

We can provide information and promotional materials, such as posters and leaflets aimed towards staff and/or patients, for display in your organisation and can also deliver free information and training sessions about the 100,000 Genomes Project to staff.

Please contact us to discuss:
- Receiving further information materials
- Finding out more about genomic education opportunities
- Having the NT GMC Education Team delivering a presentation or training sessions to your colleagues
- Or, you would like to know any more about the 100,000 Genomes Project and genomic medicine

To view the approved rare disease and cancer lists and their eligibility criteria, visit: http://bit.ly/100Kdocs

For information about the 100,000 Genomes Project in the North Thames area, visit http://bit.ly/NTGMC-info

For further information, news and updates about the 100,000 Genomes Project at a national level, visit the Genomics England website: www.genomicsengland.co.uk

For information about the Genomics Education Programme, CPPD modules and other educational resources and training opportunities, visit: http://bit.ly/NTGMC_ET

Follow us on Twitter:
@NorthThamesGMC
#genomes100k
General practice and genomics

Clinicians have always personalised patient management. There is a growing momentum to improve this further through the integration of genomic information into clinical care. This will incorporate powerful new tools through which clinicians can further tailor healthcare, improving disease prevention, prediction, diagnosis and treatment.

Advances in genetic technology and understanding, coupled with an increasing patient demand for genetic and genomic investigation, is driving this momentum. The healthcare workforce including General Practitioners (GPs) needs to be empowered to identify the opportunities for genomic medicine and feel confident in their skills to deliver personalised care effectively and compassionately. They will need to have sufficient understanding of genomics to communicate effectively, support their patients and institute appropriate management.

Making a detailed diagnosis

This requires an understanding of pathology at a molecular level, which is now made possible by rapid, affordable sequencing of the genetic code (human and microbial / viral). Deciding when to use these tests and how to interpret their results will become important parts of medical practice. Even where GPs are not using these tests directly, they need to be aware of the implications for patients and their families going through secondary and tertiary care.

Cancer

It is perhaps in the area of cancer where GPs are currently experiencing the greatest impact from genomics. With over 330,000 new cases diagnosed in the UK each year across a range of different sites, GPs have an important role to play both in supporting patients through diagnostic and treatment processes (see example 1) and in using knowledge of genomics for disease prevention. Testing of both the patient’s own genetic makeup (‘germline’ DNA) and the tumour DNA (‘somatic’ testing) are important here.

A small proportion of cancers (around 5%) are due to familial cancer syndromes such as breast or ovarian cancer associated with mutations in the BRCA1 or BRCA2 genes. This is relevant for the care of the patient with cancer, and also for the identification of risk in family members so that prevention, in the form of increased monitoring, screening (e.g. mammography) or treatment (e.g. chemotherapy or prophylactic mastectomy) can be offered. Such syndromes should be suspected in families who have multiple members affected with cancer, particularly at younger ages, with specific cancer types or with unusual cancers such as male breast cancer.
As more patients gain access to testing either through research programmes, as part of clinical care, or by direct to consumer testing from commercial companies they will turn to primary care for discussion and advice.

The detection of a tumour’s genetic signature may be used to make a precise diagnosis, enabling a more accurate prognosis and better tailored treatment. Increasingly, drugs are available that are targeted to the genetic features of a cancer, requiring genetic testing of the cancer cells to determine their potential response.

Rare diseases

Rare diseases, which are predominantly genetic in origin, affect 1 in 17 of the population and therefore make up a proportion of the clinical caseload in all specialties including primary care.

Making a detailed diagnosis is increasingly important for rare disease as there is now much greater understanding of underlying pathology, likely natural history, responsiveness to various treatments and best forms of overall management. Genomic tests now increasingly make this possible.

Once detailed diagnosis of a rare condition is made, one of the important roles in general practice is to be alert to the complications and monitor and manage appropriately. For example, in polycystic kidney disease (PKD) patients need regular and tailored surveillance involving monitoring of blood pressure and renal function, particularly close care during pregnancy and being alert to other complications such as intracranial cerebral problems.

Another important dimension of rare genetic disorders is the familial element. The GP must always be aware of the potential for disease in relatives of affected patients and should give appropriate advice.

Common complex diseases

All diseases result from a combination of genetic and environmental factors. Common disorders such as diabetes, obesity, heart disease and most cancers are all influenced by underlying susceptibility as well as the surrounding environment and the lifestyle the individual chooses to adopt. Whilst background public health measures and general health promotion are key to better health at the population level, as the potential of personalised medicine develops it is likely that prevention (for example diet or weight loss programmes) will also become increasingly tailored to the particular individual and interventions such as breast or prostate screening may be offered according to underlying risk.
Infectious diseases

In infectious disease, genome sequencing of the pathogens can, in principle, be used to diagnose infections and determine likely susceptibility to antimicrobials. For example, genomics is likely to become a key part of TB management within the next few years, as genome sequencing provides diagnostic and drug susceptibility information more rapidly (potentially within 2-3 weeks) than current laboratory methods. For most infections managed in primary care or other community settings, however, diagnosis and treatment management using genomic information will only have an impact once suitably accurate, cheap and reliable point of care diagnostic devices based on sequencing are available.

The detailed and high-resolution information obtained by sequencing a pathogen from a patient can be used to make a diagnosis and inform treatment, but is also important in a public health context for understanding how diseases spread in the community. Public Health England has already established a service using whole genome sequencing (WGS) to investigate community outbreaks of Salmonella infections, using genomic information to detect outbreaks, rule cases in and out of the outbreak and to identify the source of infections. They are also piloting this approach for enhancing the effectiveness of epidemiological investigation of potential TB outbreaks.

Pharmacogenetics and treatment

With the development of rapid sequencing assays, and multiple gene panels, it is anticipated that testing for relevant genetic variants that influence both drug efficacy and drug safety will be increasingly used to aid both drug and dosage selection and this will be relevant with different population groups.

Developing intelligent decision support systems that allow the use of genomic and clinical information to aid prescribing drugs at the right dose will be important in the future. Such information is being incorporated into the summary of product characteristics of individual drugs, and is reflected in the guidance provided by regulatory agencies such as the European Medicines Agency (see example 2).

Ethical, legal, social and organisational implications

There are a number of broader challenges that will influence the use of genomic medicine. These include:

- Developing skills and expertise in genomics within the wider health professional workforce
- Issues relating to patient communication, privacy and consent (particularly for genomic testing in children)
- Handling uncertain, unexpected or incidental findings from genomic tests in clinical practice
Further Information and Resources

The Royal College of General Practitioners. MRCGP Curriculum statement on Genetics 3.02.

Cancer genetics module on the RCGP elearning platform:
elearning.rcgp.org.uk/course/

HEE Genomics Education Programme Health Education England Information on genomics education including HEE sponsored MSc, Diploma, PG Certificate and CPPD genomics courses
0121 695 2374
genomicseducation@wm.hee.nhs.uk
www.genomicseducation.hee.nhs.uk

Online module, St George’s, University of London, The Genomics Era: the future of genetics in medicine
www.futurelearn.com/courses/the-genomics-era

UK Genetic Testing Network (UK GTN) 0203 350 4999
ukgtn@nwlcsu
ukgtn.nhs.uk

UK Pharmacogenetics and Stratified Medicine network.
www.uk-pgx-stratmed.co.uk

Statins benefit and risks drug safety update


- Implications of significant results for other family members
- Bioinformatics provision and secure genomic data storage and access within the health service
- Impact of genomics on current healthcare services, resources and patient pathways (including equity of access to genomic tests)
- Developing intelligent decision support systems that allow the use of genomic and clinical information to aid in the prescribing of drugs at the right dose
- Clarifying risks and benefits associated with using genomic tests for opportunistic screening

The future

The last two decades have seen unprecedented investment in life sciences in the UK. Advanced technologies are now available to sequence the entire genome at a cost of a few thousand pounds in as little as 24 hours, and it is envisaged that this cost will fall considerably over the next few years. More recently, the Government has signalled its confidence in the power of genomic science to produce major health benefits for the population through its investment in the 100,000 Genomes Project. However, achieving these benefits will depend on the ability of clinicians to use these new technologies effectively, efficiently and responsibly, for the population as a whole. Genomics can no longer be left to specialists and enthusiasts, but must be grasped by every clinician throughout the NHS.

Through the ‘Clinical Champions’ network, the Royal College of Physicians aims to promote education and training in genomics within every specialty. This will ensure that clinicians of the future are ready to capitalise on all of these new developments to provide personalised care for their patients.
Programme Overview

Training existing and future NHS healthcare professionals

Supporting and enhancing the 100,000 Genomes Project

Bringing better care and lasting benefits for patients

Developing people for health and healthcare

www.hee.nhs.uk
Welcome to the
Genomics Education Programme

What is the GEP?
Health Education England’s Genomics Education Programme (GEP) is the NHS initiative to ensure its workforce has the knowledge, skills and experience necessary to remain a world leader in genomic medicine, with HEE one of the five key partners involved in the 100,000 Genomes Project (bit.ly/Genomes100k).

To help achieve this, the GEP has adopted a network approach, working in conjunction with the HEE GEP funded education and training leads of both the 13 NHS Genomic Medicine Centres and the 30+ Genomics England Clinical Interpretation Partnership (GeCIP) domains.

How is the GEP transforming the workforce?
To support workforce transformation across the health service, the GEP has developed educational resources and funded courses for the NHS workforce.

Our Master’s in Genomic Medicine (bit.ly/GenomicsMSc) is a multidisciplinary and multi-professional qualification in genomics that can be applied to both clinical practice and clinical research.

It is offered through a nationwide network of 10 universities and is available as a one-year full-time or two-year part-time Master’s, as well as Postgraduate Diploma and Certificate. Individual modules can also be taken standalone as part of CPPD (continuing personal and professional development). The GEP is funding more than 550 Master’s Postgraduate Diploma and Certificate places together with 1,000 individual modules for CPPD across all NHS professions.

In addition, a large part of the GEP’s role is the development of scientific and leadership expertise at the highest level to support the introduction of genomic technologies across the healthcare system. This includes funding additional places on formal NHS training programmes as well as supporting a number of multi-professional clinical research fellowships and doctoral posts in genomics and bioinformatics.
In what ways can NHS staff learn through the GEP?
The GEP is developing a range of educational resources tailored to specific needs in the workforce, for professionals to access when and where they are needed. These include:

**Short online courses**
Engaging and interactive courses for continuing professional development, examples include:
- *Introduction to Genomics*
- *Introduction to Bioinformatics*

**Training tools**
Digital training to directly support the 100,000 Genomes Project, including:
- *Preparing for the Consent Conversation*
- *Eligibility Wheels (developed by UHL)*
- *Sample Processing and DNA Extraction*

**‘Just-in-time’ resources**
Bitesize information on genomics for busy health professionals, for example:
- *Genetics conditions information factsheets*
- *Genomics in mainstream medicine factsheets*

**Workshops and events**
The team facilitate workshops and events for our network as well as support the education and development of the genomic workforce. The team attend national and international conferences including: Primary Care & Public Health, RCGP, NHS Health & Care Innovation Expo

**Videos, images and animations**
Informative and educational multimedia for learning and teaching. Our collection includes a number of images, animations and videos on Flickr and YouTube

**Social media**
Daily updates and tailored content to more than 6,000 followers. Engage with us via:
- twitter.com/genomicsedu
- facebook.com/genomicsedu
How can I learn more?

The GEP has developed a wide range of education and training resources for health professionals available on our website: www.genomicseducation.hee.nhs.uk

Introduction to Genomics

This introductory course outlines the fundamentals of genomics and its significance for healthcare now and in the future. It shows how, through new scientific discoveries, we can examine the whole of a person’s DNA – their genome – and discover its growing importance for healthcare.

bit.ly/Intro2Genomics

100,000 Genomes Project:
Preparing for the Consent Conversation

In our comprehensive online course, learn how to navigate the key steps of the 100,000 Genomes Project’s consent process, what to consider when preparing to talk to potential participants, and how to address their questions and concerns.

bit.ly/100kconsent

Sample Processing and DNA Extraction

This series of films has been developed in line with the Genomics England protocol for handling and processing of blood and tumour samples to support Genomic Medicine Centre staff working in molecular genetics and histopathology laboratories.

bit.ly/SampleDNAExtraction

Whole Genome Sequencing: Decoding the Language of Life and Health

In our online course, learn about this cutting edge technology and its potential to transform healthcare. Go behind the closed doors of the lab to explore the sequencing and interpretation processes and the benefits to patients.

bit.ly/WholeGenomeSequencing

Tumour Assessment for Whole Genome Sequencing

This series of four educational modules is aimed at histopathologists and laboratory staff who will be processing tumour samples for whole genome sequencing and outlines the need for a standard model for tumour assessment to be adopted in order to achieve consistent results.

bit.ly/TumourAssessmentTools

www.genomicseducation.hee.nhs.uk
Career Framework: CF 2-8
Specialism: Life Sciences; physiological sciences.

Learning Package - Understanding Genomics (9 Courses)
https://www.futurelearn.com/courses/collections/genomics

Genomic Medicine Gets Personal (Georgetown University)
https://www.edx.org/course/genomic-medicine-gets-personal-georgetownx-medx202-02x

Genomic Technologies in Clinical Diagnostics: Molecular Techniques.
https://www.futurelearn.com/courses/molecular-techniques

Requirements:
This course is designed for scientists and healthcare professionals with an interest in obtaining a postgraduate-level understanding of molecular genomic techniques.
If you are new to the field, we recommend that before you start this course you complete The Genomics Era: the Future of Genetics in Medicine – the first in the Genomics in Healthcare program from St George’s.

Genomic Technologies in Clinical Diagnostics: Next Generation Sequencing
https://www.futurelearn.com/courses/next-generation-sequencing

Requirements:
This course is designed for scientists and healthcare professionals with an interest in obtaining a postgraduate-level understanding of molecular genomic techniques.
If you are completing both Genomic Technologies in Clinical Diagnostics courses, we recommend that you complete Genomic Technologies in Clinical Diagnostics: Molecular Techniques first.
And if you are new to the field, we recommend that before you start either course, you complete The Genomics Era: The Future of Genetics in Medicine – the first in the Genomics in Healthcare program from St George’s.
Clinical Bioinformatics

Clinical Bioinformatics: Unlocking Genomics in Healthcare (University of Manchester) Badged under GEP
https://www.futurelearn.com/courses/bioinformatics

Requirements:
This course is aimed at current healthcare professionals, who are interested in learning more about the role of clinical bioinformatics and will also be applicable to people with an interest in the application of genomics in healthcare.

It is not essential to have previous experience or knowledge of bioinformatics or genomics although medical terminology is used and the course is designed to be applicable to practising healthcare professionals.

Career Framework: CF 2-8
Specialism: Genomics; physical sciences; physiological sciences, nursing, medicine

Big Data: Statistical Inference and Machine Learning (Queensland University of Technology)
https://www.futurelearn.com/courses/big-data-machine-learning

Requirements:
You will enjoy this course most and benefit from the learning experience if you have a basic understanding of statistics and mathematics at an undergraduate level.
You will complete practical exercises using R Studio, so you’ll need to be familiar enough with R to:
Install a package; import data; read and run starter code; develop a solution or read through a solution and gain understanding from it.

Career Framework: CF 2-8

Big Data: Mathematical Modelling (QUT)

Requirements:
This course is designed for anyone looking to add mathematical methods for data analytics to their skill set.
We provide a multi-layered approach, so you can learn about the methods even if you don’t have a strong maths background, but we provide further information for those with a sound knowledge of first or second year undergraduate mathematics.

We will assume basic MATLAB (or other) programming skills for some of the practical exercises.

Career Framework: CF 2-8

Big Data: Data Visualisation (QUT)
https://www.futurelearn.com/courses/big-data-visualisation

Requirements:
In this course we will use a variety of tools, so that you can become comfortable with engaging with different software packages and gain confidence in trialling new packages that may better meet your particular needs.

Career Framework: CF 2-8
Master Algorithmic Programming Techniques 5 Courses £55 per course (UCL)
https://www.coursera.org/specializations/data-structures-algorithms

Requirements:
Basic knowledge of at least one programming language (C, C++, C#, Haskell, Java, JavaScript, Python2, Python3, Ruby, and Scala): loops, arrays, stacks, recursion. Basic knowledge of mathematics: proof by induction, proof by contradiction.

Machine Learning (Stanford University)
https://www.coursera.org/learn/machine-learning

Bioinformatic Methods (University of Toronto)
https://www.coursera.org/learn/bioinformatics-methods-1

Requirements:
This pair of courses is useful to any student considering graduate school in the biological sciences, as well as students considering molecular medicine. Both provide an overview of the many different bioinformatic tools that are out there.

Bioinformatics: Life Sciences on Your Computer (John Hopkins University)
https://www.coursera.org/course/bioinform

Requirements:
This course would be a fine start for those who wish to pursue formal or informal training in bioinformatics.

Bioinformatics Specialization 6 Courses £34 per course (UC San Diego)
https://www.coursera.org/specializations/bioinformatics

Data Analysis for Life Scientists 1: Statistics and R (Harvard University)

Data Analysis for Life Scientists 2: Introduction to Linear Models and Matrix Algebra (Harvard University)

Data Analysis for Life Scientists 3: Statistical Inference and Modelling for High-throughput Experiments (Harvard University)
https://www.edx.org/course/data-analysis-life-sciences-3-harvardx-ph525-3x-0

Data Analysis for Life Scientists 4: High-Dimensional Data Analysis (Harvard University)
https://www.edx.org/course/data-analysis-life-sciences-4-high-harvardx-ph525-4x-0

Data Analysis for Life Scientists 5: Introduction to Bioconductor: Annotation and Analysis of Genomes and Genomic Assays (Harvard University)

Data Analysis for Life Scientists 6: High-performance Computing for Reproducible Genomics (Harvard University)
https://www.edx.org/course/data-analysis-life-sciences-6-high-harvardx-ph525-6x

Data Analysis for Life Scientists 7: Case Studies in Functional Genomics (Harvard University)
https://www.edx.org/course/data-analysis-life-sciences-7-case-harvardx-ph525-7x
HEE GEP
Introduction to Genomics
https://www.genomicseducation.hee.nhs.uk/courses/courses/introduction-to-genomics/

Background:
As personalised medicine becomes more commonplace, it is important that health professionals have a good awareness of what genomics is. This course will introduce how genomic information is being used to support clinical practice, and its future impact on healthcare.

Suitable for?
This module has been developed for health professionals at all levels. (Wider awareness)

Introduction to Bioinformatics
https://www.genomicseducation.hee.nhs.uk/courses/courses/an-introduction-to-bioinformatics/

Background:
With advances in genomic technologies, the amount of biological data we can now generate about patients is vast. It is vital that healthcare integrates the principles of bioinformatics to process and make sense of that data in a form that clinicians can use to support patient care.

Suitable for?
This module is primarily targeted at practising healthcare scientists and at clinicians with an interest in healthcare science.

Preparing for Consent: Consent and Ethics
https://www.genomicseducation.hee.nhs.uk/courses/courses/consent-ethics/

Background:
It is vital that patients considering participating in the 100,000 Genomes Project make an informed decision. This course will prepare you, as their healthcare professional, to support them in doing so. It will also be of interest to others for whom having an understanding of the consent process would be helpful.

Suitable for?
This module has been developed for healthcare professionals working in NHS England Genomic Medicine Centres and their local delivery partners who will undertake the consent conversation with potential participants interested in joining the 100,000 Genomes Project.

Sample Processing and DNA extraction
https://www.genomicseducation.hee.nhs.uk/courses/courses/sample-processing-for-whole-genome-sequencing/

Background:
All staff involved with sample preparation and DNA extraction will need to be equipped with the relevant knowledge and skills to be able to prepare the high quality samples which are integral to the outcomes of this project.

Suitable for?
These films are primarily targeted at those working in molecular genetics and histopathology laboratories. They can also be viewed by anyone wishing to learn more about genomics and whole genome sequencing.
Genomics Education Programme Image Library
https://www.flickr.com/photos/119980645@N06/

The programme has published a collection of free-to-use genetics and genomics images on the image-hosting website Flickr.

100,000 Genomes Project: selected genetic conditions
https://www.genomicseducation.hee.nhs.uk/resources/genetic-conditions-factsheets

Background:
Participants in the 100,000 Genomes Project can opt in to be tested for a list of rare inherited conditions.

Suitable for?
Aimed at healthcare professionals, our factsheets outline the key facts of each condition, plus clinical features, diagnosis, management and treatment.

Genomics in Mainstream Medicine
https://www.genomicseducation.hee.nhs.uk/resources/genomics-in-mainstream-medicine

Background:
The Genomics in Mainstream Medicine Working Group seeks to raise awareness about genomic medicine across a range of specialties. Part of the group’s work includes the production of factsheets outlining the context and impact of genomics on key clinical specialisms

MSc in Genomic Medicine - Education Programme (CPD Modules Available to 2018)
https://www.genomicseducation.hee.nhs.uk/genomicsmsc

Background:
The Master’s in Genomic Medicine has been developed to provide a multi-disciplinary and multi-professional course in genomics that can be applied to clinical practice and medical research. The Master’s will enhance knowledge and skills in this rapidly evolving field.

Suitable for?
1) Directly supporting those professionals involved in the 100,000 Genomes Project and Microbial Genomes work.
2) Supporting the wider transformation of services to integrate genomic knowledge into healthcare.
3) Upskilling existing staff so they can make the most of genomic medicine as it applies to their work.
A
- or hypo-gammaglobulinaemia
Agranulocytosis
Amyotrophic lateral sclerosis or motor neuron disease
Anophthalmia or microphthalmia
Apparent aplastic anaemia or paroxysmal nocturnal haemoglobinuria
Arrhythmogenic Right Ventricular Cardiomyopathy
Arthrogryposis
Atypical Beckwith–Wiedemann syndrome
Atypical haemolytic uraemic syndrome
Auditory Neuropathy Spectrum Disorder
Autosomal dominant deafness
Autosomal recessive congenital ichthyosis

B
Balanced translocations with an unusual phenotype
Bardet–Biedl Syndrome
Bilateral microtia
Brain channelopathy
Brugada syndrome

C
Cardio–facio–cutaneous syndrome
Cataracts
Catecholaminergic Polymorphic Ventricular Tachycardia
Cerebellar hypoplasia
Cerebral folate deficiency
Charcot–Marie–Tooth disease
Choanal atresia
Chondrodysplasia punctata
Classical Beckwith–Wiedemann syndrome
Classical Ehlers–Danlos Syndrome
Classical tuberous sclerosis
Coarse facial features including Coffin–Siris–like disorders
Cockayne syndrome
Combined B and T cell defect
Complex Parkinsonism (includes pallido–pyramidal syndromes)
Cone Dysfunction Syndrome
Congenital adrenal hypoplasia
Congenital anaemias
Congenital Anomaly of the Kidneys and Urinary Tract (CAKUT)
Congenital hearing impairment
Congenital hypothyroidism
Congenital muscular dystrophy
Congenital myaesthenia
Congenital myopathy
Congenital neutropaenia
Congenital disorders of glycosylation
Corneal abnormalities
Costello syndrome
Craniosynostosis syndromes
Cystic kidney disease

Developmental macular and foveal dystrophy
Diabetes with additional phenotypes suggestive of a monogenic aetiology
Dilated Cardiomyopathy
Dilated Cardiomyopathy and conduction defects
Disorders of sex development
Disseminated non-tuberculous mycobacterial infection
Distal myopathies

Ear malformations with hearing impairment
Early onset and familial Parkinson's Disease
Early onset dementia (encompassing fronto–temporal dementia and prion disease)
Early onset dystonia
Early onset familial premature ovarian insufficiency
Early onset or familial intestinal pseudo obstruction
Early onset pancytopenia and red cell disorders
Ectodermal dysplasia without a known gene mutation
Epidermolysis bullosa
Epilepsy plus other features
Epileptic encephalopathy
Erythropoietic protoporphyria, mild variant
Exceptionally young adult onset cancer
Extreme early–onset hypertension
Familial and multiple pulmonary arteriovenous malformations
Familial breast cancer
Familial cerebral small vessel disease
Familial cicatricial alopecia
Familial colon cancer
Familial congenital heart disease
Familial disseminated superficial actinic porokeratosis
Familial exudative vitreoretinopathy
Familial Focal Epilepsies
Familial Genetic Generalised Epilepsies
Familial haematuria
Familial haemophagocytic lymphohistiocytic disorders
Familial hemifacial microsomia
Familial hidradenitis suppurativa
Familial Hypercholesterolaemia
Familial non-syndromic cleft lip and or familial cleft palate
Familial or syndromic hypoparathyroidism
Familial primary spontaneous pneumothorax
Familial pulmonary fibrosis
Familial rhabdomyosarcoma or sarcoma
Familial Thoracic Aortic Aneurysm Disease
Familial tumour syndromes of the central and peripheral nervous system
Familial young-onset non-insulin-dependent diabetes
Fetal hydrops
Fetal structural CNS abnormalities

Gastrointestinal epithelial barrier disorders
Generalised pustular psoriasis
Genetic Epilepsies with Febrile Seizures Plus
Genodermatoses with malignancies
Glaucoma (developmental)

Hereditary ataxia
Hereditary haemorrhagic telangiectasia
Hereditary spastic paraplegia
Holoprosencephaly
Hydroa vacciniforme
Hyperammonaemia
Hyperinsulinism
Hypertrophic Cardiomyopathy
Idiopathic hypogonadotropic hypogonadism
Idiopathic ventricular fibrillation
Infantile enterocolitis and monogenetic inflammatory bowel disease
Infantile nystagmus
Inherited complement deficiency
Inherited macular dystrophy
Inherited non-medullary thyroid cancer
Inherited optic neuropathies
Inherited white matter disorders
Insulin resistance (including lipodystrophy)
Intellectual disability
Intracerebral calcification disorders
IUGR and IGF abnormalities

Joubert syndrome
Juvenile dermatomyositis
Kabuki syndrome
Ketotic hypoglycaemia
Kleine–Levin syndrome and other inherited sleep disorders
Kyphoscoliotic Ehlers–Danlos syndrome

Lactic acidosis
Leber Congenital Amaurosis or Early-Onset Severe Retinal Dystrophy
Left Ventricular Noncompaction Cardiomyopathy
Legius syndrome
LEOPARD syndrome
Limb girdle muscular dystrophy
Long QT syndrome
Lymphoedema distichiasis
Malformations of cortical development
Meige disease
Milroy disease
Mitochondrial disorders
Moyamoya disease
Mucopolysaccharideosis, Gaucher, Fabry
Multi-organ autoimmune diabetes
Multiple bowel polyps
Multiple endocrine tumours
Multiple Epiphyseal Dysplasia
Multiple Tumours

Neonatal diabetes (diagnosed less than 6 months)
Neonatal or paediatric intensive care admission with a likely monogenic disease
Neuro-endocrine Tumours– PCC and PGL
Neurofibromatosis Type 1
Neurotransmitter disorders
Non–CF bronchiectasis
Non–Fanconi anaemia
Non–syndromic familial congenital anorectal malformations
Non–syndromic hypotrichosis
Noonan syndrome
Noonan syndrome plus other features

Ocular coloboma
Osteogenesis imperfecta
Other peroxisomal disorders

Paediatric congenital malformation–dysmorphism–tumour syndromes
Paediatric motor neuronopathies
Palmoplantar keratoderma and erythrokeratodermas
Parathyroid cancer
Peeling skin syndrome
Periodic fever syndromes and amyloidosis
Peroxisomal biogenesis disorders
Peutz–Jeghers syndrome
PHACE(S) syndrome
Primary ciliary dyskinesia
Primary Microcephaly – Microcephalic Dwarfism Spectrum
Proteinuric renal disease

Radial dysplasia
Rare multisystem ciliopathy disorders
Renal tract calcification (or Nephrolithiasis or nephrocalcinosis)
Renal tubular acidosis
Resistance to thyroid hormone
Rhabdomyolysis and metabolic muscle disorders
Rhomboencephalosynapsis
Rod Dysfunction Syndrome
Rod–cone dystrophy

SCID
Severe familial anorexia
Severe hypertriglyceridaemia
Severe multi–system atopic disease with high IgE
Significant early–onset obesity with or without other endocrine features
and short stature
Silver Russell syndrome
Simpson–Golabi–Behmel syndrome
Single autosomal recessive mutation in rare disease
Skeletal Muscle Channelopathies
Sotos syndrome
Stickler syndrome
Structural basal ganglia disorders
Syndromic cleft lip and or cleft palate
Syndromic congenital heart disease

Thoracic dystrophies

Ultra–rare undescribed monogenic disorders
Undiagnosed metabolic disorders
Undiagnosed monogenic disorder seen in a specialist genetics clinic
Undiagnosed neurocutaneous disorders
Unexplained kidney failure in young people
Unexplained monogenic fetal disorders
Unexplained skeletal dysplasia
Unexplained sudden death in the young
V
VACTERL–like phenotypes
Vein of Galen malformation
Vici Syndrome and other autophagy disorders

W
Weaver syndrome

XYZ
Xeroderma Pigmentosum–like disorders
Unless otherwise specifically excluded, all samples from invasive malignancies are eligible. Samples may be from the primary lesion, or from a metastasis. Samples collected at recurrence will only be considered for Whole Genome Sequencing if there is a primary sample available: either stored or previously submitted. Recurrent tumours without a primary sample may be submitted where advised in writing.

Approved cancer conditions to date are invasive forms of the following cancer types. Any rare malignancy within these organs are eligible unless specifically excluded:

| A | Adult Brain Tumours | L | Lung cancer |
| B | Bladder cancer | M | Melanoma |
|   | Breast cancer | O | Ovarian cancer |
| C | Cancer of Unknown Primary | P | Prostate cancer |
|   | Childhood Solid Tumours | R | Renal cancer |
|   | Colorectal cancer | S | Sarcoma (including paediatric and adult sarcoma) |
| E | Endometrial cancer | T | Testicular cancer |
| H | Haematological Malignancies | U | Upper gastrointestinal (GI) tumours |

If you would like to view the latest Eligibility Criteria documents please visit our website: [www.bit.ly/nencqmc-info](http://www.bit.ly/nencqmc-info)