



100,000 Genomes Project

Paving the way to Personalised Medicine

A message from Sue Hill, Chief Scientific Officer



The 100,000 Genomes Project is at the heart of our efforts to transform healthcare in England from a one-size-fits-all system to a targeted, personalised approach where patients will get individualised treatment based on their unique characteristics.

The project aims to sequence 100,000 whole human genomes (all the information in our DNA that makes us who we are) from patients with rare diseases and their families, and patients with common cancers. By coupling individual genome sequences with clinical information about a patient's condition we can create a better understanding of the causes of disease.

The project will identify potential new diagnostics to identify and track people's conditions, drive the development of new drugs and find new uses for existing drugs. It will also establish new relationships with academia and industry for patient benefit.

It will enable scientists and doctors to understand the underlying cause of disease, provide new insights into the prediction and prevention of disease and allow precision diagnoses to be made. This will drive the NHS into a new era, where the personalisation of treatments will lead to improved responses, fewer side-effects and better overall outcomes.

NHS England has established 13 NHS Genomics Medicine Centres to cover the country delivering an end-to-end genomic medicine pathway. They are enabling the participation of patients and family members with their informed consent; collecting samples to extract DNA; capturing clinical information to inform the interpretation of the genome sequence; and establishing the infrastructure to make genomic medicine a routine part of NHS care.

Already we've seen the NHS transform its ways of working to embrace these new technologies, through:

- active patient and public participation in designing the genomics service
- the creation of cross-specialism genomics multi-disciplinary teams
- the capture of clinical information into new and standardised datasets
- a specially-trained workforce engaged in informing and consenting participants for genomic medicine
- new laboratory services and streamlined clinical pathways focused on new approaches to ensure samples of the highest quality
- collaboration across multiple hospital trusts and with academic partners.

The UK is recognised worldwide as a leader in genomics and the unique structure of the NHS is allowing us to deliver these advances at scale and pace for patient benefit. The 100,000 Genomes Project is cementing the NHS's position as one of the most advanced healthcare systems in the world, and is providing the foundation for a new era of personalised medicine.

The radical transformation within the NHS GMCs has been unprecedented, particularly given the challenging times we are all working under, and I have found the enthusiasm of clinical staff, managers and patients for the 100,000 Genomes Project truly inspirational. We hope by sharing this work to communicate this sense of passion and our commitment to improving care for all, for now and the future.

A handwritten signature in black ink that reads "Sue Hill". The signature is fluid and cursive, with a long horizontal stroke at the bottom.

Prof Sue Hill OBE
NHS Chief Scientific Officer for England

NHS Genomic Medicine Centres

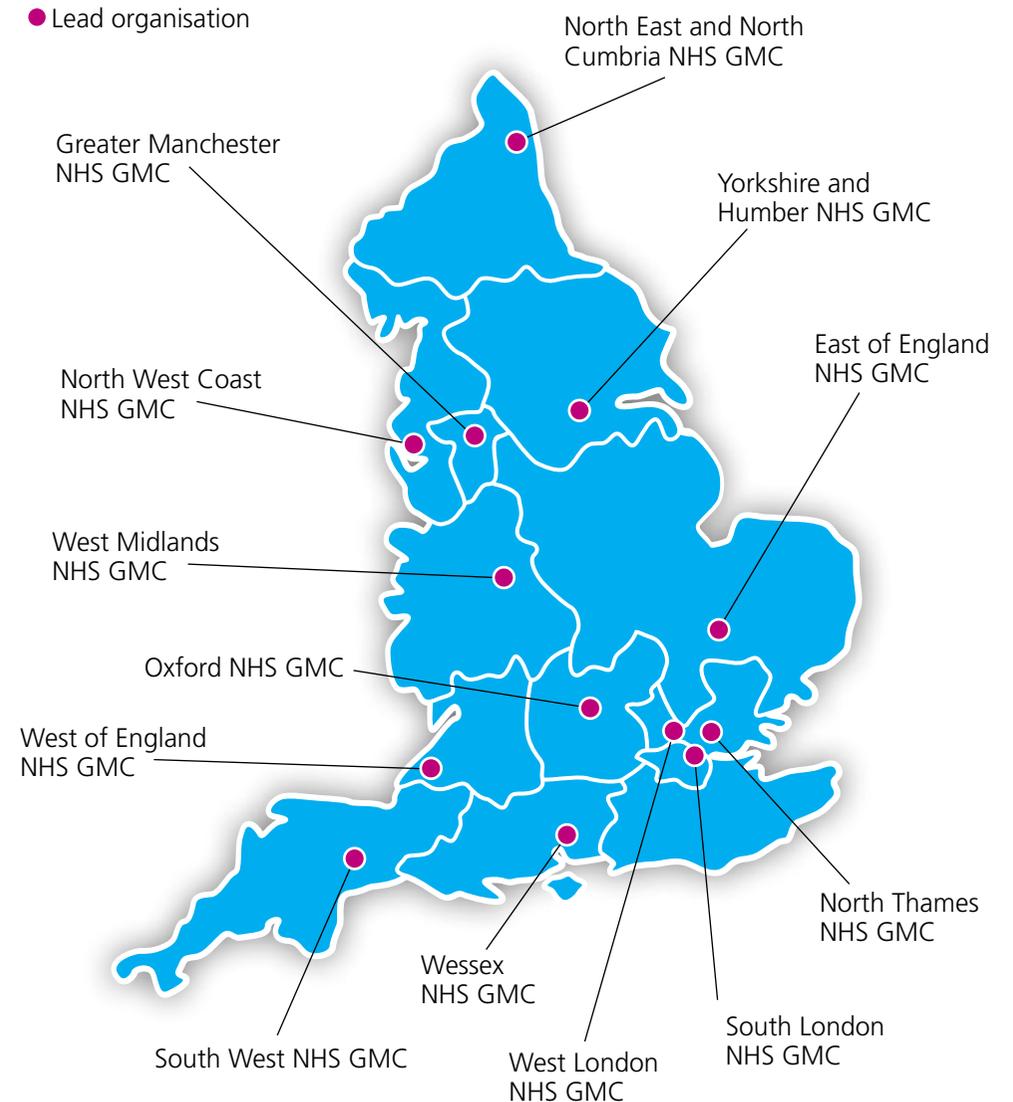
Each NHS Genomic Medicine Centre (GMC) has a lead NHS organisation responsible for a specific geography covering a population of several million. In turn, the NHS GMC works with other NHS organisations within their boundary as Local Delivery Partners to ensure that as many eligible patients as possible are able to take part in the 100,000 Genomes Project. There are 13 NHS GMCs spread across England, 11 that were formed in December 2014 and two that were formed in December 2015.

Every step of the 100,000 Genomes Project – from identification, recruitment, consent, sample collection and processing, data capture and submission through to the validation of whole genome sequencing findings, feedback of information to individuals and their subsequent treatment and management – requires the consistency and academic rigour of a huge multi-site scientific research programme.

All NHS GMCs work to a highly detailed, and constantly updated, service specification set by NHS England, which at its core is about delivering to high quality standards and establishing an infrastructure to support personalised medicine.

This common approach ensures consistency and coherence across the NHS to gain maximum benefits from this application of genomic technologies and curation of knowledge, and is laying the foundations for the future.

NHS Genomic Medicine Centres have been explicitly designed to drive integration between existing laboratory and genetics services, other diagnostic services and broader clinical specialities. This focus on genomics is already beginning to drive improvements in standards, quality and practice across many areas, delivering benefits in outcomes for all NHS patients and becoming part of the routine diagnostic pathway.



Yorkshire & Humber NHS GMC

The Yorkshire and Humber NHS GMC is pioneering the way genomic services are delivered to NHS patients, both now and in the future, to one of the largest and most geographically diverse areas in the country.

Serving a population of over 5.3 million across a mix of urban and rural locations, we are a Yorkshire and Humber-wide organisation drawing on the strengths and expertise of our key partners, including our lead organisation Sheffield Teaching Hospitals NHS Foundation Trust and joint collaborators Leeds Teaching Hospitals NHS Trust and Sheffield Children's Hospital NHS Foundation Trust.

Supported by the Yorkshire and Humber Academic Health Science Network, we are working in collaboration with our 11 local delivery partners, comprising all 22 health Trusts across the region.

We aim to create a lasting legacy to bring personalised medicine to NHS patients by:

- Engaging with clinicians across all areas of medicine, including cardiologists, neurologists and ophthalmologists, to ensure the use of genetic technologies is rapidly transferred to other areas of medicine.
- Developing a transformational informatics infrastructure where joined-up genomic data can be securely collected, shared and collated across the region.
- Providing bespoke educational and training opportunities to professionals and students from healthcare and science backgrounds. This includes an MSc in Genomic Medicine provided by the University of Sheffield.



- Undertaking research in genomic medicine to further build knowledge and expertise in the field.



- Proactively representing and embedding the views of patients, families and members of the public into our work.
- Engaging with the public about genomic medicine and its future impact by participating in key public events such as the University of Sheffield's Life: A Festival of Health, from Head to Toe and other public-facing events.

Lead organisation:

Sheffield Teaching Hospitals NHS Foundation Trust.

Partner organisations:

Leeds Teaching Hospitals NHS Trust, Sheffield Children's NHS Foundation Trust, Airedale NHS Foundation Trust, Barnsley Hospital NHS Foundation Trust, Bradford Teaching Hospitals NHS Foundation Trust, Calderdale and Huddersfield NHS Foundation Trust, Doncaster and Bassetlaw Hospitals NHS Foundation Trust, Harrogate and District NHS Foundation Trust, Hull and East Yorkshire Hospitals NHS Trust, The Mid Yorkshire Hospitals NHS Trust, Northern Lincolnshire and Goole NHS Foundation Trust, The Rotherham NHS Foundation Trust, Wellcome Trust, Yorkshire & Humber Academic Health Science Network, York Teaching Hospital NHS Foundation Trust.

For further information please contact:

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North East and North Cumbria NHS GMC

The North East and North Cumbria NHS GMC covers County Durham, Northumberland, Teesside, Tyne and Wear, northern Cumbria and parts of North Yorkshire, serving a population of 3.1 million. Our NHS GMC is located within the International Centre for Life, in Newcastle upon Tyne, and is based in the Northern Genetics Service run by the Newcastle upon Tyne Hospitals NHS Foundation Trust.

The Northern Genetics Service provides a diagnostic and genetic counselling service across the North East and Cumbria for individuals and families with problems or susceptibilities relating to genetic disorders. Working through established regional networks our NHS GMC is able to harness the collective energy of rare diseases specialists in the identification of participants who will benefit from the project, and through the additional information and value that Whole Genomic Sequencing may offer.

The 100,000 Genomes Project is already acting as a catalyst for transformation in our region:

- Driving forward the use of barcode systems for sample tracking within the Northern Genetics Service laboratories.
- Developing the role of the Clinical Genomic Practitioner, to provide information and specialist support to participants and recruiting clinicians.
- Developing a 'flag' system with the Newcastle hospitals electronic record system. This is linked to mainstream medical records, which ensures that all services across Newcastle Trust will be aware of 100,000 Genomes Project participants. This will be extended to clinical trials and other projects, and we hope to investigate the introduction of similar systems to our local delivery partner sites in the future.



- Embedding genomics into medical student education as part of the 'State of the Art' lecture series.
- Informing the genomics curriculum of nursing degree courses at local universities, preparing the future workforce for the impact of genomic medicine.
- Working with STEMNET and STEM Learning through the Regional Technology Centre to develop a series of engaging activities and lesson plans for use by STEM Ambassadors to deliver dynamic education sessions in genomics to both primary and secondary school students across the region.
- Investing in an automated liquid handling system to improve the quality and throughput of sample processing. This process will be used to confirm variants found in the 100,000 Genomes Project and the automation will increase the efficiency of the procedures carried out by the Northern Genetics Service for the benefit of future patients.



Lead organisation:

Newcastle upon Tyne Hospitals NHS Foundation Trust.

Partner organisations:

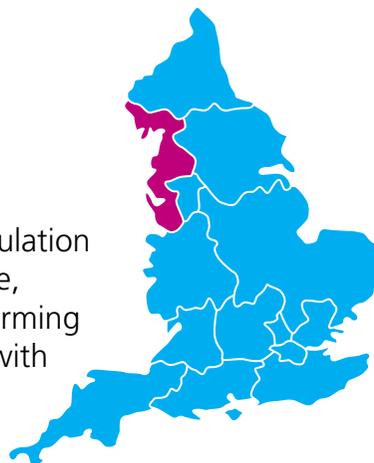
South Tees Hospital NHS Foundation Trust (FT), County Durham and Darlington NHS FT, Northumbria Healthcare NHS FT, City Hospitals Sunderland NHS Foundation Trust, City Hospitals Sunderland NHS FT, Gateshead Health NHS FT, North Tees and Hartlepool Hospitals NHS FT, South Tyneside NHS FT, North Cumbria University Hospital NHS Trust.

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#Genomes100K #NENCGMC www.genomicsengland.co.uk

North West Coast NHS GMC

The North West Coast NHS GMC covers a population of 4.3 million across West Cheshire, Merseyside, Lancashire and South Cumbria. We are transforming the way we deliver genomic services working with our Local Delivery Partners to deliver patient benefit and improve health outcomes.



Our NHS GMC is recruiting NHS patients with rare diseases and their families, in addition to patients with certain types of cancer – lung, breast, ovarian, sarcoma and brain tumours – and hope to expand this list to include paediatric tumours, haematological tumours and pancreatic tumours.

We have already begun the transformation journey, driving the delivery of a mainstreamed genomic service for all medical specialties through education, training and support, and have included our Patient and Public Involvement (PPI) forum at every step in our journey.

In addition we have:

- Developed strong Clinical Leadership and a Clinical Champions Network across our healthcare professionals' workforce that will be critical to the Multidisciplinary Team (MDT) working required for validating genomic results, clinical interpretation and reporting results back to patients.
- Optimised and standardised our laboratory processes, ensuring the highest quality at every step in all our partner laboratories processing samples, to ensure that no patient involvement is wasted.
- Integrated our partners on to one genomic informatics database (STRATA) and have a vision to create a regional Clinical Research Informatics infrastructure, linking local academia and NHS colleagues to one genomics data repository.

- Developed an education and training programme for healthcare professionals, commissioners and the public, across the NHS GMC footprint, to raise awareness of genomics and its benefits to healthcare and further facilitate mainstreaming genomics into primary and secondary care.



All of this has been delivered within a strong governance framework, with robust programme management, passionate clinical leadership and an excellent communications strategy.

Lead organisation:

Liverpool Women's Hospital NHS Foundation Trust.

Partner organisations:

Royal Liverpool and Broadgreen University Hospitals NHS Trust, The Walton Centre NHS Foundation Trust (FT), Liverpool Heart and Chest Hospital NHS FT, Alder Hey Children's Hospital NHS FT, the Countess of Chester NHS FT, Lancashire Teaching NHS FT, Aintree University Hospital NHS FT, Innovation Agency (Academic Health Science Network for the North West Coast); Liverpool CCG and Liverpool Health Partners.

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Greater Manchester NHS GMC

Greater Manchester is one of the largest metropolitan areas in the country, comprising the metropolitan boroughs of Bolton, Bury, Oldham, Rochdale, Stockport, Tameside, Trafford and Wigan, and the cities of Salford and Manchester. The Greater Manchester NHS GMC supports a population of over 3.5 million in the North West of England.



We are recruiting NHS patients with rare diseases and their families, in addition to patients with certain types of cancer, with an initial focus on ovarian and breast cancer.

In the Greater Manchester NHS GMC, we are driving change in genomic medicine by:

- Our interactions with healthcare professionals in all areas of medicine across the whole region including cardiology, neurology, respiratory medicine, oncology, renal medicine and paediatrics.
- Streamlining the way that we deliver precision medicine genetic tests in cancer so that patients can be prescribed the most effective treatments.
- Using newly designed computer-based tools to bring together clinical data, genomic test results and other clinical investigations to enhance multi-disciplinary meetings and ensure optimum patient care.
- Initial results are leading to changes in clinical care - providing personalised treatment for patients.
- Engaging with patients through 100,000 Genomes Project participant days, where updates are provided and there are opportunities for discussion about the project and its progress.

'This may not help my son now, but having the knowledge and being well informed will help future generations' (a 100,000 Genomes Project participant)



- Engaging with the public about the power of genomics in healthcare through Manchester Science events including the European City of Science, Festival of Science and Pint of Science.
- Delivering training and teaching to Health Care Professionals, including hundreds of junior doctors across our NHS GMC footprint, as well as liaising with GPs to raise awareness of genomics.

Lead organisation:

Central Manchester University Hospitals
NHS Foundation Trust.

Partner organisations:

The University of Manchester, The Christie NHS Foundation Trust, University Hospital South Manchester, Salford Royal NHS Foundation Trust and Greater Manchester Academic Health Science Network.

For further information please contact:

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West Midlands NHS GMC

The West Midlands NHS GMC covers urban centres in Birmingham, the Black Country, Coventry and Stoke-on-Trent as well as predominantly rural shires. NHS trusts across our footprint serve a population of 5.6 million.

We are recruiting patients with rare diseases, plus their families, and all eligible cancers.

Among those is seven-year-old Nile Thackery Lunn (pictured on cover), who was the first patient to be recruited at the Royal Wolverhampton NHS Trust.

Nile has a number of complex health issues which doctors have been unable to diagnose and mum Kelly hopes that taking part in the 100,000 Genomes Project may provide some answers – as well as potentially helping others in the future.

Our NHS GMC is driving change in the delivery of genomic medicine in a number of ways:

- Working in partnership, the West Midlands NHS GMC will support the IQIPS and UKAS service accreditation process for all GMC partners' diagnostic departments, to help them deliver to the same standards across the region.
- Working with the West Midlands Academic Health Science Network (WMAHSN), the Institute of Translational Medicine and the Centre for Rare Diseases will enable collaborative developments in working practice and training across all specialist areas, with clinical staff better able to support the patient journey and transform health services for a better patient experience.
- Enhancing clinical and genetic records through a new clinical data capture tool to interact with current electronic patient records (EPR). This will provide a one-stop arena for clinical and genetic



information for future cohort studies and support future clinical decision-making. Working collaboratively across the West Midlands, the supply and support of this system will encourage knowledge sharing among Local Delivery Partners to ensure a legacy that will benefit patients for generations to come.

Further to interaction with current EPR, the project team is aiming to offer genome testing not just to newly-recruited patients but also to undiagnosed patients seen over the past 25 years. Enhanced registers and classification will support tracking of this cohort.

Lead organisation:

University Hospitals Birmingham NHS Foundation Trust (UHB) in partnership with the University of Birmingham.

Partner organisations:

Birmingham Children's Hospital NHS FT, Birmingham Women's Hospital NHS FT, Heart of England NHS FT, Sandwell and West Birmingham Hospitals NHS Trust, University Hospitals of North Midlands NHS Trust, The Royal Wolverhampton NHS Trust, Royal Orthopaedic Hospital NHS FT, University Hospitals Coventry and Warwickshire NHS Trust, George Eliot Hospital NHS Trust, South Warwickshire NHS FT, Worcestershire Acute Hospitals NHS Trust, Wye Valley NHS Trust, Dudley Group NHS FT, Shrewsbury and Telford Hospital NHS Trust, Robert Jones and Agnes Hunt Orthopaedic Hospital NHS FT, Burton Hospitals NHS FT, Walsall Healthcare NHS Trust.

For further information please contact:

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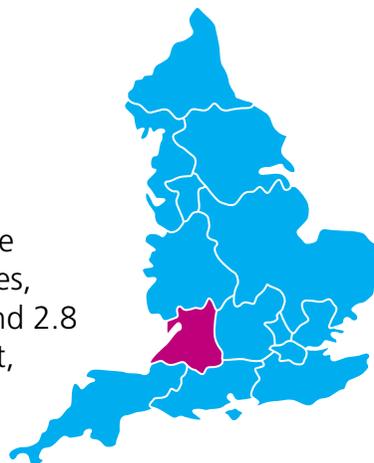
West of England NHS GMC

The West of England NHS GMC was part of the second wave of NHS Genomic Medicine Centres, established in December 2015. We serve around 2.8 million people, covering Bristol, Bath, Somerset, Wiltshire and parts of Gloucestershire.

We are recruiting NHS patients with rare diseases plus their families, and patients with certain types of cancer. Our ambition is to collect 4,650 samples by March 2018. In order to meet these aims, our NHS GMC has established collaborations with partner organisations within the region, including five NHS Provider Trusts led by University Hospitals Bristol, three universities, seven Clinical Commissioning Groups, Health Education South West, NHS England South West, Avon and Wiltshire Mental Health Partnership NHS Trust, West of England Clinical Research Network and the West of England Academic Health Science Network. The West of England NHS GMC works closely with NHS England and Genomics England to ensure national standardisation of enrolment practice and transformation programmes.

The West of England NHS GMC is driving change through the delivery of genomic medicine by:

- Working with the laboratory facilities at North Bristol NHS Trust – we received excellent feedback from NHS England on the facilities.
- Harnessing the latest technology – our Connecting Care system will be used across our NHS GMC to allow clinicians access to shared patient data.
- Breaking down barriers and working together – we were able to develop clinical pathways for the NHS GMC project quite quickly as our consultant, laboratory and consenting teams worked really well together.



- Supporting innovation – we are embracing development of a genetic variant IT tool at North Bristol Trust which NHS England/ Genomics England are already interested in supporting.



Lead organisation:

University Hospitals Bristol NHS Foundation Trust.

Partner organisations:

Gloucestershire Hospitals NHS Foundation Trust (including Gloucester Royal & Cheltenham General), Royal United Hospital Bath NHS Foundation Trust, Weston Area Health NHS Trust, North Bristol NHS Trust (Southmead Hospital).

For further information please contact:

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www.wegmc.org

South West NHS GMC

The South West NHS GMC includes seven acute trusts in Devon, Cornwall and Somerset with support from the South West Academic Health Science Network and Health Education South West. We are recruiting patients with rare diseases or cancer from a population of 2.2 million in Somerset, Devon, Cornwall and the Isles of Scilly.



We are building on an international reputation of excellence in monogenic diabetes research that has been translated into an accredited genomic testing service with full clinical interpretation and advice on diabetes management. Education of healthcare professionals through our UK network of Genetic Diabetes Nurses is a key goal in delivering this exemplar of personalised medicine. The Exeter NHS Genetics Laboratory has developed a highly successful, rare disease exome sequencing service with a diagnostic yield >50% that lays the foundations for interpretation of whole genome sequencing data in routine clinical service.

Recruitment for rare disease includes referrals from many different specialties (paediatrics, neurology, renal, endocrinology & diabetes, cardiology, ophthalmology, immunology and dermatology) in addition to clinical genetics. Patients with bladder, breast, colorectal, endometrial, sarcoma and renal cancer have been recruited and this will soon be extended to include melanoma, testicular, lung, prostate, brain, ovarian and upper GI cancers.

Creating a legacy for genomic medicine within the South West:

- The University of Exeter Medical School was appointed as the 10th provider for the Health Education England commissioned MSc in Genomic Medicine. The course has attracted considerable interest and will be delivered in collaboration with the Universities of Plymouth, Bath, Bristol and the West of England.

- In collaboration with Health Education South West we have appointed 7 Genomics Champions, specialty trainees in cancer, paediatrics, endocrinology, neurology, immunology and ophthalmology. They will follow a train the trainer model, working with the Genomics team in Exeter to develop their own expertise before disseminating this new knowledge across the region to other trainees, consultants and specialist nurses within their disciplines. They will also promote recruitment and support MDT interpretation of results as well as developing themselves as future leaders of mainstream genomic medicine.
- A highly successful engagement event in June 2016 was hosted by the SW AHSN and attended by nearly 100 participants and healthcare professionals from across the South West.
- Workforce transformation is evidenced by Cancer Nurse Specialists incorporating recruitment to the project within their job plans and histopathology dissection practitioners' involvement in tumour cut-up.



Lead organisation:

Royal Devon and Exeter NHS Foundation Trust.

Partner organisations:

Plymouth Hospitals NHS Trust, Taunton and Somerset NHS Foundation Trust, Royal Cornwall Hospitals NHS Trust, Torbay and South Devon NHS Foundation Trust, Northern Devon Healthcare NHS Trust and Yeovil District Hospital NHS Foundation Trust.

For further information please contact:

Dr Steve Johnson, South West NHS GMC Project Manager at: rde-tr.swgmc@nhs.net, tel. +44 (0)1392 408 177.

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Wessex NHS GMC

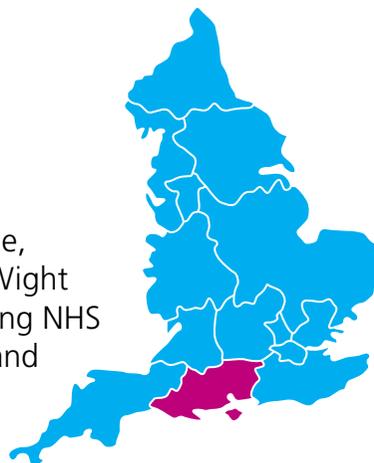
The Wessex NHS GMC serves 3.5 million people, covering Dorset, Wiltshire, Hampshire, Isle of Wight and parts of Surrey and Sussex. We are recruiting NHS patients with rare diseases plus their families, and patients with certain types of cancer.

This year Wessex NHS GMC has rapidly increased the rate at which patients and families are getting involved in the 100,000 Genomes Project. Over 600 patients have joined through the Wessex NHS GMC lead hospital at Southampton, and many more are expected to come from the hospitals in Portsmouth, Winchester and Basingstoke which are now Local Delivery Partners.

Maximising the benefits of genomics for patients and for the NHS needs a step change in awareness and education, both for clinicians and the public. Wessex NHS GMC has created an inclusive and systematic strategy to raise awareness and to educate a broad range of stakeholders, including patients, nurses, clinicians and board members. For example over 50 clinicians have already joined the Genomics MSc, and those numbers will be bolstered by a multi-media campaign to raise project awareness that includes screen savers, posters, and drop-in Genomics Cafes aimed at both the public and clinicians throughout our hospital sites. Off-site, the team has also engaged with the Café Scientifique network to schedule a series of public talks across the region, and is also hosting an event at Southampton Science Park on the commercial opportunities arising from genomics.

Our NHS GMC is driving change in genomic medicine in the Wessex area by:

- Strengthening our clinicians' awareness and knowledge of genomic medicine e.g. a new easier referral system for genomics, new medical pathways including genomics, broad engagement in the 100,000 Genomes Project across specialities.



- Informing, motivating and supporting healthcare colleagues across the region through the Wessex Academic Health Sciences Network and a web of multi-disciplinary clinical meetings, backed by comprehensive training through the University of Southampton's HEE commissioned Genomic Medicine MSc.
- Promoting a culture of multi-professional inclusion to solve complex clinical care service redesign challenges, particularly in the areas of digital data handling and information technology with new teleconferencing between Wessex Trusts.
- Aggregating expertise across the spectrum of genomic medicine's clinical, scientific, educational and societal aspects to collectively deliver an improved and sustainable service for patient and families with rare diseases and cancer.

Lead organisation:

University Hospital Southampton NHS Foundation Trust in collaboration with the University of Southampton.

Partner organisations:

Portsmouth Hospitals NHS Trust, Hampshire Hospitals NHS Foundation Trust (FT), Solent NHS Trust, Isle of Wight NHS Trust, Royal Bournemouth and Christchurch Hospitals NHS FT, Poole Hospital NHS FT, Brighton & Sussex University Hospitals NHS FT, Salisbury NHS FT and Dorset County Hospital NHS FT. This network is coordinated through the Wessex Academic Health Science Network.

For further information please contact:

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South London NHS GMC

The South London NHS GMC covers South London and reaches out into parts of Kent, Surrey and Sussex, supporting a combined population of up to 7 million. We work across South London in relation to both rare diseases and cancer, and into Kent, Surrey and Sussex for rare diseases.



We are working with NHS patients who have rare diseases and their families, as well as patients with certain types of cancer. As part of the 100,000 Genomes Project our work will contribute to a better understanding of the genetic causes of these conditions.

Our NHS GMC is driving change in genomic medicine for our patients by:

- Working in partnerships through an alliance of 11 organisations including two Academic Health Science Networks, two universities and two patient organisations – all contributing to the development of genomic medicine services in the region.
- Bringing about improvements in patient pathways – for example, streamlining sample handling and traceability, improving automated data capture, increasing genetic laboratory capacity and strengthening communications amongst those involved in treating patients.

Key achievements to date:

Delivery partners across South London NHS GMC have been working together closely to transform genomic medicine in the region and give our patients the opportunity to participate in the 100,000 Genomes Project. Collaboration and engagement across multiple clinical specialties has meant that more than 2,000 participants from over 100 rare diseases and seven common cancers have been recruited to the project at our NHS GMC, and this is continuing to expand across eligible diseases.

South London NHS GMC is also providing genomic training and education for healthcare professionals across multiple disciplines to support workforce transformation. As part of this, the NHS GMC has developed a Massive Open Online Course (MOOC) on the Future of Genetics in Medicine, which has been accessed by over 10,000 individuals. Further details on courses and education materials can be found on the South London NHS GMC website.



Lead organisation:

Guy's and St Thomas' NHS Foundation Trust.

Partner organisations:

King's Health Partners, including Guy's and St Thomas' NHS Foundation Trust (FT), King's College Hospital NHS FT, South London and Maudsley NHS FT; St George's University Hospitals NHS FT; two universities: King's College London (a partner in King's Health Partners) and St George's University of London; two major patient groups: Macmillan Cancer Support and Genetic Alliance UK; two Academic Health Science Networks (AHSNs): Health Innovation Network (South London AHSN) and Kent, Surrey and Sussex AHSN; Viapath (a pathology provider).

For further information please contact:

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West London NHS GMC

West London NHS GMC offers genetic sequencing to our NHS patients who choose to participate in the 100,000 Genomes Project. This includes patients with more than 200 different rare diseases and certain types of cancer – including bladder, renal, breast, colon, lung, ovarian and prostate.



We serve a population of 2.5 million and through our four NHS partners we cover the London boroughs of Kensington, Chelsea and Hammersmith & Fulham through to Sutton and Hillingdon.

We continuously engage and involve our patients making sure they are at the heart of what we do and who we were. We were one of the first GMCs to put together a Patient and Public Involvement (PPI) steering group, we have piloted a local evaluation and are leading on the national service evaluation to make sure patient feedback leads to on-going improvement.

Our vision is to improve the health of our community through integrating personalised medicine into healthcare. We work closely in partnership with Imperial College London and the Academic Health Science Network (AHSN) to ensure that education and transformation are at the heart of our work.

The 100,000 Genomes Project is supported by the expertise of our four internationally recognised partners to improve the way people are cared for - allowing genomic diagnoses to target investigations and treatments more efficiently and facilitating new medical research to translate into improved treatments.

Rare diseases:

Our four trusts include eight hospitals that are internationally recognised for their contributions and have strong histories of genomic research transforming the treatment of patients with rare diseases: Charing Cross, Chelsea and Westminster, Hammersmith, Harefield, Royal Brompton, Royal Marsden (London and Surrey), St Mary's and West Middlesex hospitals. These hospitals are working closely together to further integrate the benefits of genomics into their services.



For cancer: The Royal Marsden was a leading recruiting centre for the Cancer Research UK Stratified Medicine Programme. This programme laid the groundwork for current NHS GMC operations, enabling rapid implementation of genomic medicine into routine clinical care for cancer patients treated there and at Imperial College Healthcare.

Lead organisation:

Imperial College Healthcare NHS Trust.

Partner organisations:

Chelsea and Westminster Hospital NHS Foundation Trust (FT), Royal Brompton & Harefield NHS FT and The Royal Marsden NHS FT.

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North Thames NHS GMC

The North Thames NHS GMC is revolutionising the way we deliver genomic services to a population of over 6 million, primarily recruiting patients from the north and east of London, Essex, Hertfordshire and Bedfordshire but, due to the wide geographical spread of rare diseases in the UK, we will also be collecting some samples from patients across the country.



North Thames NHS GMC is a centre dedicated to the collection and study of genetic samples from patients and families with rare inherited diseases and certain types of cancer, including colorectal, lung, and breast. In addition to recruiting patients to the 100,000 Genomes Project, our centre is looking to transform genomic services in the region by increasing awareness and access to research studies for patients and families who have rare conditions.

We are working with our partners to deliver tangible benefits for patients. We have begun the transformation of services by:

- Spreading the 100,000 Genomes Project to new partner Trusts.
- Driving an increase in awareness of all research opportunities across clinical teams who are delivering care to people with conditions potentially eligible for the 100,000 Genomes Project (through the Clinical Research Network).
- Investing in a regional informatics strategy that will provide benefits beyond the 100,000 Genomes Project.
- Sharing learning with other NHS Genomic Medicine Centres.
- Actively co-designing innovative information resources for young people about genomics and to explore the best way to deliver genomic testing to maintain informed choice.



Lead organisation:

Great Ormond Street Hospital for Children NHS Foundation Trust.

Partner organisations:

Barts Health NHS Trust, London North West Healthcare NHS Trust, Moorfields Eye Hospital NHS FT, Royal Free London NHS Foundation Trust (FT), Royal National Orthopaedic Hospital NHS Trust, and University College London Hospitals NHS FT.

For further information please contact:

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 @UCLPartners #Genomes100K  www.ntgmc.nhs.uk

Oxford NHS GMC

The Oxford NHS GMC is a partnership between NHS trusts in Oxfordshire, Buckinghamshire, Berkshire, Wiltshire, and Surrey. Together, these trusts serve a population of 3 million.

The Oxford NHS GMC is recruiting NHS patients with rare diseases and their families, and patients with many different types of cancer: breast, colorectal, lung, ovarian, prostate, renal, sarcoma, endometrial, testicular, pancreatic, and bladder.

The Oxford NHS GMC is building upon the success of the existing Oxford Molecular Diagnostics Centre, part of Oxford University Hospitals NHS Foundation Trust. The Centre has pioneered the delivery of next-generation sequencing in the NHS – including whole genome sequencing – with positive benefits for patients through new diagnoses and options for treatment.

Our NHS GMC is driving change in genomic medicine for our patients in these ways:

- Through the NHS GMC, the Centre is delivering services for a wider range of diseases across regional cancer networks. It is also building upon the success of the existing Genomic Medicine Multidisciplinary Team (GM-MDT), which has pioneered the integration of whole genome sequencing into clinical processes and patient care.
- The GM-MDT brings together a range of clinicians, researchers and laboratory staff to help identify patients who could benefit from sequencing, provide advice to clinicians on prior testing and assist in the review of clinical results and incidental findings. The NHS GMC programme requires a combination of quality and scale that is difficult to achieve with manual processes for data management.
- Significant investments are being made to ensure that high-quality clinical and laboratory data is available, automatically, wherever it is needed. In particular, comprehensive informatics support is being



developed for the GM-MDT, as well as for MDTs in each of the cancer areas selected for the NHS GMC programme.

- A patient and public group for genomic medicine has been established, ensuring that the design, operation, and communication of the new genomic services is properly informed by the experiences and insights of patients and other interested members of the public.



The Oxford NHS GMC is committed to driving active engagement and participation in transformational genomic medicine. The NHS GMC is working with patients and the public to raise awareness and educate staff through a dedicated unit for training, awareness and academic learning with links to the NIHR Oxford Biomedical Research Centre, Oxford Academic Health Science Centre and Oxford Academic Health Science Network.

Lead organisation:

Oxford University Hospitals NHS Foundation Trust.

Partner organisations:

Milton Keynes University Hospital NHS Foundation Trust, Frimley Park Hospital NHS Foundation Trust, Great Western Hospitals NHS Foundation Trust, Buckinghamshire Healthcare NHS Trust and Royal Berkshire NHS Foundation Trust.

For further information please contact:

The Oxford NHS GMC Programme team at: oxford.gmc@nhs.net
@OUHospitals #Genomes100K www.ouh.nhs.uk/OxfordGMC

East of England NHS GMC

The East of England NHS GMC has the largest geographical area of all the NHS GMCs and supports a combined population of 6.1 million people, covering Bedfordshire, Cambridgeshire, Essex, Hertfordshire, Norfolk and Suffolk, Leicestershire, Nottinghamshire and parts of Northamptonshire, and extending to the East Midlands.



We are recruiting NHS patients with rare diseases plus their families, and patients with certain types of cancer, initially breast, ovary, prostate, bowel, lung and renal.

The East of England NHS GMC is delivering change in genomic medicine by:

- Setting the strategy for the responsibility and transfer of the genome sequencing processes from research laboratories to NHS genetic/genomic laboratories, to make DNA sequencing available routinely as an NHS service, and improving validation quality.
- Building up our joint Patient and Public Involvement capabilities to support the Project, by sharing ideas and best practice across our footprint.
- Improving laboratory processes and systems across the NHS GMC: working together has identified common interfaces and helped to streamline sample transfers between partner organisations and enabled agreements on what will be done where.
- Promoting genomic education and teaching with Healthcare Professionals and stakeholders across the NHS GMC footprint as well as wider network links to UK and international partnerships.



Lead organisation:

Cambridge University Hospitals NHS Foundation Trust.

Partner organisations:

Nottingham University Hospitals NHS Trust, University Hospitals of Leicester NHS Trust, and Norfolk and Norwich University Hospitals NHS Foundation Trust.

For further information please contact:

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 @_EEGMC #Genomes100K  www.eastgenomics.org.uk

For more information about the 100,000 Genomes Project, please contact:

100,000 Genomes Project
Genomics Implementation Unit
Medical Directorate
NHS England
Skipton House | 80 London Road | London | SE1 6LH

 england.genomics@nhs.net

 @NHSEngland #Genomes100k #PersonalisedMedicine

 www.england.nhs.uk/ourwork/qual-clin-lead/personalisedmedicine

For information on genomics education and training for NHS staff, please visit:

www.genomicseducation.hee.nhs.uk

You can also find out more about the 100,000 Genomes Project at:

www.genomicsengland.co.uk

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