NHS England

Informatics Solutions in NHS Genomic Medicine Centres

NHS GMC Networking Event

14 March 2017

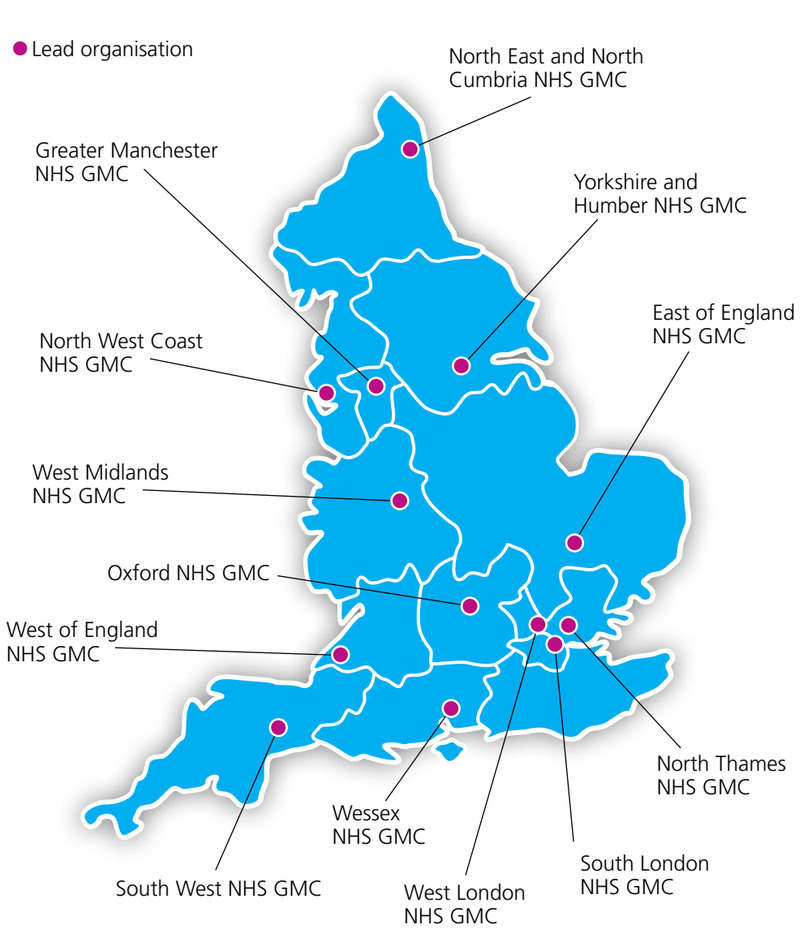
NHS Genomic Medicine Centres

Each NHS Genomic Medicine Centre (NHS 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 participants 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 more in December 2015.

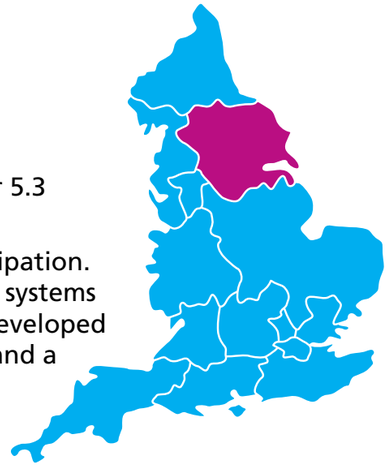
All NHS GMCs have developed a range of informatics solutions to aid identification of participants, sample collection and processing, data capture and submission, up to the validation and feedback of results.

This booklet is a snapshot of our 13 NHS GMCs' informatics successes. It is not intended as an exhaustive guide, but as an invitation to share and celebrate the NHS GMCs' achievements in harnessing the power of data for patient care. Great strides are being made to make molecular diagnostics a reality in the NHS. We would like to take this opportunity to thank our informatics colleagues in all of the NHS GMCs for their dedicated efforts and contributions to this shared endeavour.

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 requires innovative approaches to data management, interpretation and storage – enabling accurate and safe information flows across trusts and geographical boundaries to deliver new molecular diagnostic pathways.



Yorkshire and Humber NHS GMC



The Yorkshire and Humber NHS GMC serves a population of over 5.3 million across a mix of urban and rural locations.

A key priority for our GMC is to remove barriers to clinical participation. Collecting data from paper records and multiple incompatible IT systems is a very onerous task, which is why our informatics team have developed a number of innovative solutions that will have enduring value and a high impact on the interpretation of sequenced data.

Our innovative solutions include:

- The development of a web-based system, HILIS, which allows all stages of patient recruitment to be monitored and diagnostic samples to be tracked.
- The development of a solution, PPM1, that allows the data required for cancer patients to be acquired directly from existing electronic patient records.
- The development of a system, PPM+, that allows a detailed description of patients with rare genetic disease to be characterised using an internationally recognised terminology.

Lead organisation:

Sheffield Teaching Hospitals NHS Foundation Trust.

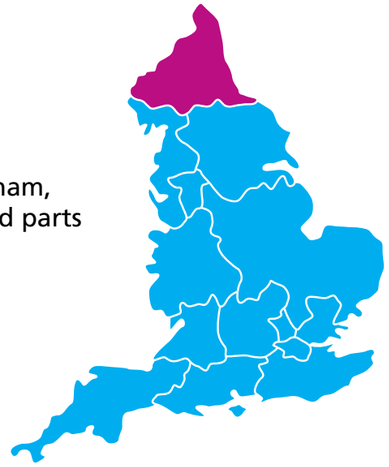
Partner organisations:

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

For further information please contact:

The Yorkshire and Humber NHS GMC Programme team at: gill.wilson@sch.nhs.uk, tel. +44 (0)7788 416 125.
@yhgmc #Genomes100K www.yorkshirehumbergmc.nhs.uk www.facebook.com/YHGMC

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 informatics successes include:

Adoption of 2D barcoded FluidX tubes for use beyond the 100,000 Genomes Project

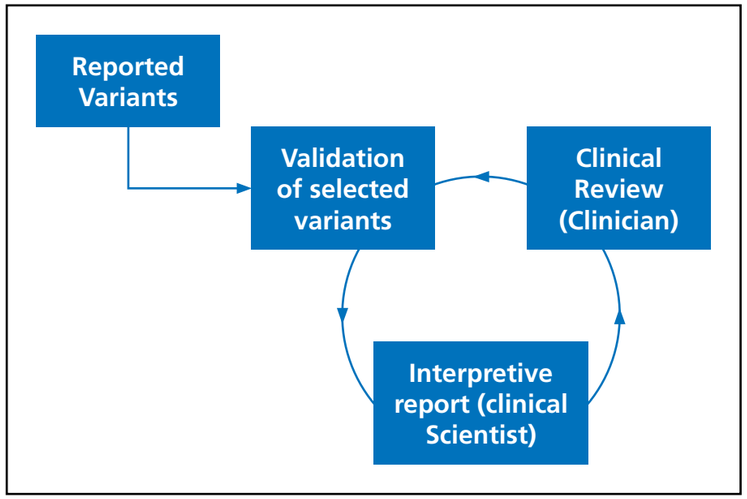
A liquid handling robot has been purchased and commissioned for the setup of PCR/Sanger Sequencing confirmations of reported 100,000 genomes variants. The system includes a module for reading 2D barcoded tubes and has been a catalyst for integrating the use of 2D barcoded FluidX tubes for Primers into the automated PCR programs, reducing the need for time-consuming checks and the potential for operator error, consequently increasing efficiency. These advantages have also benefitted all other tests in the Northern Genetics Service that use these systems, ensuring the legacy of the 100,000 Genomes Project continues beyond its completion.

Virtual Multi-Disciplinary Team (MDT)

Due to the geography and dispersed nature of the North East and North Cumbria NHS GMC catchment area, a traditional genomic MDT is not practical. Instead, a 'virtual MDT' has been established. It consists of a feedback loop involving:

- Laboratory validation
- Reporting by clinical scientist
- The referring clinician

This virtual MDT informs the validation and reporting process.



Cancer data flows

We have established a system to notify histopathology personnel when a 100,000 Genomes Project cancer participant arrives in surgery. This ensures the laboratory is prepared to receive the tissue sample, that protocol timescales for the processing of fresh tissue are met and that the required data is captured.

Our current focus is on:

- Facilitating the implementation of GS1 barcoding standards.
- Continuing work on the implementation of the GENIE software (see page 10) which will increase the effectiveness and efficiency of a number of processes within the project, such as:
  - allowing e-referral to take place across the Lead Organisation and Local delivery Partners
  - enabling more effective management and overview of recruitment
  - improving the collection of data to meet data models from various clinical systems used by the Lead Organisation and Local Delivery Partners
- Plans are in place to upgrade imaging systems in pathology.
- As a result of the success of the liquid handling robot, the use of 2D barcoded tubes is being considered for all DNA samples, which will provide further benefit to other areas of the genetics service beyond the confines of the project.

Lead organisation:

Newcastle upon Tyne Hospitals NHS Foundation Trust.

Partner organisations:

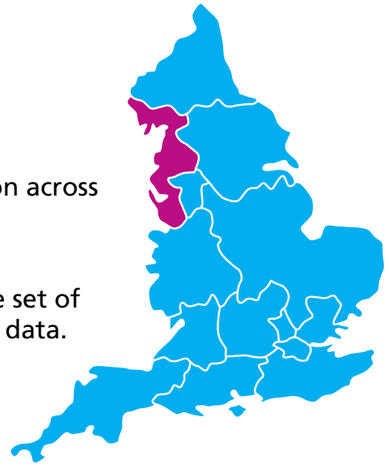
South Tees Hospital NHS Foundation Trust (FT), County Durham and Darlington NHS FT, Northumbria Healthcare NHS FT, City Hospitals Sunderland NHS Foundation Trust, City Hospitals Sunderland NHS FT, Gateshead Health NHS FT, North Tees and Hartlepool Hospitals NHS FT, South Tyneside NHS FT, North Cumbria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

For further information please contact:

Jon Stacey, North East and North Cumbria NHS GMC Project Manager, at: Jonathan.Stacey@nuth.nhs.uk, tel. +44 (0)1912 418 604.

@NewcastleHosps @NENCGMC #Genomes100K www.newcastle-hospitals.org.uk/services/NENC-GMC.aspx

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.

Our informatics successes include:

- Establishing data flows to enable the submission of a complete set of relevant patient demographic, consent, clinical and laboratory data.
- Implementing an Informational technology system, Strata Pathways, to capture clinical management data and patient-specific clinical, laboratory and imaging information – ensuring compliance with NHS national data standards.

Our current focus is on:

- Enabling automated patient demographic data synchronization with local Trust PAS systems directly into Strata Pathways, reducing manual entry by clinical staff.
- Developing interoperability with the national Somerset Cancer Registry to automatically update diagnosis information in line with Genomics England's data model.

Strata Pathways

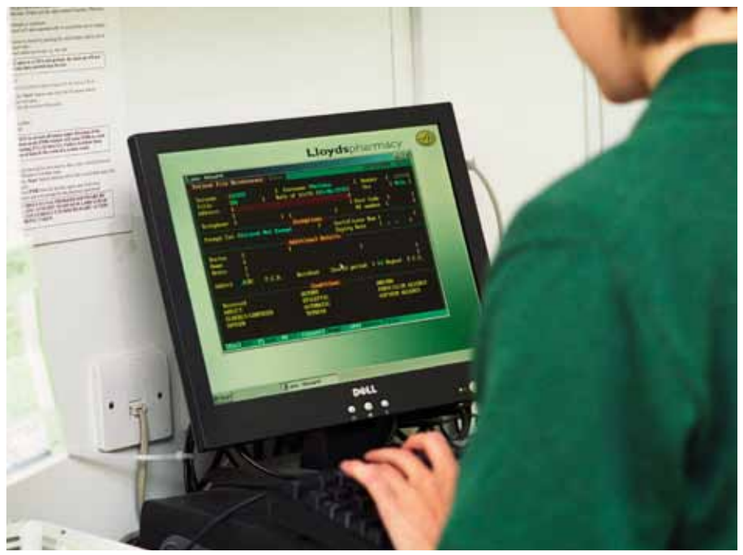
Strata Pathways is an informational technology system designed for the North West Coast NHS Genomic Medicine Centre (NWC GMC).

Using the Strata system, NWC GMC has developed the capability to capture clinical management data and patient-specific clinical, laboratory and imaging information – ensuring compliance with NHS national data standards.

A central repository has been developed that integrates patients' genomic and core data for:

- Sample information – collection and processing
- Clinical data
- Pathology data
- Consent
- Genetic test results

The transformation realised to date is that Strata Pathways has been deployed across one clinical laboratory and four Local Delivery Partners. There are plans in place to implement the system at another four partner trusts.



Value	Result	Efficiency	Effectiveness	Safety
Time saving for clinical staff, as all information readily accessible from one place	Better informed clinicians with improved end user experience	✓	✓	✓
Interoperability – enhanced information sharing	Linking and integrating data across the geography and from other clinical systems e.g. Somerset, Meditech	✓		
Automated Reporting	Reliable and accurate information generated, e.g. sample information available in a timely manner. Reduced manual burden	✓		
SNOMED CT functionality	Compliant with NIB strategy and improved clinical information sharing	✓	✓	
Reduced IT storage space	Released server capacity for other clinical systems and clinical data	✓	✓	
HL7 Messaging enabled	Improved data flows between organisations	✓		
Set up for the future	Principal system established for a centralised Genomic Data Centre for the North West Coast	✓	✓	✓

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.

For further information please contact:

Ed Millensted, North West Coast NHS GMC Programme Lead, at: ed.millensted@lwh.nhs.uk, tel +44 (0)151 702 4207.

Angela Hargreaves, North West Coast NHS GMC Programme Administrator, at: angela.hargreaves@lwh.nhs.uk, tel +44(0)151 702 4427.

@NWCGMC #Genomes100K www.nwcgmc.org

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.

Our GMC has focused on transforming laboratory services through integrating workflows and sample tracking. We are also 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.

Our successes include:

Integrated informatics and transparent workflows

Work is nearly complete on the first phase to harmonise multiple Laboratory Information Systems (LIS) into a single system, transforming how multiple laboratory disciplines inform and operate together. This will integrate with the Trust's Patient Master Index (PMI), which will facilitate electronic referrals (internally from within the Trust and in the future from external referrers). This will also allow for electronic reporting in multiple formats (PDF, XML, text) directly back into patient records and other informatics systems connected to the Trust.

This will primarily be achieved by implementation of the iGene system, which will replace the functionality currently being provided by our multiple legacy systems. As a web-based LIS, iGene can be distributed across multiple locations/sites across the Manchester GMC region and nationally. In addition, under a partnership agreement with the providers, functionality is being developed for management of genomic data.

As well as a new LIS, work has been underway since 2014 on a new Laboratory Information Management System (LIMS). This LIMS will improve sample tracking and traceability for all laboratory workflows, tracking all interactions and integrating with analytical instruments/ automation. This LIMS is also being used to automate the bioinformatics processes from instrument to scientist review, which is expected to improve turnaround times (initially in Pharmacogenetics).

This programme (G-LIMS) will involve the integration of the new LIS and LIMS to each other through their REST APIs. The first phase of the LIS project is due to go-live in April 2017 and the first phase of the LIMS project is due to go-live in March 2017.

Return of genomic results

Currently we issue genetic test results via secure email in PDF. However, work has commenced work with The Christie to share genetic test results electronically using XML data transfer, to offer more secure and accurate transfer of data. The user interface and XML file outputs development has been completed on the Greater Manchester NHS GMC side. The Christie is currently reviewing files and how they will import these into their electronic patient records (EPRs).

DataWell

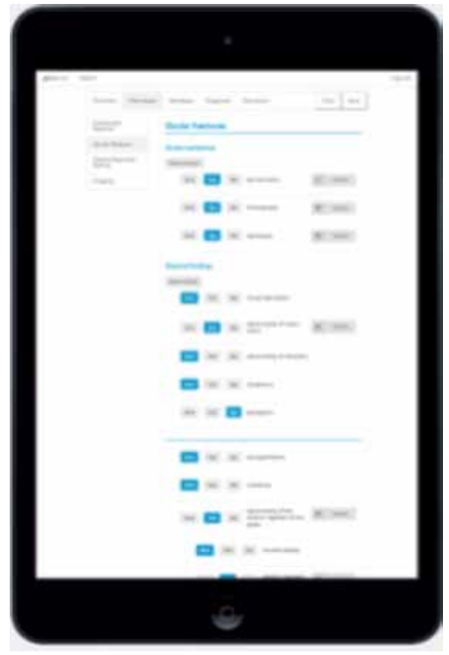
This software platform enables the exchange of NHS patient record information electronically. In Greater Manchester, two sites are fully connected –The Christie NHS Foundation Trust and Salford Royal NHS Foundation Trust, with Central Manchester NHS Foundation Trust and Trafford CCG due to complete connection by end of February 2017. The testing of cross-site pathology data sharing will complete in February 2017.

All health and social care partners in Wigan and in Tameside & Glossop are working with DataWell to link up.

Gen-o

Gen-o is a transformational web-based tool enabling mainstream clinicians to access genomic information in the care of their patients. It is iPad friendly, allowing interaction with patients during secondary care appointments. Gen-o:

- Standardises patient phenotype capture (HPO)
- Provides secure, integrated viewing of images, genomics, phenotypes
- Enables virtual MDTs
- Links HPO to gene names – for an integrated use of Genomics England's PanelApp for prioritisation of genes
- Captures clinician details and patient consent electronically
- Integrates to LIMS and bioinformatics for seamless working
- Presents back results, following expert review and genomic care decisions to secondary care, all in Gen-o.



Lead organisation:

Central Manchester University Hospitals NHS Foundation Trust.

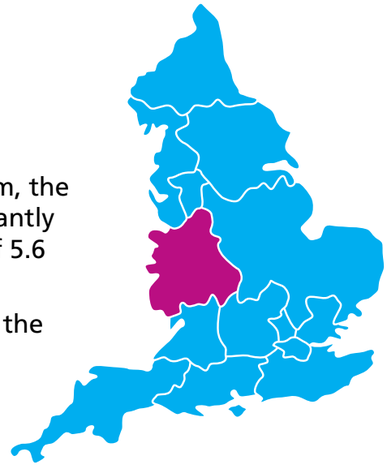
Partner organisations:

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:

Professor Bill Newman, Greater Manchester NHS GMC Clinical Director, at: William.Newman@manchester.ac.uk, tel +44 (0)161 276 4150. @CMFTNHS #Genomes100K
www.cmft.nhs.uk/saint-marys/our-services/manchester-centre-for-genomic-medicine/the-100000-genomes-project

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.

West Midlands GMC (WM GMC) took the decision at the start of the project to develop an application to support data collection and sample tracking across the region. This application, GENIE, is optimised for automated electronic data extraction from multiple systems. A MiniSpine service has been integrated in order to ensure the quality of demographic information.

A pilot project is underway in which GENIE will interoperate with an OpenEHR based system to facilitate extraction and transmission of data originating in varied formats, from multiple systems across a range of organisations.

WM GMC has the largest number of Local Delivery Partners (LDPs) totalling 18, this has meant a different way of working across the whole region, but we have successfully used GENIE in 14 out of 18 LDPs. Its success is paving the way for other region-wide systems to follow on from this project.

In the next phases of the project is a document archive and distribution system using Documentum. This was purchased for LDPs in the region, to support the dissemination of validated results, the first time this has been possible through a unified system. It will leave a legacy for sharing clinical documentation in the future.

A region-wide PACS viewer is in the final stages of this process. Integrated with GENIE, this will allow imaging files associated with 100,000 Genomes Project participants to be easily identified, stored and transmitted. It will also leave a legacy in which imaging can be accessed and shared across LDPs in the West Midlands to improve patient care, both in the emergency and non-emergency setting, for example by facilitating region-wide multi-disciplinary team meetings.

Lead organisation:

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

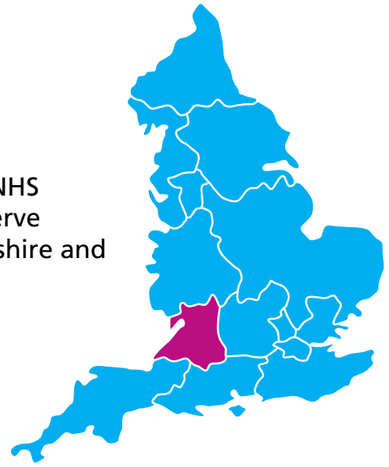
Partner organisations:

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:

Daniel Peters, West Midlands NHS GMC Operational Project Manager, at: wmgmc@nhs.net, tel: +44 (0)121 371 8619.
Rachel Hornabrook, West Midlands NHS GMC Clinical Operations Manager, at: wmgmc@nhs.net
@westmidsgmc #Genomes100K www.westmidsgmc.nhs.uk

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.

Our informatics successes include:

- Developing a bespoke Genomics module within our regional Connecting Care shared care record system. This tracks participants from referral to consent. It is actively used in MDT and clinic management.
- Streamlining data extraction from our regional LIMS system, StarLims.

Our current focus is on:

- Finalising migration of data from Access database to Connecting Care
- Re-aligning our sample selection process and data quality checks to new Connecting Care extracts
- Integrating Connecting Care genomics with regional PAS systems, Pathology and other data sources
- Phase 2 development of Connecting Care to maximise links to required 100,000 Genomes Project data, such as data feeds from pathology and radiology
- Extracting data from Somerset Cancer Register (SCR) for Bristol trusts
- Reviewing cancer diagnosis data quality and completion
- Developing a local integrated electronic solution for the collection of phenotype data for lead trust and Local Delivery Partners (LDPs). This will benefit clinicians and patient care beyond the lifespan of the 100,000 Genomes Project
- Developing XML files to meet cancer dataset version three requirements
- Finding resource for HPO entry to Open Clinica
- Start working with the new pedigree tool
- Working with our LDPs on data extraction and data feeds for cancer dataset version three submission requirements
- Developing an interoperable solution or data repository for the central collation of all 100,000 Genomes Project data not available through Connecting Care
- Strengthening links with LDP informatics specialists via the Informatics Partnership Working Group
- Further streamlining genomics data recording and extraction in our regional LIMS system, StarLims
- Potential to progress the development of a secure webserver on the N3 network to host a web portal for NHS England laboratories to design, store, track stock and report success of Sanger sequencing primers for the 100,000 Genomes Project. Development will be dependent upon the precise nature of results format and national tracking solutions.

Lead organisation:

University Hospitals Bristol NHS Foundation Trust.

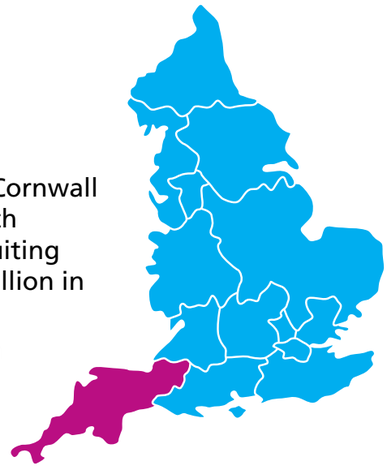
Partner organisations:

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:

Catherine Carpenter-Clawson, West of England NHS GMC Programme Manager at: ubh-tr.wegmc@nhs.net, tel. +44 (0)117 3426526.
@100k_WEGMC #Genomes100K 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.

- Purchase of GENIE informatics system, allowing data collection from all 7 Trusts that form the South West NHS GMC and development work to support the Exeter process for referrals and appointment making
- Starlims user licence and development work for GS1 barcodes
- Path XL tissue mark Software licence for tumour cellularity assessment
- Trakgene development to improve Clinical Genetics data capture
- Fluidx Genomics packs to collect and send samples
- StarLIMS developments (ddPCR integration and emailed reports enhancement)
- Biogene Ltd Mutuation Surveyor, Alamut Visual Licences HGMD licence – to aid validation of report findings
- Beckman Coulter Transfer for PC and software upgrades.

Our current focus is on:

- Purchase of a Cancer Data Management System to replace the Dendrite and Cancer Waiting Times (CWT) Tracker systems at the Royal Devon & Exeter Hospital with a single, interoperable system that has the ability to track timed patient pathways and thus ensure timely diagnosis and treatment. Implementation of the new system will improve data quality, efficiency of data capture by reducing duplication, streamline processes and improve management of MDT meetings (including the Cancer Genomic Medicine MDT meetings). The system will provide seamless integration to other hospital systems including PAS.
- Implementation of electronic patient records – the Regional Clinical Genetics Services currently relies on hard copy patient records which are stored in Exeter for all 35,000 families seen since the clinical service was set up in the 1990s.
- Integration between the Molecular Genetics StarLIMS system and the pathology LIMS system to aid data integration.
- Additional licences for variant interpretation software and a SQL server.
- GS1 barcoding - GS1 barcoded patient wristbands were implemented at the RD&E Trust in 2014. The Kidney Unit at the RD&E provides renal services for a population of around 900,000 with satellite renal clinics in South Molton, Torbay, Honiton, Taunton and Yeovil. Funding is requested to purchase printers for each of these satellite clinics to implement GS1 barcoded patient wristbands.

Lead organisation:

Royal Devon and Exeter NHS Foundation Trust.

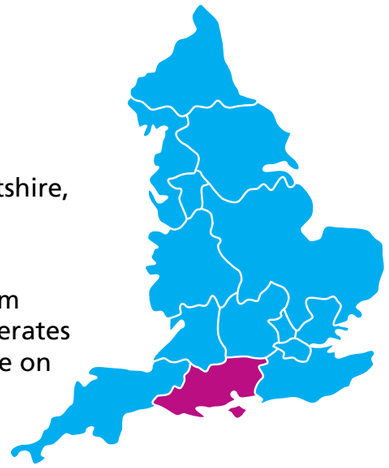
Partner organisations:

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.
@sw_gmc #Genomes100K www.swgmc.org

Wessex NHS GMC



Wessex NHS GMC serves 3.5 million people, covering Dorset, Wiltshire, Hampshire, Isle of Wight and parts of Surrey and Sussex.

Our informatics successes include:

- Developing a platform for retrieving clinical test level data from specialist systems by a specific disease profile. The platform operates across a range of operating systems and can be adapted for use on any research study.
- Creating an integrated platform with existing and new clinical systems to send cancer data directly to Genomics England. This has been available for each iteration of the dataset.
- Creating a secure regional genomics portal, currently in use at four Local Delivery Partners across five sites. This tracks KPI performance for recruitment, data completion, hold documents and application links.
- Developing a histopathology system that allows completion of structured histopathology data and mapped SNOMED CT coding.
- Developing the HL7 interface to assist external and internal partners in developing their own HL7 interfaces in turn, for both the Trakgene genetics system and the EDGE Local Portfolio Management System (LPMS).
- Refining the laboratory metadata system to integrate with the results server, Apex and Fluid-X, to automatically transmit sample metadata to Genomics England.
- Developing a pedigree XML tool – this integrates with the Trakgene system to create and send XML at the push of a button.
- Creating a GS1 barcode for the patient wristband.

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:

Ryan Beegan, Wessex NHS GMC Informatics Lead at: ryan.beegan@uhs.nhs.uk or rbeegan@nhs.net, tel. +44 (0)23 8120 4725.
@WessexGMC #Genomes100K www.uhs.nhs.uk/WessexGMC

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.

Four Trusts are involved in the informatics development:

- Guy's and St Thomas' NHS Foundation Trust
- King's College Hospital NHS Foundation Trust
- St George's University Hospitals NHS Foundation Trust
- South London and the Maudsley NHS Foundation Trust

The strategic approach of all four organisations was to ensure that, where possible, data required for the 100,000 Genomes Project was extracted from operational clinical systems, minimising the amount of data entry duplication and ensuring the highest level of data quality.

Additionally, it is intended that applications and facilities created as part of the programme will provide ongoing beneficial legacy within each of the organisations, beyond the completion of the 100,000 Genomes Project.

As a result, we have:

- Created platforms which allow the monitoring of clinical activity for recruited patients
- Are able to deliver structured phenotypic information
- Developed a pipeline called CogStac which uses OpenSource tools to process clinical and transactional data information held in clinical and administration systems, creating an Elastic Index of all patients of interest
- Redesigned and configured the existing clinical systems security and access control model to enable recording of non-trust contacts and their links with trust patients /participants
- Facilitated the electronic integration of the Genetics Laboratory system with central Patient Administration Systems and for NHS Number validation
- Created a generic research/study data management tool for use in research projects/studies beyond the 100,000 Genomes Project
- Supported clinical teams in the recruitment of participants in geographically remote satellite clinics
- Shared (with other GMCs nationally) a CSV file pre-submission validation tool.

Our current focus is to:

- Meet the requirements of the new Cancer and soon to be published Rare Disease datasets
- Enhance local data repositories to provide increased functionality.

Our future plans include:

- Electronic integration of genetics laboratory systems with local order communications systems for both test requests and results returns
- Integration of clinical genetics notes into trust-wide patient notes
- Improved reporting performance for clinical genetics clinical systems.

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:

Olaf Hartberg, South London NHS GMC Programme Lead, at: hin.southlondon@nhs.net, tel. +44 (0) 207 188 4814.
@SouthLondon_GMC #Genomes100K www.southlondon.nhs.gmc

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, also from some patients across the country.

North Thames NHS GMC (NT GMC) needs repositories for the core clinical data that will be collected from participating hospitals and sent to Genomics England. It is desirable that the repository be implemented in early 2017. Using an open data standard for the clinical data repository would be a low-risk initiative that would generate significant benefits for NT GMC and for the rest of the 100,000 Genomes Project. Using an open data standard will make it easier for NT GMC to share data, and will provide a proof of concept for other regional data sharing projects; STPs, BRCs, etc. It will complement enterprise-wide and best-of-breed approaches to an electronic patient record (EPR). It will reduce the risk of being "locked-in" to a commercial vendor, with the associated difficulties of data migration.

openEHR

- openEHR now includes more than 500 archetypes, corresponding to 7,000 clinical data points
- 70% of the Genomics England data set already exists in openEHR archetypes, the remaining 30% is in the process of being modelled
- openEHR is coordinated by the openEHR Foundation, which is based at University College London
- Although openEHR is an open standard, to implement it an execution engine is needed, and most of the execution engines are closed-source
- openEHR has been shown to work well alongside other leading healthcare standards, including HL7 messages, HL7 CDA, FHIR, SNOMED CT, and IHE standards such as XDS.

Benefits

- Data sharing
  - openEHR is the only data model that stores rich, fine-grained clinical data in an open format
  - Because most vendors use proprietary data models, it is difficult to exchange more than summary data between systems
  - openEHR allows for exchange of rich clinical data.
- Data Control
  - Vendor neutral
  - With proprietary data models, the data is locked into a format that is usually controlled by a commercial vendor
  - Switching to a different vendor requires translating the data into a different format, often with great difficulty and cost
  - By separating data from applications, openEHR allows healthcare providers to retain control of their data.

Our partners include:

- Genomics England
- NHS England
- NHS Digital
- NHS Code4Health
- Rotherham NHS Foundation Trust – SNOMED CT mapping team
- Farr Institute
- UCLH HIC
- West Midlands GMC
- Supplier community includes: Marand, Ocean Informatics, DIPS freshEHR, Operon, Ripple Health.

Next Steps

- Select an openEHR vendor
- Finalise implementation plans for openEHR at NT GMC and West Midlands GMC
- Complete detailed implementation plans for each Local Delivery Partner
- Provide Genomics England with feedback about potential improvements to the data models for future versions
- Engage with leaders at organisations that have implemented openEHR and learn from their experiences
- Share approach with other GMCs
- Contribute to the development of federated clinical records across North London for both research and patient care.

More information: http://www.openehr.org/releases/BASE/Release-1.0.3/docs/architecture_overview/architecture_overview.html

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:

North Thames NHS GMC, at: 100kgenomes@gosh.nhs.uk, +44 (0)20 7829 8867.
@UCLPartners #Genomes100K www.ntgmc.nhs.uk

West London NHS GMC

West London NHS GMC serves a population of 2.5 million. Through four NHS partners we cover the London boroughs of Kensington, Chelsea and Hammersmith & Fulham through to Sutton and Hillingdon.

Our informatics successes include:

- Developing a fully integrated research and genomic data warehouse to link to multiple health systems at Imperial and at other Local Delivery Partners. These include EPR, radiotherapy, surgery, chemotherapy, pathology (all), radiology and cancer MDT systems.
- Developing a pathology back-end system to allow direct access for all pathology data without impacting clinical activity.
- Full automation of Genomics England data flows across the GMC – registration, consent, sample and clinical data. Collection of clinical data is from primary clinical systems.
- Business intelligence reporting suite for the GMC programme board, cancer and rare disease leads at multiple sites, tissue banks and Local Delivery Partners.
- Reporting tools linked to the EPR to assist recruitment.
- Data quality reporting to allow project and clinical teams to see missing data for participants.
- Integration with tissue bank LIMS, Genomics England's Open Clinica and LabKey systems to ensure that GMC data matches Genomics England's and samples/registrations are automatically confirmed with end users.

Our current focus is on:

- Developing and testing the cancer dataset version three; implementing XML solution across all Local Delivery Partners.
- Further integration with GMC systems to allow validation sample tracking between multiple tissue banks and validation labs over varied Trust sites.
- Developing pathology reporting and decision support tools to generate structured histopathology data, fully meeting Genomics England requirements.
- EPR developments to allow genomic referrals to become business as usual.
- Developing EPR to incorporate extra clinical information required by Genomics England.
- Developing electronic consent pathways to allow the storage of tissue for project (research and 100,000 Genomes Project) to maximise the amount of potential recruitment to the programme.

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.

For further information please contact:

Ben Glampson, Research Informatics Programme Manager at Ben.glampson@imperial.nhs.uk, +44 (0) 7795 637 784.
@ImperialNHS #Genomes100K www.imperial.nhs.uk/westlondongmc

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.

Our informatics successes include:

- Developing an integrated data warehouse for health data pertaining to 100,000 Genomes Project participants and establishing data flows from a complete set of relevant clinical and laboratory systems: chemotherapy, radiotherapy, radiology, cancer tracking and reporting, EPR, national spine, biochemistry, cellular pathology, regional genetics, molecular diagnostics, and microbiology.

Our current focus is on:

- Mapping and transforming linked data from clinical and laboratory systems to automate reporting in XML format against Genomics England's Cancer and Rare Disease Data Models. These platforms and transformations will be re-used for data from our Local Delivery Partners' systems.
- Linked data from clinical and laboratory systems mapped to "enhanced" data model in LabKey and supplied to Genomics England to support "GENE 1k" data release.
- Natural language processing system being trialled for the generation of coded data from free text content in pathology reports, building on earlier work with Guy's and St Thomas' and Imperial Biomedical Research Centres.
- Integrated data service providing local access to data presented against Genomics England cancer and rare disease data models implemented using Mercury (Oxford infrastructure for Genomics England) and LabKey.
- Complete case report form system (OpenClinica) for rare diseases and cancer (dataset version three) produced as containerised (Docker) deployment and shared with South London NHS GMC.
- Online GM-MDT referral system (live since October) to improve tracking of individual referrals for Oxford University Hopspitals clinicians, allowing efficiency savings in administrative processes, facilitating clinical data collection in a centralised and referrer-led manner, and holding potential for future automated data transfer.

Lead organisation:

Oxford University Hospitals NHS Foundation Trust.

Partner organisations:

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

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.

Our informatics successes include:

Phenotips

Phenotips was already adopted by the genetics service at Cambridge because it excels in the ability to model 'families', the easy intuitive phenotyping with HPO terms and pedigree drawing capabilities. Early participation in the 100,000 Genomes Project produced a 'panel' application for the standard programme which presented standard 'panel' list of HPO terms in addition to freeform phenotyping. This is used with the project disease lists or other clinical assessments required for qualification to genetic analyses. Across our GMC there is a new enthusiasm for, and adoption of, Phenotips in the clinical genetics service at all partner sites. This has been catalysed by the project but extends to the adoption of Phenotips for all phenotypic and family profiles for both the 100,000 Genomes Project and as a general service in genetics. With the release of Capex 2016/17, implementation is underway at the various sites. In keeping with the common practice of clinical genetics Phenotips is used locally and, with the developers of Phenotips, we are exploring appropriate regional and national integration.

Resources: Phenotips is available as an open source package from the developer's website (https://phenotips.org/); the phenotype panel extensions for the 100,000 Genomes Project are available, as are other integration tools, such as Phenotips to OpenClinica (helpdesk@rdcit.org).

Rare disease clinical datasets

Rare diseases represent a hard challenge for clinical informatics – patients are managed through multiple specialities and pathways and typically are engaged in multiple encounters, in different institutions with very different information structures. Leicester have led in the use of established tools and techniques for mapping and merging disparate clinical data sets in the shape of i2b2 (a Harvard led initiative for clinical data combination and cohort analysis). As the 100,000 Genomes Project was being launched Cambridge University Hospitals (CUH) implemented EPIC, their new electronic health record, which consolidated care data for patient journeys from multiple hospital sources. The project became the testbed for the integration and interoperation of combined research and care datasets (in i2b2). EPIC data is available in 'raw' format via the CUH research gateway and integrated with other data sets available from other national rare disease initiatives in other formats. These multiple sources are pre-processed through OpenClinica, to manage the complexity of multiple encounters, and annotated with HPO and other ontology terms.

i2b2 then consolidates these very disparate data sources in a rich data warehouse format: the format separates clinical data from Patient Identifiable Data (PID) ensuring project and subject integrity and security. This richness preserves the detail of the encounter and clinician structure, and the i2b2 data format embeds the ontological terms between systems for browsing and analytical insight. Client access to these data sources is available through office tools (such as Excel) or specialist bioinformatics tools (such as Labkey or R).

Resources: i2b2 tools and technologies are available as open source from the developers (https://www.i2b2.org/software/) and from the Brisskit initiative (https://www.brisskit.le.ac.uk/), Ontological mappings for the project clinical data (Snomed CT, HPO, others) and portable high-fidelity data sets and assorted integration scripts (helpdesk@rdcit.org).

GelCI

The 100,000 Genomes Project places some unusual constraints on the provision of samples and data. In particular, DNA samples must be paired (tumour vs germline) for participants with cancer, and are grouped in families (at least parents and affected child) for those with rare diseases. This doesn't fit well with the one-at-a-time sample delivery processes that run in hospitals. To address this need, Cambridge (John Boucher) has developed a secure logistics web application that allows the entry or upload of sample-related data, and tracks data completeness to allow timely shipment of sample sets to the National Biosample Centre. In Cambridge and two of our Local Delivery Partners (Nottingham and Leicester), much of the data is uploaded to GelCI. However, for Norwich – a partner with little informatics infrastructure available to Genomics England – the application is also used as a data capture tool.

Resources: GelCI is built using the open source Django web framework (https://www.djangoproject.com/), which utilises Python and (in this case) a Python webserver (http://docs.pylonsproject.org/projects/waitress/en/latest/) and PostgreSQL database (https://www.postgresql.org/). It runs on both Windows and Linux.

Other Innovations

The joint responsibilities for the 100,000 Genomes Project has produced multiple working partnerships across the East of England NHS GMC. Nottingham were early adopters of their Achiever LIMS systems for their BioBank and this has positively influenced the adoption of the same systems for CUH and the UK BioSample Centre. Leicester were vanguard adopters of the CiviCRM system for participant communication and consent; this has been adopted at Cambridge. Cambridge developed the public facing website for our GMC, which included the eligibility criteria, a service used by other GMCs.

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:

Prof Lucy Raymond, East of England NHS GMC Programme Lead, at: flr24@cam.ac.uk, tel +44 (0)1223 762609.
@_EEGMC #Genomes100K www.eastgenomics.org.uk

Notes

Notes

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

e england.genomics@nhs.net

@NHSEngland #Genomes100k #PersonalisedMedicine

w 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

Photographs: NHS Photo Library.