NHS
South West
NHS Genomic Medicine Centre

100,000 GENOMES PROJECT

SPRING 2016

Update on progress

The South West NHS Genomic Medicine Centre (South West NHS GMC) continues to make good headway towards achieving our aim to sequence 3,457 genomes from 1,629 families by the end of 2017. This newsletter brings you up to date with the successes so far and the challenges we face.

Focus on: National Management

NHS England has a dedicated Implementation Unit tasked with overseeing delivery of the project. There are bi-monthly GMC meetings held in London; weekly teleconferences for the project managers with the Implementation Unit; weekly calls between our project manager and the NHS England account manager; weekly teleconferences for the programme directors with the NHS England Chief Scientific Officer and Genomics England Chief Scientist; plus regular teleconferences/meetings for the PPP, informatics leads and laboratory leads.

Feedback from our second quarterly review meeting in February was very positive with a green RAG rating awarded. Our recent Quarterly Review was held in May and we were rated very positively.

Communications

An event to mark the recruitment of the first 100 families and the on boarding of LDPs is being planned for June 2016 in Exeter.

Members of all LDPs will be invited to learn more about the project delivery, educational opportunities and the benefits of genomic medicine for patient care.

Focus on: Engagement of Local Delivery Partners

Recruitment at the other 6 Trusts has begun in a phased roll-out, to be completed in the next 3 months. Partnership agreements, data sharing and material transfer agreements have been sent for review and signing.

There will be reimbursement for consent/blood sampling appointments, and blood and tumour sample processing. The Clinical Research Network (CRN) is supporting the involvement of their research nurses.

Focus on: Patient & Public Participation

A Patient and Public Participation (PPP) Group has been set up in partnership with the SWAHSN. The group consists of approximately 30 members with approximately 20 regular attendees, built up from existing PPP networks at the NIHR Clinical Research Facility, staff from the Clinical Research Facility, SWAHSN and Exeter University.

Four PPP workshop and discussion events have been held to date with a recent focus on how the public can get involved in wider genomics education as well as also around the theme of ethics and social science. At the last meeting Vivienne Parry, the Head of Engagement for Genomics England came to speak to the group about progress being made with the project and Derek Sprague (LETB Director South West at Health Education South West) and some project participants were also in attendance.

Workshop topics included education opportunities, additional findings and the newly announced participant panel. This group will be part of the national service evaluation of the consent process that has been commissioned by Genomics England and will begin in May this year.

Results from this evaluation will help to improve the participant materials and the quality of the consent process. Work is underway to link in with PPP groups and charities across the South West as well as participants recruited at LDPs.

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Focus on: Cancer recruitment

After the successful completion of dry and wet runs for cancer, NHS England gave us ‘go live status’ meaning that recruitment of cancer patients could begin at RD&E. The first patient was recruited on 29th January 2016 and as of 31st March, 6 patients with breast cancer have been recruited. We are now ready to recruit patients with bowel cancer and are working with clinical teams at the RD&E to start recruitment of patients with lung, renal and bladder cancer next. The cancer ‘main programme’ began on 16th February and heralded several key changes.

Samples must now be collected ahead of administration of chemotherapy, radiotherapy, hormonal therapy or other treatments. In addition, fresh frozen (FF) tumour samples should be collected wherever biologically possible, with formalin fixed paraffin embedded (FFPE) tumour samples only being submitted where this is not possible, for example if the tumour is impalpable or too small. Pilot studies have shown that FF samples are consistently superior to fixed tissue for whole genome sequencing (WGS).

Work is on-going nationally to improve the quality of FFPE samples for WGS. A number of additional cancer types have been added to the list of eligible cancers including: adult glioma, brain tumours, renal cancer, bladder cancer, endometrial cancer, melanoma, Upper GI tumours and testicular cancer. Discussions to enable Local Delivery Partners (LDPs) in other Trusts to participate in the cancer arm of the project can now begin and this will take place in a phased approach once each LDP is actively recruiting patients with rare disease. There are plans for tumour biopsy samples, taken prior to a patient’s cancer diagnosis, to be included in the 100,000 Genomes Project.

Using biopsies would permit inclusion of patients in the project who have some of the most aggressive tumour types. It would also allow genome sequencing of chemo-naive tissue in patients to whom neoadjuvant chemotherapy is routinely administered prior to surgery and inclusion of patients whose tumours cannot be surgically removed.

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Focus on: Education and Training

The SW NHS GMC has been awarded £150k from Health Education England to support genomic medicine education and training across the south west. Two part-time Education and Training Leads (1.4FTE) have been appointed and will work closely with Health Education South West (HESW) to deliver the project objectives.

The University of Exeter has recently been named as the latest provider of the Health Education England (HEE) funded Masters programme in Genomic Medicine. This is an exciting opportunity for the University to engage with and support the on-going work of the HEE Genomics Education Programme, to upskill and transform the workforce to meet the future ambitions of the NHS.

The MSc in Genomic Medicine and associated CPPD modules have been designed to support the introduction of personalised medicine into the NHS, the work of NHS Genomic Medicine Centres and the 100,000 Genomes Project. Open to all NHS healthcare professionals, the course aims to enhance knowledge and skills in this rapidly evolving field. Details are available on the Genomics Education Programme website or on the Exeter University website.

Focus on: Rare disease recruitment

Our original target was to provide 1000 samples from 400 families by the end of 2017 but we have increased our planned recruitment for patients with rare diseases.

The first family was recruited in Exeter on April 24th 2015 and 216 family referrals have been received from clinical genetics, renal medicine, paediatrics, endocrinology, diabetes, neurology, ophthalmology, dermatology, immunology and cardiology.

As of March 31st, 258 participants (102 families) have been recruited. This represents 128% of the original contacted recruitment target and 85% of the revised recruitment target for this period.

The vast majority of referrals to date have been from clinicians in Exeter and the GMC will be working with groups of clinicians at each of the local delivery partners as they come on board so that eligible patients can be referred.

Interested clinicians in any of the 7 Trusts are encouraged to contact the project office in advance to find out more by emailing rde-tr.gmc@nhs.net. Our regular Rare Disease ReadMe and newsletters for recruiting clinicians are available on our website at: www.swgmc.org.

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Focus on: Informatics

The 100,000 Genomes project will act as a catalyst for improving informatics to support genomic medicine. This includes the collection of clinical data describing the phenotypes of patients with rare diseases, improved cancer data sets, storage of genetic data and generation of clinical genome reports. We were awarded £651k capital investment for 2015-2016.

The funding has supported data integration required for the project (via Genie); digitisation of tumour slides in histopathology; development of GS1 sample barcodes; enhancements for the Clinical Genetics Trakgene module; and improved measurement and integration of DNA concentration data and hardware for next generation sequencing data processing.

Genie is in place at the RD&E, Plymouth and Truro and access to Genie is currently being arranged for the remaining LDPs. This allows registration of patients, record keeping for the recruitment process, phenotype data entry and tracking of DNA and ”omics” samples.

USEFUL RESOURCES

WWW.GENOMICSENGLAND.CO.UK

HEALTH EDUCATION ENLAND - GENOMICS EDUCATION PROGRAMME.

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Feedback from our meeting on 29 October was positive, with a green RAG rating awarded.

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FOR MORE INFORMATION ABOUT THE PROJECT, PLEASE CONTACT:

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