

100,000 GENOMES PROJECT

JULY 2015

Update on progress

In December 2012 the Prime Minister announced the launch of the 100,000 Genomes Project. The ambitious aim of the project is to sequence 100,000 genomes of patients with cancer or rare diseases by the end of 2017. This high-profile initiative has made headlines across the world - and in the South West we are playing a key role.

After being selected in the first wave of applications in December 2014, the South West NHS Genomic Medicine Centre (South West GMC) has made great progress toward achieving our aim to sequence 3,457 genomes from 1629 families.

The project involves individuals and organisations from across the health system in the South West working together to change lives. The South West GMC includes seven acute trusts in Devon, Cornwall and Somerset, and works in partnership with the South West Academic Health Science Network (SWAHSN). This three-year project promises to be exciting, challenging and rewarding. We have produced this newsletter to provide you with an update on the current progress of the programme.

We hope that you find it useful and informative.

Overview

The 100,000 Genomes project has been commissioned by the Department of Health and is managed and delivered by Genomics England, who are working closely with NHS England. The clinical aim of the project is to improve diagnosis of patients with rare diseases and increase understanding of tumour variants that predict therapeutic response to targeted cancer drugs.

The project will also catapult the NHS to the forefront of genomic medicine, stimulating and enhancing UK industry and investment in genomics.

In the short-term, some patients may benefit by receiving a genetic diagnosis for their rare disease or identifying a mutation in their tumour that predicts response to a targeted therapy. In the longer term the expectation is that the dataset will fuel research discoveries and accelerate the implementation of genome sequencing as a diagnostic test.

Genomics England is responsible not only for delivering the project, but also for the recording and release of all performance data. Their website provides a range of information about the project – including a comprehensive FAQs section - which you may find useful. You can access the Genomics England website at: www.genomicsengland.co.uk.

100,000 GENOMES PROJECT

JULY 2015

South West GMC Progress

Over the past six months, the South West GMC has made some significant steps in the project. You can read about them here:

South West GMC identifies first patient

Following a rigorous process to establish operation readiness, the South West GMC was given permission to begin recruitment to the rare disease arm of the project on 21 April 2015. Only a few days later, on the 24 April, we welcomed our first family.

The recruitment of the patient made the news, with interviews and articles appearing on Radio Exe, BBC Radio Devon and Heart FM. The story was published in print and online by the Exeter Express and Echo and the Western Morning News, in print by View From Honiton and online by The Exeter Daily, View From publishing and Medical Xpress, as well as featuring on the RD&E and University of Exeter Medical School websites. We are encouraged by the support of the media and hope that it will raise the local profile of the study.

Focus on: Rare disease recruitment

Our target is to provide 1000 samples from 400 families. Current efforts are focused on identifying eligible patients from clinical genetics, renal and paediatrics clinics. Endocrinology, neurology and ophthalmology will be involved next.

Focus on: Cancer recruitment

Our target is to provide 2,458 samples (blood and tumour) from 1,229 patients. Pilot studies have revealed technical issues with whole genome sequencing related to the quality of DNA extracted from formalin fixed paraffin embedded tumour tissue. Work is in progress to assess the option of testing fresh frozen tumour samples.

During the initial phase of the study the project will collect tumour samples processed in both ways and is due to commence in August 2015. Cancer recruitment will begin with colorectal cancer and expand to include patients with lung, breast, prostate or ovarian cancer.

Focus on: Informatics

The 100,000 Genomes project will act as a catalyst for improving informatics to support genomic medicine. This includes the collection and integration 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 have been awarded £651k capital investment (for 2015-16) to support the data integration required for the project. Work is underway to implement "Genie", a tool developed by the West Midlands NHS GMC to manage recruitment, data collection and submission to Genomics England.

100,000 GENOMES PROJECT

JULY 2015

South West GMC Progress

Focus on: Patient and public involvement A Patient and Public Involvement (PPI) Group has been set up in partnership with the South West AHSN. It aims to engage and involve patients and the public wherever possible throughout the course of the project.

Currently the group consists of approximately 30 members built up from existing PPI networks at the NIHR Clinical Research Facility, staff from the Clinical Research Facility, South West AHSN and University of Exeter.

Work is currently underway to expand this group across the region and define its remit going forward. Progress has been made in sharing feedback from the local group about patient materials at the national PPI group meetings. In addition the group aims to communicate the aims of the project as well as provide education and information about genetics and genomics in general by having regular talks. A detailed strategic plan for PPI for the duration of the project is currently being developed which will include all regional partners.

Focus on: Engagement of Local Delivery Partners

We plan a phased roll out of recruitment to the other Trusts over the next 9-18 months. Partnership agreements will need to be in place plus data sharing and material transfer agreements with Genomics England and the Bio-Repository.

There will be reimbursement for consent/blood sampling appointments, blood and tumour sample processing and project management costs.

100,000 GENOMES PROJECT

FOR MORE INFORMATION ABOUT THE PROJECT, PLEASE CONTACT:

SOUTH WEST NHS GENOMIC MEDICINE CENTRE

EMAIL: RDE-TR.GMC@NHS.NET

TELEPHONE: 01392 408177

ADDRESS: SOUTH WEST NHS GENOMIC MEDICINE CENTRE NIHR EXETER CLINICAL RESEARCH FACILITY LEVEL 2, RILD CENTRE ROYAL DEVON & EXETER HOSPITAL BARRACK ROAD EXETER EX2 5DW

We aim to produce these newsletters 3-4 times per year and encourage you to share them with colleagues and staff across your organisation.