

# 100,000 GENOMES PROJECT

WINTER 2015

## 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.

## Education & Training

The SW NHS GMC has been awarded £150,000 from Health Education England to support Genomic Medicine Education and Training across the southwest. We will work closely with Health Education South West (HESW) to deliver the project objectives and report back in future newsletters.

## Communications

An event to mark the recruitment of the first 100 families and the on boarding of LDPs is being planned for March 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: Patient and public involvement

A Patient and Public Participation (PPP) Group has been set up in partnership with the South West Academic Health Science Network (SWAHSN). The group consists of approximately 30 members with around 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. Workshops topics included education opportunities, additional findings and the newly announced participant panel.



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## **Focus on: Informatics**

The 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 have been awarded £651k capital investment (for 2015-2016) to support the data integration required for the project.

The “Genie” tool developed by the West Midlands GMC has been installed and is now live. This will help us to manage recruitment, data collection and submission to Genomics England. Additional developments to provide the functionality required to support our in-house sample barcoding processes are now complete.

Arrangements are being made to provide web access to the Genie system for LDPs in order that they can start processing blood samples for the “omics” samples locally.

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## **Focus on: Cancer recruitment**

After the successful completion of dry and wet runs for cancer, NHS England have given us ‘go live status’, meaning that recruitment of cancer patients can now begin at the RD&E for the Initiation and Implementation Phase.

We will start to recruit patients with breast or bowel cancer, as the tumour sample is easiest to identify and has a high probability of containing tumour DNA. These samples are therefore most likely to lead to successful genome sequencing. Our current contracted target is to provide 2,458 samples (blood and tumour) from 1,229 patients, although this is likely to change as previously mentioned.

Pilot studies have revealed technical issues regarding obtaining good quality genome sequencing from FFPE tumour samples processed using standard histopathology methods. It is highly likely that fresh frozen tumour samples will be required from February/March 2016, which is when the Initiation and Implementation Phase (IIP) ends and the main cancer programme begins.

From this point our LDPs can begin cancer recruitment. We have been advised that for the main cancer programme, participants will need to be chemo naïve, a change from the original contract.

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## **Focus on: Rare disease recruitment**

As of November 30th, 141 participants (57 families) have been recruited. This represents 95% of the recruitment target for this period. However, a delayed start to the cancer arm of the project, together with the likely reduction in the number of eligible patients due to a change in the eligibility criteria and sample type, means that our cancer recruitment target (1229 patients) is very unlikely to be achieved.

To address this shortfall we hope to recruit a greater number of families with rare diseases. We are working with our Local Delivery Partners across the South West to initially roll out recruitment in Plymouth, Taunton and Truro in order to increase recruitment.

## **Focus on: National management**

NHS England has a dedicated Implementation Unit tasked with overseeing delivery of the project.

There are bi-monthly GMC meetings in London, weekly teleconferences for the project managers with the Implementation Unit, weekly calls between our project manager and 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 meeting on 29 October was positive, with a green RAG rating awarded.

## USEFUL RESOURCES

[WWW.GENOMICSENGLAND.CO.UK](http://WWW.GENOMICSENGLAND.CO.UK)

HEALTH EDUCATION ENGLAND -  
GENOMICS EDUCATION PROGRAMME.

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FOR MORE INFORMATION ABOUT THE PROJECT, PLEASE CONTACT:

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