

# 100,000 GENOMES PROJECT

AUTUMN 2015

## Update on progress

The South West NHS Genomic Medicine Centre (South West 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.

## Communications

Our website is now live at [www.swgmc.org](http://www.swgmc.org), with further development work on-going, including creation of a blog. Our first newsletter has been circulated and is also available on the website. This edition of the newsletter will also be available on the blog.

## Education & Training

The SW NHS GMC has been awarded £150k from Health Education England to support Genomic Medicine Education & Training across the southwest. We will work closely with Health Education South West (HESW) to deliver the project objectives.

## Communications

Our website is now live at [www.swgmc.org](http://www.swgmc.org), with further development work on-going, including creation of a blog. Our first newsletter has been circulated and is also available on the website. This edition of the newsletter will also be available on the blog.

# 100,000 GENOMES PROJECT

AUTUMN 2015

## 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](http://www.genomicsengland.co.uk).

## Focus on: Engagement of Local Delivery Partners

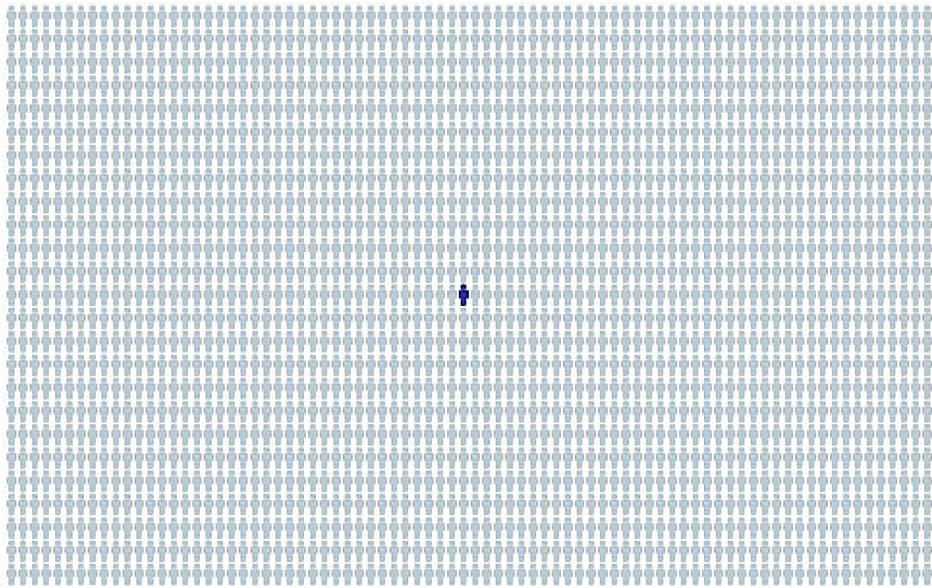
The South West Genomic Medicine Centre is one of 11 GMCs in England that are responsible for recruiting patients, providing samples and clinical data. We will validate clinically significant results and record clinical outcomes. The South West GMC includes the 7 Acute Trusts in Devon, Cornwall and Somerset, and works in partnership with the South West Academic Health Science Network (SWAHSN) to deliver the project goals between now and December 2017.

We will be initiating a phased roll out of recruitment to the other Trusts over the next nine months.

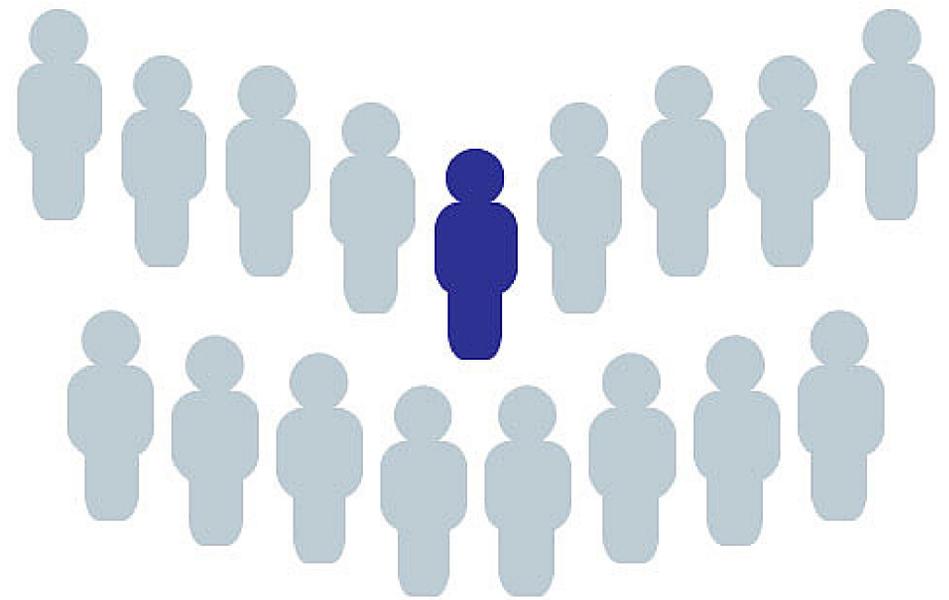
Partnership agreements will need to be in place, plus data sharing and material transfer agreements with Genomics England and the Bio-Repository. There will be reimbursement for consent/blood sampling appointments, and for blood and tumour sample processing. We are working with colleagues from Plymouth, Taunton and Truro to plan the 'wave 1' LDP roll-out. The SWAHSN is playing a co-ordinating role and the CRN is supporting the involvement of their research nurses.

# Rare diseases: facts and figures

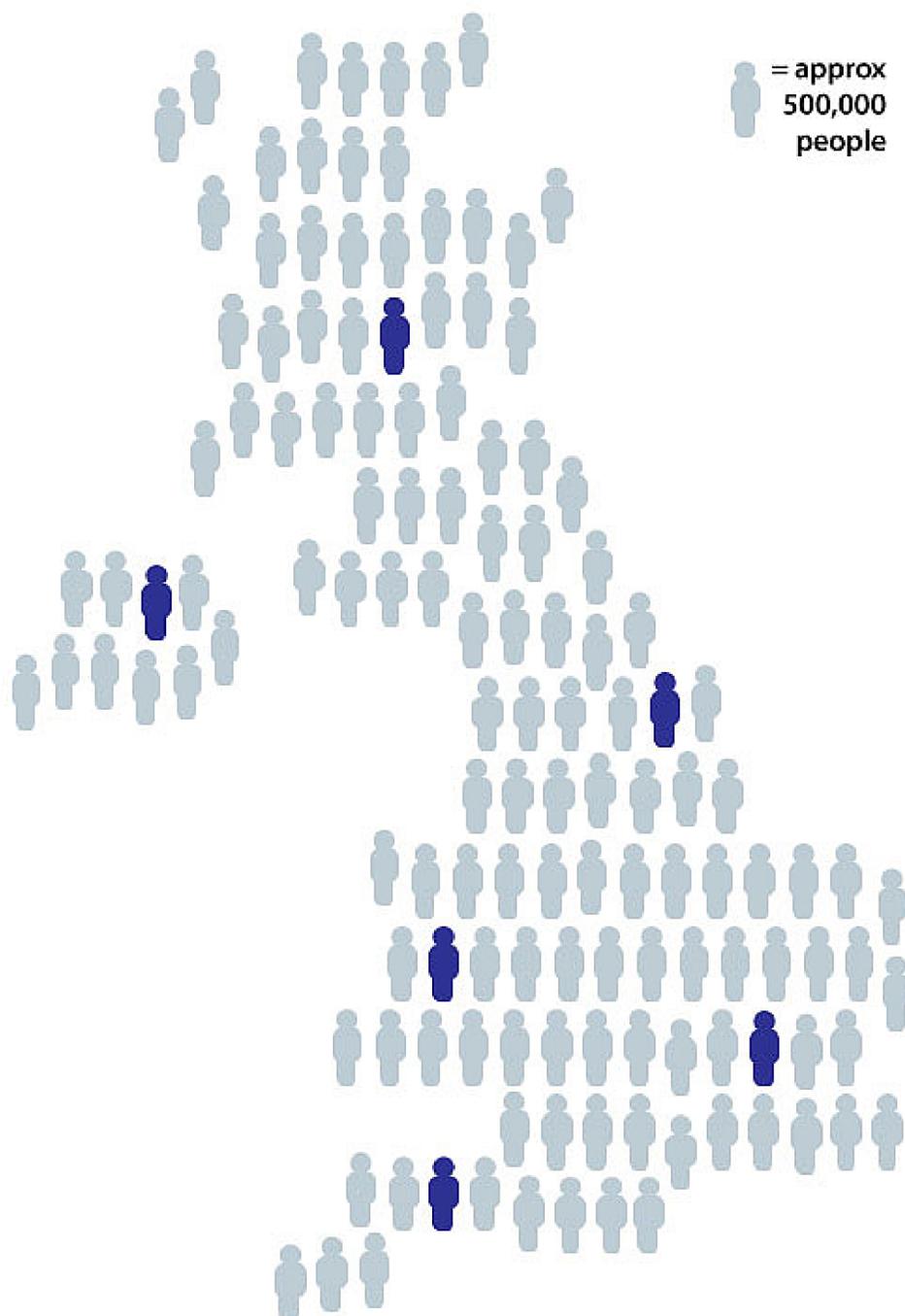
The UK defines a 'rare disease' as one that affects **1 in 2,000** or less of the population...



... so, collectively, rare disease will affect **1 in 17** of the population at some point in their life

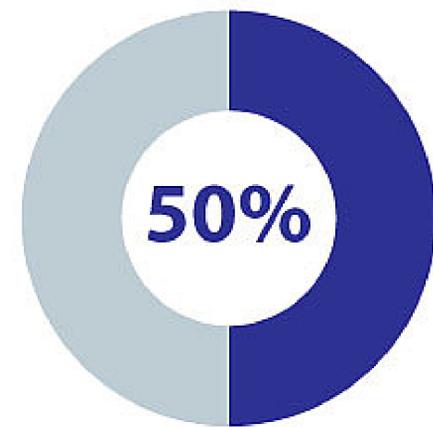


In total, that's about **3 million** people currently in the UK who will be affected by a rare disease



 = approx 500,000 people

**50%** of newly diagnosed cases of rare diseases are in children

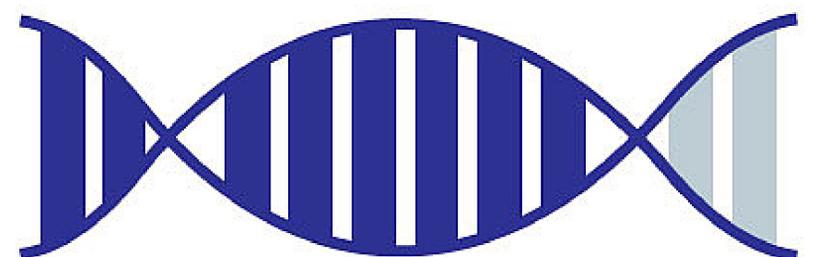


There are between **5,000** and

# 8,000

different rare diseases...

... and **80%** of them have a known **genetic origin**



# 100,000 GENOMES PROJECT

---

AUTUMN 2015

## **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. A test version of the 'Genie' tool developed by the West Midlands GMC has been installed. This will manage recruitment, data collection and submission to Genomics England. We are awaiting additional developments that will provide the additional functionality required to support our in-house sample barcoding processes.

Three 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 around the theme of Ethics & Social Science.

Participants of the project are completing post participation experience surveys to provide feedback on the project information they were given and their experience of the consenting/blood sampling appointment. They are also being asked whether they would like to receive newsletters containing project updates and if they would like to join the PPP group.

## **Focus on: Patient and public involvement**

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.

## **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 first quarterly review meeting was positive, with a green RAG rating awarded.

# 100,000 GENOMES PROJECT

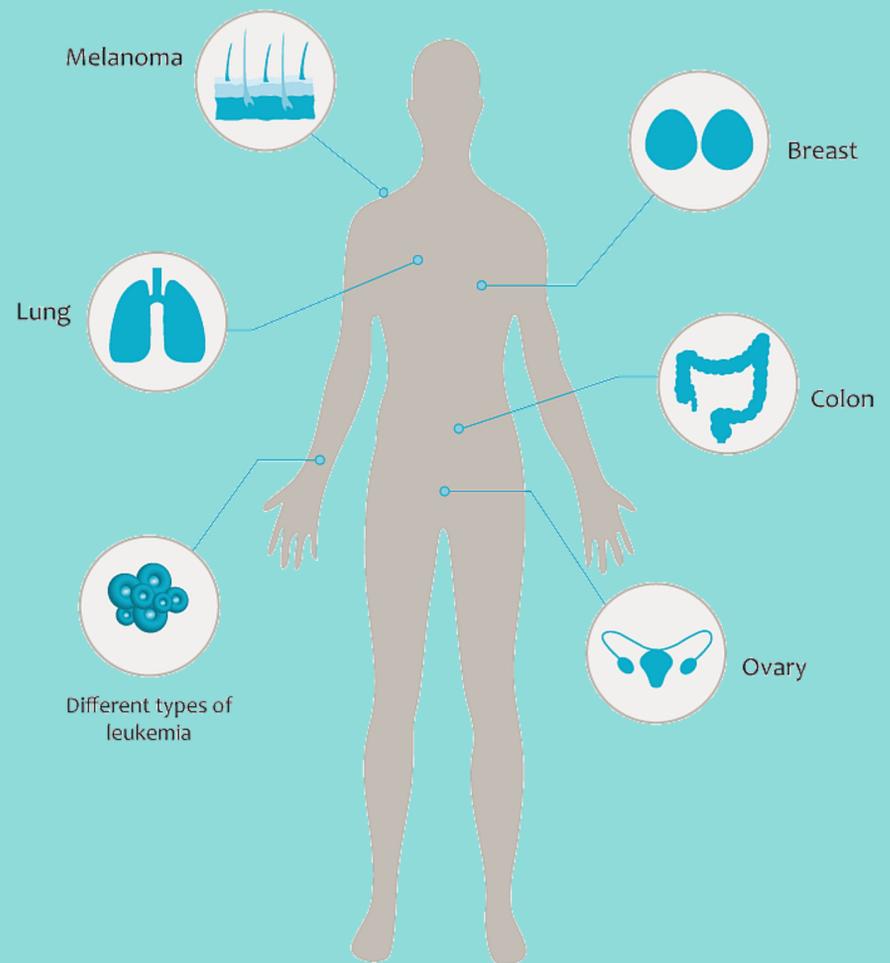
AUTUMN 2015

## Focus on: Cancer recruitment

Our target is to provide 2458 samples (blood and tumour) from 1229 patients. Pilot studies have revealed technical issues regarding obtaining good quality genome sequencing from tumour samples processed using standard histopathology methods. Work is in progress to assess the option of testing fresh frozen tumour samples.

This is the standard method used by research tissue banks. The Cancer Initiation and Implementation Phase will collect tumour samples processed in both ways and is underway in three GMCs. We are working with the NHS England team on our process maps prior to conducting dry and wet runs.

Cancers where targeted therapy is used



## Focus on: Rare disease recruitment

Our target is to provide 1000 samples from 400 families. The first family was recruited in Exeter on April 24th 2015 and 78 family referrals have been received from clinical genetics, renal medicine, paediatrics, diabetes, neurology and ophthalmology. As of September 30th 2015, 29 families, comprising 77 participants have been recruited. This represents 96% of the contracted recruitment for this period.

# 100,000 GENOMES PROJECT

---

A U T U M N 2 0 1 5

## USEFUL RESOURCES

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

HEALTH EDUCATION ENLAND -  
GENOMICS EDUCATION PROGRAMME.

## 100,000 GENOMES PROJECT

FOR MORE INFORMATION ABOUT THE  
PROJECT, PLEASE CONTACT:

SOUTH WEST NHS GENOMIC MEDICINE  
CENTRE

EMAIL: [RDE-TR.GMC@NHS.NET](mailto:RDE-TR.GMC@NHS.NET)

TELEPHONE: 01392 408177

ADDRESS: THE SOUTH WEST NHS  
GENOMIC MEDICINE CENTRE (GMC),  
LEVEL 3, RILD CENTRE, BARRACK ROAD,  
EXETER EX2 5DW

---

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