NHS
West of England
NHS Genomic Medicine Centre

# Update from your Genomic Medicine Centre: January 2018

## More than 1,500 people access state-of-the-art genome testing

**I'm delighted to report that more than 1,500 NHS patients and their family members have now enrolled to the 100,000 Genomes Project at the West of England Genomic Medicine Centre (WEGMC).**

This is a major landmark for the WEGMC which was opened in summer 2016 to enable people with rare diseases or cancer to access diagnostic genome sequencing and to transform genome medicine into mainstream NHS practice.

The WEGMC team with colleagues in NHS trusts and other partner organisations have together achieved:

- Genome-level diagnostic testing for adults and children with 218 different rare diseases
- Germline and tumour genome sequencing for 12 different types of solid or blood cancer at four NHS trusts
- Implementation of new consent clinics, bespoke informatics and cutting-edge laboratory processes
- Return of new genome sequence data for West of England patients
- Expert consent training of more than 40 local NHS staff, free access to the masters level Genomic Medicine Education programme and engagement of more than 15,000 public and healthcare workers at information events.

The WEGMC will now work with clinical teams to take full advantage of our allocated capacity by accelerating patient access until the end of the 100,000 Genomes Project in September 2018.

We look forward to transitioning this programme to a mainstream Genomic Medicine Service that will transform care for NHS patients.

***Andrew Mumford***
***Clinical director for the WEGMC***

## Louise hopes project will bring answers

**Louise James says she hopes the programme will bring hope that a firm diagnosis will be made for her son Scott.**

She said: "At birth Scott became very unwell and this was when the testing started: blood tests and scans all telling us the same thing… 'normal', yet things were clearly far from normal.

"Now eight, Scott has severe epilepsy, global developmental delay, hypermobility, a heart defect and we're still searching for answers about what his condition is.

"We know a diagnosis won't significantly change Scott, but it could give us a greater understanding.

"This project offers us hope that a diagnosis can be achieved. Without this it's impossible to know if Scott is just a one off or if our other children could be affected by the same unknown condition."

## Gregory is pleased to have signed up

**One of the patients to sign up is grandfather Gregory Fudge.**

The 69-year-old has been diagnosed with chronic lymphocytic leukaemia (CLL), a type of cancer that affects the white blood cells and tends to progress slowly over many years.

Gregory said: "I'm really pleased that I signed up to the 100,000 Genomes Project.

"If it provides any information that could help my treatment, find out more about the underlying causes of my condition or help other patients in the future, then it's worthwhile."

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