NHS
Yorkshire and Humber
NHS Genomic Medicine Centre

Genomes Update

Information for our partners, patients and staff

1,000th participant milestone reached

Over 1,000 participants in Yorkshire and Humber take part in 'game-changing' 100,000 Genomes Project

Tammy Kammin, Specialist Genetic Counsellor and Maria Edwards, a Research Nurse in ophthalmology highlight the 100,000 Genomes Project

Over 1,000 people who have a rare disease are now taking part in research to understand how genomics could be used to help develop new treatments and possible cures for a number of conditions.

The Yorkshire and Humber NHS Genomic Medicine Centre has recruited its 1,000th rare disease participant – just 18 months after becoming an NHS Genomic Medicine Centre.

The national '100,000 Genomes Project' is the first and only project of its kind in the world and one of the national centres recruiting patients to the project is the Yorkshire and Humber NHS Genomic Medicine Centre.

Dr Gill Wilson, Programme Manager for the Centre, said: "This is fantastic progress, and demonstrates the key role we are playing in the development of personalised medicine in our region as part of the national '100,000 Genomes Project'. By comparing the genomes from lots of people, we will gain a better understanding of diseases, how they develop and which treatments are likely to be effective."

For years Holly Wood and her husband, from Middlecroft, Chesterfield, were told that their son Anthony, who was born with global development delay, suffered with "challenging" behaviour and that it was a phase he would grow out of.

Antony, who is 8 years old, is the 1,000th person to sign up to the '100,000 Genomes Project'. His mum Holly hopes the research could change his future by helping them to find out more about his condition.

Holly explains: "Any results could make a big difference to his future, and even if there's some result, no matter how big or small, it would be great news for us.

"We didn't understand anything about gene codes, but the staff explained all the positives to us. Being the 1,000th participant opens your eyes up to just how many people are in the same situation as us, even though the circumstances might be slightly different. It's fantastic what they can do, and if they can do this now, imagine what they can do in the future."

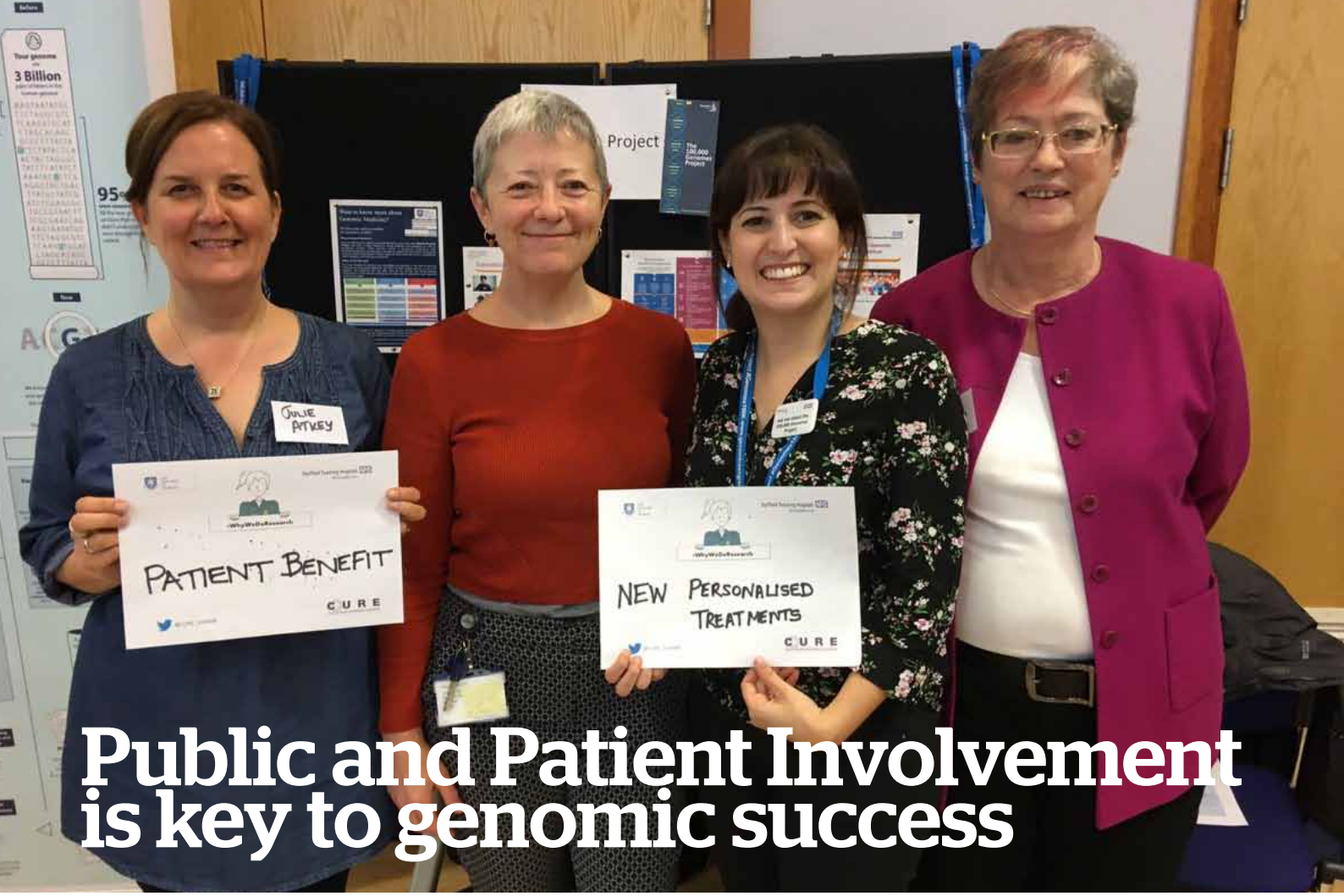
In Yorkshire and Humber, the Centre's team are recruiting participants with rare diseases and certain types of cancer, including breast, colorectal, lung, prostate, and ovarian, so that their DNA samples and cancer tumour samples can be sequenced as part of the national initiative.

Yorkshire and Humber GMC win regional innovation accolade

The Yorkshire and Humber NHS Genomic Medicine Centre has won a sought-after regional award for helping to spread vital knowledge about the '100,000 Genomes Project' in the Yorkshire and Humber region.

The team won the Best Example of Adoption and Diffusion category at The Yorkshire & Humber Academic Health Science Network's Innovation, Improvement and Impact Conference.

Gill Wilson, Programme Manager of the Yorkshire and Humber Genomic Medicine Centre.



Public and Patient Involvement is key to genomic success

Our Public and Patient Involvement group has been vital in helping the Yorkshire and Humber NHS Genomic Medicine Centre deliver its goals for the introduction of genomic testing in the NHS, acting as a critical friend and public representative.

We are very fortunate to have an active and committed group of patients and members of the public who contribute to our Public and Patient Involvement (PPI) group.

Many of the members have direct experience of a rare disease or cancer and they are keen to champion the potential genomics could have on future diagnosis, treatments and decision making for patients. For example; initially their focus was on equity of access to the programme, how we would raise awareness amongst both the public and NHS staff, preparing them for this new development and its impact on care, data security and processes. Most recently, our public and patient involvement members have been looking at the way results will be fed back to patients and what this experience will feel like.

Members contribute virtually via email and in person by attending regular two to three monthly meetings. They are an integral part of the whole Yorkshire and Humber Genomic Medicine Centre, and the views they provide to us is well received, acknowledged and also fed back nationally where appropriate. One of our panel members also sits on the national PPI panel for Genomics England and is able to provide a link between the regional and national group.

Getting to know... John Rouse

What do you do?
I'm a patient advocate, with a particular interest in melanoma, a type of skin cancer. Since retirement, I can spend up to five days a week attending meetings and conferences about cancer research, patient advocacy, and the use and security of data.

How did you get involved in the Yorkshire and Humber's 100,000 Genomes' Public, Patient Involvement Group?
I am a member of the Yorkshire and Humber Consumer Research Panel, and we were invited to attend an inaugural meeting of the Public, Patient Involvement group. I have a personal interest in both rare diseases and in cancer.

Why do you think Genomics is important?
Personalised medicine is going to become the core of the NHS in the future. If we can select the right drug for the right patient, it will save a lot of money.

What advice would you give to anyone wanting to find out more about the genomics programme?
Visit the website: www.yorkshirehumbergmc.nhs.uk or talk to one of the Public, Patient Involvement panel members.

Spreading the word about genomics

The Yorkshire and Humber Genomic Medicine Centre have had a busy time proactively engaging with members of the public, NHS staff, patients and patient representatives at a number of key events in the region. Here's a quick look back at some of the events we've taken part in over the past few months:

Healthcare Science Week 2017

Staff from the Pathology Department and Genomic Medicine Centre, including some NHS Science Training Programme trainees in Genomics and Genomic Counselling, talked to members of the public and hospital staff about the 100,000 Genomes Project and how important the role of the healthcare science workforce is in delivering this important project. Tammy Kammin (Genetic Counsellor at Sheffield Teaching Hospitals) and David Walker (Trainee Genetic Counsellor at Sheffield Children's Hospital) used a fantastic analogy of 23 different pairs of socks hung on a washing line, to teach people about chromosomes.

Socks say it all at Healthcare Science Week

Sheffield Hospitals' research event

A special research event showcasing the difference medical research makes to NHS patients' lives took place at Sheffield Teaching Hospitals NHS Foundation Trust. During the day representatives from the Yorkshire and Humber Genomic Medicine Centre ran an informative stand where attendees could ask questions and collect further information about the project.

Dr Gill Wilson, Programme Manager for the Yorkshire and Humber NHS Genomic Medicine Centre, delivered a keynote speech to the audience and explained how the sequencing of 100,000 Genomes could transform future care for patients with rare diseases and cancers.

Promoting research into rare diseases

Rare Disease Day

Julian Hartley, Chief Executive of Leeds Teaching Hospitals NHS Trust, joined the Centre's team on Rare Disease Day, to promote research into rare conditions and and patient recruitment into the 100,000 Genomes Project.

About 80% of rare diseases have a genetic cause. The cause is often a single changed 'letter' amongst the 3.2 billion letters of DNA that make up the human genome. Through the 100,000 Genomes Project we hope to break the mould for patients in the NHS, and are sequencing DNA from patients with rare conditions and their relatives. We aim to find the cause of these conditions which medicine has so far been unable to diagnose.

Local delivery partners engagement event

The first local delivery partners engagement event focused on genomics and personalised medicine. The event was attended by staff from nine NHS Trusts throughout the region.

The event proved a great success for networking and provided an opportunity to discuss how best to implement local pathways, overcome potential challenges and identify education and training opportunities.

Our milestones

Dec 2015
- Yorkshire and Humber Genetic Medicine Centre established

March 2016
- Rare Disease recruitment in Leeds and Sheffield begins
- Recruitment of rare disease patients in Clinical Genetics commences

April 2016
- Presentation on the GMC and the Future of Genomic Medicine at the Sheffield Life Festival.

July 2016
- 100th participant landmark achieved

Aug 2016
- Cancer recruitment begins in Leeds

Sept 2016
- 250th participant landmark achieved

Nov 2016
- Cancer patient recruitment commences in Sheffield

Dec 2016
- Y&H GMC's 1st Birthday Celebration
- 500th Participant Landmark Achieved

Jan 2017
- Recruitment of Neurology Patients commences in Sheffield
- Award for Best Example of Adoption & Diffusion

Feb 2017
- 750th Participant Landmark Achieved

March 2017
- Participation in Healthcare Science Week 2017

May 2017
- 1,000th patient milestone reached

June 2017
- 13 mainstream and 13 cancer teams now recruiting

New hope for Marcia

Rare disease sufferer, Marcia Bates, says the initiative to decode complete sets of genetic information in specific diseases could transform care for future generations like her.

Marcia Bates, 60, of Chesterfield, suffers with cerebellar degeneration, a rare disease which causes an unpredictable 'lag' in the way messages are sent from her brain to other coordinating parts of her body. This can affect her balance and coordination, memory and concentration, certain aspects of vision, speech, writing and even lapses in doing day to day things that we all take for granted.

Now she hopes that by taking part in the Yorkshire and Humber NHS Genomic Medicine Centre's '100,000 Genomes Project' others will be able to find out more about the genetic mutations that cause her condition. "I feel it's important to make people aware of the condition. My dad had it for many years before it finally debilitated him. People thought he was drunk, but he wasn't, he just stumbled a lot. Maybe if some research had been done then it may have extended his life. I've got a grandson and a daughter, so taking part in the 100,000 Genomes Project was a no-brainer, and it is only since I started talking to the specialists in Sheffield, that I have realised that I have had this for many years, like him."

Through the '100,000 Genomes Project' doctors and scientists are now able to sequence a person's genome – or the full blueprint of their DNA – in two days compared to 13 years ten years ago.

For Marcia, it could transform care for future generations: "Messages from my brain don't get to places they need to. I get confused easily and can't take too much information in at one time. I can start a conversation, or be carrying out an everyday task, but if I am interrupted, it may take a few moments to regain my momentum. I feel very apprehensive about walking out on uneven ground. There's so many times I would have fallen if I hadn't had my husband with me. This type of research may not be able to help me, but it could help a lot of people in future from the bank of evidence they can get from the DNA."

Recruitment update

Over 1,500 individuals living in the Yorkshire & Humber region, including both cancer patients and families living with rare diseases, have now signed up to participate in the project.

The initial recruitment of families with rare disease has been through the clinical genetics teams at the Northern General Hospital in Sheffield and Leeds Teaching Hospitals, but we are now in a position to offer support to other specialist teams at partner hospitals.

Training available for NHS clinical teams

One of the major objectives of this project is to encourage patient recruitment to the '100,000 Genomes Project' as part of routine practice across the breadth of clinical teams, to support healthcare professionals to do this, there are a number of training and education opportunities available free to NHS clinical staff.

Please contact Denise, Gill or Tammy to get started in your team:

- Denise Hancock (GMC Specialist nurse, Leeds) – **denise.hancock3@nhs.net**
- Tammy Kammin (YH GMC Specialist Recruiter, Sheffield) - **Tamar.Kammin@sth.nhs.uk**
- Gill Wilson (YH GMC Programme manager) – **gill.wilson@sch.nhs.uk**

We will arrange to meet with you and your team, provide any training required, and work with you to start recruitment in your specialist area.

Getting talking about genomics

The last Wellcome Trust Monitor survey said only 12% of the population would say they had a good understanding of what a genome is. The **'Socialising the Genome'** project is an engagement venture funded by **Genomics England**, the **Wellcome Trust** and **Wellcome Trust Sanger Institute**, with the aim of encouraging more people to talk about genomics. A series of six animations have been produced to inspire people to communicate more about genomics. **The animations can be seen at www.genetube.org**

The Yorkshire and Humber NHS Genomic Medicine Centre

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