Winter 2017

NHS
Yorkshire and Humber
NHS Genomic Medicine Centre

Genomes Update

Information for our partners, patients and staff

First patients sign up to the 100,000 Genomes project in Yorks and Humber

Hundreds of patients in the Yorkshire and Humber have now been recruited into the 100,000 Genomes Project – one of the largest gene mapping studies in the world.

Through the groundbreaking project scientists and researchers hope to find and share vital information that could unlock the genetic roots of how rare inherited diseases and some forms of cancer develop.

A genome is an individual's unique sequence of DNA, 3.2 billion letters long, and found in almost every cell in the body.

Donna Proctor, a 49-year-old domestic supervisor at Seacroft Hospital was the first person to provide a blood sample for the Yorkshire & Humber GMC's rare diseases programme because she has a predisposition to cancer. She said: "This could make all the difference for people in the future if it helps researchers find different treatments or cures for cancers. I think it is a really positive thing to do and I would encourage everyone who is eligible to take part if they can."

Anyone taking part in the 100,000 Genomes Project gives a sample of their DNA, along with their relatives, to one of the 13 NHS Genomic Medicine Centres. This is usually taken from a small blood sample. Cancer patients also donate a small piece of their tumour. After DNA samples are collected, they are sent securely to be sequenced. Genomics England then analyse the data. Results are then sent back to the local Genomics Centre for validation and clinical action.

The '100,000 Genomes Project' intends to deliver the sequence of 100,000 whole human genomes by 2017, from patients with rare inherited diseases and patients with a range of cancers, and will involve more than 4,000 participants from the Yorkshire and Humber region.

Did you know?

Genetic changes which increase the risk of developing diseases such as diabetes, coronary heart disease and several types of cancer have been discovered thanks to genomics

What is the 100,000 Genomes Project?

It is a landmark project which is helping bring personalised medicine to NHS patients and new knowledge and insights to health.

The Yorkshire and Humber is very proud to have been established as one of 13 NHS Genomics Medicine Centres across England.

The project involves looking at the genomes of patients with certain rare diseases and patients with certain cancers. By comparing the genomes from lots of people, the NHS Genomic Medicine Centre will help to give a better understanding of the diseases, how they develop and which treatments may provide the greatest help to future patients.

What is a genome?

Your genome is the instructions for making and maintaining you. It is written in a chemical code called DNA. All living things have a genome; plants, bacteria, viruses and animals.

Your genome is all 3.2 billion letters of your DNA. It contains around 20,000 genes. Genes are the instructions for making the proteins our bodies are built of – from the keratin in hair and fingernails to the antibody proteins that fight infection.

Genes make up about 1-5% of your genome. The rest of the DNA, between the genes, used to be called 'junk' DNA. It wasn't thought to be important. But we now know that DNA between genes is important for regulating the genes and the genome. For example, it can switch genes on and off at the right time. There is still much more to learn about what it all does.

What is DNA?

DNA (deoxyribose nucleic acid) is a long molecule. It has a twisted, double helix shape. DNA is made up of four different chemicals, or bases. These are represented by the letters A, T, C and G. The bases are attached to two phosphate backbones.

The bases are paired together; A with T, G with C. The two backbones twist around each other to give the characteristic double helix.

As well as being helix shaped, DNA is tightly packed so it takes up less space. If you stretched the DNA in one cell all the way out, it would be about 2m long.

What is genome sequencing?

Sequencing is a technique that is used to 'read' DNA. It finds the order of the letters of DNA (A, T, C and G), one by one. Sequencing a human genome means finding the sequence of someone's unique 3 billion letters of DNA.

Why sequence a genome?

Learning more about genomes can help us to identify the cause of genetic diseases.
Some rare diseases are caused by as little as a single change (variant), like a spelling mistake, in someone's DNA. Looking at the genome of a person affected by a rare disease can help find which DNA changes might be causing the problem.

In cancer, the tumour cells have developed a different genome to the healthy cells. Comparing the normal and cancer genomes may give clues about ways to treat the cancer. For some patients, knowing more about their genome may mean that a particular treatment can be recommended.

When the genome sequences of patients with the same condition are compared, it is possible to see patterns. These patterns can be put together with health information. Once this is done we may be able to link particular patterns with whether people are likely to become ill and, if so, how severe their illness is likely to be.

What is cancer genomics?

Cancer can be described as a genomic disease. It is caused by changes to DNA. The changes cause cells to divide and grow uncontrollably. Sometimes, the genomic changes are inherited from parent to child. But most of the time (about 90%) they happen naturally in cells.

We are sequencing DNA from a patient's tumour and healthy cells. We compare the two sequences. This helps uncover the exact genomic changes causing an individual's cancer.

This information can improve diagnosis. It can also help doctors choose treatments most likely to be effective for each person

Did you know?

The discovery of the human genome now makes it possible for scientists to read every letter of our genetic code in 2 days instead of 13 years

A world of opportunity for health professionals too

The 100,000 Genomes Project is a hugely exciting project which could fundamentally change the way medicine is delivered. It relies on partnerships across the health system and with Genomics England, and offers many opportunities for clinicians and nurses to develop new skills – all of which will be vital in bringing life-changing opportunities to patients both now and in the future. We'll be talking about these opportunities and developments and what they mean for patients and practitioners in the Yorkshire and Humber region in future editions.

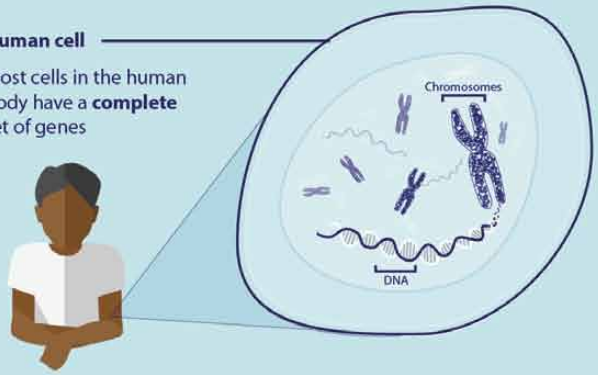
The 100,000 Genomes Project

Genomics england

What is a genome?

Human cell

Most cells in the human body have a **complete** set of genes



Your **genome** is one whole set of all your genes plus all the DNA between your genes.

There are around **20,000** genes in your **genome**

What is genomics?

Genomics is the study of the whole genome and how it works, but has also come to have a broader meaning to include the way that the genome is interpreted and the technologies that have been developed because of it.

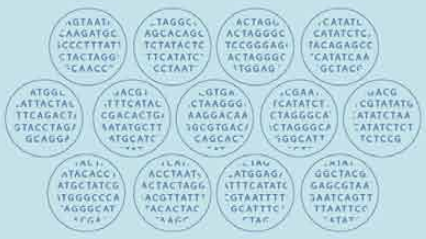
Why?

Looking at the whole genome will help us understand how disease develops and which treatments will be most effective.

80% of rare diseases are genomic

Around **40,000** people with cancer and rare diseases will take part in the project

over **330,000** new cases of cancer reported every year and growing



We compare books (that is, other patients' genomes) and try and find letter relationships that match to better understand gene activity.

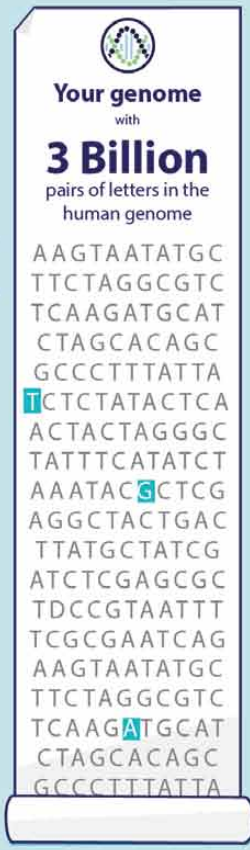
Sequencing

This means reading the human genome letter by letter (all 3 billion pairs of letters)

In the past it has taken
13 years & £2 billion
to read every letter of our genetic code

Today it only takes **2 days & £1k** to read every letter

Before



5%
were analysed
Only the active genes were looked at

95%
were unused
All the non-gene sections that we didn't understand were disregarded as useless

Now

We know that the non-gene (non-coding) parts of your genome may have a role to play so we look at the whole thing, every single letter, and how the different parts work together.

Think of it as reading a book, **every letter counts.**

About the Yorkshire and Humber Genomic Medicine Centre

The Yorkshire and Humber NHS Genomics Medicine is one of 13 centres involved in the national genome project and serves 5.3 million people from across the region.

We work with all specialists in the hospitals and active patient groups, to identify patients who could benefit from a genome test. A genome test is useful where a disorder runs through a family or if there is a strong likelihood of a change in a gene being a major risk factor for the development of a condition. Sometimes the gene sequence is useful to classify conditions into different subtypes that need different treatment, as is the case in cancer.

Once the project has been introduced to the patient by their healthcare professional, our team will approach the patient and explain the project in detail. It involves consent, sample taking and medical data input. Results will be fed back to the patients' clinicians like any other NHS test.

By doing this we are contributing to the national project and everyone who takes part helps us to build up a picture of the way genomes differ between us all. As a result we will start to understand what all the variations mean in terms of health and disease.

The Yorkshire and Humber NHS GMC is led by Sheffield Children's NHS Foundation Trust, Sheffield Teaching Hospitals NHS Foundation Trust and Leeds Teaching Hospitals NHS Trust. It is supported by the other 11 acute trusts in the region along with the Yorkshire & Humber Academic Health Science Network and our major Universities. It was also supported by direct input from patients and the public, which places the people of Yorkshire and Humber at the heart of crucial developments and improvements in healthcare.

Our aim in the Yorkshire and Humber is to collect samples from 4,700 participants with rare diseases plus their families, and patients with certain types of cancer, including breast, colorectal, lung, prostate, ovarian and sarcoma by December 2017.

Did you know?

A genome is an individual's unique sequence of DNA, 3.2 billion letters long, and found in almost every cell in the body

Did you know?

The work that the 100,000 Genomes Project does today will form the foundation for genomic medicine in the NHS and healthcare systems around the world.

Do you want to find out more or take part?

If you or a family member are living with a rare inherited disease and are interested in taking part in the 100,000 Genomes Project contact your clinical consultant to discuss further or visit **http://www.genomicsengland.co.uk/taking-part/** for further information.

The Yorkshire and Humber GMC is led by Sheffield Teaching Hospitals NHS Foundation Trust in partnership with Sheffield Children's Hospital Foundation Trust and Leeds Teaching Hospitals NHS Trust.

The Yorkshire and Humber NHS Genomic Medicine Centre

www.yorkshirehumbergmc.nhs.uk
Email: yhgmc@nhs.net
@yhgmc_org

NHS
Yorkshire and Humber NHS Genomic Medicine Centre