

Who is involved?

The project is being run by Genomics England, a company wholly owned by the Department of Health. Genomics England is working together with NHS England, Health Education England and Public Health England to deliver the 100,000 Genomes Project. NHS Genomic Medicine Centres (GMCs) have been set up across the country to recruit patients, take the samples and provide medical information to the project.

Who is eligible?

At the moment, patients with certain inherited rare diseases and some common cancers may be able to take part. Patients are referred to their local GMC by their clinical geneticist or hospital consultant. The project is in the early stages. In the future, it is possible that patients with other conditions, and those in other parts of the country will be able to take part too.



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The 100,000 Genomes Project



About the 100,000 Genomes Project

The project aims to sequence 100,000 whole genomes from about 70,000 people. Participants are NHS patients with certain types of cancer and patients with rare disease plus their families.

With their consent their genomes will be sequenced and linked to details about an individual's medical condition. Doing this may help medical teams provide better diagnosis or treatment. But it may not because not enough is known yet about the meaning of all the genomic data.

By adding key information from medical records too and putting all the data in one highly secure place, scientists can compare the data from tens, hundreds or even thousands of people with the same condition. Allowing approved scientists to have monitored access to this data is a very powerful way to understand the causes of ill health and how to treat it.

What is a genome?

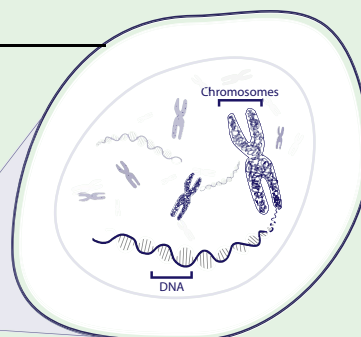
Your genome is one whole set of your genes, plus all the DNA between your genes. There's a copy in almost every cell of your body and it's the instructions for making, running and repairing you.

Genes account for less than 5% of your genome. It used to be thought that the rest of the DNA was junk but now we know all of your genome is important. That's why we're doing whole genome sequencing.

Sequencing means reading all the 3 billion DNA letters in your genome one by one. And now we know. Every letter counts.

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**

Aims of the project

Improving care for patients: Some patients with rare disease may get a diagnosis for the first time or discover what treatment might work best.

Setting up an NHS genomic medicine service:

By the time it finishes, this project will have put everything in place to enable the NHS to offer genomic medicine to those patients who need it. Genomic medicine will mean personalised treatment for patients.

For future generations: By looking at all the 100,000 Genomes Project data, scientists will gain new insights and understanding about the causes of disease and find better ways of helping patients, especially those who have conditions that are currently hard to treat.

To kickstart a UK genomics industry: Participants generously agree that researchers from industry such as pharmaceutical companies can look at their data. This is the best and quickest way to ensure that understanding from the project is turned into new medicines and diagnostics for patients. It will also help establish new jobs and companies in this important field in the UK.