

Understanding experiences of recruiting for, and participating in, genomics research and service transformation: the 100,000 Genomes Project, 2015-17

Lay summary

We know that many common and rare conditions have genetic causes but until a few years ago it had not been possible to use this knowledge to its maximum potential. In 2012, the 100,000 Genomes Project was announced which sought to collect 100,000 human genomes in England. A human genome is the complete set of genes or genetic material present in each individual. The goal of the Project was then to use the information derived from the genome for research, and to help improve the diagnosis and treatment of people with rare diseases, as well as those with common cancers. By 2018, the Project had reached its goal of collecting and examining 100,000 genomes. However, ultimately, the Project was about the people behind the data. With this in mind, in 2015 the Policy Innovation Research Unit (PIRU) had been asked to explore what donating DNA meant for the individuals concerned, as well to understand the experiences of the health care staff involved and the wider public's perception of the Project. The idea was that the findings of the PIRU research could be used to improve the process of recruiting and informing donors during the Project, as well as improving the implementation of similar genomics programmes in the future.

Specifically, the research looked at why participants had donated their DNA and what the process was like for them. It also asked a range of NHS staff, some directly involved in the Project and some not, for their views and experiences. To do this, in-depth interviews were conducted with 34 patients (some with rare diseases and some with cancer) and a range of genomics professionals. Group discussions were also held with members of the public and non-specialist healthcare workers to find out what they thought of the 100,000 Genomes Project and what they knew about it. Some of the patients that were interviewed for the study made videos talking about their experience of being involved in the project – you can see them here: healthtalk.org/experiences-participating-100000-genomes-project

The interviews and discussions were revealing in many ways. Most members of the public were supportive of the Project, were willing to be involved in related research, and had a trust in medical science and the NHS to do what is 'right' with the information. The people who donated samples were also surprisingly content with the process, how the samples were taken and how long it could take for their results to be available. It was as if any 'problem' with the Project was not the Project itself but rather the anxiety discussions about it raised in the public. Concerns were raised by healthcare staff but these were primarily about the pressure to hit recruitment targets and the competing demands of their usual work, and, as expected, some people raised concerns over the use of the genetic information but these concerns did not seem to undermine participants' and the public's support for the Project.

The positive message from this work is that there is general public support for the collection, storage and use of genetic samples on the grounds that people with rare genetic conditions and some common cancers are likely to gain more from genomic research and genomic medicine than they have to lose.

If you want to find out more, visit:

piru.ac.uk/projects/current-projects/understanding-participation-in-genomics-research.html

