Pooling genomic data into a "national data lake": the first steps towards establishing a UK learning healthcare system

Dr Sandi Dheensa, Clinical Ethics & Law, Faculty of Medicine

University of Southampton

Introduction

The UK 100,000 genomes project was launched by Genomics England in 2014. Participants are National Health Service (NHS) patients with a rare disease or cancer who want information about a diagnosis or treatment. However, personal benefit is not guaranteed.

The project is creating a biorepository of genomic and other health data for research and industry.

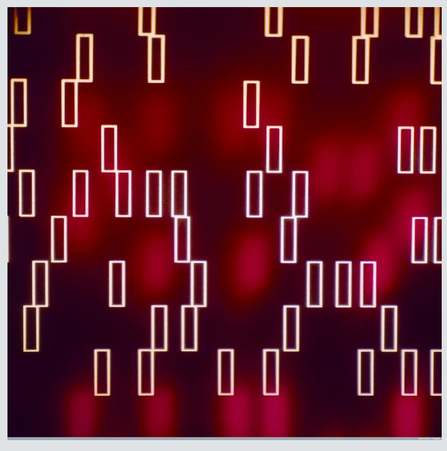
The decision over what happens to the biorepository once the project closes rests in the hands of the UK Secretary of State for Health.

The NHS Chief Information Officer recently proposed integrating genomic data, and data from the general UK from digitised health records, wearables, and apps, population, into a "national data lake". Pseudonymised and identifiable data would be made available for research and analytics by pharma and tech companies.

These are the first steps towards establishing a learning healthcare system, a model that hybridises research and clinical care, in the UK.

A principle that is professed to underlie the proposed approach is for information-sharing to be based on the local trusted relationship between citizens and healthcare professionals.

Given this, we aimed to investigate patients and family members' views about the uses of the data generated as part of the 100,000 genomes project. Findings help to shed light on views about how data ought to be used in any learning healthcare system.



Findings

Trust can sometimes be assumed

"[The data is] not going to be used for evil in any way is it…all the medical experts…you just trust that…things that are for the good of [others]. Only good can come of it." [P15, parent of child with undiagnosed dermatological condition]

But broad consent not always acceptable

"The only concern of it would be how much they knew. You're never going to know, that's the trouble, and how much of it goes to [for example] the Ministry of Defence and how much the other agencies are involved. You're never going to know, and that's the trouble. If it's got a civilian use, it's got a military use as well, And how much [of the data] will be made public? Can I look at it?" [P19, parent of child with undiagnosed neurological disorder]

Hopes for benefit-sharing and equality of access to healthcare

"I don't necessarily agree with the costs that [pharma companies] pass back to people. They make money out of my blood! A lot of money out of my blood. When you see on the news what they charge for producing these thing… but then at the same time I guess initially getting to the point of creating treatment has probably taken years." [P17, hereditary breast cancer]

"Maybe further down the line the pharmaceutical companies would have access and maybe the treatment will be tailor-made to the person." [P10, parent of child with undiagnosed growth disorder]

Altruism: more than just rhetoric?

"[Participants] enrolled on the principle that this is altruistic, and they don't expect any personal benefit. They're doing it because they want someone else to have a better chance than they did" [Genomics England board member]

I: "What are your hopes for and expectations about taking part in the 100,000 genomes project? One of the hopes is that you find a genetic diagnosis?"
P3: "Exactly. Find out more about the disorder and maybe even what you think caused it." [P3, undiagnosed neurological disorder]

Method

1) Analysis of media coverage of the 100,000 genomes project
2) Focus groups with healthcare professionals
3) Surveys with 300+ patients/family members with rare diseases or hereditary cancers
4) In-depth semi-structured interviews with 20 of these participants

We present data from (4) here. Analysis was thematic.

Discussion

Meaningful, non-tokenistic, public and patient involvement is needed to ensure that patient-generated data are used and shared in ways that benefit the common good. As the 100,000 genome project closes and genome sequencing moves into routine healthcare, deference to bureaucratic approaches to oversight are less useful than focusing on building trustworthiness, being transparent, and enabling on-going communication between patients and professionals. This should be the foundation of any learning healthcare system.

wellcome trust

www.soton.ac.uk/cels
Email: s.dheensa@soton.ac.uk
Contact author for references