

## Patient and Public Involvement (PPI) in Research – Case Study

**Provided by:**

Dr Jenny Taylor (Programme Director, Genomic Medicine Theme) and Dr Jude Craft (Project Manager)

**How patients and the public were involved**

- The Genomic Medicine Theme has been involved in several whole exome sequencing (WES) and whole genome sequencing (WGS) programmes over the last four years. WES is a cost-effective alternative to WGS as, while the exome represents less than 2% of the human genome, it contains ~85% of known disease-causing variants. Our work began with WGS500 and culminated in our current Health Innovation Challenge Fund (HICF) funded project to translate WGS into the clinic. These programmes have involved patients and families with suspected genetic disease, and those enrolled in the last 18 months have been recruited through an ethically-approved informed consent process developed through the project. Results are validated clinically and fed back through the clinician responsible for the family, providing diagnoses for patients where standard testing has failed.
- A new project run as part of the HICF programme aims to assess the significance of additional findings (potentially pathogenic genetic variants identified in participants with no symptoms). Results from this project have potential to influence the way this type of finding is viewed in terms of likely significance, but the area is highly sensitive and much debated ethically. For this reason, participants are asked to consent specifically for this part of the project.

**The impact of involving patients and public in the study**

Through these projects, we have shown the value of genomic testing for patients with genetic disease for whom standard testing has been inconclusive.

The National 100,000 genomes programme (of which Oxford is a centre) will test this further on a large scale, but our current research has potential to influence and establish the way genomic testing is integrated into NHS systems at a local level when this programme finishes.

**The importance of involving patients and the public in research**

The ultimate goal of the research we are involved in is patient benefit (whether in the immediate future for the individual families involved directly in our programme, or the longer term in establishing better and more comprehensive testing through the NHS). Genomic testing is a complex area ethically, and the patients and families involved in our programme are in a unique position to help shape the way we do things for the future. Being able to understand patient priorities will help us build systems that take into account the patient perspective and arrive at solutions that suit everyone.

**The biggest challenges in PPI and what might help to overcome them**

Finding ways to assess what is important to patients and their families, and the level of comprehension by the public generally of the benefits and limitations of genomic testing.

We have, through the programmes we are involved in, a reasonable number of patients and family

members, some of whom have indicated willingness to help with the qualitative aspects of the study. Routes to engage with the public on this very specific topic and methods of doing so in a meaningful way are less clear.

Suggestions:

- Access to organisations and people who can help us to define our questions clearly so that we are able to get the most from our patient research.
- Assistance with designing and running targeted public events.