100,000 Genomes Project

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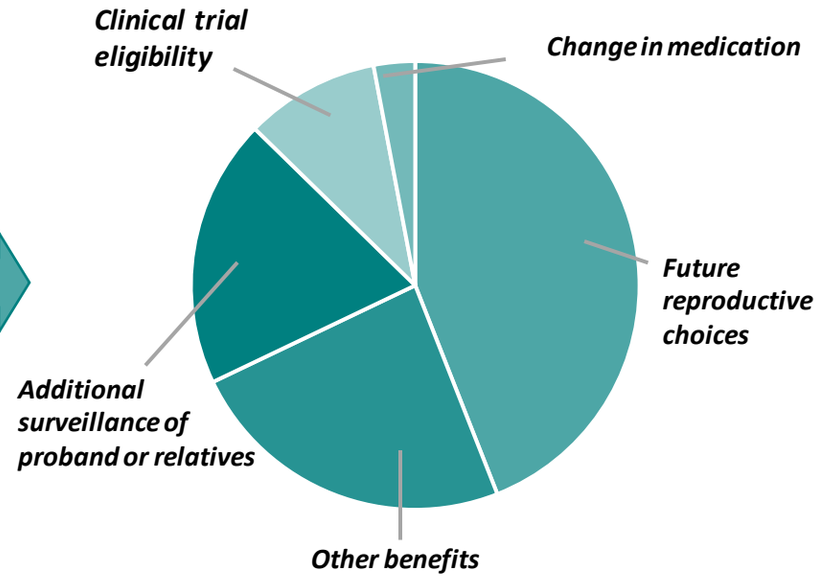
What?

The 100,000 Genomes project was a UK initiative set up in 2013. In essence, it combined **whole genomic sequencing (WGS) of over 100,000 genomes** with phenotypic NHS patient data to investigate the role of WGS in patients with **undiagnosed rare diseases, cancer and infection**.[1]

Why?

Aside from a pilot study already showing that WGS can improve the genetic diagnosis of rare diseases [2], the project has created the largest genomic healthcare data resource in the world and has initiated the **integration of genomics into routine healthcare** via the Genomics Medicine Service. [1]

Clinical impact of genetic diagnoses during the 100,000 genomes pilot-study on rare-disease diagnosis [2]



Who?

As of December 2021, participants who took part in the project will have received or are awaiting results for a specific disease. Some participants will have given **consent to receive further results on genomic variants** which may increase their risk of developing other diseases. Ultimately, everyone has the potential to be affected as genomic medicine becomes part of patient care.[1]

Author Comments

Genomics can improve patient care, for example by **reducing time waiting for a diagnosis** or being able to **identify which drug is likely to work best**. However, there are controversies around genomics medicine, including uncertainties around genome data sharing, storage and access, resistance to NHS infrastructure change and limited number of positively affected patients.[3]

References

1. Genomics England. 100,000 Genomes Project. Available from https://www.genomicsengland.co.uk/initiatives/100000-genomes-project [Accessed 31st March 2022)
2. The 100,000 Genomes Project Pilot Investigators. 100,000 genomes pilot on rare-disease diagnosis in health care – preliminary report. *N Engl J Med*. 2021; 385: 1868-1880
3. Barwell JG, O'Sullivan RBG, Mansbridge LK, et al. Challenges in implementing genomic medicine: the 100,000 Genomes Project. *Journal of Translational Genetics and Genomics*. 2018;2:13