Challenges in implementing genomic medicine: the 100,000 Genomes Project

In 2012, the then Prime Minister David Cameron announced funding for the 100,000 Genomes Project, to be organised by Genomics England (GE), a private company formed by the Department of Health in 2013. Through this project, GE works in partnership with NHS England (itself a non-departmental public body of the Department of Health) to integrate WGS into the NHS. The 100,000 Genomes Project aims to sequence 100,000 genomes from NHS patients with cancer and rare diseases. Data collected from the 100,000 Genomes Project can inform research on rare diseases, or benefit patient care potentially by streamlining the diagnostic process and tailoring care to the individual.

The project has strict inclusion criteria, to ensure data of clinical and research benefit is gathered. For over 300 rare diseases, specific criteria[1] are applied to maximise chance of recruiting individuals whose disease may have a Mendelian basis. **The project also requires submission of phenotypic data using SNOMED[2] (standardised healthcare terminology used for electronic health records and coding in over fifty countries https://www.snomed.org/snomed-ct/what-is-snomed-ct) terms**, and evidence of previous genetic testing (to screen out previously known mutations). A patient's whole blood samples must pass quality assurance and quality control tests. When relevant, close relatives (usually the parents) of the patient also undergo WGS. In the case of autosomal dominant conditions all affected members may be sequenced.

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