Personalised Medicine
All-Party Parliamentary Group



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# The 100,000 Genomes Project: transforming the NHS

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This document was produced for an APPG on Personalised Medicine event on the 100,000 Genomes Project. It sets out why the Project is important for embedding genomics in the NHS.

## The central role of genomics in delivering personalised medicine in the NHS

The 100,000 Genomes Project, which is being delivered by Genomics England, aims to sequence 100,000 genomes from approximately 70,000 patients with rare diseases and their families, and from patients with cancer.

## Why genomics?

This project is intended to provide the infrastructure, knowledge and catalyst to enable the NHS to establish an integrated genomic medicine service. By sequencing patients' genomes, wholly or in part, analysing these data and combining them with other medical information, clinicians can potentially offer more precise diagnoses and a greater degree of treatment personalisation for some conditions. An important part of this process is the development of analytical techniques and databases to store and share this information, such that the clinical impact of genetic changes on disease state can be fully understood, and acted upon.

## Specialist use: cancer and rare diseases

For many patients with rare but undiagnosed diseases, the insights gained from genome sequencing could be invaluable. The data collected from patients and their families could identify genetic mutations that have caused the disease and whether they were inherited or not. This may also give clues to potential features of disease and any therapies that might be available. Genomic and genetic testing is becoming an increasingly important part of the clinician's toolkit in a range of specialities, as sub-groups of patients with underlying rare diseases can be detected and offered tailored care.

Sequencing tissue from a patient's cancer and comparing it to their normal, healthy tissue allows us to identify the genetic changes that have contributed to the development of the cancer, and also which treatments might be most suitable for a patient based on the tumour's genetic profile. We can already test tumour tissue in a range of cancers and personalise therapy based on mutations in certain genes – for example, breast cancers with mutations that make cells overproduce a protein called HER2 can be treated with Herceptin. In the future, tumour genetic profiling could be used to further personalise treatment by determining which type of chemotherapy or radiotherapy would be most suitable for a patient, for how long, and at what dose.

## Looking to the future: mainstream use of genomics

Genomics also has the potential to benefit more mainstream areas of medicine, for example endocrinology, respiratory medicine and cardiology. In these areas, one of many potential applications is that of pharmacogenetics, using knowledge of how a patient's genome determines their response to drugs to decide which treatment and dose is most suitable for them. A better understanding of the genetic basis of disease will also help in the development of new diagnostic tests, and predictive testing, for

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example to discover whether couples are at risk of passing on inherited conditions to any future children, or antenatal tests to see if a baby is affected by a genetic disease. Genome sequencing technology is already invaluable for the detection of new disease threats, control of hospital infections and outbreaks, and tracking and controlling antibiotic resistance.

## Preparing the medical system for genomics

The expertise and infrastructure developed as a result of the 100,000 Genomes Project will play an important role in preparing the NHS for widespread clinical roll-out of genomic services. The new systems and procedures devised for the project, in conjunction with the current planned modernisation of NHS genetics laboratories, are a vital part of this process.