Personalised Medicine
All-Party Parliamentary Group

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What is genomic medicine?

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This document was produced for a joint event with the All Party Parliamentary Group on Health: ***Implementing a National Genomic Medicine Service for the NHS: building on the 100,000 Genomes Project***. It sets out what genomic medicine is and how embedding genomics across the NHS could improve care for patients.

A genome is a complete, unique set of DNA from an individual; genomic differences (variants) can affect people's characteristics, including their risk of, and response to, disease. Genomic medicine is an emerging medical discipline that involves using information about an individual's DNA to improve their clinical care through precise diagnosis, risk prediction, prognosis or treatment. Recent advances in DNA sequencing technology and bioinformatics (data science) have made it feasible to sequence vast human genomes at high speed and relatively low cost, and to analyse and make sense of that information for clinical use, though this is still a complex and challenging task. Genomics is also a crucial element in personalised medicine, a move away from a 'one size fits all' approach to using genomic and other forms of scientific and clinical data to inform more personalised care and targeted treatments.

The 100,000 Genomes Project

The world leading project was established to sequence 100,000 genomes from families with rare diseases and patients with cancer to identify genomic variants that are linked to disease, enabling more accurate diagnosis and prediction of disease and potentially also new treatments. Genomics England also aims to increase public understanding of and support for genomic medicine; accelerate uptake of genomic medicine into the NHS; and stimulate UK life sciences industry growth in genomics.

Improving care for patients

A genomic diagnosis for patients with rare diseases can both inform their direct medical care and also risks to other family members. Cancers arise when cells develop DNA mutations that drive their uncontrolled growth; comparing the genomes from a patient's cancer cells and normal, healthy cells can reveal which treatments will be most effective for the cancer. In both cases, the genomic information may also help the development of new diagnostic tests, devices or treatments that could help to deliver personalised care for future patients.

Genome sequencing is already playing a vital role in **infectious disease control**, detecting outbreaks and helping to track and combat antibiotic resistance. Genomics is also becoming an increasingly important tool for health professionals in a range of **mainstream clinical specialities**, such as endocrinology, respiratory medicine and cardiology. Sub-groups of patients with underlying rare diseases can be detected and offered tailored care. Pharmacogenetics (knowledge of how a patient's genome determines their response to drugs) can also help in decisions about the type and dose of drug treatments.

Embedding genomics across the NHS

NHS England plans to build on the 100,000 Genomes Project to make personalised medicine a reality in the NHS.[1] The Chief Medical Officer for England has strongly endorsed[2] the importance of creating a scalable, cost-effective NHS genomics service accessible by all patients who need it. To achieve this, she noted critical steps including the need to streamline genomics laboratory services, embed national standards, explain the necessity for sharing patient data in a secure environment, and change health professional attitudes to genomics.

1. *Improving Outcomes Through Personalised Medicine* - NHS England (2016)
2. *Generation Genome: Annual Report of the Chief Medical Officer 2016* – Department of Health (2017)