Genomic medicine: a tool for population health?

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Authors

Cecilia Price, NHS graduate management trainee

Genomic medicine has been hailed as the magic bullet for prevention, diagnosis and personalised treatment. But what can we really expect from this field?

The 100,000 Genomes Project (https://www.genomicsengland.co.uk/about-genomics-england/the-100000-genomes-project/) was established in 2012 with the aim of improving our understanding of the causes and development of diseases by sequencing the whole genome of individuals with cancer or a rare disease or their relatives. The 100,000th genome was sequenced in December 2018 and the results continue to roll in.

What is genomic medicine?

The development of a disease can be affected by one to hundreds of genetic variants – or none. What genomic medicine aims to do is identify areas of variation that might relate to disease. One method is whole-genome sequencing, which maps out the genome – all the genetic material of an organism, including the ~20,000 genes (DNA sequences that act as code for making proteins) and non-coding DNA. To identify variation, every cancer patient will contribute two genomes for comparison (one from a cancer cell and one from a healthy cell) and each rare disease case will involve 3 genomes (one from the affected person, two from biological relatives). Another type of genomic analysis is polygenic risk scoring, which combines information from variants across a genome to identify people with the highest risk of developing a disease.

For those with cancer or rare, undiagnosed and potentially genetic conditions, genomic medicine has the potential to improve diagnosis by uncovering variation not identified by genetic testing and other diagnostic tools. As a result, a patient might be offered more effective, personalised treatment: for example, a drug known to improve one specific type of cancer. Genomic medicine also presents the possibility of identifying predispositions to diseases, allowing for prevention: for example, the use of statins by someone with a genetic risk of coronary heart disease.

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To date, between a quarter and a fifth of rare disease cases and around half of cancer cases in the project have 'actionable findings (https://www.genomicsengland.co.uk/about-genomics-england/the-100000-genomes-project/)', which means participants might have received a diagnosis (https://www.genomicsengland.co.uk/alexs-story/), been able to join a clinical trial or even begun or altered treatment (https://www.genomicsengland.co.uk/about-genomics-england/participant-stories/) as a result of having their genome sequenced. The benefits of this go beyond medicine and into wellbeing; for example, receiving a diagnosis after years of uncertainty can provide a huge sense of relief. Indeed, how we as individuals interpret findings is a key consideration when it comes to the impact of genomic medicine.

Building on the successes of this project, which forged a path for the new NHS Genomic Medicine Service (https://www.england.nhs.uk/genomics/nhs-genomic-med-service/), the headline genomic ambition of the long-term plan is to sequence 500,000 (https://www.longtermplan.nhs.uk/online-version/chapter-3-further-progress-on-care-quality-and-outcomes/better-care-for-major-health-conditions/research-and-innovation-to-drive-future-outcomes-improvement/) whole genomes by 2023/4. To achieve this, from 2019 whole-genome sequencing will be offered to all children with cancer, seriously ill children who are likely to have a rare genetic condition, and some adults with rare conditions and specific cancers. The long-term plan also aims to offer genetic testing to a quarter of people who have inherited high levels of cholesterol in order to diagnose and treat those at genetic risk of a sudden heart attack. *Advancing our health: prevention in the 2020s*, the government's consultation (https://www.gov.uk/government/consultations/advancing-our-health-prevention-in-the-2020s/advancing-our-health-prevention-in-the-2020s-consultation-document) on prevention, outlines an ambition to accelerate the pace and carry out 5 million genomic analyses on not only patients, but also seemingly healthy individuals not displaying symptoms by 2023/24.

So what potential does genomics actually present for healthy individuals and how can we avoid misinterpreting the benefits? Before we realise its potential – as with any new medical technology – there are various practical and ethical challenges that must be overcome. For some cancer cases on the 100,000 Genomes Project, actionable findings from their results came after they have finished treatment; genomic analysis must be sped up before it becomes a mainstream diagnostic or preventative tool. The logistical challenges to embedding the genomic medicine service are uncovered in the House of Commons Science and Technology Committee's report (https://publications.parliament.uk/pa/cm201719/cmselect/cmsctech/349/349.pdf). Questions around who owns the data, how individuals give informed consent, how we can ensure equity of access and so on must be answered. These ethical issues are well summarised in the former Chief Medical Officer's 2016 annual report, *Generation Genome (https://assets.publishing.service.gov.uk/government/uploads/system/uploads/attachment_data/file/631043/CMO_annual_report_generation_genome.pdf)*.

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The next question is how we interpret this data once we have it. For some, discovering a genetic predisposition to a disease might feel like a death sentence. For others, understanding their predisposition to a disease might prompt them to reduce other risk factors they have control over. Portraying genomic medicine as health care's 'magic bullet' can be problematic because it promotes genetic determinism: the belief that genes alone can determine one's health, behaviour or personality, a view seen as both harmful and false (http://www.councilforresponsiblegenetics.org/Projects/PastProject.aspx?projectId=11) . Although genomic medicine might improve diagnosis and treatment for particular cases – such as rare disease and cancer – for healthy individuals there are other factors that influence our health that carry more weight and we have more control over.

Population health research (https://www.kingsfund.org.uk/publications/vision-population-health) indicates our physiology and genetics accounts for the smallest aspect of our health compared to other determinants, so it is essential that we think about the potential of genomic medicine within the wider context of a population health approach (https://www.kingsfund.org.uk/topics/population-health) . This approach gives us a fuller understanding of how genomics contributes to our health, alongside socio-economic, environmental and behavioural factors. Genomic medicine should be remodelled from a 'magic bullet' to a source of data, giving us a framework for acting on and contextualising this data. It's what we do with, and the value we ascribe to, this data that matters.

As the NHS's new Genomic Medicine Service kicks off and training programmes (https://topol.hee.nhs.uk/wp-content/uploads/HEE-Topol-Review-2019.pdf) are established for all health care professionals, could the focus on genomic literacy be part of a larger push for the NHS on wider population health literacy? Both as individuals and collectively, we cannot lose sight of the importance of what we already know about the socio-economic, environmental and behavioural determinants of our health.

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