The UK's plan to sequence the genomes of 200,000 newborn babies

Written by Jacob Dykes Published in **Development (/people/development)**

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Genomics England are poised to a launch a pilot project which will see the genomes of newborn babies sequenced on their very first day of life

Back in 2001, when a complete set of human genes (the genome) was sequenced for the first time, Francis Collins, then director of the National Human Genome Research Institute, predicted that it would be 'feasible' within 20 years to use genomic data to produce 'a kind of report card analysis' for a newborn baby's health. 'Well, here we are 20 years later,' says Simon Wilde, public engagement director at Genomics England. The organisation is poised to launch a publicly-informed pilot project to sequence the genomes of a large proportion of newborn babies within the NHS.

If it catches on, the practice will transform medicine. Armed with complete maps of babies' DNA, doctors could screen for a much wider range of genetic diseases than the nine conditions they currently look for. Rare diseases could be identified and diagnosed earlier, helping to find babies that may benefit from early treatment before

the damage is done. Moreover, if the genetic information is stored, it would be a powerful tool for doctors to tailor treatment to individuals across their entire lives. That information would also contribute significant data to research studies, enabling clinicians to deepen society's understanding of health and disease.

In July, Genomics England released the **results of a public consultation (https://www.genomicsengland.co.uk/public-dialogue-genomics-newborn-screening/)**, which indicated widespread support for newborn screening in the case of diseases for which there are existing treatments or management options. Now, Genomics England's pilot project, called **The Newborn Genomes Programme (https://www.genomicsengland.co.uk/newborn-sequencing/#:~:text=The%20Newborn%20Genomes%20Programme%20will,treatments%20for%20rare%2** is poised to begin enrolling 200,000 newborns over several years. The Newborn Genomes Programme will run an ethics-approved research pilot embedded in the NHS to explore how, and whether, to offer whole genome sequencing (WGS) to all newborns and accelerate diagnosis and access to treatments for rare genetic conditions. Enrolling a large proportion of the 600,000 babies born annually in England, it will screen for up to 600 genetic diseases for which treatment and care is 'available and actionable', such as vitamin B6–dependent epilepsy, or familial Diamond-Blackfan anemia – a red blood cell disorder.

'The focus of the pilot is exclusively on screening for rare diseases that present in early-life,' says Wilde. However, the genetic data that is harvested is powerful, and could be used in other ways across the patient's life. Genomic data could **aid diagnosis (https://www.frontiersin.org/articles/10.3389/fped.2021.663752/full)** for any diseases that emerge (if they have a genetic component), identify which treatments might be more beneficial, or those that the patient might react badly to.

It could also help other patients in the future. With parents' consent, Genomics England will de-identify and add babies' genomes to the National Genomic Research Library, alongside their health information. This could enable researchers to better understand the way that our genes influence health and disease throughout our lives.

There are significant ethical considerations, which are being handled with sensitivity. If the prospect is realised, healthcare bodies will need to safeguard genetic information. It must be kept away from third parties such as insurance or marketing companies. Then, there are broader considerations about what information is appropriate to share with patients and their families, and at what time in their lives.

It gets ethically thornier still, when we consider how the data might be used to assess for diseases that could present later in the babies' life. By assessing whether the baby has genetic variations commonly associated with bowel or breast cancer, for example, clinicians could paint a predictive picture of lifelong health prospects. 'There's still a huge amount of listening to be done. We're currently evaluating people's appetites for that kind of information,' says Henrietta Hopkins, who helped to oversee Genomics England's public consultation. 'Later-onset conditions are a tricky ethical area. As a newborn, you're not the one giving consent for your DNA to be sequenced. Once you reach the age of consent, what if you don't want to know whether you're likely to get cancer in your fifties or sixties?'

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Genomics England's consultation identified a spectrum of views here, 'which ranged from "knowledge is power" to "ignorance is bliss",' says Hopkins. Many, knowing that information on their genetic risk for diseases is within reach, will want to empower and inform their healthcare and lifestyle choices. 'Many people felt that, "actually, this is my health, my future, or my child's future, and we need to know as much as possible to inform ourselves". 'Others simply say "I just don't want to know".'

It is complicated by the fact that many genetic variants, even those associated with disease, never actually lead to the condition. In the USA, under a **pilot project called BabySeq (https://www.science.org/content/article/sequence-every-newborn-s-dna-despite-obstacles-uk-pushes-ahead)**, a team co-led by Robert Green from Brigham and Women's Hospital found that across 1,500 genes in 127 healthy and 32 sick babies, 8 per cent of apparently healthy babies had mutations for a childhood genetic disorder, and some 88 per cent were carriers of a genetic disease.

There are also questions around who the information should be shared with. When, for example, should indication of later-life predisposition to disease be shared with the children? 'People were very clear that we can't just consider the nuclear family – families come in a variety of structures. And for the later-life aspect, what are the implications if it's on my record that I am at risk of something that won't affect me until my 50s? Does that make me less employable for some jobs? Could an employer get hold of that information and see that I'm less genetically suited to becoming a pilot, for example?' Hopkins asks.

'Some people were even saying "what if you could analyse your genome and see that you are genetically suited to a particular profession?",' adds Wilde. Should that information be accessible? Should it trickle down to steer the way we design our lives?

The considerations go on. Studies show that disease-causing gene variants are not always uniform across ethnic groups, meaning the technique could lead to misdiagnoses if datasets are not broadly representative. Reference genetic databases, used to determine what a 'healthy' genome looks like, must therefore incorporate data from a balanced mixture of ethnic groups. 'There's a moral imperative here, and a scientific one,' says Wilde. 'If you have a variation in your genome, but the reference dataset is skewed toward the White European, diagnostic issues could occur. That's why it's so important that we have a diverse data initiative.'

There will also inevitably be questions around global equitability. During Covid-19, there has been many ethical debates around 'vaccine nationalism'. Do rich countries have an obligation to assist the developing world? Similar concerns may emerge as whole genome sequencing becomes part of routine newborn care in the developed world – a trend the UK is now at the vanguard of. 'There's a pride that the UK is one of the world's leaders in genomics, but there's a responsibility that comes with it to share the benefits equitably,' says Wilde.

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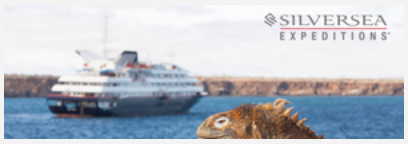

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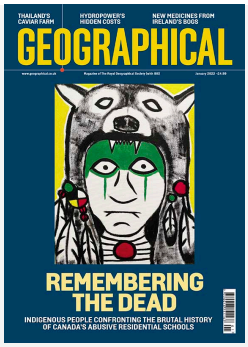

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