# NHS initiative uses whole genome sequencing to diagnose rare diseases in newborns



Cancer survivor and Health Secretary Wes Streeting has heralded a “landmark moment” in the NHS with the rollout of whole genome sequencing for newborns, a pioneering DNA test that can diagnose rare diseases, including cancers, at birth. Streeting described how the test, part of the Generation Study, recently helped identify a rare eye cancer in a newborn baby boy, Freddie Underhay, allowing life-changing treatment to begin before the cancer could cause serious damage or spread. This breakthrough exemplifies the NHS’s shift towards prevention and earlier intervention, aiming to save lives through cutting-edge genomic medicine.

The Generation Study, an ambitious initiative led by Genomics England in partnership with NHS England, seeks to screen up to 100,000 newborns in England for over 200 rare genetic conditions by using whole genome sequencing of blood samples taken from the umbilical cord shortly after birth. The project has already enrolled more than 20,000 families and collected over 500 samples from newborns at 13 NHS hospitals, with plans to expand to around 40 hospitals nationwide. Hospitals such as Imperial College Healthcare NHS Trust, Norfolk and Norwich University Hospital, and University Hospitals Bristol and Weston NHS Foundation Trust are actively participating, underscoring the programme's wide reach and impact.

This initiative aims to overcome the longstanding challenge many families face, where children with rare diseases may wait years or remain undiagnosed as their conditions worsen. By identifying more than 200 treatable conditions early, such as Metachromatic leukodystrophy (MLD) and rare cancers, the study offers earlier access to care, better health outcomes, and, in some cases, the possibility of preventing the onset of serious symptoms. Health Secretary Streeting emphasises how this innovation is a leap towards modernising the NHS, embedding genomics into routine care, and ensuring England leads the world in life sciences and medical technology.

Beyond newborn screening, the NHS is also involved in broader genomic research partnerships to advance understanding of complex diseases like cancer and dementia. These include collaborative efforts with organisations such as Oxford Nanopore, Genomics England, and UK Biobank to analyse tens of thousands of biological samples and create a world-first ‘epigenetic map’. Such research holds promise for new diagnostic and therapeutic breakthroughs, reinforcing the NHS’s commitment to combining scientific excellence with patient care.

Wes Streeting, a kidney cancer survivor himself, underlines the optimistic future this technology heralds—not just for families like Freddie’s, but for the future of medicine in the country. The NHS's 10 Year Health Plan envisages this genomic revolution as a cornerstone in transforming healthcare towards prevention-driven, innovative, and personalised medicine, giving children the best possible start in life and potentially eradicating some diseases before they manifest.

### 📌 Reference Map:

* Paragraph 1 – [[1]](https://www.mirror.co.uk/news/health/wes-streeting-heralds-landmark-moment-36087649)
* Paragraph 2 – [[1]](https://www.mirror.co.uk/news/health/wes-streeting-heralds-landmark-moment-36087649), [[2]](https://www.standard.co.uk/news/health/nhs-wes-streeting-england-genomics-england-children-b1185611.html), [[4]](https://www.independent.co.uk/news/health/nhs-wes-streeting-england-children-josh-b2622966.html), [[7]](https://www.uhbw.nhs.uk/p/latest-news/uhbw-leads-the-way-in-national-genomics-research-with-ground-breaking-newborn-s), [[5]](https://www.eastgenomics.nhs.uk/about-us/news-and-events/newborn-babies-tested-for-over-200-genetic-conditions-as-world-leading-study-begins-at-nnuh/), [[3]](https://www.imperial.nhs.uk/about-us/news/babies-tested-for-over-200-genetic-conditions-as-trust-joins-world-leading-study-in-nhs-hospitals)
* Paragraph 3 – [[1]](https://www.mirror.co.uk/news/health/wes-streeting-heralds-landmark-moment-36087649), [[2]](https://www.standard.co.uk/news/health/nhs-wes-streeting-england-genomics-england-children-b1185611.html), [[3]](https://www.imperial.nhs.uk/about-us/news/babies-tested-for-over-200-genetic-conditions-as-trust-joins-world-leading-study-in-nhs-hospitals), [[5]](https://www.eastgenomics.nhs.uk/about-us/news-and-events/newborn-babies-tested-for-over-200-genetic-conditions-as-world-leading-study-begins-at-nnuh/), [[4]](https://www.independent.co.uk/news/health/nhs-wes-streeting-england-children-josh-b2622966.html)
* Paragraph 4 – [[6]](https://www.gov.uk/government/news/landmark-genetics-partnership-to-probe-causes-of-cancer-and-dementia)
* Paragraph 5 – [[1]](https://www.mirror.co.uk/news/health/wes-streeting-heralds-landmark-moment-36087649)

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## Bibliography

1. <https://www.mirror.co.uk/news/health/wes-streeting-heralds-landmark-moment-36087649> - Please view link - unable to able to access data
2. <https://www.standard.co.uk/news/health/nhs-wes-streeting-england-genomics-england-children-b1185611.html> - The Generation Study, led by Genomics England in partnership with NHS England, aims to screen up to 100,000 newborns in England for over 200 rare genetic conditions. This involves whole genome sequencing using blood samples taken from the umbilical cord shortly after birth. The study has already collected more than 500 samples from newborns at 13 NHS hospitals, with plans to expand to around 40 hospitals. ([standard.co.uk](https://www.standard.co.uk/news/health/nhs-wes-streeting-england-genomics-england-children-b1185611.html?utm_source=openai))
3. <https://www.imperial.nhs.uk/about-us/news/babies-tested-for-over-200-genetic-conditions-as-trust-joins-world-leading-study-in-nhs-hospitals> - Imperial College Healthcare NHS Trust is participating in the Generation Study, testing hundreds of babies for over 200 rare genetic conditions. The study aims to identify conditions like Metachromatic leukodystrophy (MLD) early, enabling earlier diagnosis and treatment to slow disease progression and improve or extend lives. ([imperial.nhs.uk](https://www.imperial.nhs.uk/about-us/news/babies-tested-for-over-200-genetic-conditions-as-trust-joins-world-leading-study-in-nhs-hospitals?utm_source=openai))
4. <https://www.independent.co.uk/news/health/nhs-wes-streeting-england-children-josh-b2622966.html> - The Generation Study, led by Genomics England in partnership with NHS England, is screening up to 100,000 newborns for over 200 rare genetic conditions. Whole genome sequencing is used to identify these conditions early, allowing for earlier diagnosis and treatment that could slow disease progression or extend lives. More than 500 blood samples have been taken from newborns at 13 NHS hospitals, with plans to expand to around 40 hospitals. ([independent.co.uk](https://www.independent.co.uk/news/health/nhs-wes-streeting-england-children-josh-b2622966.html?utm_source=openai))
5. <https://www.eastgenomics.nhs.uk/about-us/news-and-events/newborn-babies-tested-for-over-200-genetic-conditions-as-world-leading-study-begins-at-nnuh/> - Norfolk and Norwich University Hospital (NNUH) has joined the Generation Study, offering newborns whole genome sequencing to identify over 200 rare genetic conditions. This early detection enables families to access appropriate support, monitoring, and treatment sooner, potentially preventing long-term health problems and promoting healthier lives for children. ([eastgenomics.nhs.uk](https://www.eastgenomics.nhs.uk/about-us/news-and-events/newborn-babies-tested-for-over-200-genetic-conditions-as-world-leading-study-begins-at-nnuh/?utm_source=openai))
6. <https://www.gov.uk/government/news/landmark-genetics-partnership-to-probe-causes-of-cancer-and-dementia> - A partnership involving Oxford Nanopore, Genomics England, UK Biobank, and NHS England aims to create a world-first 'epigenetic map' by analysing 50,000 samples from the UK Biobank. This initiative seeks to better understand the causes of diseases like cancer and dementia, potentially leading to breakthroughs in new diagnoses and treatments. ([gov.uk](https://www.gov.uk/government/news/landmark-genetics-partnership-to-probe-causes-of-cancer-and-dementia?utm_source=openai))
7. <https://www.uhbw.nhs.uk/p/latest-news/uhbw-leads-the-way-in-national-genomics-research-with-ground-breaking-newborn-s> - University Hospitals Bristol and Weston NHS Foundation Trust (UHBW) is leading in national genomics research with the Generation Study. Since October 2024, St Michael’s Hospital has been the first site in England to recruit families and complete testing for the study, enrolling over 800 mothers and successfully taking over 500 whole genome sequencing tests. ([uhbw.nhs.uk](https://www.uhbw.nhs.uk/p/latest-news/uhbw-leads-the-way-in-national-genomics-research-with-ground-breaking-newborn-s?utm_source=openai))