

Baby diagnosed ‘completely out of the blue’ with rare cancer in genetic screening

The study will help inform the Government’s long-term ambition to offer genomic sequencing to all newborns



Scientists make breakthrough that could halt spread of aggressive cancer

A [baby](#) was found to have a rare eye [cancer](#) and his sight is expected to be saved thanks to a new screening tool which is being assessed for widespread use.

Families across England are taking part in a “world-leading research study” which is screening 100,000 new born [babies](#) in England for dozens of genetic conditions.

One mother, whose son was diagnosed with a rare cancer picked up by the study, described how the results came “completely out of the blue”.

Vicky Underhay’s son Freddie was enrolled into the Generation Study which sees newborn babies offered whole [genome sequencing](#) using blood samples, usually taken from the umbilical cord shortly after birth.

Just four weeks after he was born at Sheffield Teaching Hospital in April, Freddie was diagnosed with a rare and aggressive form of eye cancer, which was picked up as a result of screening.

Freddie’s father Joey Underhay said that taking part in the study had changed his son’s life “phenomenally”.

The youngster, now six-months-old, was diagnosed with hereditary retinoblastoma, caused by a change in his RB1 gene.

Some 44 children are diagnosed with retinoblastoma every year in the UK.

Only around 44 children are diagnosed with retinoblastoma every year in the UK.

Catching the cancer early is crucial to save the sight of affected children.



Vicky and Joey Underhay with their son Freddie who was found to have a rare eye cancer and his sight is expected to be saved thanks to a new screening tool which is being assessed for widespread use (*Family handout/PA Wire*)

When there is no known family history of the disease, the cancer often goes undetected until later when the disease has progressed.

“There were no signs that anything was wrong – he was a normal little baby, so the results came totally out of the blue,” said Mrs Underhay.

Within four days of finding the genetic abnormality Freddie was seen by experts at Birmingham Children’s Hospital, where a a tumour was found.

Freddie was given a combination of laser treatment and chemotherapy and his doctors are hopeful they can save his vision.

Mrs Underhay, 36, told the PA news agency: “They said we are incredibly lucky to have caught it this early.

“It was such an amazingly quick turnaround.”

She said that the couple had spoken to other families of children with the same condition who spent months trying to get a diagnosis.

Decision to join the Generation Study

Mr Underhay, 37, said: “Our decision to join the Generation Study has changed Freddie’s life phenomenally.

“We were told that the first six months is vital in diagnosing and treating the condition.

“There is no telling at what point it would have been discovered if we had not taken part and what might have happened.

“The care that Freddie has received, and us as parents as well, has been second to none. We cannot thank everyone enough for all that they are doing for Freddie.”

He told PA: “If we had not taken part in the study there could have been a completely different outcome.

“He was just a normal little baby, you would have not known there was anything wrong with him.”



Freddie was given a combination of laser treatment and chemotherapy and his doctors are hopeful they can save his vision (*Family handout/PA Wire*)

Mrs Underhay added: “We’re hopeful that this kind of testing will be readily available to any parent who wants it for their child in the future.”

Dr Joe Abbott, Freddie’s ophthalmologist, said: “The chances of protecting Freddie’s eyesight have been greatly improved by finding out about his eye tumours earlier, thanks to the family participating in the Generation Study.”

The research project, led by Genomics England in partnership with NHS England, is available at 51 hospitals in England.

The study will help identify more than 200 rare conditions which causes a progressive loss of physical and mental skills.

By detecting these rare genetic conditions sooner, experts say hundreds of children could benefit from earlier diagnosis and treatment that could help slow progression of the disease or extend their lives.

Now, these conditions can be hard to diagnose, leading to delays in care.

Whole genome sequencing provides a readout of a person’s entire genetic code and looks for changes that relate to specific health conditions.

The Generation Study

The Generation Study, which was launched in 2024, screens for these conditions in babies who appear healthy but whose symptoms may not become apparent until later in life.

The findings of the study will help inform the Government's long-term ambition to offer genomic sequencing to all newborns as part of routine care.

The Generation New Era study will track the lives of 30,000 babies and their families (PA)

It will generate evidence on whether and how whole genome sequencing should be offered to every baby as part of screening for rare genetic conditions at birth, alongside the existing heel prick test – which looks for 10 rare but serious conditions.

Over 20,000 families joined the study

So far, over 20,000 families have joined the study and more than 60 “condition suspected” results have been returned by Genomics England to the NHS for confirmatory testing.

Dr Rich Scott, chief executive of Genomics England, said: “We believe genomics can transform healthcare in this country and be used to get ahead of serious illness.

“It's been incredibly moving to see the life-changing impact the Generation Study is having for families like Freddie's, who have been able to access treatment that makes a world of difference soon after being born.

“Every year thousands of babies are born in the UK with rare genetic conditions, but they can be hard to diagnose.

“On average, it takes around five years for a rare condition to be diagnosed, at which point it has often progressed to the point where treatment, if it exists, is far less effective.

“We are fortunate the UK is uniquely placed to test and – if the evidence supports it – roll out genomic innovations that help us move towards preventing sickness, not just treating it.

“The Generation Study is an example of just that – a national-scale research study developing evidence to inform whether every baby should be offered genomic sequencing. It's an important step towards a future of healthcare that is more preventative, with genomics playing a key role in that throughout someone's life.”

Professor Dame Sue Hill, chief scientific officer for England, said: “Today's announcement is a major step forward for the NHS in the use of whole genome sequencing in newborns.

“Freddie's amazing story highlights the strength of our world leading partnership with Genomics England, showing it is possible to identify rare genetic conditions like these earlier so children can start treatment faster meaning they have the best chance of a cure.”

-The Independent, October 17, 2025