NEWS FEATURE | 19 August 2026

Screening babies' genomes could save lives. Here's how it would work

Massive genomic newborn-screening studies are under way all over the globe. But questions about scalability, feasibility and net benefit remain.

By Uzma Rentia

Justin Ghattas and Scarlett Morwood enrolled their daughter, Giselle, in a study to screen for genetic disorders. Credit: Nadir Kinani Photography/Newspix

Two-year-old Giselle Ghattas is fearless, funny and affectionate, according to her parents. She loves diving headfirst down playground slides and climbing onto anything she can reach. Giselle seems like any other lively toddler, despite having a rare genetic disorder that threatens to rev her immune system at full throttle.

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The condition, known as familial haemophagocytic lymphohistiocytosis (HLH), causes fever and inflammation and can spiral into organ failure, neurological damage and death in as little as months if it goes untreated. And that's the case for many children with the disease. Because HLH is rare and variable, clinicians often misdiagnose it or fail to catch it early.

But Giselle is not like most people with HLH. Her parents, Justin Ghattas and Scarlett Morwood, enrolled her in BabyScreen+, a study in Australia, which uses whole-genome sequencing to screen newborns for genetic variants associated with severe, treatable diseases. Ghattas and Morwood came across the study on social media.

"If I had just kept scrolling on Facebook and not joined, then we'd probably still be, potentially even now, working out: 'What's wrong with her?'" says

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Ghattas. Instead, Giselle received a bone-marrow transplant at six months old, and despite some complications, she has recovered and begun to thrive. According to her physicians, she will probably not need any more treatment beyond routine monitoring, says Ghattas.

BabyScreen+ is just one of dozens of initiatives around the globe that is assessing the feasibility of expanded genomic newborn screening. Early results have shown that these approaches can flag treatable conditions that aren't covered by conventional newborn screening, which checks for up to a few dozen conditions. If the trials prove successful more broadly, genome sequencing could revolutionize current practices for newborn screening, providing in-depth information about a range of deadly and debilitating conditions, including some cancers.

For Wendy Chung, a physician-scientist at Boston Children's Hospital in Massachusetts, the promise of genomic newborn screening was evident long before she became a principal investigator on GUARDIAN, one of the largest genomic newborn-screening studies so far. "Newborn screening is, I would argue, one of the most, if not the most, successful public-health initiatives in the sense that it leaves no one behind," she says. "GUARDIAN is really adding another modality to enhance what already is a very successful public-health initiative."

But, for some, optimism is tempered by practical questions about the process, which is currently costly and difficult to scale up for broader implementation. Some also have ethical questions about applying genome sequencing to thousands of people, says Robert Green, a medical geneticist at Harvard Medical School in Boston. "There's a lot of controversy around this," he says, including privacy issues and the potential for discrimination by insurance companies. And not everyone has had the positive experience with genomic newborn screening that Giselle Ghattas's family has.

Early results

Current newborn-screening practices in many parts of the world use a dried blood spot taken from the heel shortly after birth. Laboratory tests screen the blood for a number of congenital disorders, mostly through chemical analysis of proteins and metabolites rather than through gene sequencing. US guidelines recommend testing for 66 conditions, which are mainly metabolic disorders. Many countries screen for fewer. France tests for 16 conditions, for example, and the United Kingdom screens for 10. Of the nearly 3.6 million infants born in the United States each year, 98% undergo this kind of screening, and it has been predicted that roughly 6,600 — about 1 in 600 — will test positive for a condition[1].

Genomic newborn screening would drastically expand what can be detected. Using DNA from the same dried blood spots collected for conventional screening, pilot studies are sequencing hundreds of genes or

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even the entire genome, with some screening for more than 700 disorders. If implemented broadly, this approach could identify thousands — perhaps millions — of children worldwide with rare genetic diseases.

Green co-led the BabySeq Project. Initiated in 2013, it was one of the first studies to evaluate genomic sequencing in healthy newborns. Across two independent BabySeq trials, around 1,045 infants were enrolled, including 432 who were chosen at random to undergo genomic sequencing. Among the infants whose genes were sequenced, approximately 11% had disease-associated genetic variants, and roughly one-third were already showing early signs of disease[2,3].

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More than a decade later, the BabySeq Project has been joined by a growing number of larger genomic newborn-screening studies. These programmes provide results — not diagnoses. Potential issues require confirmatory testing, with some findings ultimately being confirmed and others ruled out.

As genomic newborn screening expands, scientists are beginning to see how this process unfolds on a larger scale. At a conference last October, researchers shared preliminary results[4] from the GUARDIAN study from 15,000 newborn participants out of a planned 100,000. Whole-genome sequencing identified 411 infants (2.7%) whose screening results were subsequently confirmed through diagnostic testing. The vast majority were not identified through current newborn screening because the conditions they have are not included in those tests. In some cases, the findings prompted life-saving interventions, including bone-marrow transplants[5].

Several studies published early findings in 2025 with comparable results. The BabyScreen+ study[6], in which Giselle Ghattas was enrolled, screened 1,000 newborns and reported confirmed findings in 1.6% of participants. In Belgium, the BabyDetect study[7] confirmed genetic conditions in 1.8% of nearly 4,000 infants, including 0.8% whose conditions would have been missed by conventional newborn screening.

For researchers leading these initiatives, the findings provide important evidence that the technology is effective and that the approach is acceptable to families and consistent. "Even though we're based in different health-care systems, and we've taken some slightly different approaches to some of the components, many of the results are actually quite similar, which is reassuring," says Zornitza Stark, a clinical geneticist at the Murdoch Children's Research Institute in Parkville, Australia, and co-leader of the BabyScreen+ study.

Gene-list considerations

In their effort to maximize the benefits of genomic screening, researchers must first decide which genes should be on the panel — a question that has proved surprisingly contentious. Existing studies vary considerably. BabyScreen+ analyses 605 genes, whereas the BabyDetect study screens 405. The GUARDIAN study began with about 250 genes before expanding to 450, whereas North Carolina's Early Check programme evaluates 169. Most programmes focus on severe childhood-onset disorders that have some form of intervention.

As the number of studies has grown, however, researchers have begun to uncover the limits of current knowledge about the relationship between genetic variants and disease. In the GUARDIAN study, for example, infants with variants in the epilepsy-associated gene *SCN1A* differed notably from one another in the age of onset for seizures. Even variants in well-characterized genes don't always reliably predict disease, and disease databases do not always agree on how harmful a given variant is.

"Understanding genotype–phenotype correlations and fine-tuning reporting requires very large numbers of individuals to be tested," says Stark. "We're not going to get there unless we actually test thousands, if not millions, of individuals."

One of the central aims of genomic newborn-screening studies is determining which genetic changes cause disease and which prove benign. In the GUARDIAN study, 64 of 475 infants who were flagged initially as potentially having a genetic disorder showed no signs of disease at the time of confirmatory testing. The Early Check study reported 22 such cases among 50 flagged infants, whereas the BabyScreen+ study reported none.

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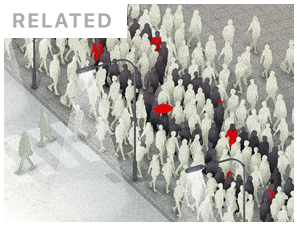

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The second challenge is deciding which conditions are sufficiently actionable to justify screening. "You only do screening if detecting it before it becomes clinically diagnosed leads to better outcomes," says Ned Calonge, a physician at the Colorado School of Public Health in Aurora and the outgoing chair of a disbanded advisory group that made newborn-screening recommendations in the United States. But what constitutes a meaningful health benefit remains open to debate.

Accordingly, some genomic newborn-screening studies have taken an exploratory approach to designing their gene lists. Some studies enable parents to opt in for tests for which the clinical utility is less well established than it is for the standard panel. In the GUARDIAN study, for example, all participating parents who consented to their child

undergoing screening for a primary list of treatable conditions were given the option to add a second panel of neurodevelopmental disorders associated with seizures. Although many of these conditions have no cure, investigators say that identifying infants who are affected might enable earlier treatment of seizures, which could improve outcomes.

Parental buy-in

For parents, the effects of such broad screening panels can vary drastically. For Dorka Nemes, the results were transformative. Her daughter, Safi Ford, participated in the UK's Generation Study and screened positive for isolated growth-hormone deficiency, a condition that limits growth. Nemes has the same condition, as do her brother and father.

Safi started growth-hormone therapy at just 6 months of age, whereas her mother was not treated until the age of 17, after much of the critical period for maximizing growth had already passed. Stories like Safi's are one reason that many patient advocates see the potential of broader genomic screening. Jennifer Handt, whose son has Duchenne muscular dystrophy and who helped to advocate for its inclusion in current newborn-screening panels in the United States, emphasizes that early symptoms rarely go unnoticed by families.

"You're not living in some blissful existence where you think your child is fine," she says. Watching a child struggle and then facing a delayed diagnosis can feel like a "double injury", she says, one that expanded newborn screening could help to prevent.

Safi Ford, daughter of Dorka Nemes and Cameron Ford, was able to start treatment for a genetic condition earlier than most children.Credit: Mel Yeneralski/Cambridge University Hospitals NHS Foundation Trust

But not every family leaves satisfied. Drew Villano gave birth to a healthy baby boy, Harmony, in April. According to Villano, a programme

coordinator approached her about enrolling in the GUARDIAN study shortly after delivery. She signed up.

Five weeks later, the phone rang. A genetic counsellor told her that her son carried a variant in a gene associated with the rare genetic disorder Smith–Magenis syndrome. Villano says the genetic counsellor struggled to explain the significance of the finding over several phone calls and ultimately told her she could look up the condition online.

Villano, a writer and owner of a real-estate company, said that the information she received wasn't very reassuring.

Ultimately, more testing showed that the variant was unlikely to be disease-causing, but the lack of clear, digestible explanations throughout the process, she says, left her shaken.

"When the margin of error involves human lives and not just data, there's a certain level of care that you should be taking," she says.

Chung, who leads the GUARDIAN study, says the programme's coordinators and genetic counsellors are well trained and should advise families not to go down an "online rabbit hole" when questions arise. Although she says she does not wish to minimize the stress that Villano describes, she adds that the response falls at the far end of a spectrum of experiences with the study.

More broadly, Alban Ziegler, a clinical geneticist at Toulouse University Hospital in France, who previously worked on the GUARDIAN study, says that parents are resilient in the face of findings that are troubling but uncertain. "We see that parents are able to cope with the news," he says.

Rolling out screening

Roughly 130 million babies are born worldwide each year, yet most genomic newborn-screening studies still take years to recruit only a few thousand participants, sequence their genes and interpret and return results.

"We have not made any inroads in terms of being able to answer, 'How do we get from this small pilot demonstration to that sort of scale?'," says Stark.

For conventional screening, many US states have yet to implement fully every condition recommended by federal guidelines, partly owing to stretched lab capacity. Incorporating large-scale genomic sequencing alongside existing screening programmes requires substantial investments in lab infrastructure, data analysis and workforce.

And costs would be higher than they are for conventional screening. At the launch of the GUARDIAN study, Chung estimates that sequencing and analysis cost just under US$1,000 per child. By comparison, Minnesota — one of the highest-spending states for newborn screening — earmarks

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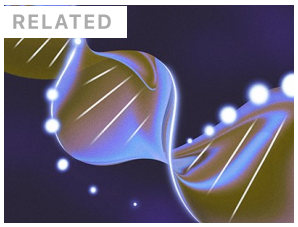

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roughly $250 per infant. As sequencing capacity expands and data interpretation becomes increasingly automated, Chung expects the cost of genomic newborn screening to eventually fall by at least half. Even so, whether state public-health programmes would be willing — or able — to absorb the costs remains uncertain.

Florida might hold the answer to that question. In 2025, lawmakers passed the Sunshine Genetics Act, creating a five-year pilot programme that aims to enrol 100,000 newborns and screen for more than 750 genetic conditions. "Hopefully, we can be the model for other states," says Pankaj Agrawal, a physician-scientist at Holtz Children's Hospital in Miami, and co-leader of the programme.

The United Kingdom has taken a similarly ambitious approach. Backed by £650 million (around $880 million) in UK government funding for genomics, the Generation Study is now more than halfway towards its goal of recruiting 100,000 newborns. "I think the government, at the moment, is very supportive of the use of genomics in health care," says Meekai To, a maternal- and child-health clinician who is involved in the study, which is run by the London-based company Genomics England.

On a national level, the United States is also beginning to shift from small research studies towards implementation. In 2025, the National Institutes of Health awarded funding for BRIDGES-NBS, a project co-led by Green that plans to sequence the genes of 30,000 newborns over three years, across six states and Puerto Rico. The initiative engages public-health labs to determine whether genome sequencing can be integrated into existing newborn-screening programmes.

Even if these studies prove successful, researchers expect implementation to happen gradually. Chung says that broad genomic sequencing should not replace the dried-blood-spot tests. Instead, she expects individual genes or groups of similar genes to be incorporated incrementally in current newborn screening.

Green says that newborn screening might ultimately become only the first chapter in a much broader vision. Rather than viewing a genome sequence as a one-time test, he imagines it as a lifelong clinical resource that could be reanalysed as individuals develop symptoms later in life. In that future, the value of genomic sequencing would extend well beyond the newborn period.

That vision remains years away, but many researchers think that the field is now taking its first meaningful steps towards it. "We had really this feeling when launching the study that it was a potential breakthrough. It's not so

frequent, at least in my career, having this feeling," says Ziegler. "I hope my grandchildren will benefit from it."

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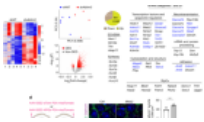

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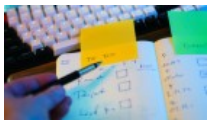

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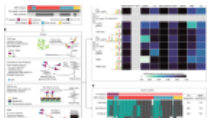

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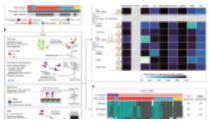

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