The New York Times https://www.nytimes.com/2025/06/05/health/babies-incurable-diseases.html

The Ethical Minefield of Testing Infants for Incurable Diseases

Screening can now determine their risk for an ever-growing list of conditions — including ones we can't do much about.

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June 5, 2025

In every postpartum hospital unit across the country, 1-day-old babies undergo the same ritual: A nurse pricks the newborn's heel and stamps tiny drops of blood onto a paper filter, which is then sent off for a standard screening panel.

Today, that panel checks for unusual bio-markers that may indicate a rare but treatable disease like sickle cell anemia or cystic fibrosis. But what if that same dried blood spot could tell you about the baby's risk of developing certain conditions later in life — some with no method of prevention or cure?

What if that heel prick could tell you that the baby was almost certainly going to be diagnosed with autism by the time they turned 5? Or that the child would be more likely to develop breast cancer as an adult?

Would you want to know? Would she?

These questions are no longer hypothetical. Tens of thousands of parents have sought such insights by enrolling their newborns in research projects that examine the baby's genome — the full blueprint for her growing body. As the cost of

sequencing plummets, the practice of analyzing hundreds of genes in healthy babies is quietly on the rise, ushering in new questions about where to draw the boundaries of knowledge — and who should get to decide.

Scientifically speaking, the possibilities are almost endless. Since virtually every disease has some basis in our genes, the full genome — with three billion base pairs, coding some 20,000 genes — contains a wealth of data to be mined for lifesaving intel and gut-wrenching secrets.

But the experts are divided. Some say that revealing a risk of an incurable illness will only put parents in distress, bombarding them with despairing predictions for their young child's life. Others believe any data about diseases that arise in adulthood, like breast or colon cancer, must be excluded, since they violate that future adult's privacy and autonomy — in other words, the right *not* to know.

Still others are of the mind-set that genetic forecasting is the future of medicine, and that knowledge is power — if used wisely.

A baby receiving a neonatal heel prick, or a Guthrie Test, an analysis for the detection of rare diseases. Astier/Science Source

Just Because We Can, Should We?

In a sense, the debate over newborn screening is as old as the tests themselves. In the 1960s, doctors began using dried blood spots to screen babies for a rare metabolic disorder called phenylketonuria and, soon after, a few other conditions.

By the end of the decade, the World Health Organization had published a list of 10 principles to guide whether a condition was appropriate for population-based screening. The guidance stated, for example, that there should be a consensus about what constitutes a positive case, and also an available treatment for it.

In the 1990s, when laboratories began to use new devices called tandem mass spectrometers to run many different tests on one blood sample, there was yet another burst in both scientific potential and ethical discourse: *Just because we can, should we?*

Officials in the United States ultimately settled on the idea of a federal committee that would scrutinize the evidence for each biomarker test — how grave the disease was, how precise the test was and whether there was some way to act on it — before deciding whether to add it to the "recommended uniform screening panel" it advises states to adopt.

But there is no such oversight system for whole genome sequencing, which hardly existed two decades ago but is now available to anyone with curiosity and a few hundred dollars. In some hospitals where research is underway, parents are even paid to participate.

The current administration has disbanded the federal committee, but even so, its approach to the recommended screening panel clearly wouldn't have scaled to meet the challenge of the genomic revolution. The committee pored over the data one disease at a time, and only nine conditions were added to the 29 original recommendations since they were established in 2010. At that rate, the team would have been hard-pressed to evaluate the hundreds of potential disorders that genome sequencing can already expose in a single test.

Congress has now commissioned a group of experts to help the government plan for a new era in baby screening. To some of those scientists, caution is key. Genetics has long been considered a uniquely delicate field of study, because it deals not with the present but with the future — and it's dangerous, these skeptics believe, to throw away long-held principles of what to look for just because we've gotten better at looking.

So what should be the new framework? Most experts agree on one conviction, at least: If examining a gene can reliably prevent a devastating outcome, we should do it.

An infant born with mutations in both copies of the SMN1 gene, for example, will develop Type 1 spinal muscular atrophy — a condition in which nerve cells in the spinal cord waste away, killing the child by age 2. But if the baby is given a therapy starting at 15 days old, she can meet all developmental milestones and stave off symptoms indefinitely.

A genomic sequencing machine. As the cost of sequencing plummets, the practice of analyzing hundreds of genes in healthy babies is quietly on the rise. Martin Krzywinski/Science Source

The debate lies in the many genes that aren't so straightforward. To unpack the nuances, it is helpful to think of genetics in terms of baking. While traditional blood-spot tests look for physical indications of disease in the body — unusual density in a loaf of fresh bread, for example — gene sequencing goes far upstream, reading the recipe and looking for errors in the original instructions that could one day lead to disease.

Now, here is where the drama comes in. Recipes usually lead to very specific dishes, but — as beginner bakers quickly discover — not inevitably. Plenty of genes linked to disorders can vary in both how likely they are to cause disease and how severe the symptoms might be. The environment matters, too — much like the way elevation can completely change how some recipes pan out.

In essence, a gene mutation in a healthy baby indicates a level of risk, not a diagnosis. Parents might be able to learn about a mutation days after their child's birth, but it could take months, years or even decades to see what bearing the mutation has on the child's life. That level of subtlety is a challenge for any of us to grasp, let alone a new parent who hasn't slept in weeks and is staring down at her bundle of hope as he coos in her arms.

'Maximize My Child's Outcome'

Many of the leading researchers in baby genomics are not intimidated by this uncertainty. They are also not worried about a condition being curable, so long as it can be acted upon in some way, and they argue that knowing a child's predispositions can help parents get support faster once they need it.

Consider Dr. Wendy Chung, a pediatric clinical and molecular geneticist, whose study, called "GUARDIAN," offers sequencing results for about 450 conditions to parents of babies born in NewYork-Presbyterian hospitals. More than 90 percent of parents who have enrolled have opted into results from a panel of conditions that have no cures, including neurodevelopmental ones associated with autism.

The rationale is that the data could hopefully grant a child access to speech and occupational therapy while the brain is still plastic, enabling them to become more integrated at an early age. It could also help them get earlier treatment for various conditions that often accompany autism, such as epilepsy, gastrointestinal issues and sensory challenges, like vision and hearing problems.

“What many parents told us is: If it’s going to be what it’s going to be, then I’d rather feel empowered to be able to potentially maximize my child’s outcome,” Dr. Chung said. They understood that “it’s there, regardless of whether you read it out.”

For all the potential benefits of such an approach, the study has been criticized bitterly by autism advocacy groups. Many of them see newborn testing for autism-associated genes as one step closer to prenatal testing to prevent the births altogether — one step closer to eugenics.

Bioethicists, too, have brought their own questions: How will the shortages in pediatric specialists and genetic counselors meet the demands of a deluge of families? Isn’t this going to draw out worrisome prognoses in children who may never manifest major developmental delays? And won’t it worsen inequities for children who actually turn out to be autistic, since well-connected parents will have gotten ahead of the curve and advocated early services?

But those are largely questions for the public health system to resolve. For parents who learn that their child is at risk of autism, the more immediate challenge is digesting the news. Dr. Chung and her team were concerned enough about parents’ emotional health that they designed a parallel project to study it.

A blood draw of a baby with spinal muscular atrophy, in Madison, Wis., in 2020. Parents might be able to learn about a mutation days after their child's birth, but it could take much longer to see what effect the mutation has on the child's life. Amber Arnold/Wisconsin State Journal, via Associated Press

How Early Is Too Early?

The BabySeq program in Boston takes genetic forecasting to another level — all the way into middle age. It reports results from more than 4,000 genes, including those that code for some forms of adult-onset conditions like breast and ovarian cancer.

When Robert C. Green, a medical geneticist at Mass General Brigham and a professor at Harvard Medical School, began BabySeq in 2013, it was the first program in the world to sequence healthy babies, and it was "totally radioactive," he said.

"People would publish academic papers that basically had the title, 'BabySeq is unethical.' People would stand up in meetings and yell at me." Then he paused and added: "They still sometimes do."

Critics of the program say that learning early about the likelihood of an adulthood illness does not offer any immediate benefit to the child and that labeling a healthy child as at-risk can actually negatively affect her life, leading to undue stigma, overprotection by parents, or even a self-fulfilling prophecy. (There is a clinical term for this phenomenon: "Vulnerable Child Syndrome," or V.C.S.)

But Dr. Green sees that worry as paternalistic. Parents are perfectly capable of adapting to nuanced data — even incomplete data — if that information might help their child thrive. They can encourage certain dietary choices or earlier colonoscopies, for example. The risk of catastrophic distress among parents is a "false narrative," he said, since those who would be particularly upset by a finding are self-aware enough to opt out.

What undergirds the support for projects like BabySeq is an opposition to the entire concept of genetic exceptionalism — this idea that our DNA is fundamentally different from the rest of our medical data and should be treated with extra tact just because it peers so far down the road. In an era when people track their own vitals on their smartwatches and experiment with longevity regimens, our genomes are just a new clause in the evolving contract between doctors and patients — a new space for shared knowledge and responsibility.

Kaitlin, a mother of two in the Boston area, was the ideal target for BabySeq. On the spectrum of parents who want to live "blissfully unaware" to those who want "every single iota" about their child, she said, she was deep in the latter camp. When a researcher came into her hospital recovery room and offered to have her 1-day-old son's genome sequenced from his heel prick, she asked whether anybody had ever said no.

Eventually she got a call: Her son had a variant of the BRCA2 gene that put him at heightened risk of pancreatic, prostate and even breast cancer when he grew up. And he had inherited the mutation from his mother.

Kaitlin, who asked to be identified only by her given name, took her son's screening results to her own doctor, who told her to go take out a life insurance policy. Then she saw a cancer specialist, had her uterus and ovaries removed, and prepared for

a double mastectomy while she went into early menopause, putting her at risk of brittle bones and other issues.

Kaitlin believes BabySeq most likely saved her life. But the answers it provided seemed to cascade into new questions. Kaitlin wondered whether her older son also carried the mutation. She wondered how much to tell the younger one, who would eventually discover the abnormality in his own medical records. She grappled with what to share with relatives — brothers, aunts and cousins who might also have inherited the BRCA2 variant, but who had most certainly not expressed any desire to know.

The dawn of widespread genome sequencing will unleash troves of quandaries like this one. Still, officials say, it has become an inevitability: The National Institutes of Health recently put out a call for projects to test the feasibility of population-wide baby genome sequencing — essentially touching off a scientific competition to envision the next era of newborn screening.

The tidal wave is coming no matter what, and the question is whether it will arrive systematically and responsibly — becoming a new pillar of a health care infrastructure that serves families well — or thoughtlessly and recklessly, creating an ethical morass for doctors and an emotional one for parents.

In the meantime, judgment calls remain in the hands of individual parents. Kaitlin ultimately sent an email to relatives about her genetic findings. It ended: “I’m sorry to be the one to bring this to light within the family in case it brings on more distress than intended.” Several family members decided to undergo sequencing and found the dangerous variant in their own genes. Others chose not to test.

Kaitlin’s younger son is now 8. She and her husband still have not decided when and how to tell him.

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