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Improving Infant Lives through the Power of Rapid Precision Medicine

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Decoding the human genome is driving a revolution in precision medicine, especially for newborns and children fighting to overcome a rare disease.

When it comes to pinpointing the exact cause of an unexplained serious medical condition, the diagnostic technology known as rapid whole genome sequencing (rWGS) can provide fast, precise and often life-changing answers to medical mysteries.

In a pilot study funded by the State of California called Project Baby Bear, we demonstrated that using rWGS in neonatal intensive care units produced better health outcomes and reduced suffering for the infants while saving millions in health care costs.

Among high-risk infants with rare genetic diseases, time to treatment is crucial. Project Baby Bear provided doctors with vital diagnoses that empowered them to make life-altering medical decisions with certainty.

Over the course of 18 months, we successfully sequenced babies at five hospitals across the state to show that not only did rWGS provide crucial information to guide individualized care, but resulted in shorter hospital stays and fewer unnecessary procedures for a net cost savings.

It's very rare to be able to use the most advanced, innovative technology medical science offers to both improve a child's life and save money, yet Project Baby Bear proved it's possible.

Using rWGS, we analyzed the entire genetic code of a total of 178 babies in intensive care. The results provided diagnoses for 43% that explained why the infant was hospitalized and led to a change in medical care for 31%.

In July 2018, California lawmakers allocated $2 million to fund Project Baby Bear as a pilot program to help improve outcomes for babies enrolled in Medi-Cal, the state's public health program, and examine the health benefits and cost effectiveness of rWGS.

Project Baby Bear more than paid for itself by saving $2.5 million above the state's investment. But diagnoses and savings are not the end of the story. Babies with rare diseases need expert care and follow-up.

Through Project Baby Bear, we built a statewide precision medicine guidance practice that provides sequencing laboratory services with expert telemedicine consultation for physicians about treatment options. In this way, we support intensive care physicians at hospitals near and far in providing rapid precision medicine to their youngest, most vulnerable patients.

In fact, in addition to Project Baby Bear, we are already collaborating with some three dozen clinical and research partners nationwide to provide these services, allowing them to deliver comprehensive rapid precision medicine.

What this has shown is that we are now in a position—across California and the nation—to be able to adopt rapid whole genome sequencing as first-line action, instead of a last resort.

Across the country, there are thousands of children born annually who become critically ill and are admitted to intensive care with a rare, undiagnosed disease. In the absence of rapid whole genome sequencing, the cause of their diseases may go undetected causing irreversible harm to many and, in some cases, proving fatal.

Here's the catch. We have the technology and expertise available to save them, but sadly rWGS is not currently covered by public health programs and only in a few exceptions by private medical insurance. As a result, babies are in jeopardy of missing out.

We urge policymakers and private medical insurance providers to meet this need and make rapid whole genome sequencing a covered benefit for all infants with rare and undiagnosed diseases.

We're not saying to sequence every newborn. But let's make this available to every child admitted to intensive care who is gravely ill and struggling to survive with an unexplained condition.

Rather than taking the traditional approach of searching for answers by ordering a series of tests, let's use the best-in-class option first to get the diagnosis with a single test on the first try.

Rapid whole genome sequencing offers life-changing benefits. Now it's up to lawmakers and private insurers to recognize that this not only improves and saves lives, but pays for itself too. Recently, Blue Shield of California agreed to cover rapid and ultra-rapid whole genome sequencing for critically ill infants and children in intensive care who have unexplained medical conditions. Learn more.

Our hope is that this will become the standard of care for babies and children all across America. The children are waiting.

Join us online for our virtual Frontiers in Pediatric Genomic Medicine Conference on September 24-25.

About Dr. Stephen Kingsmore

Stephen Kingsmore, MD, DSc, leads a multidisciplinary team of scientists, physicians and researchers at Rady Children's Institute for Genomic Medicine who are pioneering the use of rapid whole genome sequencing to enable precise diagnoses for critically ill newborns. Among his accomplishments, Dr. Kingsmore holds the world record for achieving the fastest molecular diagnosis using whole genome sequencing in 19.5 hours.