BeginNGS™

NEWBORN GENOMIC SEQUENCING

to end the diagnostic odyssey

Birth...

the beginning of a family's story. But for families of babies born with a rare genetic disease, the story is one fraught with challenges on their journey to an accurate diagnosis and appropriate care.

Imagine if we could change the narrative for those families from the very start.

Rady Children's Institute for Genomic Medicine® (RCIGM) is helping to **rewrite the story** for infants with rare diseases through **BeginNGS™, a novel health care delivery system** designed to screen newborns for genetic diseases — and connect their doctors with effective interventions.

Sometimes the solution is as simple as a vitamin supplement; other times, the baby can receive cutting-edge gene therapy and grow up to be a typical healthy child, like baby Fitz Kettler, shown right (radygenomics.org/fitz).

> By rewriting the beginning, we can help families end their diagnostic and therapeutic odysseys **so they can fill their stories with hope**.

Stephen Kingsmore, MD, DSc,
President and CEO of RCIGM

Genetic diseases are chronic, progressive, and can be life-threatening. They may not appear until later in childhood; however, the effects may begin before symptoms manifest. The effects may be irreversible if not diagnosed and treated. The combination of rWGS and a new treatment guidance system — Genome-to-Treatment (GTRx) — helps ensure each baby is screened for treatable genetic conditions, and helps physicians deliver Rapid Precision Medicine.

Like traditional newborn screening, BeginNGS will screen only for early childhood-onset, severe diseases for which there are effective interventions today and for which early intervention will improve outcomes. Since the whole genome is sequenced, the screening can be rapidly expanded to include new disorders as effective interventions become available.

Screening
Screening newborns for ~400 genetic diseases with known intervention options using rWGS

Diagnostic Report
Diagnosing rare disease earlier

Rapid Precision Medicine™ Treatment
Employing Genome-to-Treatment (GTRx) to provide immediate intervention guidelines to clinicians

Why Now?

For over 50 years, newborn screening (NBS) has successfully identified conditions that may affect a child's long-term health or survival.

Current NBS tests for a few dozen conditions. As our understanding of genetic diseases grows, and the pace of therapeutic innovation accelerates, NBS by rWGS may provide a more dynamic way of expanding the number of conditions as new interventions are approved. Technological advances in rapid whole genome sequencing (rWGS) have made it possible to return test results in just a few days at a lower cost.

"Expanding rapid WGS to newborn screening is important work with the potential to end agonizing years-long diagnostic odysseys while yielding critical data on the true prevalence of rare disorders. **Early diagnosis of rare disease patients will also open the door to ongoing monitoring of symptoms and gathering of critical natural history data.**"

Charlene Son Rigby
CEO of Rare-X

BeginNGS uses rapid whole genome sequencing (rWGS®) **to identify and address genetic conditions *before* a child gets sick. BeginNGS is not intended to replace current newborn screening programs.** It will complement the existing processes.

RCIGM is forming a consortium of leading organizations to shape and scale BeginNGS.
Partners include:

- Non-profit research organizations
- Advocacy groups
- Life sciences
- Diagnostic & labs
- Health systems
- Public & private payors
- Government (state & federal)
- Regulatory bodies
- MDs & genetic counselors

Consortium members will together address important challenges such as privacy, diversity and data governance.

To learn more about BeginNGS,
go to **Begin-NGS.org**
or email us at
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