Rady Children's Hospital Foundation

INNOVATION
BELONGS IN EVERY MOMENT

Rady Children's Institute for Genomic Medicine
Project Baby Bear Bridge Funding Philanthropic Opportunity

Background

For children with genetic diseases, timely diagnosis via genomic sequencing enables specific, targeted treatments that save lives, prevent suffering, empower families, and cut costs associated with misdiagnosis.

Maverick Coltrin, a Rady Children's NICU patient whose life was saved thanks to rapid Whole Genome Sequencing

Over the next five years, the greatest opportunity by far to realize these benefits is for infants less than a year old who live in neonatal and pediatric intensive care units (NICUs and PICUs, respectively). There are more than 8,000 known genetic diseases; approximately 30% of the infants admitted to a Level 4 NICU exhibit symptoms that could be associated with genetic disease. These symptoms include acute physical distress, developmental disorders, and/or other severe chronic illness. **For this fragile patient population, genomic medicine has the potential to transform both babies' health outcomes and the healthcare economics that determine access to this life-saving and life-changing service.**

Genetic diseases, including chromosomal defects, genomic structural defects and single gene diseases, are the leading cause of death in newborns and NICU and PICU infants in North America. **For a baby in the NICU or PICU, quickly identifying a precise diagnosis for a genetic disease is critically important for parents and physicians.** This information allows for optimal decision-making, particularly when contemplating weighty decisions around major surgeries, suitability for organ transplant, use of extracorporeal membrane oxygenation, institution of protein replacement therapies, and/or implementation of palliative care.

Furthermore, the 10% of children who suffer from severe chronic illness incur 70% of all healthcare costs at tertiary pediatric healthcare organizations across the United States. The care of infants in Level 4 NICUs and PICUs is among the most expensive in healthcare. Costs can vary widely across various hospital systems, but the average cost is around $3,000 per day for infants hospitalized in neonatal intensive care units. These high costs are typically associated with protracted hospital stays and ineffective treatments that are often directed at the alleviation of symptoms rather than targeted at their root causes. **Indeed, most of the diagnostic methods currently utilized in these advanced care centers are based on the symptoms or effects of disease, and not their underlying causes.**

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Rady Children's Institute for Genomic Medicine (RCIGM) offers a unique solution: a combination of state-of-the-art tools and services enabling our expert team to provide rapid Whole Genome Sequencing (rWGS) for the diagnosis and management of genetic disease among infants in NICUs and PICUs.

At RCIGM, our focus on implementation science integrates process engineering – the design, operation, control, optimization and scalability – of rWGS to enable rapid diagnosis and implementation of precision medicine. Through close partnership among expert physicians, scientists, and technologists, RCIGM is able to provide children and families with diagnoses and interventions within the critical therapeutic window – often saving and transforming lives before it is too late.

PROJECT BABY BEAR

Project Baby Bear is a state-sponsored research project under which our rapid Whole Genome Sequencing (rWGS) approach is being implemented for acutely ill children enrolled in the Medi-Cal program. We are sequencing babies at five neonatal and pediatric intensive care sites in California:

- Rady Children's Hospital – San Diego;
- Valley Children's Hospital;
- UC Davis Medical Center – Sacramento;
- UCSF Benioff Children's Hospital Oakland; *and*
- Children's Hospital of Orange County.

In this study, Rady Children's Institute for Genomic Medicine is evaluating the extent to which rWGS changes both patient health outcomes and the cost of patient care.

Our goal is to demonstrate to Medi-Cal the health and economic benefits of providing rWGS as a frontline, reimbursable, covered service across the state of California. Furthermore, this proof of concept will advance parallel conversations with private insurance providers in California and beyond. **Ultimately, our aim is to make rWGS available to every child and family across the United States who could benefit from this service, and for insurance providers to cover the cost.**

In June 2018, the State of California appropriated $2 Million in funding for the Whole Genome Sequencing Pilot Program. The State Department of Healthcare Services in turn provided this grant funding to Rady Children's Hospital – San Diego to sequence 100 Medi-Cal babies across our five partner sites, in an effort called Project Baby Bear. At the conclusion of our state grant, we will provide California's State DHCS a report on the cost of care and outcomes of children receiving rWGS compared to those not receiving rWGS.

As of July 2019, Rady Children's has completed sequencing the initially promised 100 children (*see figure)*. We have begun preparing our report to the State detailing our compelling results, which include a 51% diagnostic rate and a 45% change in medical management for those babies who received diagnoses.

PROJECT BABY BEAR PILOT SITE ENROLLMENT

PROJECT BABY BEAR SITES	1ST PATIENT SAMPLE RECEIVED	TOTAL # OF CASES RECEIVED TO DATE
Valley Children's Hospital	11/20/18	25
Rady Children's Hospital – San Diego	11/20/18	25
Children's Hospital Oakland	1/12/19	14
UC Davis – Sacramento	1/15/19	25
Children's Hospital Orange County	2/6/2019	11
	Total Project Baby Bear Cases to date	**100**

Our partners have expressed great enthusiasm for Project Baby Bear, citing great benefit to patients and physicians when a genetic diagnosis was returned (or ruled out); and underscoring the tremendous impact in outcomes they were able to deliver to children and families.

Current State

Due to this enthusiastic and effective partnership, we reached the 100-sequence benchmark well ahead of the original anticipated time line. As such, our state funding has been depleted and our work with these hospitals must cease while we wait for the next state budget cycle, which opens in January 2020. We plan to apply at this time for additional state funding, which would be allocated in June 2020 – at which point we can resume sequencing patients. In the meantime, we will be finalizing our report to the State proving the efficacy of our method. **Upon providing our report to the State, we anticipate that the overwhelming evidence of clinical efficacy and healthcare cost saving will lead legislators to provide more funds for an "all state go forward" status for rWGS of Medi-Cal children.**

Philanthropic Opportunity

Bridge funding would allow RCIGM to continue sequencing and providing results for eligible babies and families during the window between state funding cycles. Continuing this program without interruption would ensure that our partners continue to have their clinical leadership and internal resources devoted to this enterprise, while momentum at their hospitals in support of our approach has been building. RCIGM will be able to generate stronger, more iron-clad proof that our approach provides irrefutable healthcare outcomes and cost benefits. Medi-Cal will then have greater incentive to implement this service as a covered benefit and will serve as the first domino in a series of insurers likely to follow suit. Most importantly, babies' lives across California will be saved and their families' lives transformed.

Thank you for considering an investment to sustain this transformational study. You have the opportunity to change lives today, and to change children's healthcare in perpetuity.

Total Philanthropic Investment Opportunity: $1 Million