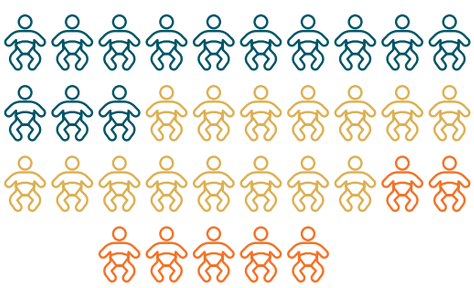
TIMELY AND SUSTAINABLE ACCESS TO DIAGNOSTIC TESTING

SOLUTION: Reduce costs and limit poorer outcomes associated with prolonged diagnosis by providing broader newborn and family screening and access to exome or genome sequencing for rare disease patients in instances where (a) no/few alternative testing options are available or (b) broad testing provides advantages to sequential/multiple single marker testing or help reduce likelihood of unnecessary downstream treatments or procedures by helping to render a more informed diagnosis[1,2].

Earlier diagnosis reduce costs and the number of unnecessary testing—often invasive—and specialty consultations. A genetic diagnosis also provides information about risks to other family members, including reproductive risks. Potential changes in standard of care will result in reduced costs and yield savings, as was demonstrated by **Project Baby Bear**.

project baby bear

$2M appropriated[4]



178 babies enrolled

76 babies diagnosed[7]

35 rare diseases identified[7]

Each icon above represents 5 babies.

$2.5M in healthcare savings[7]

$750K min. ROI[7]

SOURCES

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ABOUT US

Global Genes is a 501(c)(3) non-profit organization dedicated to eliminating the burdens and challenges of rare diseases for patients and families globally. In pursuit of our mission we connect, empower, and inspire the rare disease community to stand up, stand out, and become more effective on their own behalf -- helping to spur innovation, meet essential needs, build capacity and knowledge, and drive progress within and across rare diseases. We serve the more than 400 million people around the globe and nearly 1 in 10 Americans affected by rare diseases. If you or someone you love has a rare disease or are searching for a diagnosis, contact Global Genes at 949-248-RARE or visit our resource hub at globalgenes.org.

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