Timely and Sustainable Access to Diagnostic Testing

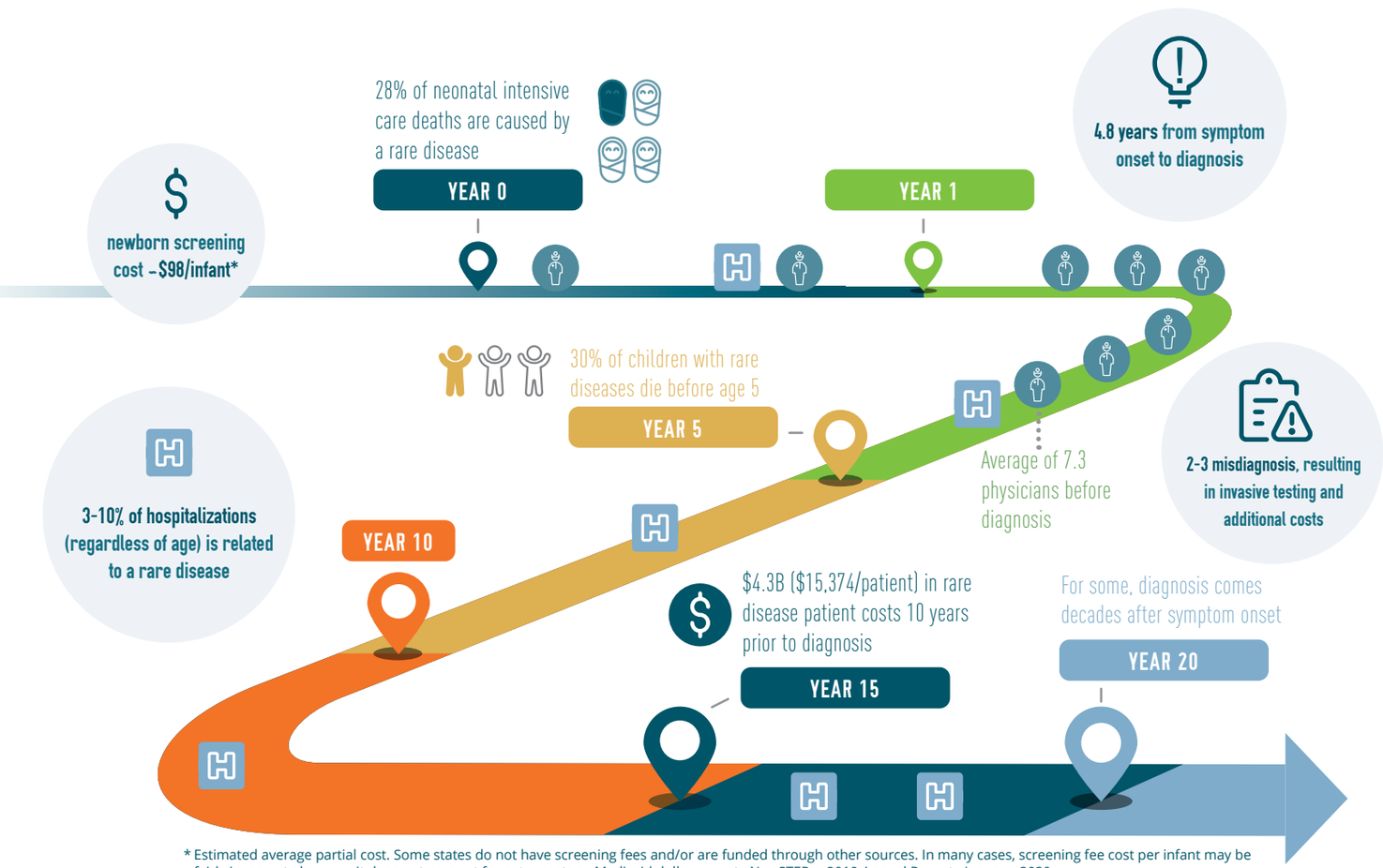
Rare diseases are defined as conditions that affect fewer than 200,000 patients in the US, and 50% of rare conditions affect children[1]. The patient journey to a diagnosis in rare disease, often known as the diagnostic odyssey, is complex and often lengthy. According to a study done by Global Genes, the average time from symptoms onset to diagnosis is 4.8 years. However, some patients report delays of up to 20 years[2].

Over the course of that diagnostic odyssey patients may: experience worsening symptoms, develop additional medical complications[3], miss out on critical, early interventions[4], undergo unnecessary testing and procedures, or the entire family unit may experience significant psychological stress during the search for a diagnosis[5,6].

50% of known rare diseases affect children

65% of rare diseases are associated with a reduced lifespan

⚠ PROBLEM: Patients with rare diseases and their caregivers often endure an extended, frustrating, and costly diagnostic odyssey' on the way to an accurate diagnosis and a potential treatment.



* Estimated average partial cost. Some states do not have screening fees and/or are funded through other sources. In many cases, screening fee cost per infant may be fairly inaccurate because it does not account for extra costs or Medicaid dollars spent. NewSTEPs. 2019 Annual Report. January, 2020.

SOURCES

1. Acess to Critical Therapies White Paper https://globalgenes.org/resources/guiding-principles-of-rare-disease-care-and-patient-access/

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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