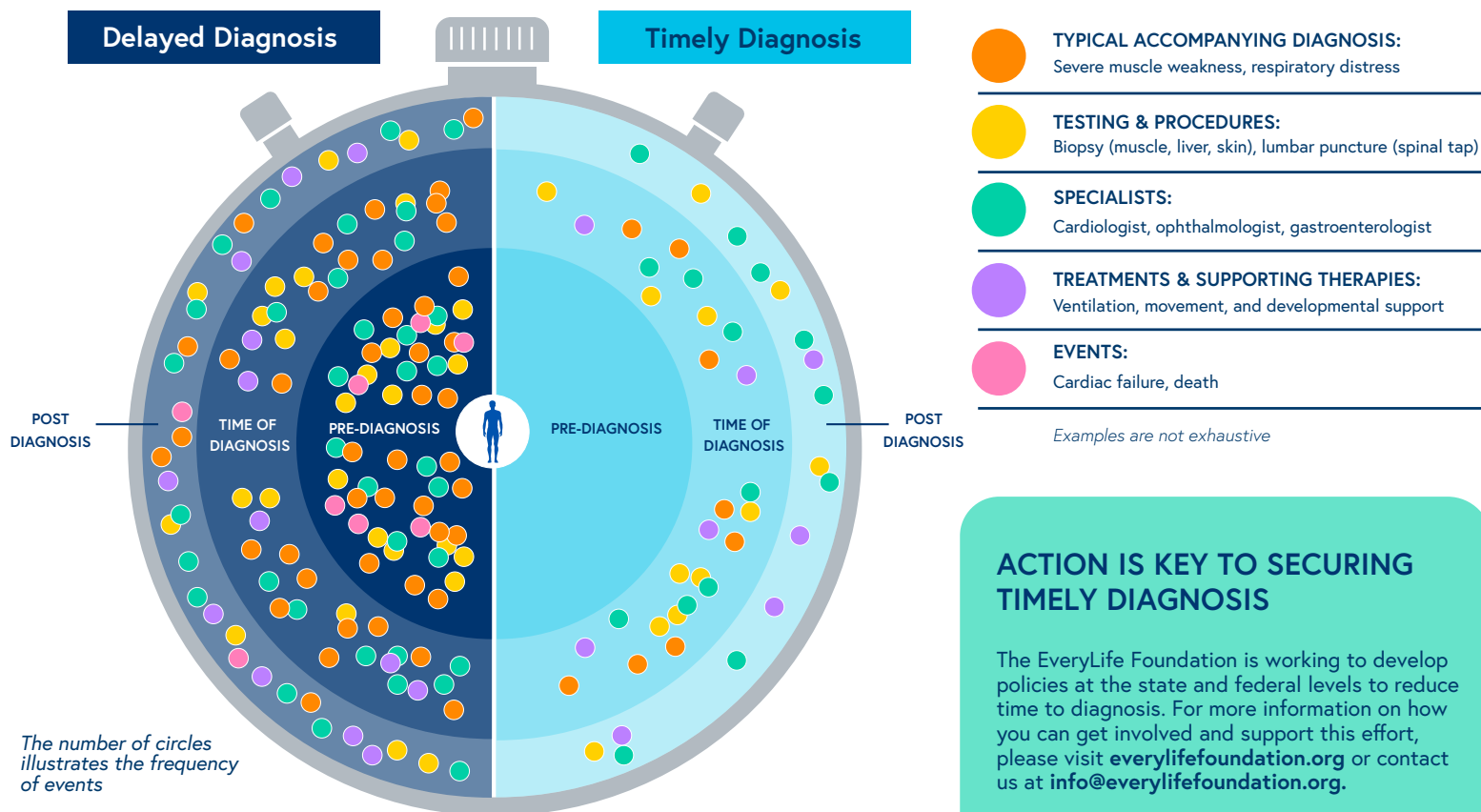


The Cost of Delayed Diagnosis in Rare Disease: *A Health Economic Study*

The Impact of Delayed Diagnosis in Pompe

Navigating a rare disease diagnosis can require more than 6 years and 17 medical interventions, on average, after symptoms begin, including emergency room visits and out-of-state specialist appointments.¹ The EveryLife Foundation and our partner the Lewin Group took an in-depth look into Pompe disease, a rare neuromuscular condition, to illustrate the significant economic impact of this diagnostic odyssey.

Every Minute Matters



ACTION IS KEY TO SECURING TIMELY DIAGNOSIS

The EveryLife Foundation is working to develop policies at the state and federal levels to reduce time to diagnosis. For more information on how you can get involved and support this effort, please visit everylifefoundation.org or contact us at info@everylifefoundation.org.

DELAYED DIAGNOSIS DRAMATICALLY INCREASES COSTS FOR FAMILIES*

\$219,081 Total medical cost and productivity loss per family

9+ Lost days of work for caregivers each year

41 Additional hospital visits, physician appointments, and other medical interventions

**Across the diagnostic odyssey*

How Did We Collect These Data?

- Medicare and Commercial insurance claims data, including patients with Pompe
- Results from The Rare Disease Financial and Social Impact Survey, completed by 1,409 community members

BENEFITS OF TIMELY DIAGNOSIS

Timely diagnosis, defined as before age one for children with Pompe,² can improve health outcomes by:

- Providing earlier access to supportive therapies and treatment
- Delaying disease complications and physical disabilities
- Lowering costs for patients and families by reducing or eliminating costly and unnecessary services or procedures

In states where newborn screening has been implemented for Pompe, timely diagnosis eliminates the diagnostic odyssey, resulting in no costs incurred before the year of diagnosis.

References:

1. Yang G, Cintina I, Zhou M, et al. The National Economic Burden of Rare Disease Study. The EveryLife Foundation for Rare Diseases. https://everylifefoundation.org/wp-content/uploads/2021/02/The_National_Economic_Burden_of_Rare_Disease_Study_Summary_Report_February_2021.pdf. Published February 25, 2021.
2. Infantile-onset Pompe disease: Diagnosis and management. Arch Argent Pediatr. 2019;117(4). doi:10.5546/aap.2019.eng.271