

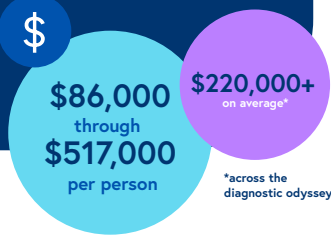
# The Cost of Delayed Diagnosis in Rare Disease:

*A Health Economic Study*

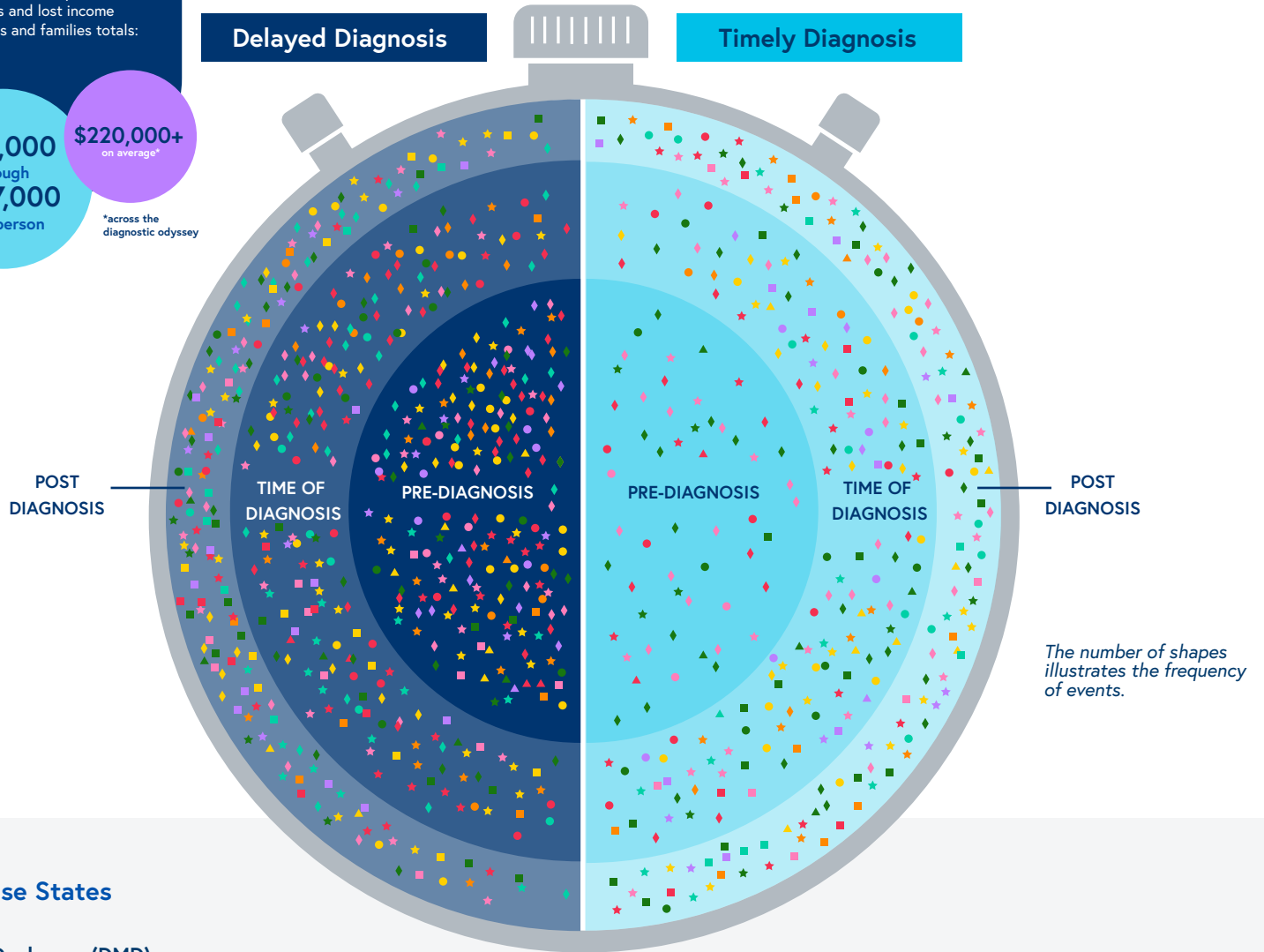
# The Impact of Delayed Diagnosis in Rare Disease

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.<sup>1</sup>

Our in-depth look at the diagnostic odyssey across seven rare diseases found the economic impact of medical costs and lost income for individuals and families totals:



## Every Minute Matters



### Disease States

- Duchenne (DMD)
- Pompe
- Adrenoleukodystrophy (ALD)
- Severe Combined Immunodeficiency (SCID)
- Fragile X Syndrome (FXS)
- Wilson Disease (WD)
- Generalized Myasthenia Gravis (gMG)

### Journey Map Categories

- ◆ **Typical Accompanying Diagnosis:**  
Anemia, liver failure (Wilson), cognitive impairment, seizures (ALD), Gait issues (Duchenne)
  - **Testing & Procedures:**  
Cognitive deficit testing (Fragile X), muscle biopsy (gMG)
  - ▲ **Events:**  
Cardiac Failure (Pompe), Hospitalization, Death (multiple diseases)
  - ★ **Specialists:**  
Palliative care, total care team (ALD), orthopedist (gMG)
  - **Treatments & Supporting Therapies:**  
Auditory therapy, feeding tube (SCID)
- Examples are not exhaustive*

# DELAYED DIAGNOSIS DRAMATICALLY INCREASES COSTS FOR FAMILIES

This study shows the positive economic impact of newborn screening and early diagnosis, particularly for diseases in which early intervention is key to optimizing long-term health outcomes.

An average **5-year delay in diagnosis** resulted in

**4x**

more likelihood of seeing **3+ specialists**

Delayed diagnosis shifts healthcare spending away from treatment and supportive therapies to unnecessary procedures

Cost of the diagnostic odyssey can be eliminated for diseases like SCID, ALD, and Pompe where newborn screening is routine in many states.

## How Did We Collect These Data?

- Medicare and Commercial insurance claims data, including patients with Duchenne, Pompe, ALD, SCID, Fragile X, Wilson, and gMG
- Results from The Rare Disease Financial and Social Impact Survey, completed by 1,409 community members

## BENEFITS OF TIMELY DIAGNOSIS

Timely diagnosis improves health outcomes by:



Providing earlier access to supportive therapies and treatment



Reducing or eliminating expensive and unnecessary services, tests, and treatments



Preventing deaths and delaying disease complications and physical disabilities



Enabling opportunity to evaluate future family planning based on diagnosis

## 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](https://everylifefoundation.org) or contact us at [info@everylifefoundation.org](mailto:info@everylifefoundation.org).

### Reference:

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](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.