Early Diagnosis: The Importance of State Newborn Screening Programs

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TEXAS HILL COUNTRY SUMMIT
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WOMEN IN GOVERNMENT

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

Economic Impact of Timely Diagnosis in Three Rare Diseases

Women In Government
November 3rd, 2023

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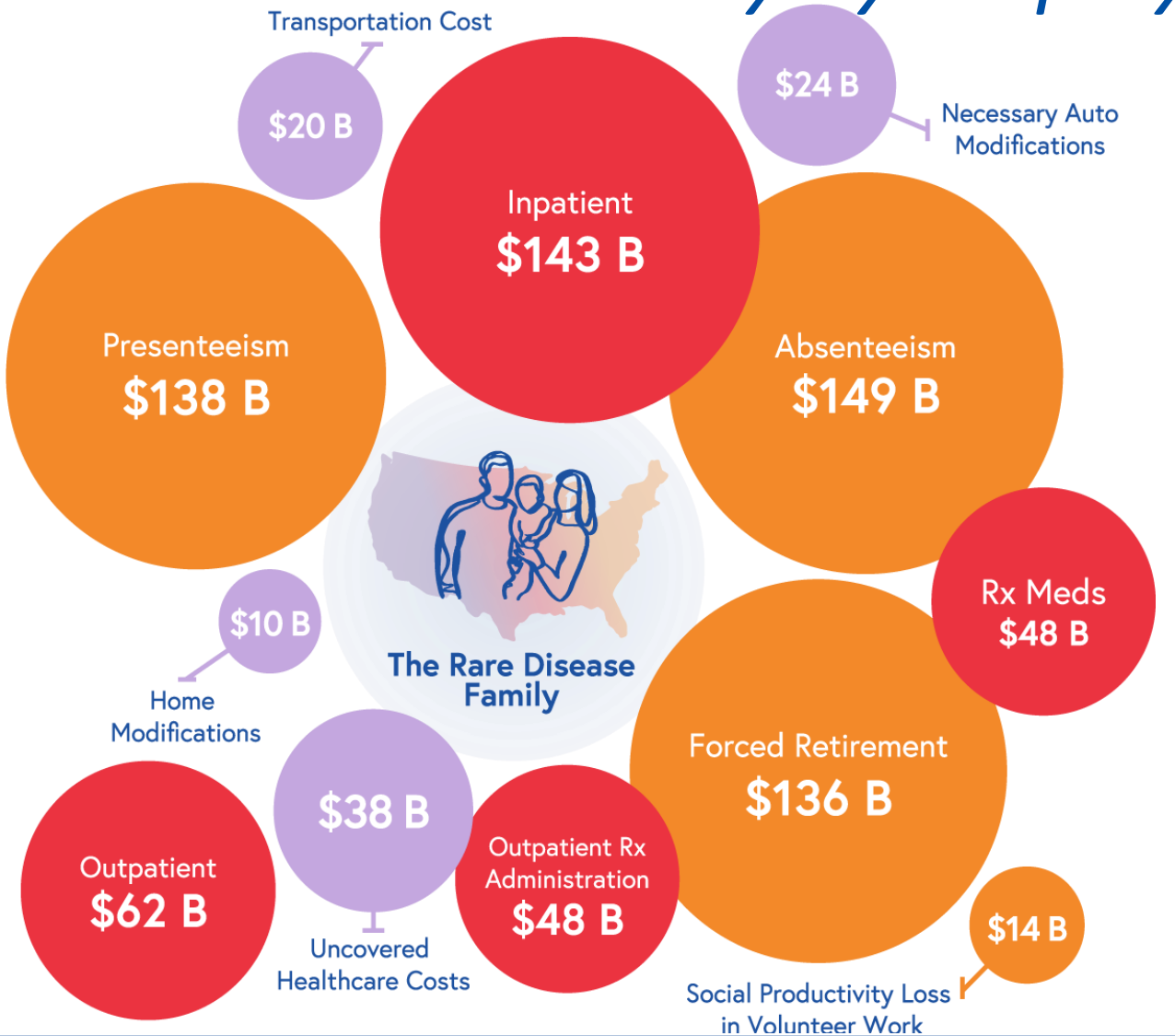
Public Health Urgency of Rare Disease

60% of Costs Shouldered Directly by Employers & Families

$437 Billion
INDIRECT COSTS: PRODUCTIVITY LOSS

$418 Billion
DIRECT MEDICAL COSTS

$111 Billion
NON-MEDICAL & UNCOVERED HEALTHCARE COSTS



Total Economic Burden of Rare Disease in the U.S in 2019:

$966 Billion

Estimated prevalence of 379 RDs

15.5 million

The National Economic Burden of Rare Disease Study

Conservative Estimate of Economic Burden Based on 379 of 10,000 RDs

Study Results:

RD Impact Survey Captures Medical Burden, Long Diagnostic Odyssey

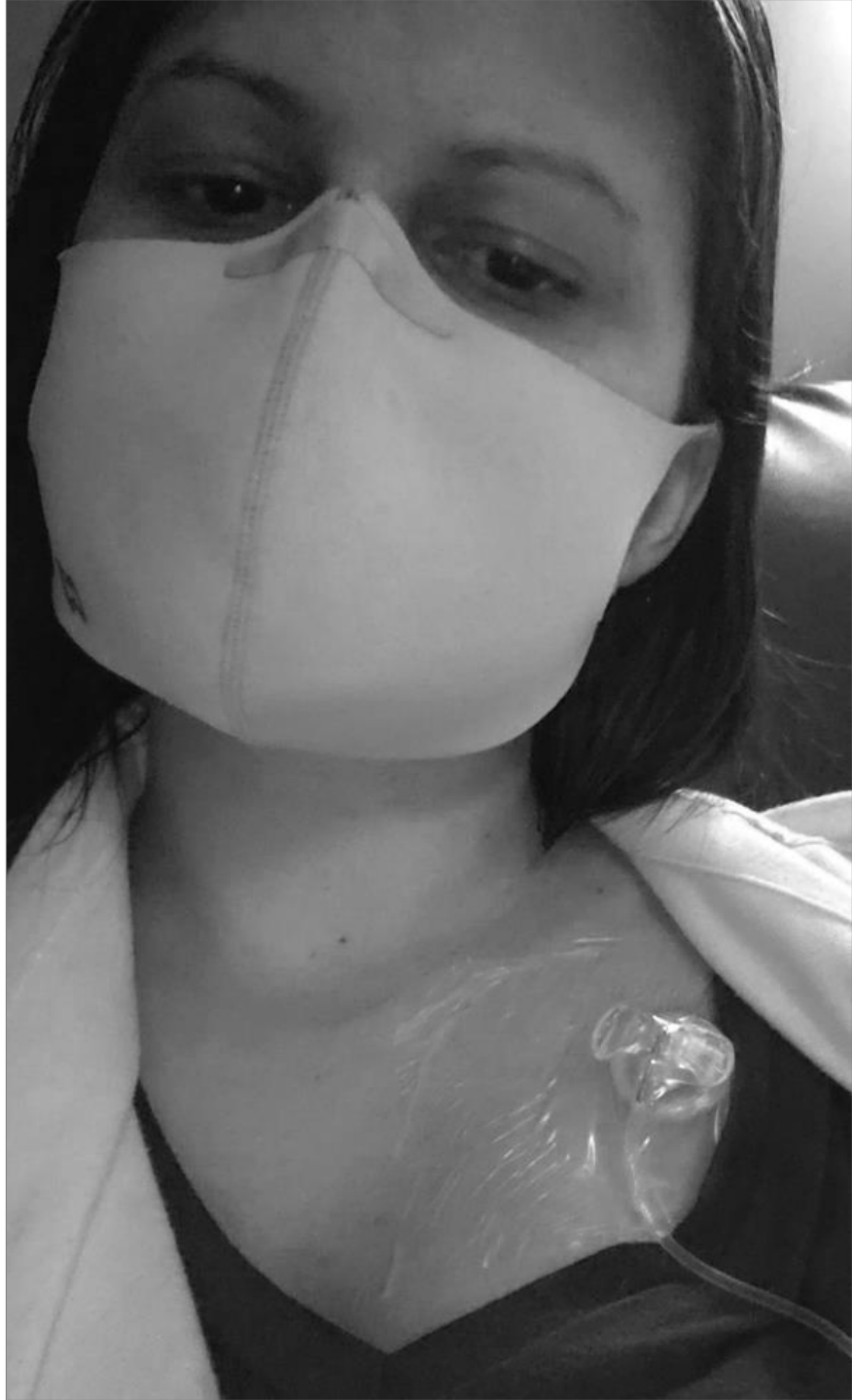
16.5 YEARS
Since first RD symptom (mean)

10.2 YEARS
Years since RD diagnosis (mean)

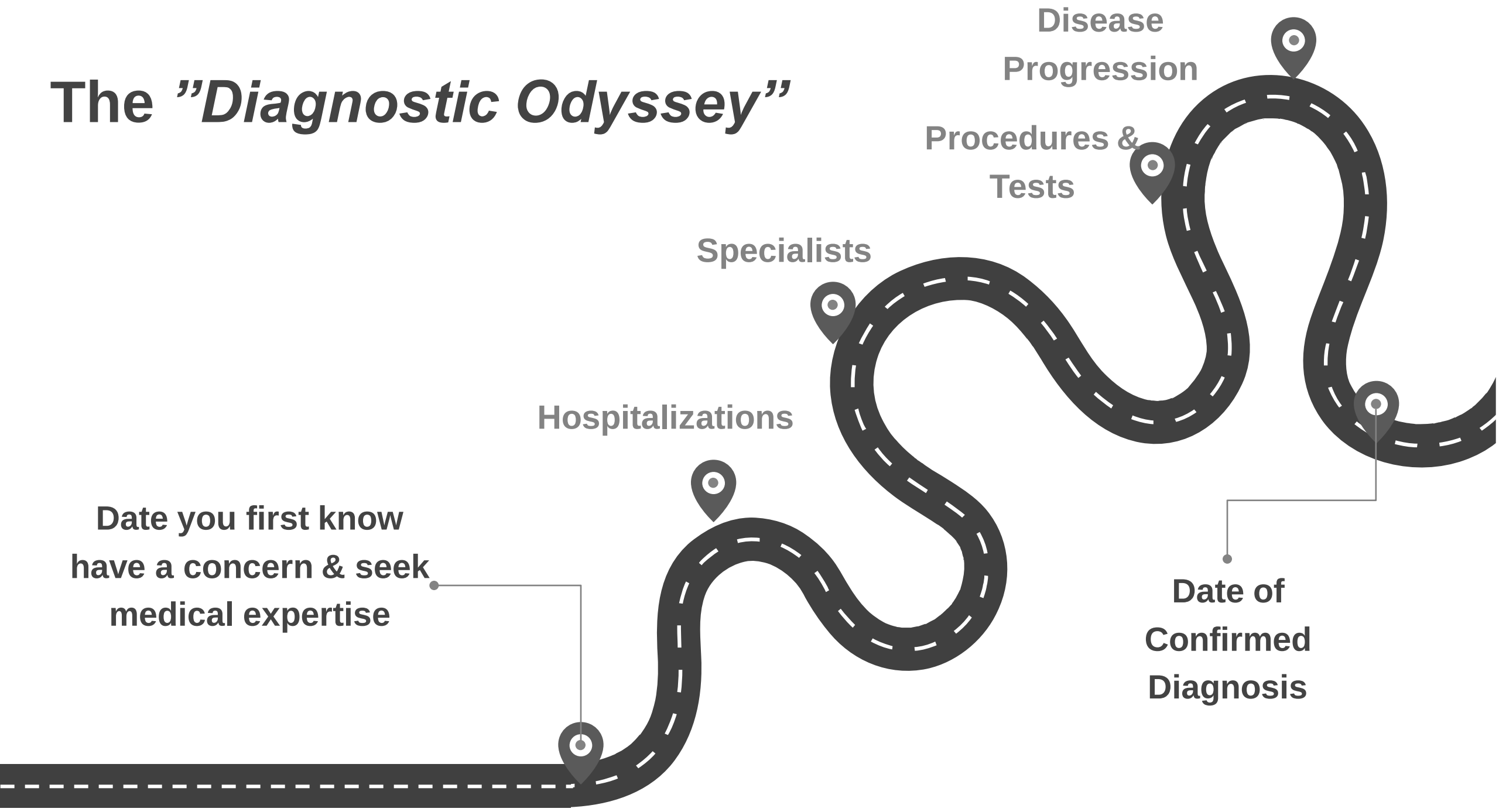
6.3 YEARS
Navigating without RD diagnosis (mean)

16.9
Average number of specialists seen since first RD symptom

Based on final analysis sample of 1,360 completed responses



The *"Diagnostic Odyssey"*



What We Know About the Diagnostic Odyssey --

- The rare disease diagnostic odyssey results in delayed or missed diagnoses.
- Delays in accurate diagnosis can yield inappropriate clinical management, unnecessary medical interventions, and missed treatment windows for therapeutic interventions to optimize health outcomes or prevent disease symptom onset.
- The diagnostic odyssey may contribute to increased probability of irreversible disease progression and complications, **ultimately putting patients at risk for preventable mortality and disability.**

We Also Know --

- **We have the technology to do better.**
- Advancements in technologies to screen, diagnose, treat, and manage rare disease have the potential to dramatically shorten — and even eliminate — the diagnostic odyssey.

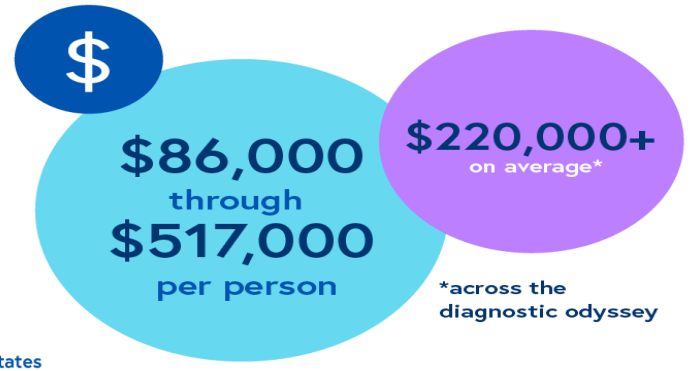
Study Overview

- We selected **7 rare diseases** as sampling, intended to establish methodology – **SCID, Pompe, ALD**, Fragile X, Duchenne, gMG, Wilson Disease
- Analysis was informed by detailed **patient journey maps** based on literature review & consultation with **disease-specific experts**
- A comprehensive literature review and KOL engagement informed disease epidemiology, disease milestones and progression, testing and procedures that can be part of the diagnostic odyssey, and economic impact.
- 27 experts served as a part of the **technical advisory panel,** representing the research, clinical, and patient advocacy communities.
- **Productivity loss methodology** was developed

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.[1]

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:



Disease States

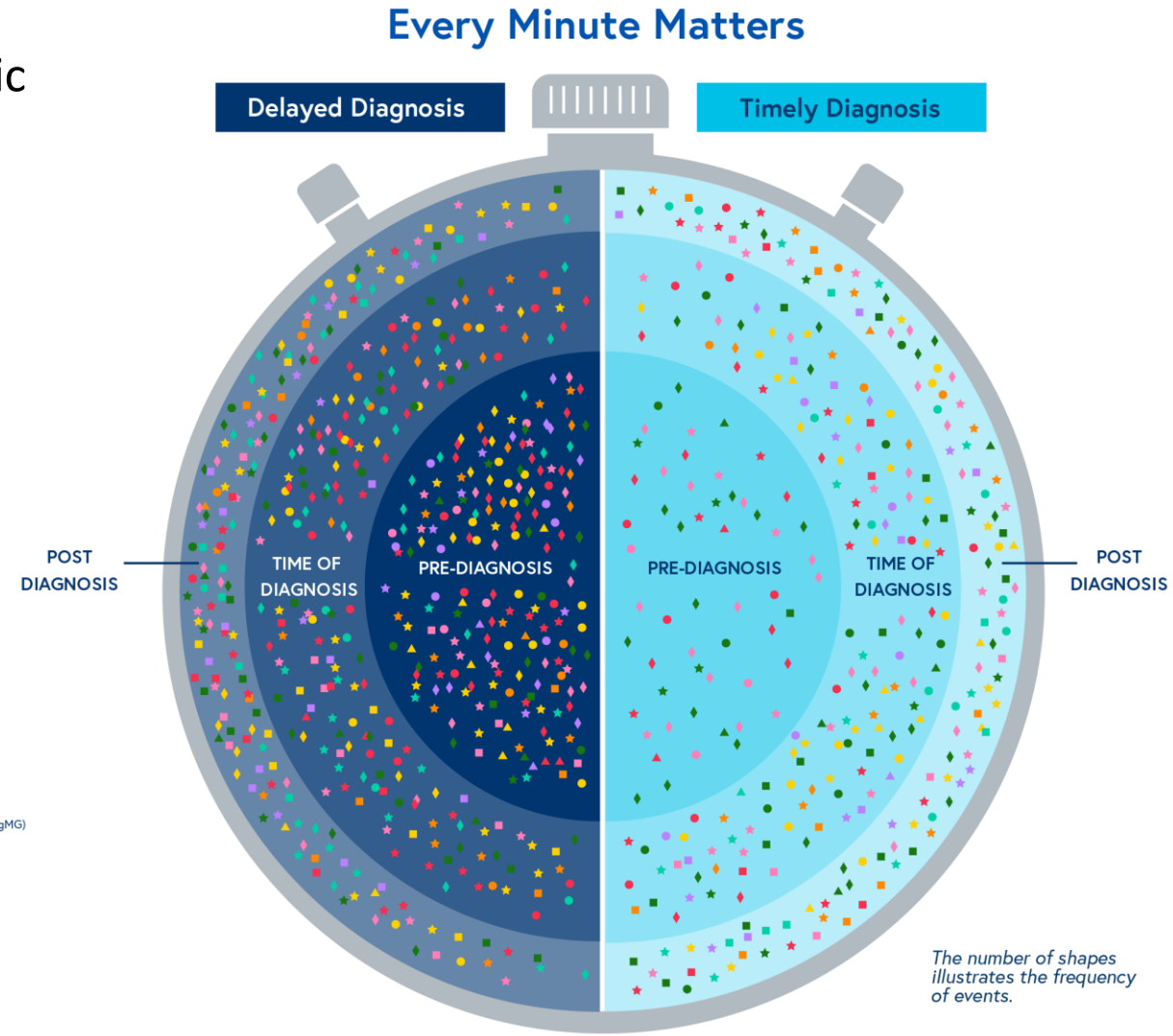
- Duchenne (DMD)
- Pompe
- Adrenoleukodystrophy (ALD)
- Severe Combined Immunodeficiency (SCID)
- Fragile X Syndrome (FXS)
- Wilson Disease (WD)
- Generalized Myasthenia Gravis (gMG)

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

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.[1]

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** or contact us at **info@everylifefoundation.org.**

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

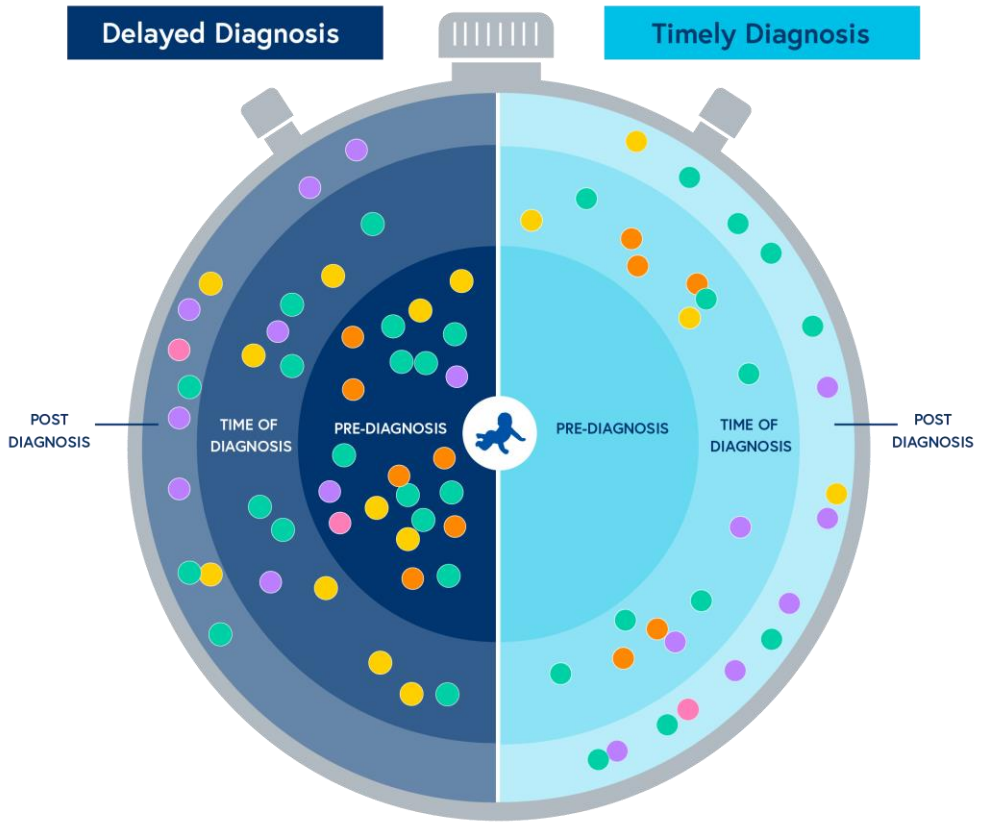
Reference:

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. Published February 25, 2021.

The Impact of Delayed Diagnosis in Duchenne

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] We took a detailed look into Duchenne muscular dystrophy, a rare neuromuscular condition, to illustrate the significant economic impact of this diagnostic odyssey.

Every Minute Matters



TYPICAL ACCOMPANYING DIAGNOSIS:
Gait issues and muscle weakness

TESTING & PROCEDURES:
Genetic testing, liver biopsy

SPECIALISTS:
Orthopedist, Hepatologist, Gatroenterologist

TREATMENTS & SUPPORTING THERAPIES:
Ankle-foot orthotics (AFO)

EVENTS:
Unexpected hospitalizations

Examples are not exhaustive

ACTION IS KEY TO SECURING TIMELY DIAGNOSIS

The EveryLife Foundation and leading partners, including Parent Project Muscular Dystrophy (PPMD), are working to develop state and federal policies to eliminate the diagnostic odyssey in Duchenne. For more information on how you can get involved and support this effort, please visit **everylifefoundation.org** and **parentprojectmd.org.**

DELAYED DIAGNOSIS DRAMATICALLY INCREASES COSTS FOR FAMILIES*

$211,229 Total medical cost and productivity loss per family

20 Lost days of work for caregivers each year

17 Out-of-state trips for medical care across the 7 communities studied

*Across the diagnostic odyssey

How Did We Collect These Data?
- Medicare and Commercial insurance claims data, including patients with Duchenne
- 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 five years old for children with Duchenne[2,3], 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

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

Parent Project Muscular Dystrophy

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. Published February 25, 2021. 2. Ciafaloni E, Fox D, Pandya S. Muscular Dystrophy Surveillance, Tracking, and Research Network (MD STARnet). J Pediatr. 2009;155(3):380-385. 3. Families Continue to Experience Delay in Getting a Diagnosis of Duchenne Muscular Dystrophy. National Center on Birth Defects and Developmental Disabilities, Centers for Disease Control and Prevention; 2023. https://www.cdc.gov/ncbddd/musculardystrophy/features/delay-in-diagnosis-of-duchenne-muscular-dystrophy.html

Implications

Avoidable costs attributable to delayed diagnosis represent:

- Economic burden on patients and families searching for a diagnosis
- Healthcare dollars that could have been better spent on treatment and supportive therapies that improve patient quality of life and may even increase workforce productivity

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Implications

Timely diagnosis

- Provides opportunity for optimal disease management and timely treatment initiation for patients
- Prevents prolonged diagnostic odyssey and prevents both avoidable medical costs and intangible costs such as time spent managing financials, time off work for health care visits, and in many cases, out-of-state trips to see specialists for diagnosis and treatments

Newborn screening makes timely, presymptomatic diagnosis possible with profound benefits to affected newborns and families

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

- Challenge of ICD codes and variable subtypes
- Majority of rare diseases have significantly longer diagnostic odysseys; findings are extremely conservative
- Productivity loss methodology extremely conservative

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Study Supporters

Rare Disease Innovations Institute

Newborn Screening in Rare Disease

Tara J. Britt
President & Founder
Rare Disease Innovations Institute, Inc.
https://rdii.org/

Newborn Screening Testing Saves Lives

- About 4 million babies are born and their families benefit from newborn screening through early detection of disease and the delivery of life saving treatments. 12,000 of these babies will be identified with a newborn screening condition each year.
- Newborn screening is widely accepted as one of the largest and most successful disease prevention programs in the history of the US. CDC recognized NBS as 1 of the 10 most important public health achievements of the century.
- Currently 33 states do not screen for all of the federally recommended conditions leaving hundreds of babies undetected without timely treatment each year that often contributes to the early death of those babies.
- RUSP: list of disorders that the Secretary of the Department of Health and Human Services (HHS) recommends for states to screen as part of their state universal newborn screening (NBS) programs.

Recommended Uniform Screening Panel (RUSP)

- The RUSP is a list of disorders that the Secretary of the Department of Health and Human Services (HHS) recommends for states to screen as part of their state universal newborn screening (NBS) programs.
- Disorders on the RUSP are chosen based on evidence that supports the potential net benefit of screening, the ability of states to screen for the disorder, and the availability of effective treatments. It is recommended that every newborn be screened for all disorders on the RUSP.
- Most states screen for the majority of disorders on the RUSP; newer conditions are still in process of adoption. Some states also screen for additional disorders.
- Although states ultimately determine what disorders their NBS program will screen for, the RUSP establishes a standardized list of disorders that have been supported by the Advisory Committee on Heritable Disorders in Newborns and Children and recommended by the Secretary of HHS.

NBS North Carolina Case Study

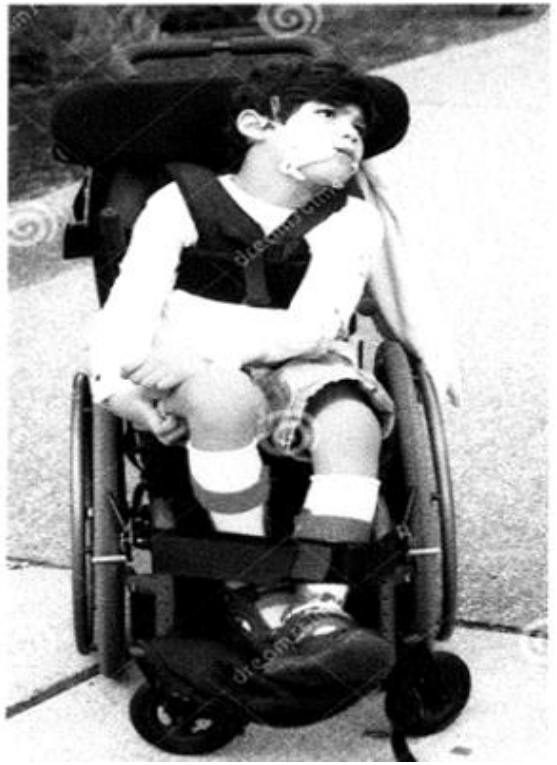
- In 2017-2018 4 indications were added to North Carolina's NBS panel through legislation.
- Diseases added for testing were: Pompe, MPS 1, X-ALD..
- If these diseases are not caught through NBS or in early life, these are deadly diseases and patients live a very poor quality of life. This affects not only the patients but the family and caregivers.
- Diseases not caught through NBS are not only deadly for the patient but the economic impact to the state is far greater than the cost of testing in most all cases.
- NC is home to Centers of Excellence for treatment of Pompe, MPS 1, X-ALD.

North Carolina Case Study cont'd

- Early diagnosis of these conditions through NBS and treatment with bone marrow transplant or enzyme replacement therapy or both prevents death or lifelong disability
- Pompe early diagnosis and treatment results in 100% survival but untreated MPS 1 or X-ALD leads to permanent lifelong disability or death in childhood.
- During NC's set up of the state lab to accommodate these NBS tests, COVID hit and the testing of these diseases took much longer to implement.
- While we don't know the exact numbers; deaths & lifelong disability were certain during this time period of not being tested.
- New legislation introduced in 2021 to limit the time NC's state lab has to test for any new indications added to the NC NBS panel.
- Legislation mandates testing to take place within 3 years of ratified legislation.

Early Diagnosis

Late Diagnosis



Samuel (left) was diagnosed with ALD at age 5 and received early treatment. Jonathan (right) suffered severe brain damage before he was diagnosed with ALD at age 7.

Adrenoleukodystrophy, or ALD, is a deadly genetic disease that affects 1 in 18,000 people. It most severely affects boys and men. This brain disorder destroys myelin, the protective sheath that surrounds the brain's neurons, the nerve cells that allow us to think and to control our muscles. In 2018 the Haley Hayes Newborn Screening bill was submitted and passed into law to add three new diseases to North Carolina's newborn screening panel,(Pompei, MPS-1 and X-ALD). The Council submitted a white paper on the importance of early intervention and newborn screening to the Deputy Secretary of NC Department of Health and Human Services. This document was instrumental in the passage of newborn screening legislation. Early detection and intervention can change, and even save a life.

Newborn Screening Saves Lives!!!

Rare Disease Innovations Institute

THANK YOU!

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