Rare Disease Overview

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EveryLife Foundation for Rare Diseases

The EveryLife Foundation for Rare Diseases is a 501(c)(3) nonprofit, nonpartisan organization dedicated to empowering the rare disease patient community to advocate for impactful, science-driven legislation and policy that advances the equitable development of and access to lifesaving diagnoses, treatments and cures.

“We do not speak for patients. We provide the training, education, resources and opportunities to make their voices heard. By activating the patient advocate, we can change public policy and save lives.”

-Julia Jenkins, Executive Director

U.S. Rare Disease Prevalence Estimates

7,000+
rare diseases

~30
million
affected

- In the U.S., a disease is considered rare when it affects fewer than 200,000 people
- Researchers estimate there are more than 7,000 RDs
- RDs affect an estimated 30 million Americans
- Actual RD prevalence could exceed this estimate
- 93-95% of RD have no approved treatments

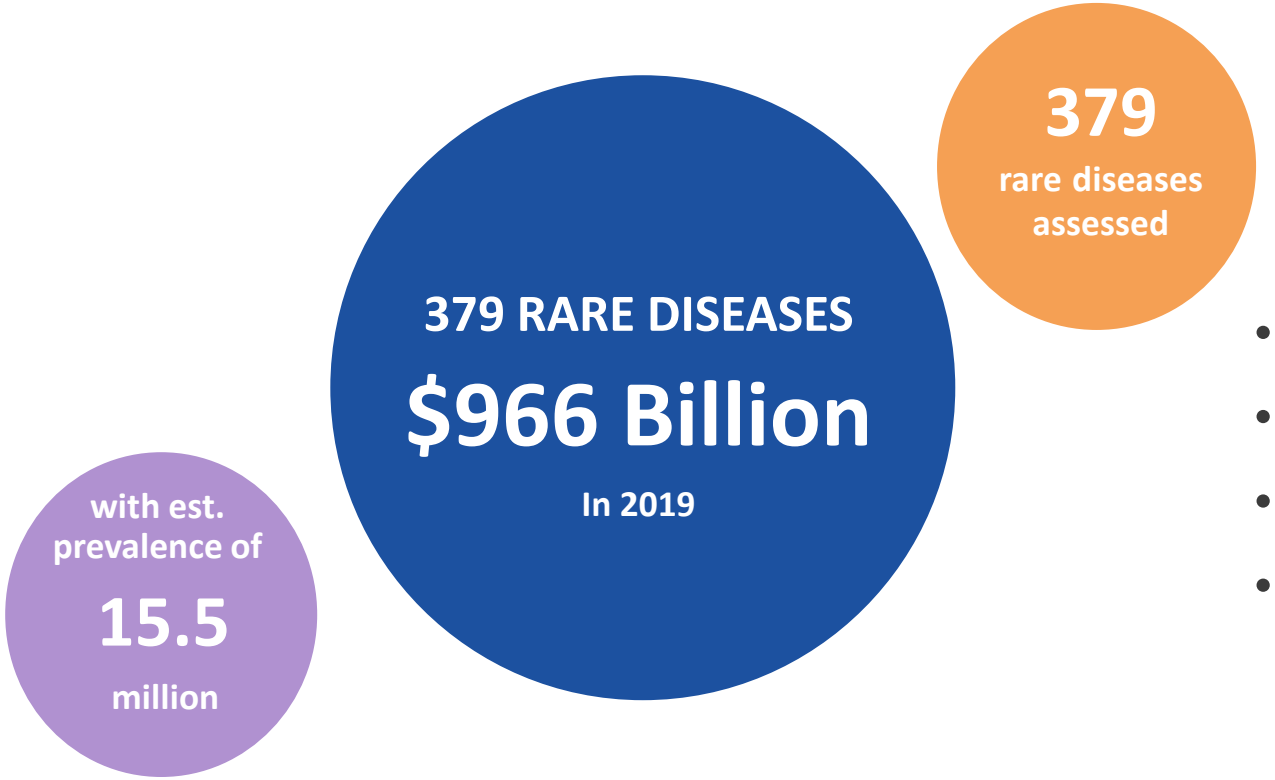
The National Economic Burden *of* Rare Disease Study

The National Economic Burden *of* Rare Disease Study

EVERYLIFE FOUNDATION FOR RARE DISEASES

LEWINGROUP®
Part of OptumServe

Study Results:
Total Economic Burden of 379 RDs Was Nearly **$1 Trillion** in 2019



- The study assessed 379 out of 7,000 RDs
- Estimated prevalence of 379 RDs = 15.5 million
- Economic burden is not generalizable to all RDs
- Further research is needed to understand prevalence and economic burden of *all* RDs

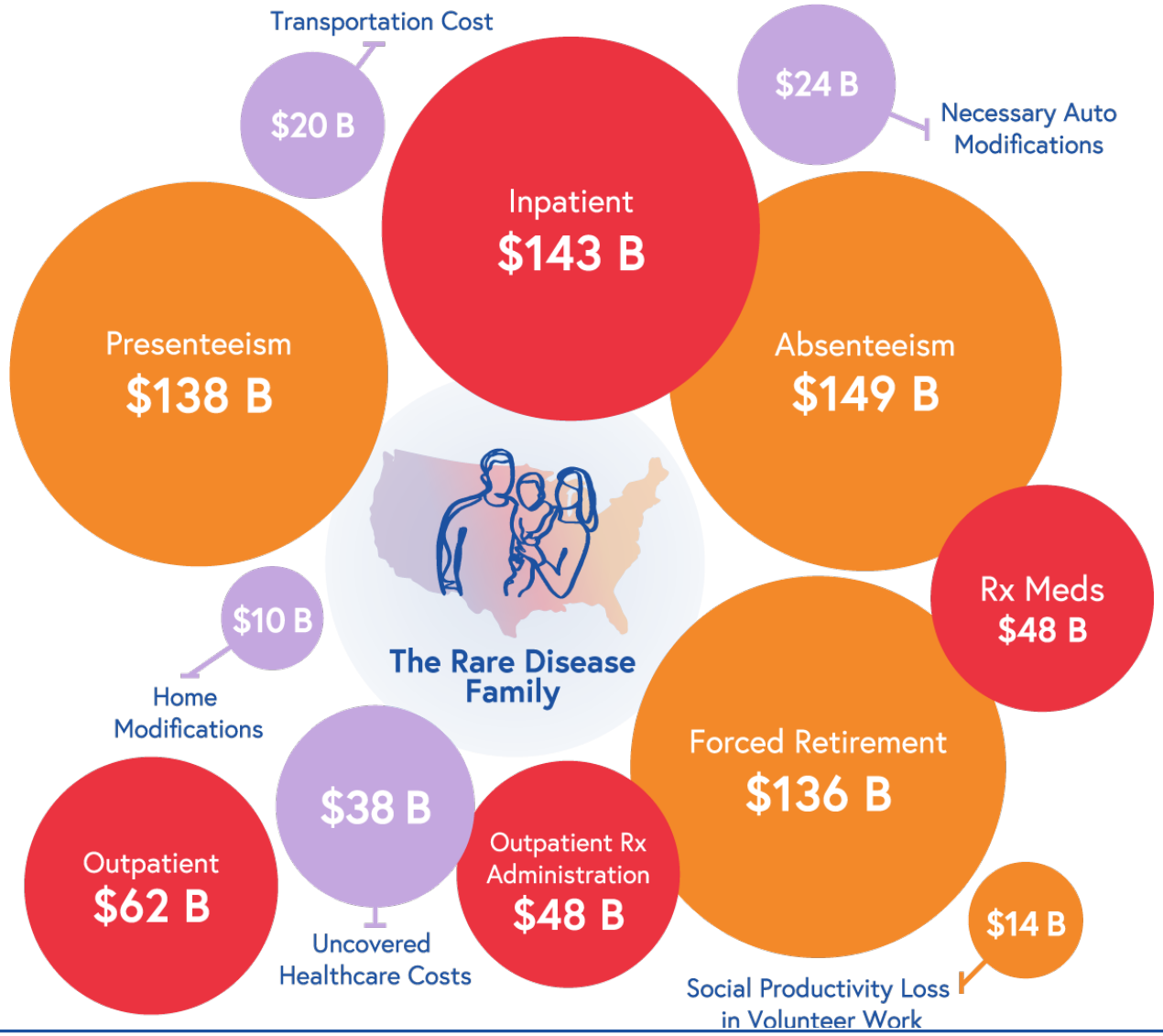
Conservative Estimate of Economic Burden Based on 379 of 7,000 RDs

What is the Impact on the Average Rare Disease Family?

$418 Billion
DIRECT MEDICAL COSTS

$437 Billion
INDIRECT COSTS: PRODUCTIVITY LOSS

$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

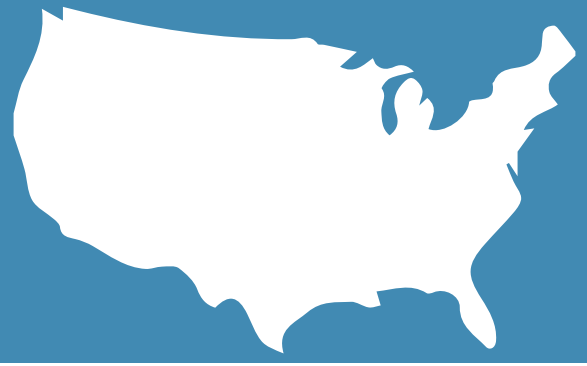
15.5 million

The National Economic Burden *of* Rare Disease Study

Rare Diseases Are Hard to Diagnose

7.6 years

in the United States to obtain a diagnosis for a rare disease



40%

of patients receive a misdiagnosis more than once

7.3

Physicians seen before receiving the proper diagnosis

70%

of physicians believe it would be helpful to receive additional training in rare diseases

- http://www.fromhopetocures.org/fighting-rare-diseases
- https://www.raregenomics.org/rare-disease-facts Accessed September 2019.
- https://www.who.int/medicines/areas/priority_medicines/Ch6_19Rare.pdf Accessed September 2019.

Rare Disease Challenges to Optimizing Diagnosis and Treatment

- Clinical Research: small numbers, dispersed populations, uncertainty around outcomes
- Natural History Knowledge: cause of disease, pathophysiology, natural course of disease, epidemiological data all limited in many cases
- Medical expertise is limited and not available in many areas of the country
- Limited classification systems (many rare diseases have no ICD Codes for example)

What Types of Access Issues do Rare Disease Patients Face?
EveryLife Foundation Community Congress Access Subgroup Topics

Subgroup 1: Barriers to Accessing Out of State Care

- **Addressing restrictions in out of state care** for Medicaid patients
- Reducing **complex documentation and agreement requirements** with out of state rare disease experts
- Assessing extent of challenges among commercially insured or among ACA plans

Subgroup 2: Rare Disease Expert Representation & Engagement in Medicaid DUR & P & T Processes

- Patients & expert providers are **under-represented** in Medicaid DUR/P&T process
- Coverage policies may **not be public**

Subgroup 3: Addressing ERISA/Self-Funded Plans' Ability to Laser Out Treatments for Specific Diseases & Identify Employees or Their Families With Rare Diseases

- Understand and address the extent of challenges rare disease patients or employees with insured children with rare diseases face in ERISA/self-funded plans to guide solutions.
- Prevent workplace discrimination based on perceived costs associated with rare diseases.

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Subgroup 4: Use of Value Assessments and Implementation of Value Based/Alternative Payment Arrangements

- Ensure that the outcomes used to develop value assessment frameworks and value based/alternative payment arrangements are inclusive of outcomes that are meaningful to patients
- Need to advocate for value based/alternative payment arrangements to reflect the unique considerations for rare disease therapies

Subgroup 5: Access to Affordable Genetic Testing Services

- Coverage for recommended genetic testing, especially whole genome and whole exome testing, is lacking and requirements for obtaining coverage are lengthy and complex, even for panels in many cases
- Genetic testing as a diagnostic tool is often viewed as an experimental method which creates wide gaps in coverage across the states.

Subgroup 6: Inappropriate Utilization Management Practices

- Address the lengthy timelines rare disease patients face in the prior auth and appeals processes that result in negative health outcomes
- Fix processes that create unnecessary hurdles, streamline and simplify
- Ensure the appropriate rare disease expert and patient engagement in utilization review processes and decision making

Ad-hoc Subgroup: Addressing Threats to the Accelerated Approval Pathway

- Address recent policy threats to the viability of the accelerated approval pathway
- Address broad misunderstandings of what accelerated approval is, its impact and its cost

Rare Disease Research

CDMRP

NIH National Center for Advancing Translational Sciences

RARE DISEASES CLINICAL RESEARCH NETWORK

Currently Funded Research Programs

- Alcohol and Substance Abuse Disorders
- Amyotrophic Lateral Sclerosis
- Autism
- Bone Marrow Failure
- Breast Cancer
- Chronic Pain Management
- Combat Readiness-Medical
- Duchenne Muscular Dystrophy
- Epilepsy
- Gulf War Illness
- Hearing Restoration
- Joint Warfighter Medical
- Kidney Cancer
- Lung Cancer
- Lupus
- Melanoma
- Military Burn
- Multiple Sclerosis
- Neurofibromatosis
- Neurotoxin Exposure Treatment Parkinson's
- Orthotics and Prosthetics Outcomes
- Ovarian Cancer
- Pancreatic Cancer
- Peer Reviewed Alzheimer's
- Peer Reviewed Cancer
- Peer Reviewed Medical
- Peer Reviewed Orthopaedic
- Prostate Cancer
- Rare Cancers
- Reconstructive Transplant Research
- Scleroderma
- Spinal Cord Injury
- Tick-Borne Disease
- Traumatic Brain Injury and Psychological Health
- Tuberous Sclerosis Complex
- Vision

EveryLife Foundation Policy Goals

1. Close the innovation gap for the 93% of rare diseases that have no FDA-approved treatment

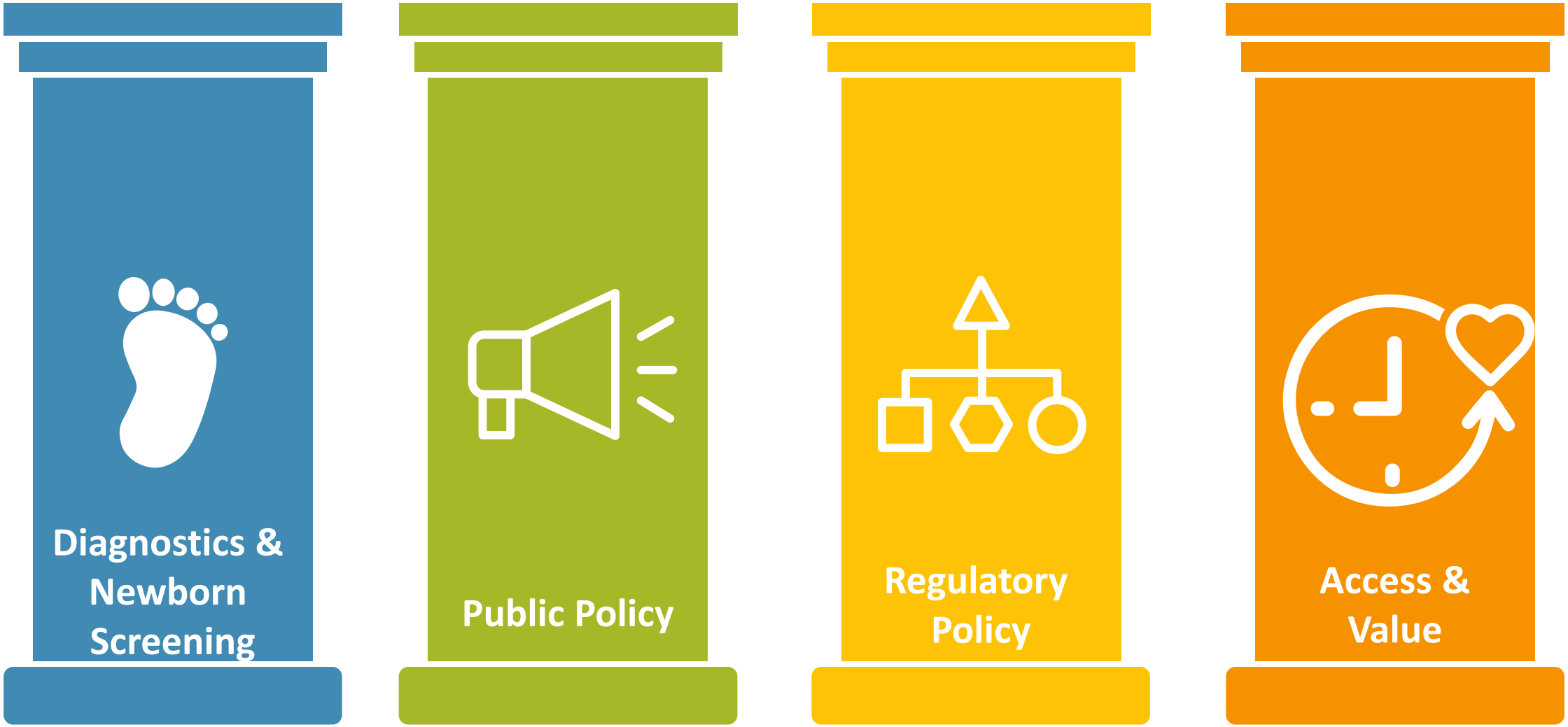
2. Eliminate the diagnostic odyssey for rare disease patients

3. Improve the regulatory process & advance regulatory science for rare disease therapies

4. Ensure patient access to safe & efficacious therapies and cures at the earliest moment possible

5. Empower patients to develop an impactful voice in policymaking, drug development, and regulatory decision-making

EVERYLIFE POLICY PILLARS



Rare Disease Caucus

180 total—27 Senators and 153 Representatives

2021 Caucus Briefings:

- Economic Cost of Rare Diseases (ELF and NCATS Studies)
- STAT Act
- Newborn Screening Study
- Appropriations/Undiagnosed Diseases
- Scientific Workshop case studies and COVID survey results

For 2021:

Regularly communicate rare disease community priorities, perspectives, and stories to caucus staffers

- Create, write, and share quarterly updates with the Caucus staffers including highlighting a patient story and relevant policy updates (create a new banner for the “newsletter effect”).
- Provide caucus staffers with a “Rare Disease Day/Week” communications toolkit that they can use to promote rare diseases at the beginning of February including a sample social media post, sample newsletter article on the caucus, etc.

RDLA
Rare Disease Legislative Advocates

Rare Disease Advocacy Opportunities

RARE ACROSS AMERICA
EVERY VOICE, IN EVERY DISTRICT, MATTERS

RARE DISEASE WEEK ON CAPITOL HILL

YARR · YOUNG ADULT REPRESENTATIVES OF RDLA

- **Rare Across America**
  - More than 600 advocates from 48 states, plus D.C and P.R met with their Members of Congress
  - Save the date for 2022!
- **Rare Disease Week on Capitol Hill**
  - Advocates from 49 states, plus D.C and P.R met with their Members of Congress last week
  - EveryLife Foundation also hosted a 2-day legislative conference, diversity roundtable, caucus briefing and more.
- **YARR, Young Adult Representatives of RDLA**
  - For advocates ages 16-30

RDLA
Rare Disease Legislative Advocates
POWERED BY THE EVERYLIFE FOUNDATION