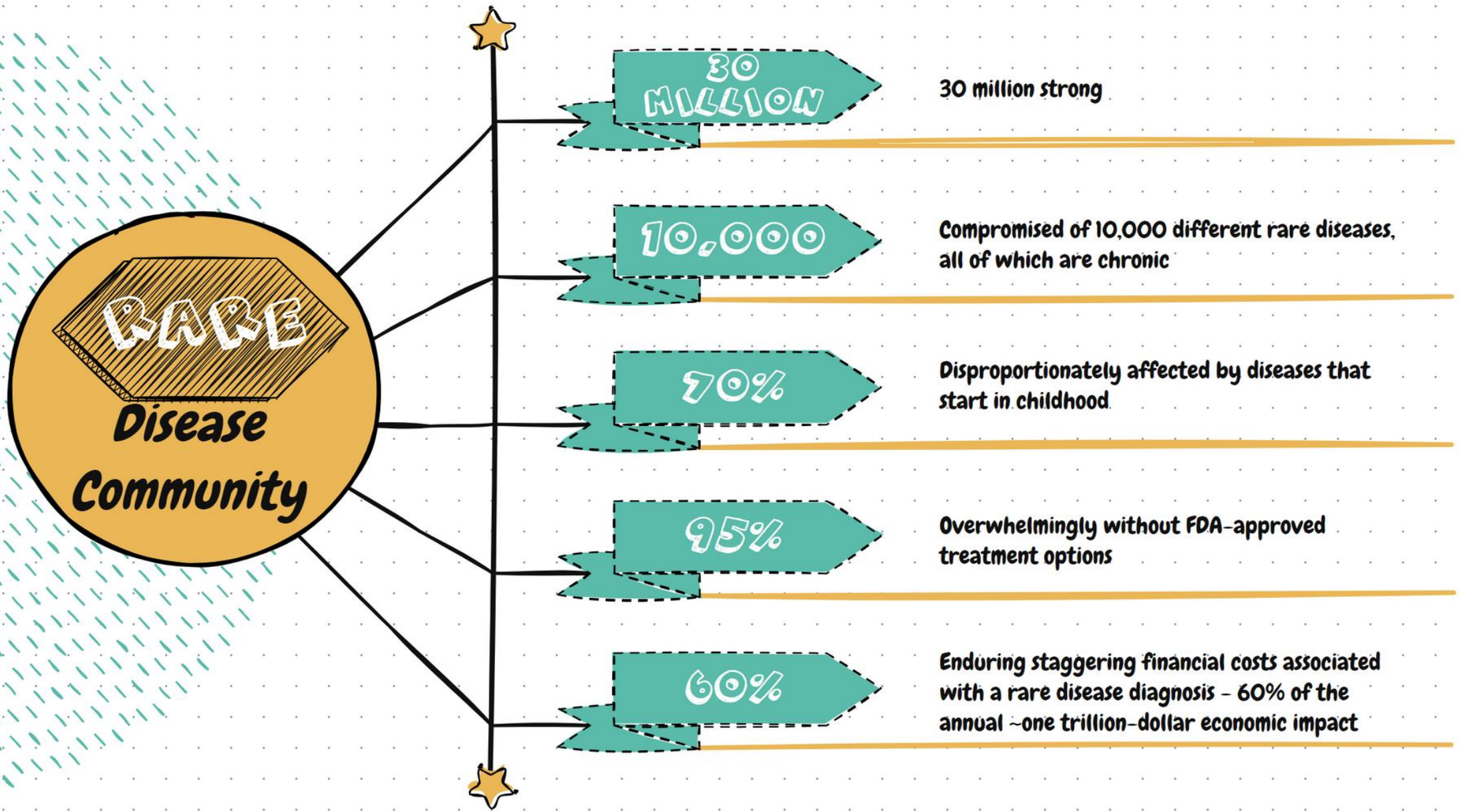


A large group of people, including patients and staff, are posing for a group photo in a grand, multi-story hall. The hall features high ceilings, large columns, and numerous flags hanging from the upper levels. The group is arranged in many rows, filling a large open space. Some individuals are seated in wheelchairs in the front rows. The overall atmosphere is one of a significant community gathering.

Stronger Together – Rare Disease Federal and State Policy & Advocacy

***Annie Kennedy
Chief of Policy, Advocacy & Patient Engagement
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.*

COMMUNITY CONGRESS

POWERED BY THE EVERYLIFE FOUNDATION



POWERED BY THE EVERYLIFE FOUNDATION

RARE
DISEASE WEEK
ON CAPITOL HILL



RARE
ACROSS AMERICA

EveryLife Policy Priority Areas

Everything we lead, support, monitor, and engage in maps to one or more of these 3 policy areas



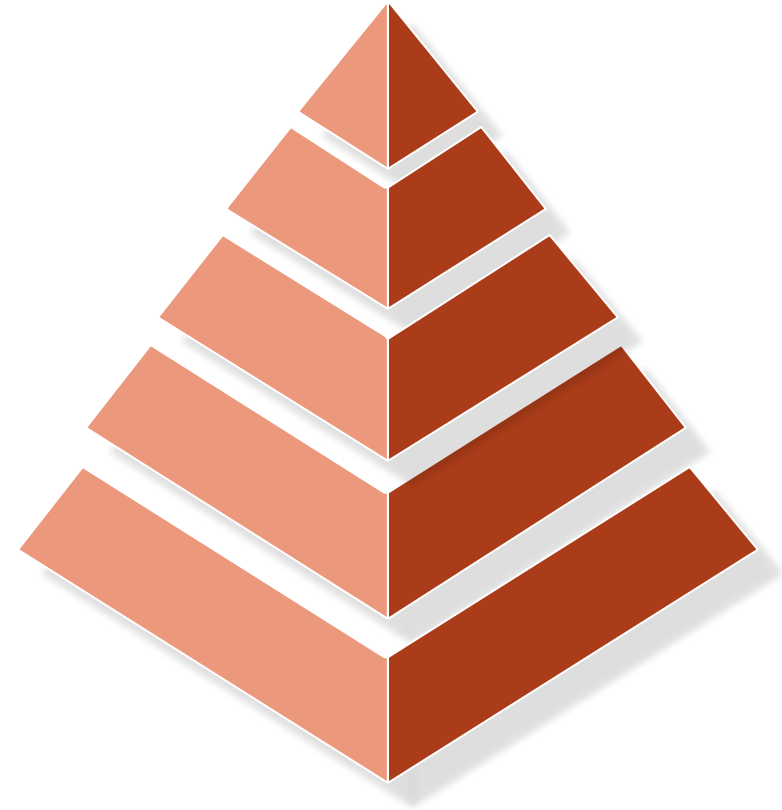
Elimination of Dx Odyssey

**Reduction & elimination
of the diagnostic odyssey.**



Therapy Development

**Development of expedient
pathways for discovery
and approval for effective
therapies for rare
diseases**



Access & Value

**Access to approved
therapies**

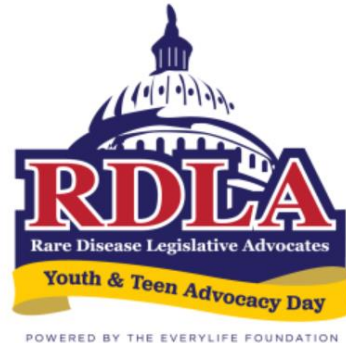
Rare Disease Legislative Advocates (RDLA) Programs

RARE
DISEASE WEEK
ON CAPITOL HILL




Rare
Advocacy
Learning
EMPOWERING EVERY VOICE

RARE
ACROSS AMERICA



EveryLife Policy Resources

www.everylifefoundation.org



ABOUT ADVOCACY POLICY PROGRAMS EVENTS RESOURCES N

DIAGNOSTICS

INNOVATION

REGULATORY

ACCESS

COMMUNITY CONGRESS

PUBLICATIONS/COMMENTS

ECONOMIC BURDEN

POLICY IMPACT

TAKE ACTION

ACCELERATED APPROVAL


CO-PAY ACCUMULATORS

DRUG PRICING

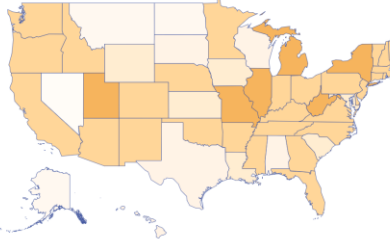
EXPERT CARE

ICD CODE ROADMAP

VALUE ASSESSMENTS



SHARE THE WHITE PAPER SHARE YOUR STORY SUBMIT A LETTER TO THE EDITOR REACH OUT TO LOCAL MEDIA



What is newborn screening?

How are newborn screening programs funded?

Where is testing conducted?

Where are specimens stored?

COPAY ADJUSTMENT PROGRAMS

SEE IF YOUR HEALTH INSURANCE PLAN HAS A COPAY ACCUMULATOR POLICY

Have you ever been in a situation where your deductible was not satisfied even though you thought it should be?

SHARE YOUR STORY

Has the cost of your medication impacted you or your family? Have copay assistance coupons or cards helped you?

ASK YOUR FEDERAL AND STATE REPRESENTATIVES TO RESTRICT COPAY ACCUMULATORS

The National Economic Burden of Rare Disease Study



FEBRUARY 25, 2021

PREPARED FOR: EVERYLIFE FOUNDATION PREPARED BY: LEWIN GROUP

NEW

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



EveryLife Foundation for Rare Diseases
in Partnership with:
The Lewin Group, part of Optum Serve
Expert Consultants
The Rare Disease Community
September 14, 2023

EVERYLIFE FOUNDATION LEWIN GROUP

2025 FEDERAL LEGISLATIVE POLICY PRIORITIES



Lead

- **Newborn Screening: Federal Reauthorization & Modernization Efforts**
- **Protect & Strengthen Accelerated Approval Pathway**
- **Orphan Drug Incentives**
 - Rare Pediatric Disease PRV Reauthorization (Give Kids a Chance Act H.R 1262/ S. 932) >>
 - Protect & Restore Orphan Drug Tax Credit (Cameron's Law H.R 1414) ▶
- **Appropriations**
 - FDA Rare Disease Innovation Hub
 - Federal Biomedical Research and Public Health Programs



Engage

- BENEFIT Act ○
- Scientific EXPERT Act (H.R 1532/ S. 822) >>
- Medicaid Coverage
- Genetic Testing Coverage



Support

- HELP Copays (S. 864) ▶
- Accelerating Kids Access to Care Act (H.R 1509/ S. 752) >>
- PROTECT Rare Act ○
- PASTEUR Act ○
- MVP Act ○
- Joe Fiandra Access to Home Infusion Act ○
- RARE Act ○
- Safe Step Act ○
- MINI Act ○
- SOAR Act ○
- ENABLE Act (H.R 1436/ S.627) ▶



Monitor

- Clinical Trial Diversity Policies
- Drug Pricing Policies
- Telehealth & Data Security Policies
- Prior Authorization Reform
- Laboratory Developed Test Regulation
- PBM Reform
- QALY Bans
- NIH Reform
- Alternative Funding Programs

Status:

Not yet Introduced ○

Introduced in One Chamber ▶

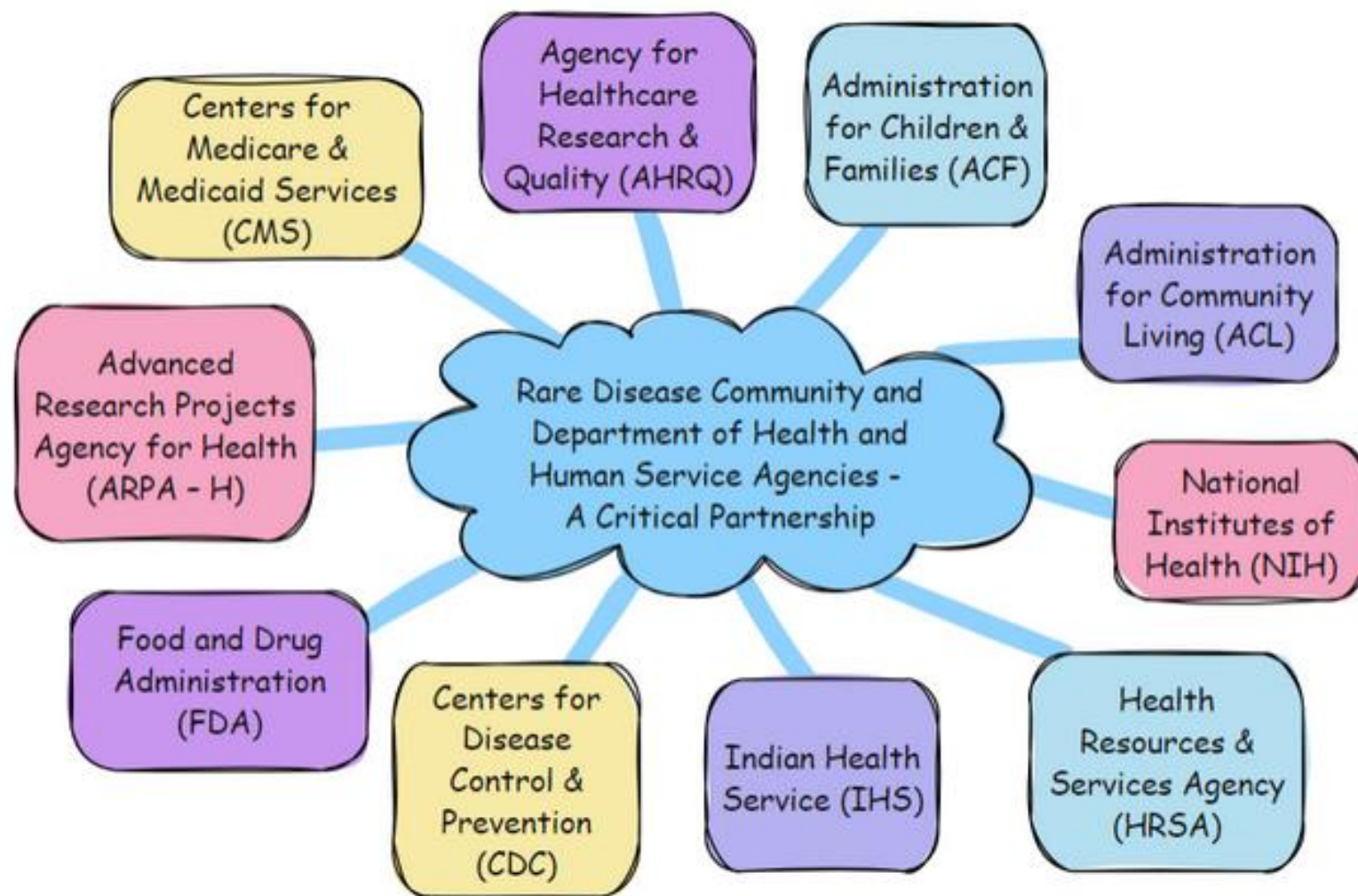
Introduced in Both Chambers >>

Advanced Out of Committee ➡

Passed the House +

Passed the Senate ★

Enacted ☑



Reauthorize the Rare Pediatric Disease Priority Review Voucher Program

- Led to new treatments for 40 rare diseases
- No cost to the tax-payer
- Program expired on December 20, 2024, despite reauthorization from the House.



Patty Mevis | Green Bay, Wisconsin
Mother of Rett Syndrome Patient and Advocate

“When we see people are invested in our kids, it gives us hope in the midst of the weight of a diagnosis.”

#Cures4RareKids

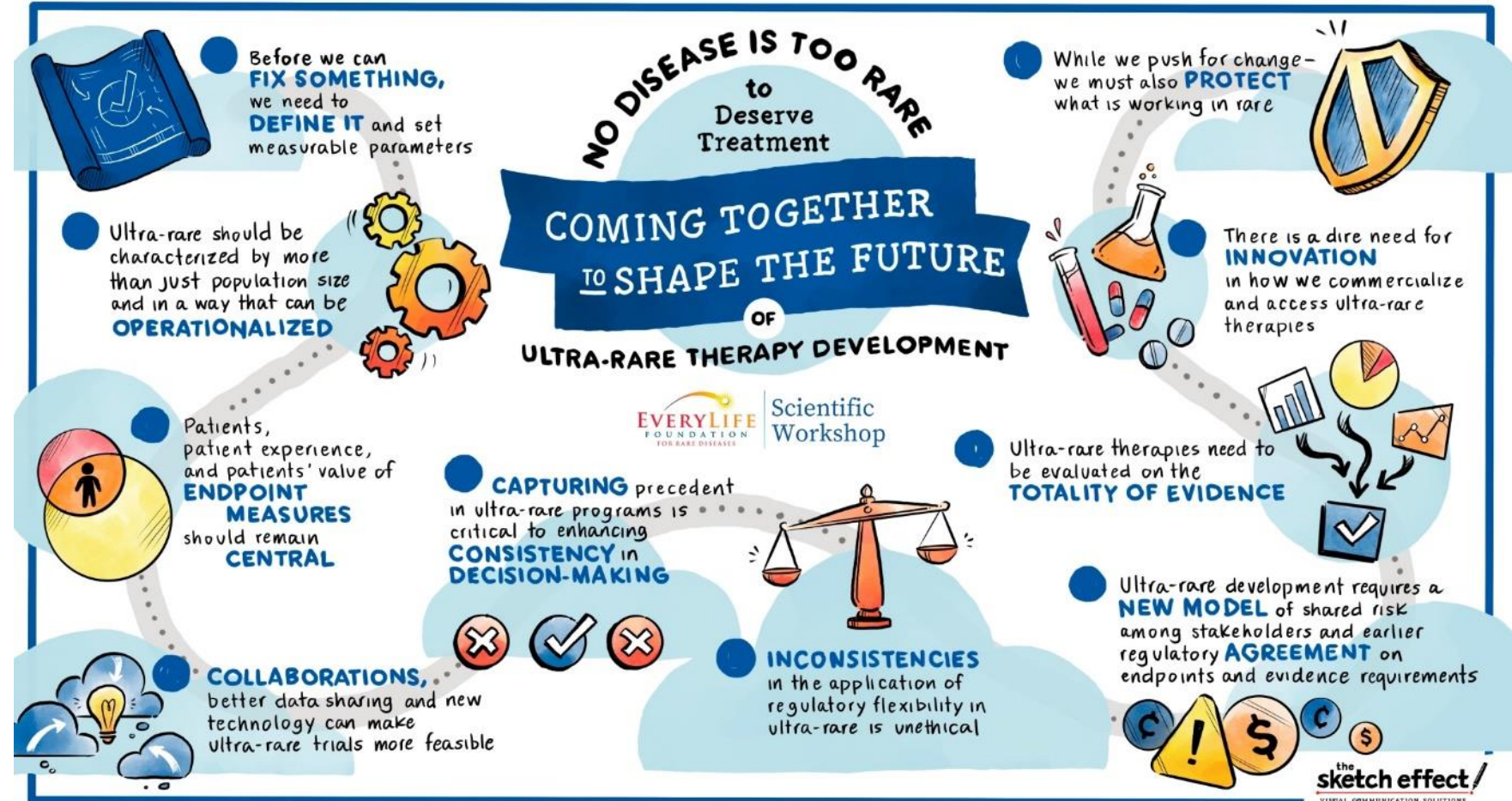
Support Successful Implementation of the Rare Disease Innovation Hub at the FDA

- Improve consistency and predictability of therapy approaches
- Building FDA regulatory science expertise in rare disease
- Advocating for resources for the RDIH



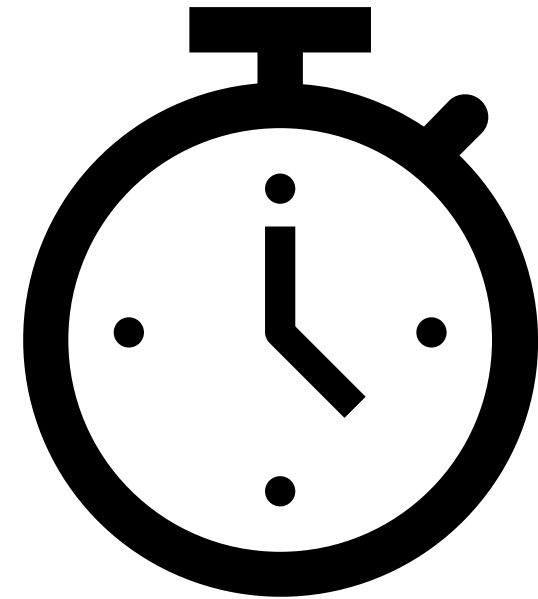
Developing Treatments for Ultra-Rare Diseases

- Address concerns surrounding limited natural history data and costly clinical evaluations
- Enable pipelines for small populations to thrive



Support policies that facilitate timely and affordable access to approved therapies

- Eliminated time and cost as patient access hurdles
- Ensure access to appropriate FDA-approved therapies without burdensome and medically inappropriate utilization management requirements
- Enable innovative payment models that incorporated meaningful community input



NATIONAL RARE DISEASE COMMUNITY PETITION



More than 11,000 rare disease advocates from all over the country signed a petition **urging Members of Congress to encourage the President and the Secretary of Health and Human Services to support strong federal agency leadership, sustained biomedical research funding, and public health agency resources.**

The petition was delivered to every member of Congress on February 28 in honor of Rare Disease Day.

SCAN TO
SIGN PETITION



2025 STATE POLICY PRIORITIES



Lead

Represents policies that the EveryLife Foundation will initiate and drive. Activity may include crafting strategy, conducting direct lobbying activities, activating advocates to support, and more.

- Newborn Screening RUSP Alignment



Engage

Represents policies that the EveryLife Foundation will drive in collaboration with other entities, coalitions, or organizations. Activity may include representing rare disease perspectives, crafting strategy, conducting direct lobbying activities, activating advocates to support, and more.

- Payer Decision-Making: Transparency, Accessibility, and Rare Disease Expert & Patient Representation



Support

Represents policies led by existing entities, coalitions, or organizations that the EveryLife Foundation will proactively act on. Activity may include joining sign-on letters, submitting testimony, engaging state legislatures, participating in joint meetings, or sharing opportunities to provide input with advocates.

- Access to Genetic Testing Services
- Genetic Data Non-Discrimination
- Interstate Telehealth
- Copay Adjustment & Maximizer Programs
- Interstate Medical Licensure Compact
- Prior Authorization Requirements



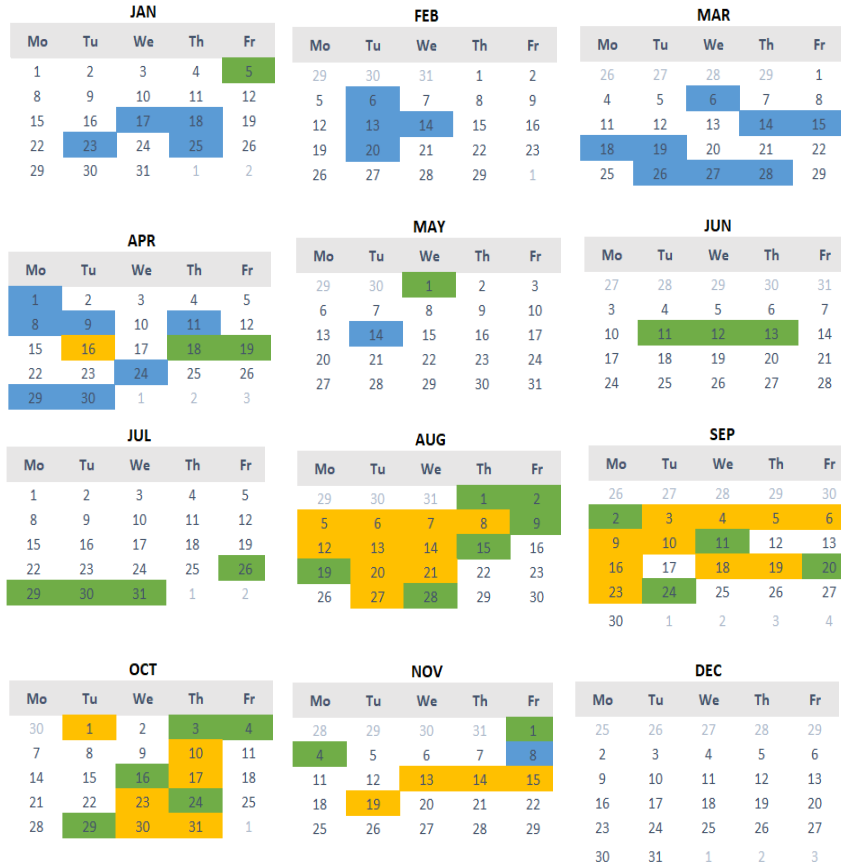
Monitor

Represents policies that are relevant to the rare disease community. The EveryLife Foundation will actively consider proposals from Community Congress members or changes in the policy landscape that necessitate further consideration or action.

- Access to Genetic Testing Services
- Biomarker Testing
- Medical Foods Equity
- Step Therapy
- Access to Therapies Approved via Accelerated Approval
- Cost-sharing
- Personal Data Privacy
- Value Based Payments & Assessments
- Alternative Funding Programs
- Intrastate Telehealth
- Prescription Drug Affordability Boards
- Rare Disease Advisory Councils
- Medical Debt

2024 - State Policy Activity

- Meeting w/ Legislator
- Submit Testimony/Letter
- Share Policy Resource w/Legislator



420

Bills Tracked



48

Meetings with
Legislators



28

Testimonies
or Letters
Submitted



183

Legislators
Contacted



COMMUNITY CONGRESS
Annual Meeting

COMMUNITY
CONGRESS

15
EVERY LIFE
FOUNDATION
FOR RARE DISEASES

An Example of Advocacy Activation in Action

State Newborn Screening Legislation

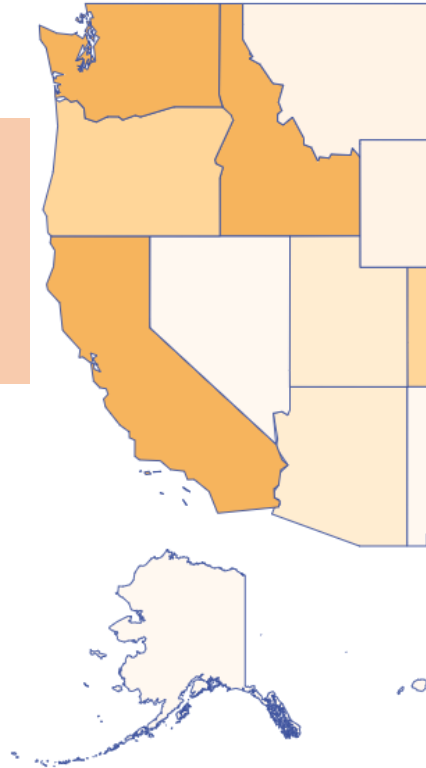
Programs vary widely by

What is RUSP Alignment Legislation?

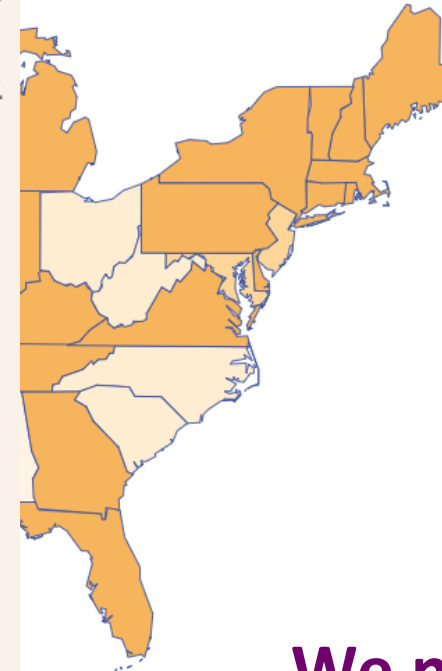
- 1 Requires that states consider screening newborn babies for any disorder on the RUSP.
- 2 Implements a timeline for states to begin screening for new disorders added to the RUSP.
- 3 Ensures resources are available to facilitate the addition of new disorders.

disparate health outcomes.

Some states screen for
fewer than
30 diseases.



Other states screen for
more than
60 diseases.



We must do better.

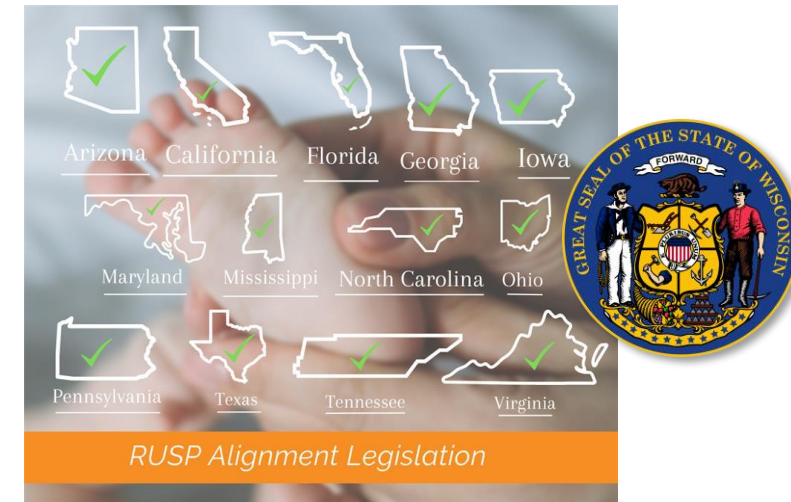
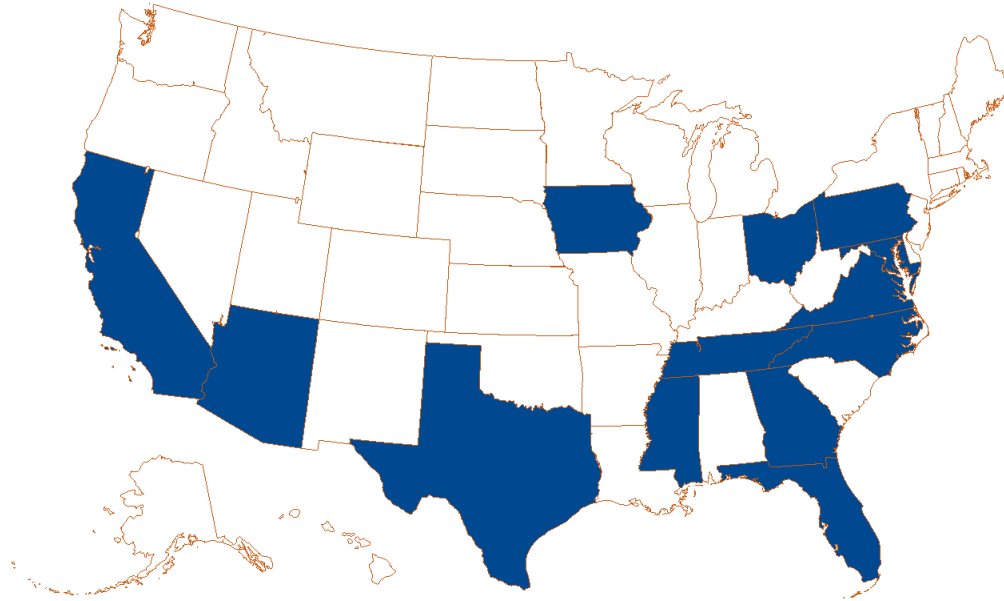
2025 State Policy Accomplishments

Virginia HB 1782- signed by the governor

- > 95,000 babies born in VA Annually
- > 300 annual diagnoses through newborn screening
- > 40 patient organizations supported

52% of babies are born in RUSP aligned states

- Virginia is the 13th state to become RUSP aligned
- RUSP alignment = timeline, funding, and screening language



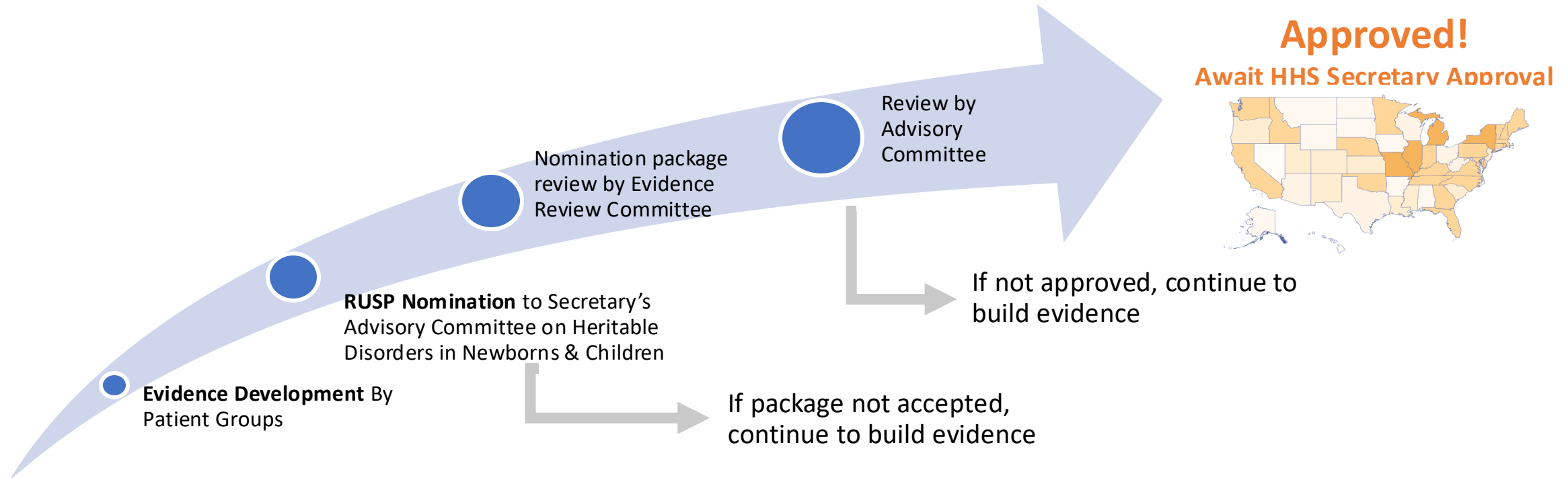
Newborn Screening Across The States

The Recommended Uniform Screening Panel (RUSP)

- **A list of disorders** recommended for states to screen
- 37 recommended conditions

The Advisory Committee on Heritable Disorders

- **Provides RUSP recommendations** to the Secretary of Health and Human Services.
- Doctors, scientists, parents, ethicists, and researchers



Restore & Modernize our Nation's Newborn Screening Ecosystem

- Restore & Update Mechanism for Evidence Review of new Conditions to RUSP
- New policies are needed that address:
 - The role of genetic testing
 - increased investment in public health infrastructure, education, and data systems to support a more effective and equitable newborn screening program

Newborn Screening (NBS)

NOUN

A public health program that screens newborns for serious, treatable, disorders. NBS includes a heel prick, hearing test, and pulse oximetry test.

#RAREdictionary

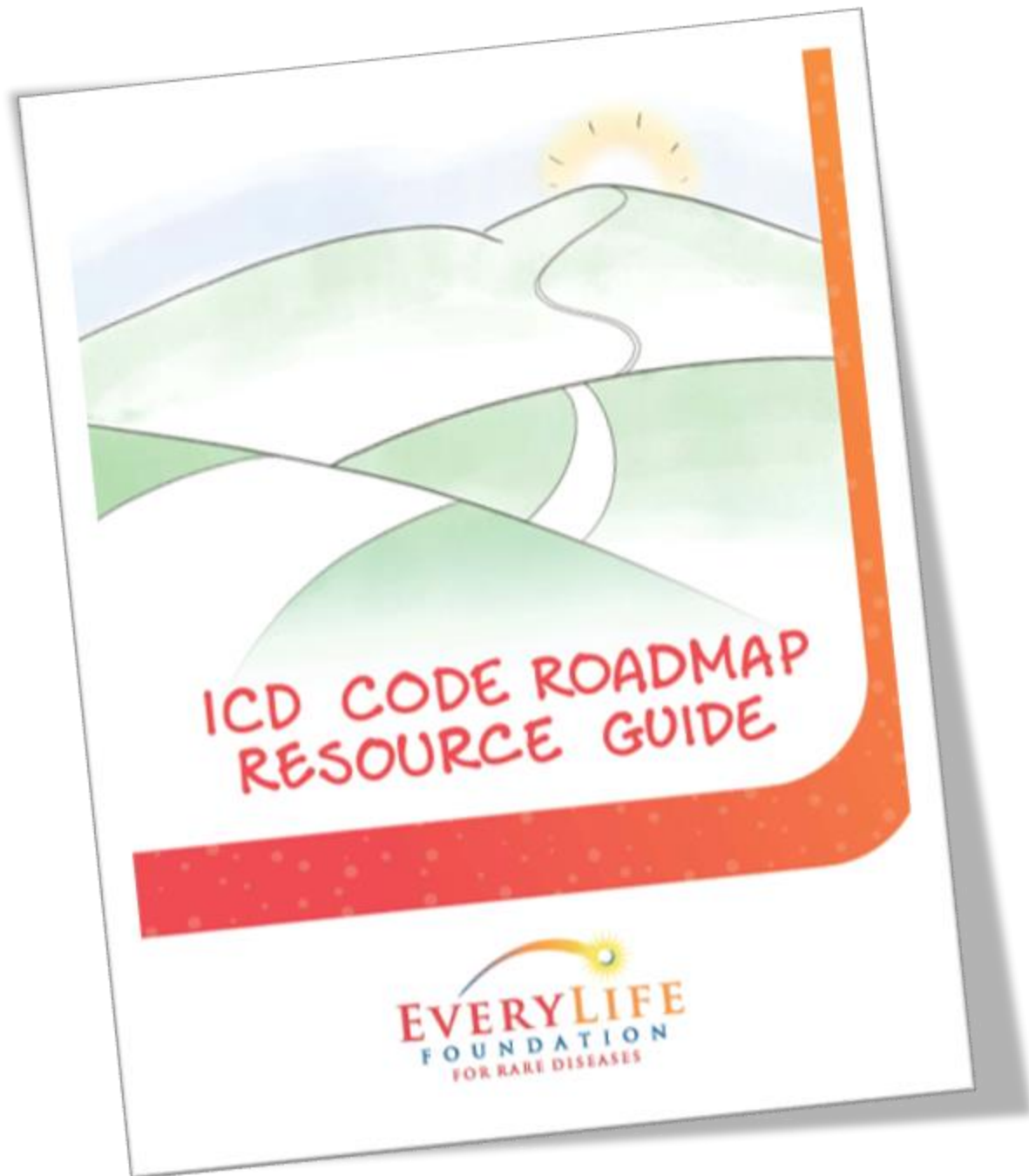


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For all who seek to better understand how diagnosis codes are assigned, updated and revised in the United States' health information system, this guide is for you.

It starts with basic information about the coding system and then describes the process by which patient advocacy organizations and others can propose changes to existing codes or introduce new codes, including the necessary components of a proposal.

Case examples and links to additional resources provide the information you will need to determine whether and how to get involved in the continuous process of refining the diagnostic coding system used in the United States.

Guide To Patient Involvement In Rare Disease Therapy Development

A PUBLICATION OF THE RARE DISEASE PFDD COMPENDIUM WORKSHOP SERIES

HOSTED BY:



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8 FOCUS AREAS

BY THE NUMBERS



23

Leadership and Steering Committee Members



88

TOTAL

Subject Matter Experts

8 Academia
40 Industry
28 Patient Advocacy Organizations
5 Payer Organizations
7 Other



20

FDA Guidances



12

Hours of Workshops



8

Cross-Cutting Topics and Sets of Action Steps



4

Workshop Summaries




112

Resources Linked in This Guide

Accelerated Approval Resources

<https://everylifefoundation.org/accelerated-approval/>



ABOUT ISSUES PROGRAMS EVENTS RESOURCES NEWS [DONATE](#)

ACCELERATED APPROVAL IS...


safe, effective, necessary, and working to deliver better health to those in dire need.

[About Accelerated Approval](#) | [Policy Activity](#) | [Resources](#)

About Accelerated Approval

The accelerated approval (AA) pathway was established in 1992 when HIV/AIDS was destroying lives and communities. The AA pathway allows FDA to use a surrogate endpoint (also called a biomarker) to evaluate the safety and efficacy of therapies for serious conditions with unmet needs. By evaluating a surrogate endpoint that is reasonably likely to predict clinical benefit, the AA pathway allows patients to

"The accelerated approval pathway is working as intended: delivering better health to those in dire need. We must value and protect it for the benefit of patients where the opportunities of accelerated



Policies that regard treatments approved via the AA pathway as experimental **exacerbate health inequity** among communities eligible for potentially life-altering and lifesaving medications.

THE ACCELERATED APPROVAL PATHWAY AND RARE DISEASES

Accelerated Approval Overview:

- In 1992, in response to the HIV/AIDS crisis, the U.S. Food and Drug Administration (FDA) instituted the Accelerated Approval pathway to improve the time to approve drugs that treat serious conditions that fill an unmet medical need based on substantial evidence of safety and efficacy and a surrogate endpoint* that is reasonably likely to predict outcomes like irreversible mortality or morbidity.¹
- By 1996, antiretroviral drugs approved under the Accelerated Approval pathway helped transform HIV/AIDS from a fatal disease into a manageable chronic illness. Since 1992, 278 drugs have been approved under the Accelerated Approval pathway with nearly 50% going on to traditional approval.
- In 2012, the Food and Drug Administration Safety Innovations Act (FDASIA) was passed by Congress. FDASIA amended the Food, Drug, and Cosmetic Act to allow surrogate or intermediate clinical endpoints to be used in Accelerated Approval trials.
- Accelerated Approval is NOT the same as the Conditional Approval pathway in the E.U. Conditional approvals may use a clinical endpoint and must be renewed annually based on satisfactory progress in demonstrating the treatment's effectiveness and a review of safety issues.

MYTH VS FACT

The accelerated approval pathway is not as rigorous as traditional approval.	Accelerated approval is NOT a lower standard. Treatments approved via the AA pathway are subject to the same statutory standards for proving safety and efficacy as traditional drug approvals.
Surrogate endpoints are inferior to clinical outcome measures.	Surrogate endpoints can have advantages over clinical outcomes, especially in cases where they can more accurately capture real-time disease progression or improvement.

Importance for Rare Disease Patients:

- Between 93% and 95% of the 7,000 known rare diseases have approved therapy.
- The development timeline from clinical studies to FDA approval for a rare disease drug takes an average of 15 years.
- In clinical trials, endpoints are used to determine whether the investigational therapy has a clinically meaningful benefit for patients. Endpoints include survival, functional capacity (like ventilator-free), and/or development of new or worsening disease symptoms.
- Proving that a drug has clinical benefit which is required for traditional approval can be challenging due to:
 - Small number of patients available for clinical trials
 - Variability in disease presentation among patients
 - Slow/variable progression of some diseases
 - Multiple organ impact

*Surrogate endpoints:

- Are markers of expected benefit from a drug. These can include enzymatic changes, radiologic imaging, or other relevant measures.
- Are not direct markers of clinical benefit but are intermediate signs of an and/or likely benefit to patients.
- Allow for shorter clinical trials that substantially shorten the length of time to FDA approval.



When it really comes down to it, the important question that too many keep overlooking is: wouldn't you want a life preserver of a treatment if this was your child?"

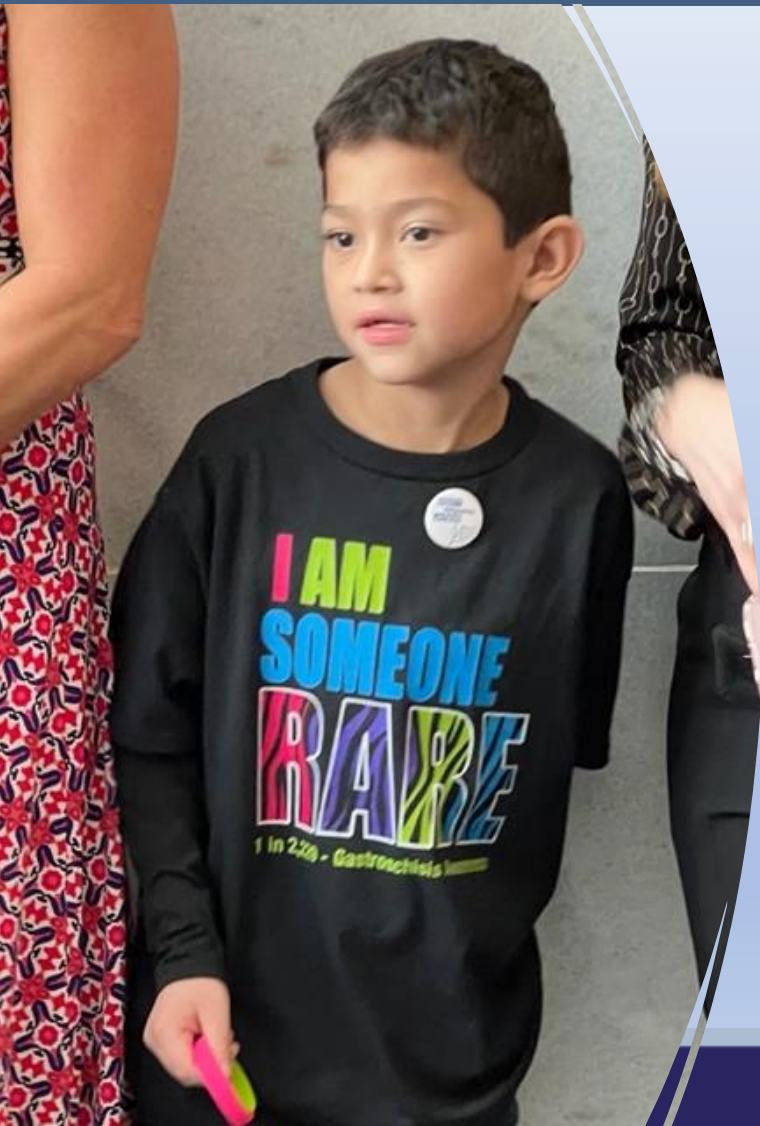
—Terri Ellsworth, Billy's Mom



PARTNERSHIP TO FIGHT CHRONIC DISEASE

#FacesofAcceleratedApproval

Thank you!



**“Surround yourself with relentless humans.
People who plan in years,
but live in moments.
Who work like savages,
but create like artists.
People who know this is finite,
and choose to play infinite games.
Find people going up mountains.
Climb together.”**

~ Original Source Unknown, Loved by Annie