In May, we will celebrate the 10th anniversary of the EveryLife Foundation for Rare Diseases. When launched, the Foundation established goals to remove regulatory barriers for rare disease treatments and increase investment in their development. In 2009, fewer than twenty biotechnology companies invested resources in rare disease, and disease-specific nonprofits worked independently, struggling to gain traction. The term ‘rare disease’ was virtually unknown to Members of Congress.

The landscape has improved significantly over the last decade, thanks to EveryLife’s leadership and the engagement of rare disease advocates. Today, more than 100 companies focus on rare disease drug development. A bipartisan Rare Disease Congressional Caucus now exists, with 135 Members and counting. Numerous organizations with thousands of advocates now work collaboratively, united behind common commitment. Most remarkably, we now talk not just of developing treatments for rare diseases, but of developing cures.

While we are now witnessing a “golden age” of rare disease advocacy and scientific advancement, we stand at a critical tipping point. Ninety-three percent of the 7,000 known rare diseases still have no FDA-approved therapy. The U.S. Food and Drug Administration (FDA) process for approval of a rare disease drug (referred to as “orphan”) can cost hundreds of millions and take an average of 15 years. For the 30% of children with a rare disease who will not live to see their fifth birthdays, that wait is unthinkable.

Thirty million Americans suffer from a rare disease, making it a public health crisis. Prolonged diagnosis and delayed treatment of rare diseases actually increase healthcare costs. A rare disease patient must wait an average of six years after symptoms present before receiving a proper diagnosis, having received several incorrect diagnoses during that time. Untreated disease causes caregivers to miss work and patients to lose their ability or potential to work, creating emotional and financial crises for families.

As the Foundation enters its second decade, we are determined to continue our mission, hand in hand with rare disease patients and their caregivers, to advocate for policies that will pave the way toward treatments – and cures - for rare diseases.

We hope you join us in this fight. United together – 30 million strong – we will continue to change policy and save lives.

Sincerely,

Julia Jenkins
Executive Director,
The EveryLife Foundation for Rare Diseases

Mark Dant
Chairman of the Board of Directors,
The EveryLife Foundation for Rare Diseases

In the United States, a disease is defined as rare when it affects fewer than 200,000 people.

One in 10 Americans suffers from one of 7,000 known rare diseases.

50% of rare disease patients are children, 30% of patients die before the age of five.
Because Every Life Matters

The EveryLife Foundation for Rare Diseases is a 501(c)(3) nonprofit, nonpartisan organization dedicated to advancing the development of treatment and diagnostic opportunities for rare disease patients through science-driven public policy.

“‘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, EveryLife Foundation Executive Director

- Thirty million Americans suffer from a rare disease, making it a public health crisis.
- 93% of the 7,000 known rare diseases have no U.S. Food and Drug Administration-approved therapies.
- Rare disease patients must wait an average of six years after symptoms present before receiving a proper diagnosis, having received several incorrect diagnoses during that time.
- The U.S. Food and Drug Administration process for approval of a rare disease drug (referred to as “orphan”) can cost hundreds of millions and take an average of 15 years.
- Thirty percent of children with a rare disease will not live to see their fifth birthdays.

THE EVERYLIFE BEGINNING

The EveryLife Foundation for Rare Diseases was founded by Dr. Emil Kakkis in 2009. Kakkis, a renowned geneticist, has dedicated his life to the development of novel drug treatments for rare diseases. These treatments have saved the lives of thousands of children, children like Ryan Dant.

In 1991, doctors told Ryan’s parents that he would not live to see adolescence due to a rare disease called mucopolysaccharidosis I (MPS I), which had no known treatment. The Dants were determined to change that. Starting with a bake sale netting $342, they raised over $3 million to further research for MPS. The funds proved critical to Dr. Kakkis in the development of Aldurazyme.

When a biotech startup took a risk on the new therapy, a clinical trial was approved. In 1998, Ryan became one of the first patients. The response was dramatic. Ryan regained motion in his joints and the swelling in his liver and spleen subsided. Ryan went on to graduate from The University of Louisville.

Despite positive results, the FDA required a second trial, adding years and millions of dollars in funding before Aldurazyme was approved in 2003. In 2009, Dr. Kakkis started the EveryLife Foundation to drive legislative and policy change and bring needed treatments to rare disease patients. In 2017, Mark Dant became chairman of the EveryLife Foundation Board and continues to advance this critical mission.
Access to the Accelerated Approval Pathway

Accelerated Approval (AA) pathway was created by FDA regulation in 1992, intending to help shorten the drug development and approval timeline and make new therapies available more rapidly. However, during the first 16 years of AA, only one rare genetic disease therapy was approved. More had to be done.

With this intention in mind, the Unlocking Lifesaving Treatments for Rare Diseases Act (ULTRA) was introduced in 2011, requiring the FDA to issue guidance on the use of biomarkers as surrogate endpoints for rare diseases. The Foundation worked with Congress to draft the bill’s language and secure the support of more than 300 patient organizations.

The ULTRA language was later incorporated into the Faster Access to Specialized Treatments Act (FAST) and was ultimately included in the Food and Drug Administration Safety and Innovation Act (FDASIA). However, the House and Senate versions of the FDASIA bill each included separate provisions specific to the rare disease community. Rare disease patient groups were divided in their support between the two bills.

In response, the Foundation led an innovative strategy: encouraging organizations to sign a letter urging Congress to include all five provisions benefiting rare diseases: ULTRA, Patient Focused Drug Development, Pediatric Priority Review Voucher Program, EXPERT Act and the Breakthrough Therapy Designation. Within 24 hours, more than 120 organizations had signed the Foundation’s letter.

The approach paid off. When FDASIA was signed into law in July 2012, the bill included all five rare disease provisions.

Although FDASIA was a watershed moment for the rare disease community, the challenges for rare disease drug approval remain. The Foundation continues to call for the use of biomarkers through its Scientific Workshops, publications and advocacy efforts.

Alternative Clinical Study Designs

To date, the Foundation has held ten Rare Disease Scientific Workshops, providing guidance which has helped improve the development process for rare disease treatments. Recognizing the unique challenges faced by small rare disease patient populations, the Foundation has focused several of these workshops on clinical trial designs. The Foundation’s 2019 workshop will also address this critical issue and is entitled “The Science of Small Trials in the Age of Biological Plausibility.” Following the workshop, a guide will be published on successful approaches to small clinical trial designs.

Responding to an Urgent Crisis

Shortly after the passage of FDASIA, the FDA began applying stricter standards for toxicity studies, the process of evaluating the safety of potential drugs. The standards, more rigid than had ever been applied in the agency’s history and stricter than Europe’s, drove clinical trials overseas, leaving U.S. patients unable to participate in life-saving clinical trials.

In order to address this crisis, the Foundation held its 2014 Annual Rare Disease Scientific Workshop on “Rationalizing Safety Testing to Enable Clinical Studies and Approval in the U.S. for Rare Disease Treatments.” The workshop, attended by FDA policy makers, featured experts who presented case studies to illustrate the critical need for flexibility in toxicity requirements.

The following year, the FDA followed the Foundation’s recommendations in its guidance on enzyme replacement therapy, allowing for flexibility in toxicity requirements and resulting in the reduction of U.S. drug approval time by nearly a year.

More Specialized FDA Drug Review

The development and review of therapies for rare diseases faces unique challenges, requiring unique expertise. To address this need, the Foundation provided recommendations to Congress that were included in the 21st Century Cures Act, the most significant rare disease-related legislation since the Orphan Drug Act in 1983.

The Cures Act (passed in 2016) works to improve the FDA’s ability to recruit and retain staff, strengthens Patient Focused Drug Development, enhances the use of drug development tools such as the use of biomarkers and alternative clinical trial designs, and provides critical funding to the FDA and NIH.

The Foundation partnered with hundreds of rare disease organizations and individual advocates across the country to host days of action and a social media campaign in support of the legislation which led to the bill being signed into law in December 2016. The Foundation was honored for its leadership efforts by Research!America with the 2018 Paul G. Rogers Distinguished Organization Advocacy Award.

Even more progress was made in 2018, when the FDA announced a restructuring plan for the way its Office of New Drugs evaluates new medicines including a new division for rare diseases. The Foundation has since expanded its goal for specialized FDA drug review, calling for an FDA Center of Excellence for Rare Diseases. The proposal, modeled after the Cancer Center of Excellence, would allow for knowledge sharing across the FDA’s Center for Drug Evaluation and Research and Center for Biologics Evaluations and Research divisions.
After receiving calls from patients about the challenges of gaining access to off-label therapies, the Foundation worked with Congress to draft the OPEN ACT (Orphan Product Extensions Now, Accelerating Cures and Treatments). The bill was introduced in December 2013 and was designed to bring hundreds of safe, effective, and affordable medicines to rare disease patients within a few years by providing a small incentive to biopharmaceutical companies to repurpose major market drugs for the treatment of rare diseases.

The Foundation secured the support of more than 150 patient advocacy organizations. These efforts were successful in having the legislation included in the 21st Century Cures Act which passed the House of Representatives with broad bipartisan support in July 2015. Unfortunately, the legislation was not included in the final version of the Cures Act when it was signed into law. The Foundation is currently developing a new legislative solution to help spur the development of new and repurposed treatments for rare diseases.

Increased NIH & FDA Funding

As a member of Research!America and the Alliance for a Stronger FDA, the Foundation has helped the rare disease community successfully advocate for increased funding for the National Institutes of Health (NIH) and U.S. Food and Drug Administration (FDA). While Congress has provided the NIH and FDA with crucial funding increases in recent years, the NIH is still forced to turn away 80% of the promising research grant proposals it receives, and FDA resources are stretched dangerously thin as it works on multiple fronts to speed medical progress and approve treatments for rare diseases.
Newborn screening detects rare genetic conditions at birth. It enables timely diagnosis and access to treatments that can save lives. Programs vary widely by state, leading to disparate health outcomes. While some states screen for less than 30 diseases, other states screen for more than 60.

In 2016, California unanimously passed the EveryLife Foundation-sponsored newborn screening legislation (SB1095) in just seven months. The Foundation coalesced 120 patient organizations to support the bill. This law requires newborn screening for a condition within two years of its addition to the federal Recommended Uniform Screening Panel (RUSP). This legislation provides newborns with early access to vital, and sometimes life-saving, diagnosis.

In 2017, Florida unanimously passed newborn screening legislation (SB1124) in just three months. The Foundation led the lobby efforts of more than 86 patient organizations that supported the bill. This law requires the state’s Genetics and Newborn Screening Advisory Council to consider conditions within one year of them being added to the RUSP, and implement the Council’s decision within 18 months. In August 2018, the Foundation organized patients to testify before the Council in support of adding Pompe and mucopolysaccaridosis 1 (MPS 1) to the list of screened conditions. As a result of this effort, the Council voted to add these conditions, after years of delay. In February 2019, the Committee voted to add spinal muscular atrophy (SMA) to the list of screened conditions, less than seven months after the disease was added to the RUSP.

In partnership with California Action Link for Rare (CAL RARE), the Foundation helped launch the bipartisan Rare Disease California Caucus in 2017. The Caucus helps to bring public and legislative awareness to the unique needs of the rare disease community—patients, physicians, scientists, and industry. During its first legislative briefing, the Caucus introduced a budget proposal, providing $2 million dollars for a whole genome sequencing pilot program for newborns in California. The line item was included in the final budget. The program will submit a report on the outcomes and the Foundation is actively encouraging advocates to lead similar efforts in their states.
Rare Disease Legislative Advocates (RDLA) is a program of the EveryLife Foundation for Rare Diseases created in 2009 to support the advocacy of all rare disease patients and organizations. RDLA is committed to growing the patient advocacy community and working collaboratively, thereby amplifying the patient voice to be heard by local, state and federal policy makers.

RDLA provides free grassroots advocacy resources such as action alerts, monthly webinars and newsletters and legislative scorecards. It also hosts a variety of educational events which are free to patients and caregivers.

In 2012, 70 advocates traveled from around the country to participate in RDLA’s first Lobby Day event. They advocated in support of provisions that were later included in the Food and Drug Administration Safety and Innovation Act of 2012 (FDASIA). The bill was signed into law later that year.

Over the years, Lobby Day has evolved into a series of events called Rare Disease Week on Capitol Hill. In 2019, more than 800 advocates participated. Events included a rare disease documentary screening, legislative conference, lobby day, Rare Disease Congressional Caucus Briefing, Rare Artist Reception and the National Institutes of Health Rare Disease Day.

“Attending Rare Disease Week on Capitol Hill is a great way to engage and take back power that our disease took.”

“I was motivated to attend to speak for those with a rare disease that could not be here.”

“I love the supportive atmosphere and opportunities to network and the ability to be resources for each other. Thank you for this. This gives so many of us a certain happiness we often feel like we’ll never have.”

In 2010, RDLA led the effort to form the bipartisan Rare Disease Congressional Caucus. The Caucus provides a vital platform for discussing pressing policy issues and gives rare disease patients a voice on Capitol Hill. RDLA coordinates quarterly Caucus briefings that address issues impacting the Food and Drug Administration and the National Institutes of Health and legislation affecting the rare disease community. The Caucus has attracted 135 Members in the 116th Congress thus far (17 Senators, 118 Representatives), making it one of the largest health-related legislative caucuses.

In 2014, RDLA launched In-District Lobby Days to build on the relationships created during Rare Disease Week on Capitol Hill and allow advocates unable to travel to Washington, D.C. to make their voices heard. RDLA organizes meetings for advocates with their Members of Congress and/or the Member’s staff during August recess. In 2019, the program was rebranded as Rare Across America, reflecting the program’s diverse and engaged community.

“In my meeting I was able to share my story about living with sickle cell disease and how these policies can benefit the rare disease community. I want to thank the RDLA for preparing me and giving me such a great opportunity to share my thoughts and opinions.” - Kadeem, Somerville, MA

Since 2012, the RDLA has hosted the Rare Voice Awards, an annual celebration to honor advocates who have spoken on behalf of patients, especially children who might not otherwise be heard. Each year, more than 300 patient advocates, industry executives, and Congressional and government agency staff gather to honor these outstanding advocates at Arena Stage in Washington, D.C. The Rare Voice “Abbey” Award was named after Abbey Meyers, the founder of the National Organization for Rare Disorders (NORD). Mrs. Meyers received the Lifetime Achievement Award at the inaugural Rare Voice Awards for her vital role in the passage of the Orphan Drug Act.
Since November 2015, the Foundation has given patient organizations and industry partners an equal seat at the table to work together on shared goals as members of the Community Congress. The Congress acts as a strategic advisory council, providing advice and insight on urgent policy issues and Foundation programs and initiatives. The Congress has three working groups that work on self-selected projects to advance policy for rare disease patients, including: Public Policy, Regulatory Science and Newborn Screening. In 2018, the Community Congress reached more than 215 members representing diverse stakeholders from the rare disease community.

**Community Congress**

**Rare Artist**

The Rare Artist Contest was established in 2010 to exhibit the unique gifts of individuals affected by rare diseases and to promote the expression of their stories through art. All Rare Artist awardees are presented with gift cards and invited to speak during Rare Disease Week on Capitol Hill. Additionally, their artwork is showcased throughout the year at various patient and biopharmaceutical conferences. The contest attracts hundreds of submissions each year from around the country to be considered for one of the four award categories: Children, Teen, Adult and Adult Digital and Photography. The contest’s popular vote, whereby the rare disease community chooses their favorite artwork via a social media platform, has reached more than one million people.

**Rare on the Road**

In 2017, the Foundation launched Rare on the Road, a rare disease leadership tour, in partnership with Global Genes. The tour is designed to build and activate the rare disease community at the local level, by hosting regional one-day training sessions for rare disease patients, caregivers and other advocates. Dozens of stipends are awarded each year, enabling more advocates to attend. These events have cultivated hundreds of advocates in ten cities around the country.

**Rare Hub**

As part of its commitment to cross-disease collaboration and legislative advocacy, the Foundation introduced the Rare Hub in November 2018. Located in the heart of Washington, D.C., the Rare Hub provides a shared office space for rare disease organizations and also serves as the Foundation’s new headquarters.

**Young Adult Representatives of RDLA**

The Young Adult Representatives of RDLA (YARR) was established in July 2018 to foster the next generation of rare disease advocates. In less than a year, YARR has attracted 41 members, ages 16-30, who have worked to educate other young adults about rare disease and provided energetic support of RDLA’s advocacy efforts. The group hosts meetups throughout the year and communicates via the organization’s growing social media outlets. Several YARR members have represented the group as guest speakers at rare disease events around the country.

**Rare Giving**

Rare Giving provides travel stipends to rare disease advocates, enabling them to meet with Members of Congress, the FDA and NIH, and ensuring that policymakers hear directly from patients. Rare Giving also sponsors patient organization’s events that train advocates about legislation. The program gives more than $100,000 annually to support patients and patient organizations.

"Because of the Everylife Foundation and the people I have met through them, I have become a more effective advocate for Sarcoidosis and expanded my capacity to advocate for all rare diseases.”
- Mary, Rare Disease Advocate

"I became one of the first Moyamoya Disease patients to join Rare Disease Week on Capitol Hill and share my personal story with legislators. I learned so much about the rare disease world and how my small voice could make such a difference. Since I underwent bilateral brain bypass surgeries just a few months before, I wasn’t in a financial position to attend without the travel stipend.”
- Lisa Deck, Rare Disease Advocate
The Foundation receives funding from philanthropic organizations, government, individual donors, and corporations to support its activities. Sponsors are offered recognition for their support, but do not influence Foundation policy priorities or event program content. To view our complete funding policy, visit: everylifefoundation.org/fundingpolicy

Thank You to our 2018 Supporters

- **$350,000**
  - Emil D Kakkis
  - Italian Street Painting Marin

- **$100,000 - $140,000**
  - Manis Community Foundation
  - Retrophin
  - Shire HGT
  - Horizon Pharma

- **$55,000 - $90,000**
  - BioMarin
  - AveXis
  - Sanofi
  - Gene Spotlight
  - Vertex Pharmaceutical Incorporated

- **$25,000 to $50,000**
  - George A Weiss
  - GlaeserSmithKline
  - BioGen
  - Genzyme
  - Mallinckrodt
  - Pfizer Inc
  - Amicus Therapeutics
  - Genentech
  - PhrMA
  - Sarepta
  - Bridge Bio
  - Novartis
  - Recordati Rare Diseases Inc.
  - Bluebird Bio
  - Gilead Sciences, Inc.

- **$10,000 - $24,000**
  - PTC Therapeutics
  - Actelion
  - Johnson & Johnson
  - MT Pharma
  - Sobi
  - Biotechnology Innovation Organization
  - Abeona Therapeutics, Inc.
  - Audentes Therapeutics, Inc.
  - CSL Behring
  - Gribbs
  - Ritu Baral
  - William Aliski
  - Akcea Therapeutics
  - Aurxure Family
  - RegenXBio Inc.
  - Stealth Biotherapeutics
  - J.P. Morgan
  - Center-View Partners
  - Cowen Group
  - Dohmen Life Science Services
  - Eli Lilly and Company
  - Fibrocell
  - John Crowley
  - Perkin Elmer Foundation
  - United Therapeutics
  - Lung Biotech
  - Harbor Point Charitable Foundation
  - Fenwick Foundation

- **$350,000**
  - Emil D Kakkis
  - Italian Street Painting Marin

- **$100,000 - $140,000**
  - Manis Community Foundation
  - Retrophin
  - Shire HGT
  - Horizon Pharma

- **$55,000 - $90,000**
  - BioMarin
  - AveXis
  - Sanofi
  - Gene Spotlight
  - Vertex Pharmaceutical Incorporated

- **$25,000 to $50,000**
  - George A Weiss
  - GlaeserSmithKline
  - BioGen
  - Genzyme
  - Mallinckrodt
  - Pfizer Inc
  - Amicus Therapeutics
  - Genentech
  - PhrMA
  - Sarepta
  - Bridge Bio
  - Novartis
  - Recordati Rare Diseases Inc.
  - Bluebird Bio
  - Gilead Sciences, Inc.
PATIENTS HAVE BEEN AND ALWAYS WILL BE THE KEY TO ADVANCING INNOVATION.

BECAUSE EVERY LIFE MATTERS

501(c)(3) nonprofit organization (Tax ID 26-4614274)

1012 14th St, NW, Suite 500, Washington, D.C. 20005
(202) 697-RARE (7273) info@everylifefoundation.org

everylifefoundation.org

facebook | linkedin | twitter | @EveryLifeOrg