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FARA Friedreich's Ataxia Research Alliance

RARE DISEASE WEEK EXTRAVAGANZA!

This Rare Disease Day, February 28, was historic—for the first time in history, FA has a treatment! The FDA notified Reata Pharmaceuticals that their New Drug Application for omaveloxolone, now SKYCLARYS, was approved.

It took many stakeholders to make this possible, including patients, FA families and friends, clinicians, researchers, industry partners, and regulators. Additionally, FA was the recipient of the incredible work by our Congressional Members and their staffers in bolstering this outcome.

The Orphan Drug Act, celebrating 40 years this year, began the legislative path only to be strengthened by PDUFA I-VII, 21st Century Cures Act, and Act for ALS, to name a few. The flexibility Congress granted to the FDA when reviewing a rare disease drug candidate permitted them to consider one clinical trial with confirmatory evidence, in this case, a comparison to years of natural history data, and enabled access to a treatment years earlier than would have been possible under the traditional model. **These legislative efforts were instrumental in reaching this important milestone and would not have been possible without patient advocates working with Congressional Members.**

As the FA community celebrated our first drug approval on Rare Disease Day, FARA also continued the important advocacy work toward more approved treatments. FARA, and many in the FA community, joined thousands of stakeholders throughout the world in raising awareness, developing Congressional relationships, and supporting legislation that will continue to expedite drug approvals. **Here are some of the highlights!**

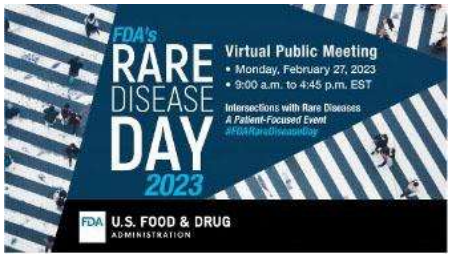
Rare Disease Week on Capitol Hill

EveryLife Foundation held its **12th Rare Disease Week on Capitol Hill**, which was in person for the first time since 2020. From February 28 to March 2, over 600 advocates representing 300 patient organizations, including FARA, gathered to advocate on behalf of the rare disease community. For the first time, all 50 states, plus Puerto Rico and the Cherokee Nation, were represented.

Advocates covered four legislative asks:

- Support Rare Disease Appropriations Priorities
- Support the Better Empowerment Now to Enhance Framework and Improve Treatments (BENEFIT) Act, H.R. 1092 and S.526
- Join the Rare Disease Caucus. To view a full list of Caucus members, click here.
- Join a Congressional sign-on letter to the FDA requesting the formation of an internal FDA task force to review and inform agency-wide rare disease activities

Please save the date for the next Rare Disease Week on Capitol Hill, February 25–28, 2024.



FDA and NIH Host Rare Disease Day Events

On February 27 and 28, the FDA and the NIH respectively hosted events to celebrate Rare Disease Day. These events helped to raise awareness of the challenges faced by rare disease patients and caregivers and highlighted programs to accelerate rare disease research and therapy development.

NORD LIGHTS UP THE WORLD IN BLUE, GREEN, PINK, AND PURPLE

With thousands using the **#ShowYourStripes** or **#RareDiseaseDay** hashtags across social media platforms, 650+ personal stories and sentiments shared to NORD's Rare Disease Day Dedication Wall, and 120+ pledges to **#LightUpForRare** (not to mention a rare disease rap song from Philly and an exclusive Rare Disease Day beer release in Wisconsin), NORD is proud to say that Rare Disease Day 2023 was a thrilling success!

FARA arranged for the Pennsylvania Capital Lt. Gov. Balcony Lights to be lit from Feb 23 to Mar 1, as well as facilitated getting all four bridges in Boston lit on Feb 28. FARA also participated in the International Global Chain of Lights event at Notre Dame.

Thanks to thousands of posts, **#RareDiseaseDay** was on Twitter's top five trending list on February 28. Along with Twitter, individuals showed their stripes and posted their Rare Disease Day celebrations on LinkedIn, Facebook, Instagram, and TikTok. Together, millions of individuals in the US and beyond became aware of rare diseases!

EURORDIS Partners with EveryLife Foundation for Brussels Rare Disease Week 2023

EURORDIS-Rare Diseases Europe, partnering with the EveryLife Foundation for Rare Diseases, launched the second annual Brussels Rare Disease Week, to raise awareness among EU policymakers of the high unmet needs of the 30 million people living with a rare disease in Europe.

After a series of virtual meetings and training sessions as part of the first Rare Disease Week in 2021, EURORDIS staged its first in-person iteration of the program in Brussels from February 6–9, 2023. View the press release here.

FARA AMBASSADORS HIT THE ROAD AND AIRWAVES

State of Vermont
Executive Department
A Proclamation

WHEREAS, there are nearly 7,000 diseases and conditions considered rare (each affecting fewer than 200,000 Americans) in the United States, according to the National Institutes of Health (NIH); and

WHEREAS, while each of these diseases may affect small numbers of people, rare diseases as a group affect almost 30 million Americans; and

WHEREAS, many rare diseases are serious and debilitating conditions that have a significant impact on the lives of those affected; and

WHEREAS, while more than 450 drugs and biologics have been approved for the treatment of rare diseases according to the Food and Drug Administration (FDA), millions of Americans still have rare diseases for which there is no approved treatment; and

WHEREAS, individuals and families affected by rare diseases often experience problems such as diagnosis delay, difficulty finding a medical expert, and lack of access to treatments or ancillary services; and

WHEREAS, while the public is familiar with some rare diseases, many patients and families affected by less widely known rare diseases bear a large share of the burden of funding research and raising public awareness to support the search for treatments; and

WHEREAS, residents of Vermont are among those affected by rare diseases because nearly ten percent of Americans have rare diseases; and

WHEREAS, the National Organization for Rare Disorders (NORD) is organizing a nationwide observance of Rare Disease Day on February 28, 2022.

NOW, THEREFORE, I, Philip B. Scott, Governor, hereby proclaim February 28, 2022 as

RARE DISEASE DAY

in Vermont.

Given under my hand and the Great Seal of the State of Vermont on this 25th day of February, A.D. 2022.

Philip B. Scott
Governor

Brittney L. Wilson
Secretary of Civil and Military Affairs

In addition to covering all of her Vermont Congressional meetings, Ambassador Mary Nadon Scott secured a Vermont State Proclamation recognizing February

28 as Rare Disease Day. Thank you, Governor Phil Scott!

FARA Ambassadors Noah Griffith, Adriana Capri, Mekayla Holm, Shandra Trantham, Kyle Waterman, and Jake Tompkins were guest speakers on Rare is Us, Raising Rare, and LEMS Aware podcasts.

Capitol Hill Updates

President Biden Supports Research in his FY 2024 Budget

President Biden released his $6.9 trillion budget proposal for Fiscal Year 2024 on March 9th. The proposed budget has spending levels for basic and applied research that top $100 billion for the first time in history and spending for federal research and development that tops $200 billion. To view a fact sheet on the President's FY 2024 proposed budget, click here.

Here is a great summary from our colleagues at Research!America:

FY24 President's Budget Request for federal research agencies

RESEARCH AMERICA

AGENCY	FY22 ENACTED	FY23 OMNIBUS	FY24 PRES. BUDGET REQUEST	CHANGE FROM FY23 TO FY24	% INCREASE OVER FY23
NIH	$44.96	$47.50	$48.265	$0.81	1.70%
CDC	$8.46	$9.22	$11.58	$2.36	25.64%
FDA	$3.32	$3.54	$3.96	$0.42	11.86%
AHRQ	$0.35	$0.37	$0.45	$0.08	20.00%
NSF	$8.84	$9.54	$11.31	$1.78	18.61%
ARPA-H	$1.00	$1.50	$2.50	$1.00	66.67%

(all $ in billions) www.researchamerica.org

CDMRP
DEPARTMENT OF DEFENSE
CONGRESSIONALLY DIRECTED MEDICAL RESEARCH PROGRAMS

CDMRP/PRMRP FY24 Appropriations

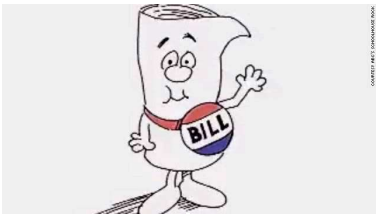
Now that the President has submitted the proposed budget, the Congressional Appropriations Committee will work to draft twelve appropriations bills for Congress to pass before the end of the fiscal year on September 30th. Each Member will collect, review, and submit various Appropriations requests to the appropriate subcommittee for consideration.

Each year, a Congressional request must be made to remain on the Congressionally Directed Medical Research Program (CDMRP)/Peer Reviewed Medical Research Program (PRMRP). FARA, in collaboration with the National Ataxia Foundation (NAF), submitted 35 Senate and

22 House requests for the continued inclusion of “Hereditary Ataxia” on the CDMRP/PRMRP.

This effort was made possible by the many community members who have initiated and developed relationships with their Congressional Members. As your Members learn more about FA, they are better situated to evaluate and support legislation that might help the FA community. You can help too! It all starts with an introduction. You can do it by email, attending an organized event, or just stopping by their local office.

BENEFIT Act Reintroduced to Congress



The BENEFIT Act (S. 526/HR 1092) was reintroduced to Congress by Senators Klobuchar and Wicker, along with Representatives Matsui and Wenstrup. This legislation will require FDA to provide a description of how patient-experience data was considered in its risk-benefit framework. Patient experience data can include patient-reported outcomes, testimonials, patient preference data, and natural history studies. Support for the BENEFIT Act was selected as a community ask for Rare Disease Week.

Senate Leaders Respond to CMS Regarding Accelerated Approval Drug Coverage

A group of 18 Republican senators sent a letter to CMS expressing concern over the proposal from the Center for Medicare & Medicaid Innovation Center (CMMI) to cut Medicare payments for drugs approved via the Accelerated Approval pathway. The senators highlighted that the proposed change could stifle innovation from manufacturers and force clinicians to prescribe alternative treatments. Moreover, the proposal calls into question the regulatory standards imposed upon Accelerated Approval drugs, despite those drugs being subject to the same strict standards as drugs traditionally approved.

Upcoming Advocacy Events

May 1–3, 2023 Philadelphia, PA

The RARE Drug Development Symposium, hosted by Global Genes and the Orphan Disease Center of the University of Pennsylvania, equips advocates with the knowledge, skills, and connections they need to advance therapy development for their communities. You will meet individuals, advocacy leaders,

industry, and research experts who have been there and done it, over two days of in-person-only panel discussions, hands-on workshops, and expert office hours. Click here to learn more and register.

All Wisconsin advocates are invited to join RDLA for a State Advocacy Day at the Wisconsin State Capital on May 17th! To learn more, please read the information below. To register, click here. If you have questions, please contact the State Advocacy Manager, Amanda Houdeschell at: ahoudeschell@everylifefoundation.org.

What: A day of action for Wisconsin residents impacted by rare disease to join together and meet your state legislators, share your stories, and help advance the policy priorities of the rare disease community.

Why: Many healthcare policies that affect the rare disease community are made at the state level. These policies impact diagnosis, treatment, access to care, and more. One of the most powerful ways that you can influence policymakers is by meeting with them in person. We need you and every resident of Wisconsin who is impacted by rare disease to join us for Rare Disease State Advocacy Day and make your voice heard. If you are new to advocacy, there is no better opportunity to get involved than by joining other patients and caregivers to speak for a common cause.

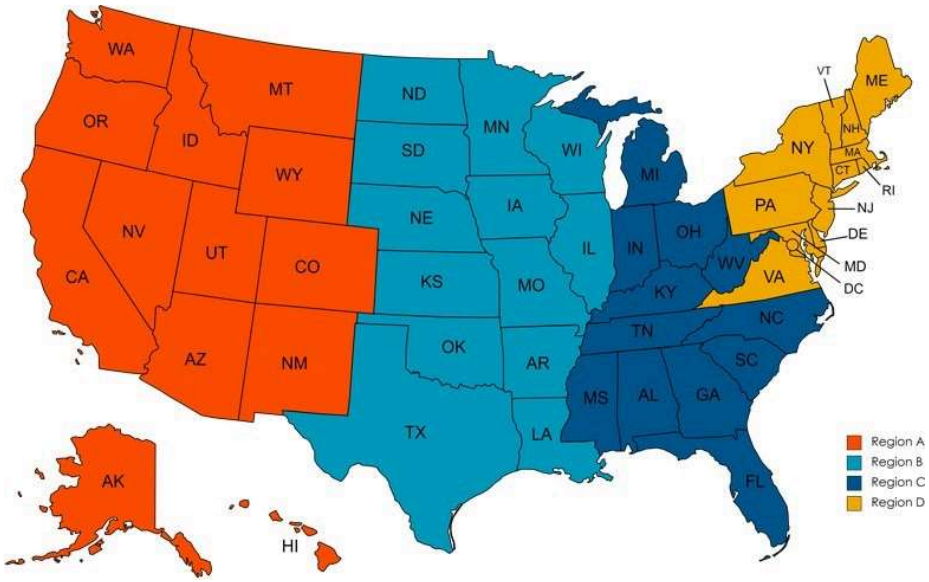
Who: This event is open to Wisconsin residents only. No prior advocacy experience is necessary. Registration for this event and all RDLA events are free for all rare disease advocates. Registration is required for all participants, including spouses and children.

Where: Wisconsin State Capitol, 2 E Main St, Madison, WI 53703

When: Wednesday, May 17th, 2023 8:30 a.m. – 5:00 p.m. CT

Mandatory Training Webinar: How to Advocate with Your State Legislature on April 26th, 2023 2:00 p.m. – 3:00 p.m. CT

NORD's National Policy & Advocacy Taskforce



Join NORD's Policy & Advocacy Taskforce! This program will provide a platform for individuals to work with other rare disease advocates in their region to advance policies that benefit the rare disease community.

Region A: Washington, Oregon, California, Idaho, Nevada, Utah, Arizona, Montana, Wyoming, Colorado, New Mexico, Alaska, Hawaii

Region B: North Dakota, South Dakota, Nebraska, Kansas, Oklahoma, Texas, Minnesota, Iowa, Missouri, Arkansas, Louisiana, Wisconsin, Illinois

Region C: Michigan, Indiana, Ohio, Kentucky, West Virginia, Tennessee, North Carolina, South Carolina, Mississippi, Alabama, Georgia, Florida

Region D: Virginia, District of Columbia, Maryland, Delaware, New Jersey, Pennsylvania, New York, Connecticut, Rhode Island, Massachusetts, Vermont, New Hampshire, Maine

No prior advocacy experience is necessary to join Taskforce meetings. NORD will provide you with the training and tools you need to be a successful rare disease advocate. You can learn more about the Taskforce, how to participate, and which region you live in on this webpage.

Next National Taskforce meeting is Tuesday, May 23 at 4:00 pm ET.

RARE ACROSS AMERICA

SAVE THE DATE!
Rare Across America 2023 will take place from August 7th through the 18th.

During that time, rare disease advocates will have the opportunity to meet with their Members of Congress at the Member's in-state, in-district office. To

learn more about Rare Across America, click here.

Announcements

CDER Director Peter Marks Lays Out Plan of Action for Pilot Rare Disease Project

In a statement to the Biopharma Congress, Peter Marks, the Director of the FDA's Center for Biologics Evaluation and Research (CBER), announced the center's intention to pilot a program similar to Operation Warp Speed. Operation Warp Speed was the federal effort to accelerate the development of COVID-19 vaccines. Director Marks noted that "…the goal is to take a drug with some promise in the rare disease space for diseases that don't have alternatives … and then take these products, maybe they have a breakthrough or advanced therapy designation, they have promise and there is a product in development and not just a concept. And then give them the opportunity with not just chemistry, manufacturing, and controls, which we have a pilot for, but allow the clinical development to happen in constant communication and sharing of potential results before submission of an NDA or BLA. That's the idea, is to move things as fast as possible." Read a summary here.

FDA/CBER Public Listening Meeting

CBER Office of Therapeutic Products (OTP) is hosting a virtual public listening meeting to solicit input on methods and approaches for capturing post-approval safety and efficacy data for cell and gene therapy products. With an increasing number of cell and gene therapy products in development and the potential for more of these treatments to become available to patients in the future, it is important to understand the full spectrum of long-term effects and collect accurate, timely, and comprehensive data to ensure these products remain safe, effective, and of high quality.

More information about the event is below:

- **Date:** Thursday, April 27, 2023
- **Time:** 12:00–4:30 pm ET
- **Location:** Virtual Zoom Meeting
- **Registration:** Required. Visit this webpage to register.

Interview with Director PATRIZIA CAVAZONNI of CDER

Watch BioCentury's interview with Patrizia Cavazonni, director of FDA's Center for Drug Evaluation and Research (CDER) here.

Here is the EveryLife Foundation's summary of the topics covered:

- the future of FDA's regulation of products for neurological conditions (she says Billy Dunn's departure will not change FDA's direction)
- accelerated approval (enrollment and launch of confirmatory trials prior to accelerated approval will be the default assumption)
- description of the circumstances warranting "regulatory flexibility"
- thoughts about advisory committees (FDA is thinking about how to have meetings to discuss endpoints decoupled from consideration of specific products)
- FDA's hybrid "new normal" work environment

FDA/CDER and Johns Hopkins Webinar on Clinical Trials for Rare Diseases

On Tuesday and Wednesday, May 2–3, 2023, the FDA/CDER and Johns Hopkins University will cohost **a virtual workshop that is open to the public** titled, "Addressing Challenges in the Design and Analysis of Rare Disease Clinical Trials: Considerations and Tools Virtual Workshop."

Topics will include the use of natural history studies and registry data to inform rare disease drug development, as well as tools and approaches to design, conduct, and analyze trials for rare diseases. To register for the webinar, click here.

ARPA-H Releases First Round of Funding

On March 15th, ARPA-H announced its first broad agency announcement to seek funding proposals for research that utilizes unconventional approaches. The first round of funding will focus on four areas: health science futures, scalable solutions, proactive health, and resilient systems. You can read more about the funding announcement here.

NORD's annual State Report Card is a tool for state advocates and lawmakers to assess how all 50 states are serving the rare disease community, and to aid in advocating for meaningful policy change. This year's State Report Card was compiled using data through November 2022 and provides a detailed analysis of each state's performance on nine major policy areas of importance such as newborn screening, Medicaid eligibility, RDAC formation, and prescription drug out-of-pocket cost protections.

Curious how your state measures up? Visit the NORD website to read the full report card now.

State Update on Newborn Screening

North Carolina Adds Conditions to Screening Panel

The North Carolina Department of Health and Human Services announced that the state would begin screening all newborns for two additional diseases, Pompe Disease and MPS I. Each year, more than 200 babies in North Carolina are identified through newborn screening. You can read a full statement here. The state now screens for 35 of the 37 federally recommended conditions. See a comprehensive view of North Carolina newborn screening here and a map of newborn screening by state here.

Texas RUSP Bill Introduced

Texas Representative Stephanie Klick and Senator Blanco recently introduced H.B. 2478 and S.B. 1697. The bills implement reporting language that will help to ensure that Texas is screening for core conditions on the Recommended Uniform Screening Panel (RUSP) within two years. The reporting language will focus annually on implementation steps, anticipated completion dates, and potential barriers related to adding recommended newborn screening tests not currently included in the state's regimen. This legislation will eliminate the unnecessary delay in screening for diseases, ensuring that Texan babies born with debilitating and life-threatening diseases are diagnosed and treated at the earliest age possible and without devastating delays.