Trip report for Rare Disease Lobby Day on Capitol Hill Rare Disease Legislative Advocates thanks to a travel grant from the National Niemann-Pick Disease Foundation.

Prepared by Margo Frey, PhD, PE
Background

RARE / ORPHAN DISEASE

- A rare or orphan disease is defined as disease affecting fewer than 200,000 people in the US. More than 80% of rare disease of rare diseases are considered ultra-rare, affecting fewer than 6,000 people. Some rare diseases affect fewer than 100.
- Rare diseases include rare cancers, tropical or neglected diseases, genetic disease and many pediatric diseases including cancers.
- Rare disease affects more than 30 million Americans and their families. One in ten Americans have a rare disease and 50% of those are children. Nearly 1/3 of children with a rare disease die before the age of 5.
- There are more than 7,000 known rare diseases but the vast majority (~93%) do not yet have a treatment approved by the FDA.
- On average, it takes between 5.6 and 7.6 years to correctly diagnose a rare disease patient in the US, with 25% of patients with the most “common” rare diseases wait between 5 and 30 years to receive a correct diagnosis, with 40% receiving an incorrect initial diagnosis.

ORPHAN DRUG ACT

- Enacted in 1983, creating the orphan drug designation and providing needed incentives for researchers and manufacturers to develop therapies for rare diseases.
- Prior to 1983, only 38 drugs had ever been approved for the ~7,000 orphan/rare conditions. Since then, the FDA has approved over 500 orphan drugs. In 2018 alone, 90 rare disease indications were approved and 34 novel treatments for rare diseases approved, accounting for 58% of all 2018 FDA drug approvals (see table below). This has created new challenges at the FDA.

- The formation of a Rare Disease Center of Excellence (COE) to help address these challenges is a logical next step.

<table>
<thead>
<tr>
<th>Year</th>
<th>Drugs Approved by FDA CDER</th>
<th>Number (%) for Rare Diseases</th>
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<td>46</td>
<td>18 (39%)</td>
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<tr>
<td>2018</td>
<td>59</td>
<td>34 (58%)</td>
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<tr>
<td>2019</td>
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RARE DISEASE LEGISLATIVE ADVOCATES (RDLA)

Rare Disease Legislative Advocates is a program of the EveryLife Foundation for Rare Diseases designed 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 scorecard. It also hosts a variety of events which are free to patients and caregivers.

RARE DISEASE LOBBY DAYS HAVE HAD SIGNIFICANT PRIOR IMPACT

- Rare Disease Lobby Day was first organized by RDLA in February of 2012
- At the time, the 5-year reauthorization of funding for the FDA (via the Prescription Drug User Fee Act, or PDUFA 5) was up for reenactment
- As part of that effort, advocates lobbied for the following, which were ultimately included and collectively called the Food and Drug Safety and Innovation Act (PDUFA 5 or FDASIA):
  - **Section 901: Accelerated Approval in Rare Disease** (enabling orphan drug applications to take advantage of the accelerated approval pathway originally created during the AIDS epidemic as the new norm for approval).
  - **Section 902: Breakthrough Therapy Designation** (process designed to expedite the development and review of drugs that are intended to treat a serious unmet need)
  - **Section 903: Rare Disease Experts Provision**
  - **Section 905: New Benefit-Risk Framework**
  - **Section 1137: Incorporate Patient Views in Medical Decision Making**
- Additional gains for the rare disease community were made as part of 21st Century Cures Act, passed in 2016:
  - **Additional funding** for NIH and FDA outside of PDUFA structure
  - **Advancing Targeted Therapies for Rare Diseases Act** – allowed use of extrapolated data from previously approved drugs that use the same or similar approach (backbone or chemistry) for drugs that target rare conditions
  - **Expanded Access Policy** – required drug companies to make available its policy regarding expanded access (once approved)
  - **Pediatric Voucher** – Creating Hope Act – continued Priority Review Voucher Program
  - **Patient Focused Impact Assessment Act (PFIA)** – feedback loop from FDA about how reviewers are or are not using patient focused data and created guidance on generating such data. This includes the following:
    - Benefit Risk Data
    - Patient Reported Outcome Data
    - Qualitative Patient Data
RARE DISEASE LOBBY DAY 2020

- Patient Guidance Documents
- Patient Experience Data
- The BENEFIT Act is a logical next step to this journey.

RARE DISEASE LOBBY DAY 2020

Rare Disease Legislative Advocates (RDLA), a program of the EveryLife Foundation for Rare Diseases, brought together patients, caregivers, and others in Washington, DC for a week of events dedicated to empowering patients, families, friends, and healthcare professionals to become legislative advocates. During the week of February 25-28, 2020, rare disease advocates had an opportunity to meet with Members of Congress and to learn about policy updates and best practices for successful advocacy.

Rare disease advocates (myself included) attended the all-day Legislative Conference on February 26th to learn about federal legislation and policies that affect the rare disease community. Policy experts from Capitol Hill, non-profit organizations, and industry shared their expertise with advocates. In addition, advocacy professionals provided opportunities for participants to refine techniques for effective advocacy on the Hill and build strong relationships with their Members of Congress.

On February 27th, over 500 rare disease advocates went to Capitol Hill and met with their Senators, Representatives, and Congressional staffers to discuss key legislation, policies and the Rare Disease Congressional Caucus.
2020 Legislative Asks

Ask #1 - Support Creation of the Rare Disease Center of Excellence (COE) at the U.S. FDA

Background

- A Center of Excellence (COE) within the FDA would elevate rare disease expertise within the agency and help to remove communication barriers across centers and divisions to accelerate and improve the rare disease therapy development process at the U.S. Food and Drug Administration (FDA).
- The FDA already has authority under the 21st Century Cures Act to establish Centers of Excellence.
- Three years ago, the FDA established the first FDA Center of Excellence focused on oncology (the Oncology Center of Excellence) which has been extremely successful in bringing new cancer therapies to patients.
- Two years ago, the EveryLife Foundation hosted a Scientific Workshop on this topic with case studies from patient organizations and industry that demonstrated the value of having such a Center of Excellence.
- Given the challenges and, therefore, the unique expertise needed to advance the development and review of products for rare diseases, innovators have long believed that a Rare Disease Center of Excellence would provide the necessary resources and support to allow patients across FDA to more consistently and efficiently review novel products for these rare conditions.

Vision

- The COE would serve as a consultative and cross-cutting body to build knowledge and expertise, and to consult with review divisions in considering applications for rare disease therapies.
- The COE would identify and address current and emerging challenges and opportunities in rare disease therapy development, including the development of therapies for individual or very small populations.
- The COE would not supplant any authorities held by FDA review divisions.
- The Rare Disease Center of Excellence would also include a dedicated program focused on the many unique needs associated with developing treatments for individual or very small populations (e.g., N of 1).
March XX, 2020

The Honorable Stephen Hahn
Commissioner
Food and Drug Administration
10903 New Hampshire Avenue
Silver Spring, MD 20993

Dear Commissioner Hahn:

We write to encourage the Food and Drug Administration (FDA) to consider establishing a Center of Excellence for Rare Diseases in an effort to improve treatments and health outcomes for those suffering from these conditions.

In December 2016, Congress authorized the FDA to establish Centers of Excellence to coordinate actions to address major diseases in an effort to improve treatments for patients as part of the 21st Century Cures Act. These Centers of Excellence are intended to combine expertise from a range of areas to leverage the FDA’s ability to facilitate the development of life-saving medical therapies.

It is critical that we work to increase the number of safe, effective, and affordable treatments that are available for people with rare diseases. Developing therapies for small patient populations presents unique challenges, and we believe that a Center of Excellence for Rare Diseases could help to improve treatments for some of the patients who need them most. We also recognize that a Center of Excellence for Rare Diseases has the potential to leverage statistical analysis, systems biology, trials design, toxicology, and other areas of expertise across the FDA.

It is for these reasons that we ask you to consider establishing a Center of Excellence for Rare Diseases. We also respectfully request that you provide us with information concerning the expected cost of and timeline for establishing such a Center of Excellence. In addition, we ask that you provide more information on how a Center of Excellence for Rare Diseases could strengthen the FDA’s ability to treat and prevent rare diseases, as well as how the FDA would work with other federal agencies to establish and develop such a Center of Excellence.

Thank you for your consideration of this important matter. We look forward to your response.

Sincerely,

Amy Klobuchar
United States Senator

G.K. Butterfield
Member of Congress

Roger Wicker
United States Senator

Gus M. Bilirakis
Member of Congress
Ask #2 – Enact the Newborn Screening Saves Lives Reauthorization Act, HR2507/S2158

Please Enact S. 2158/ H.R. 2507
Reauthorization of the Newborn Screening Saves Lives Act

Diagnosis through newborn screening saves lives, improves healthcare outcomes, and reduces long term healthcare costs by allowing for detection and intervention at the earliest moment possible.

Please contact Chairman Lamar Alexander (202-224-9616) and Ranking Member Patty Murray (202-242-6211) to share your support.

Background
- In 2008, Congress passed the original Newborn Screening Saves Lives Act (P.L. 110-204), which established national newborn screening guidelines and helped facilitate comprehensive newborn screening in every state. The Act was first reauthorized in 2014.
- Prior to this act, the number and quality of newborn screening tests varied greatly by state.
- In 2007, only 10 states and the District of Columbia required infants to be screened for all of the recommended disorders. Today, all 50 states and the District of Columbia require screening for at least 31 treatable conditions, as recommended by the Department of Health and Human Services.

Current Status of Bill
- Federal newborn screening programs expired on September 30, 2019.
- The House has done its job, passing the Newborn Screening Saves Lives Reauthorization Act in July 2019. Currently, the bill is held up in the Senate.
- The Senate bill is held up due to a proposed amendment that would require parents to opt-in to allow their newborn’s unidentifiable dried blood spot (DBS) to be used for research, which would break down the entire newborn screening system.
- Public health laboratories and scientific researchers need DBS to conduct life-saving research to improve the current tests and work to develop new treatments for the thousands of rare diseases still without a cure.
- Complying with this amendment would place a high burden on hospitals that would likely choose not to participate in collection of DBS. Studies have demonstrated that 90-99% of parents would choose to opt-in, but at times only half of parents would be asked by hospital staff due to the compliance burden.

Key Bill Provisions
- Reauthorizes the Health Resources and Services Administration (HRSA) state grants to expand and improve screening programs, provide educational resources to parents and health care providers, and improve follow-up care for infants with a detected condition.
- Reauthorizes the Secretary’s Advisory Committee on Heritable Disorders in Newborns and Children, which provides states with a Recommended Uniform Screening Panel (RUSP), helping to ensure every infant is screened for conditions that have a known treatment.

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Please contact Shannon von Felden (vonfelden@caregroupons.org) to learn more about RDLA.
Newborn Screening Facts

- Of the four million babies born in the U.S. each year, one in 300 are found to have a severely
devastating condition through newborn screening.
- 12,000 newborns benefit from the early detection and delivery of life-saving treatments.
- Newborn Screening is the practice of testing every newborn for certain genetic, metabolic, hormonal,
and behavioral conditions that are not otherwise apparent at birth.
- Diagnosed through newborn screening never lives, improves health outcomes, and reduces long-term
healthcare costs by allowing for detection and intervention at the earliest moment possible.
- Newborn screening is the most cost-effective public health program in the history of our country.
Legislative Ask #3 – Support Increased Funding in the US FDA Orphan Products Clinical Trial Grants Program and the Natural History Grants Program

**Fiscal Year 2021 Appropriations Request**

Bill: Agriculture, Rural Development, Food and Drug Administration, and Related Agencies
Sections: Food and Drug Administration, Office of the Commissioner, Office of Orphan Products Development

**PLEASE INCLUDE SUPPORT FOR THESE PROGRAMS IN YOUR APPROPRIATIONS REQUEST TO THE AGRICULTURE, RURAL DEVELOPMENT, FDA APPROPRIATIONS SUBCOMMITTEE**

**Background**

- Thirty million Americans suffer from a rare disease, making it a public health crisis.
- 50% of rare disease patients are children, 30% of these children will not live to see their fifth birthday.
- 92% of the 7,000 known rare diseases have no U.S. Food and Drug Administration (FDA)-approved therapy.
- Due to the small patient population of any individual rare disease it is often not financially viable for private sector investment in research and therapy development.
- The Office of Orphan Products Development (OOPD) within the FDA administers provisions of the Orphan Drug Act to promote development of therapies for rare diseases patients. OOPD manages two vital and successful grant programs - the Orphan Product Clinical Trial Grants Program and the Natural History Grants Program. These programs help to bring therapies to patients that save lives and lower long-term healthcare costs. Clinical trial cost increases have not been matched by increases in FDA-appropriated grant funds. The capacity of the programs to provide support to clinical trials that will lead to life-saving and life-improving therapies for patients with rare diseases has been significantly reduced.

**Programs**

**Orphan Products Clinical Trial Grants Program**

This program supports new and continuing extramural research projects that test the safety and efficacy of promising new drugs, biologics, devices, and medical foods through human clinical trials in extremely vulnerable populations often with life-threatening conditions. Over 700 new clinical trials have been funded through this program to date. Orphan Products Clinical Trial Grants have supported the marketing approval of more than 60 orphan products for serious or life-threatening orphan indications. In FY 2018, OOPD received 79 clinical trial applications but was only able to fund 12 new grants.

**Natural History Grants Program**

This grant program supports studies that advance rare disease therapy development through characterization of the natural history of rare diseases and development and/or validation of clinical outcome measures. The natural history of a disease is the course a disease takes from its onset to a final outcome in the absence of treatment. This information isextremely valuable to assessing the efficacy of a therapy in comparison to natural history and is often lacking for rare diseases. OOPD received 89 applications in the first cycle of this program, but was only able to fund 13 new grants.

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*Please contact RDLA via web (rarediseaselegislative.org) to learn more about RDLA.*
Legislative Ask #4 – Join the Rare Disease Congressional Caucus

Please Join the Rare Disease Congressional Caucus

The bipartisan and bicameral Rare Disease Congressional Caucus is led by Representatives G.K. Butterfield (D-NC) and Gus Bilirakis (R-FL), and Senators Roger Wicker (R-MS) and Amy Klobuchar (D-MN) to promote awareness of rare disease issues.

Background: There are more than 7,000 known rare diseases that together affect more than 30 million Americans and their families. One in ten Americans has a rare disease. Rare or orphan diseases are defined as diseases affecting fewer than 200,000 people in the U.S. More than 90% of rare diseases are considered ultra-rare, affecting fewer than 6,000 people; some affect fewer than 100 people. Rare diseases include rare cancers, tropical or neglected diseases, genetic diseases and many pediatric diseases including cancers. Many of these diseases are life-threatening and have no treatment options.

The Orphan Drug Act was enacted in 1983 to encourage pharmaceutical companies to develop drugs for diseases that have relatively small patient populations. Despite the success of the Orphan Drug Act, there have been fewer than 700 treatments for less than 550 diseases approved for marketing by the Food and Drug Administration (FDA) in the last 30 years.

The science exists for many of these diseases to be treated; however, treatments may never be developed because of roadblocks in the development process, such as a lack of investment and a challenging regulatory environment. Additionally, while relatively few treatments have become available, patients struggle with insurance companies and government programs to afford these lifesaving treatments.

Solution: The Rare Disease Congressional Caucus helps bring public and Congressional awareness to the unique needs of the rare disease community (including patients, physicians, scientists, and industry), and creates opportunities to address roadblocks to the development and access to crucial treatments. The Caucus gives a permanent voice to the rare disease community on Capitol Hill. Working together, we can find solutions that turn hope into therapies and cures.

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Contact: RDLA: vonfelden@curetheprocess.org; Senator Wicker’s office: Sally_Farrington@wicker.senate.gov or Rep. Butterfield’s office: Caitlin.VanSant@house.mail.gov
Senator Co-Chairs: Senators Roger Wicker (MS) and Amy Klobuchar (MN)  
House Co-Chairs: Representatives G.K. Butterfield (NC-1) and Gus Bilirakis (FL-12)

HOUSE:  
Mark Amodei NV-2  
Chad Hays AZ-6  
Andy Sanfilippo NY-6  
Joyce Beatty OH-13  
Anita Davis CA-9  
Tedd P. Edwards TX-25  
Donovan Brown, Jr. VA-8  
Gus M. Bilirakis FL-12  
Sanford Bishop, Jr. GA-2  
Lisa Blunt Rochester DC  
Suzanne Benjamin OR-4  
Mo Brooks AL-5  
Julie Benmlash CA-20  
Vera H. Hagen FL-15  
Michael Burgess TX-26  
Cheri Bustos IL-7  
G.A. Davis NC-1  
Sandra Jacobs CA-54  
André Carson IN-7  
John Conyers, Jr. MI-11  
Sara Carnahan IL-6  
Steve Chabot OH-1  
Jody Hice GA-37  
David Cicilline RI-1  
Lucy C. McBath GA-13  
Steve Cohen TN-9  
James Cooper KY-1  
Glenda Collins GA-53  
Jim Cooper KY-1  
Danny Davis TX-23  
Rodney Davis IL-13  
Peter DeFazio OR-4  
Katie DelBene WA-1  
Rosa DeLauro CT-10  
Ted Deutch FL-21  
Bobbie Ewing, Jr. MS-2  
Mike Doyle PA-14  
Chuck Estes NY-16  
Amy Eshleman CA-18  
Steve Fahringer IA-1  
John Faso NY-18  
Hale"ie Eimerman NH-1  
Vansyngroh Maiy AB-6  
Seth Moulton MA-6  
Richard Neal MA-1  
Donald Norcross NJ-1  
Evan Holsten New Jersey DC  
Steven Palazzo NY-5  
Paul Pallucco NJ-6  
Jeremy Peters CA-20  
Chris Pappas NH-4  
Bill Pascrell NJ-9  
Donald Payne, Jr. NJ-10  
Scott Perry PA-5  
Collin Peterson MN-7  
Chellie Pingree ME-1  
Bill Posey FL-8  
David Price NC-4  
Mike Quigley IL-5  
Jamie Raskin MD-8  
Kathleen Rice NY-4  
Mark Rose NC-11  
David Rouzer NC-7  
C.A. Dutch Ruppersberger MD-2  
John Sarbanes MD-7  
John Sarbanes MD-7  
Brad Scherer IL-11  
David Scott GA-13  
Mikie Sherrill NJ-11  
Mike Thompson IL-2  
Alvin B. Sáenz TX-30  
Beto O'Rourke TX-13  
Lindsey Graham SC-9  
Trey Hollingsworth IN-9  
Earl Blumenauer OR-7  
Catherine Twitchell PA-8  
Will Hurd TX-22  
Mark Murphy MA-7  
Fred Upton MI-8  
Jared Viger HT-14  
Nydia Velázquez NY-7  
Pete Visclosky IN-3  
Ann Wagner MO-2  
Greg Walden OR-2  
Debbie Wasserman Schultz FL-23  
Samantha Warren Vermont VT-1  
Lucy Wadsworth VT-2  
John Yarmuth KY-3  
Jesse White WV-4  
Lacy Zeigler WV-1  

SENATE:  
John Bar stool WV  
John Boozman AR  
Noël Corrigan WA  
Stacy Davis Capote WY  
Christopher Coons DE  
Tom Cotton AR  
John Hoeven ND  
Cheryl Hyde-Smith MS  
James Inhofe OK  
John Kennedy LA  
Angus King ME  
Amy Klobuchar MN  
Edward Markey MA  
Jeff Merkley OR  
Gary Peters MI  
James Risch ID  
Kyrsten Sinema AZ  
Jeanne Shaheen NH  
Tom Smith MN  
Debbie Stabenow MI  
Chris Van Hollen MD  
Roger Wicker MS  

RARE DISEASE LEGISLATIVE ADVOCATES (A PROGRAM OF THE EVERY LITTLE FOUNDATION) 
1612 14TH STREET NW, SUITE 500, WASHINGTON DC 20005 
PHONE: 202-499-RAKL (7273) WWW.RAILADVOCATES.ORG @RAILADVOCATES
Legislative ask #5 – Support the Better Empowerment Now to Enhance Framework and Improve Treatments (BENEFIT) Act

While much progress has been made on incorporating patient experience data in the drug approval process through FDA implementation of previous legislative efforts, some significant gaps remain. One such gap is the lack of any requirement in law today that the FDA include, as part of its benefit-risk framework, any patient experience or patient-focused drug development (PFDD) data. This means that the agency’s signature tool for evaluating benefit-risk does not have to include data from the patient perspective that could be critical to informing the agency’s evaluation and, ultimately, decision on whether or not to approve a product.

To address this gap, Senators Roger Wicker (R-MS) and Amy Klobuchar (D-MN) introduced the Better Empowerment Now to Enhance Framework and Improve Treatments or the BENEFIT Act (S. 1057) which passed in the Senate in August of 2017.

This legislation would amend the Food, Drug, and Cosmetic Act (FDCA) to ensure that patient experience, PFDD, and related data — including information developed by a product sponsor or a third party such as a patient advocacy organization or academic institution — be considered as part of the benefit-risk assessment. This action would send an important signal to all stakeholders that patient experience and PFDD data will be fully incorporated into the agency’s review process and would encourage such entities to develop scientifically rigorous and meaningful tools and data.

The BENEFIT Act would also enhance an important transparency and accountability provision from the 21st Century Cures Act by requiring the FDA to say how such patient experience and PFDD data was considered within the benefit-risk assessment for any approved therapies. This will provide additional learning to all stakeholders, particularly patients, and help further refine and develop such tools going forward. PPMD is now working with Congressional champions to reintroduce in the Senate and introduce a companion bill in the House.
Legislative ask #6 – Co-Sponsor the Medical Nutrition Equity Act, HR 2501

Specialized medical nutrition is medically necessary for the safe and effective management of digestive and inherited metabolic disorders. Medical foods are formulated to be consumed as a standard of care for certain digestive conditions and diseases (e.g., low-protein modified foods for those with PKU). The cost of medical foods are ~8X cost of grocery store foods. They must be ordered through a doctor and accessed through special manufacturers. The out of pocket costs are ~$2500 for infants and ~$25000 per year for a grown man or pregnant woman. Most fall through cracks in coverage, as coverage sometimes depends on route of administration (e.g., tube feeding is covered, enterally is not). There are currently 56 co-sponsors on this bill. Need to ask Representative and Senators to sign on as co-sponsor and introduce respectively.
PEOPLE WITH PKU NEED MEDICAL NUTRITION TO THRIVE

Thousands of children and adults in the United States live with inborn errors of metabolism (IEMs) that prevent their bodies from properly metabolizing and absorbing normal, everyday food. For these patients, medical nutrition is the primary treatment for the effective management of these conditions. Unfortunately, many health insurance plans in the United States do not provide reimbursement for medical nutrition despite their proven efficacy in the treatment of IEMs, causing medical nutrition therapy to be cost-prohibitive for many patients.

Necessity of Medical Nutrition for PKU:

- For more than 50 years, the United States, early medical nutrition intervention has resulted in near normal or normal development of individuals with PKU.
- Without access to medical nutrition, children with PKU can lose 4 IQ points each month and will suffer severe and irreversible intellectual disabilities before reaching adulthood.
- Adults who are not on treatment experience severe developmental, behavioral, and mental health consequences that result in difficulties in school, work, and relationships.
- Children carried by women with poorly-controlled PKU may have maternal PKU syndrome which causes small brains, intellectual disabilities, birth defects of the heart and low birth weight.

Cost of Medical Nutrition:

- While medical nutrition is medically essential for PKU patients, it is not uniformly reimbursed by health insurance, creating a massive financial barrier in accessing treatment for many patients.
- The cost of correct care for medical nutrition products is up to 8 times the cost of normal groceries. i.e.:
  - Metabolic Formula: ≥ $300/00 (per case of 6)
  - Low-protein modified foods:
    - Loose of Breast: $13.99
    - Box of Pasta: $11.49
    - Unbaked Slices: $3.40

The Medical Nutrition Equity Act of 2019 (H.R. 2510) provides for the coverage of medical formula and low-protein modified foods, as well as individual amino acids for children and adults with PKU and other metabolic disorders under Federal health programs and private insurance.

The Medical Nutrition Equity Act currently has 56 cosponsors with bipartisan support.

For more information, contact Kylie Barber at kbarber@eur-theprocess.org

Please contact the offices of Representative Mc Govern or Representative Herrera Beutler to co-sponsor HR 2510, or Senator Gary to become an original sponsor of the Senate bill.
HOW COVERAGE OF MEDICAL FOODS FOR INBORN ERRORS OF METABOLISM (IEM) CAN SAVE LIVES & COSTS

Inborn Errors of Metabolism (IEMs) are metabolic conditions in which specific enzymes defects interfere with the normal metabolism of protein, carbohydrate, or fat. Federal newborn screening policy and state screening programs identify this majority of IEMs. Medical nutrition intervention is a mainstay of patient management and must begin shortly after birth to prevent death, mental retardation, and other adverse health outcomes.

Individually, IEMs are rare. For example:
- Glutaric acidemia type 1 (GA-1) occurs in 1 in 90,000 live births
- Very Long-Chain Acyl-CoA Dehydrogenase (VLAD) Deficiency 1 in 63,000 live births
- Phenylketonuria (PKU): 1 in 10,000 live births

There are two types of IEMs: diet-treated with medical food, foods modified to be low in protein, and/or supplemented with individual amino acids, and drug treated by enzyme replacement or substrate inhibition.

The consequences of not treating these conditions are devastating. For example:
- GA-1: Metabolism can be fatal. For survivors, irreversible brain damage that can affect the ability to walk, talk, or even see or hear, often with uncontrollable, painful movements called dystonia.
- VLAD: Metabolic crisis that can be fatal. For survivors, poor growth, liver failure, heart failure, and episodes of painful muscle breakdown called rhabdomyolysis that can cause kidney failure.
- PKU: irreversible cognitive impairment, hyperactivity, autistic behavior, autism.
- Methylmalonic acidemia (MMA): Affected infants of women with prenatally tested PKU and may include microcephaly (small head), irreversible intellectual disabilities, congenital heart defects and other birth defects, and low birth weight.
- Homocystinuria (HCY): Strokes that can be fatal. For survivors, paralysis, cognitive impairment, sensorial vision because of dislocated lenses, scoliosis, and skeletal deformities.

The annual total cost to treat IEM with medical nutrition ranges from $2.26k for an infant to almost $28,000 for an adult male or pregnant woman. Without coverage, treatment is unaffordable for the majority of patients. However, the cost of NDI, providing accessible and appropriate treatment for these patients is much greater.

The lack of medical nutrition coverage across the United States has been, and continues to be, detrimental to individuals with IEM, their families, and society. In 2009, the Newborn Screening Saves Lives Act passed with overwhelming support. The Medical Nutrition Equity Act is the best way to achieve the aims of that act: healthy lives for those with IEM.

Please contact the offices of Representative McGovern or Representative Herrera Beutler to co-sponsor HR 2301, or Senator Casey to become an original sponsor of the Senate bill.
THE LOW-PROTEIN DIET FOR PKU & OTHER METABOLIC DISORDERS IS EXPENSIVE & DIFFICULT TO MANAGE

Medical formula must be re-authorized regularly and ordered monthly. Low-protein foods must be specially manufactured, ordered online, and shipped. They cost on average 5 TIMES MORE than regular products.

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<td>Metabolic Formula</td>
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<td>Primary Source of Nutrition</td>
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<tr>
<td>Loaf of Bread</td>
<td>$1.99</td>
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<tr>
<td>Box of Rice</td>
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<tr>
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<td>$9.99</td>
</tr>
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Please contact the offices of Representative McGovern or Representative Herrera Beutler to co-sponsor HR 2501, or Senator Casey to become an original sponsor of the Senate bill.
MEDICAL NUTRITION HELPS PEOPLE SURVIVE AND THRIVE

A TIMELINE OF PKU TREATMENT

1934: PKU was discovered
1850s: The low-protein diet for PKU was developed, providing effective and efficient treatment
1960s: Newborn screening was implemented throughout the United States to ensure diagnosis in infancy
1990s: Diet for Life becomes the medical standard practice to ensure lifelong protection of the body and brain
2020: People with PKU and similar metabolic disorders are still fighting for coverage for the primary (and often only) treatment for their disorders.

The Medical Nutrition Equity Act

- Levels the playing field
- Closes the loopholes in state laws which include restrictions based on:
  - Age
  - Diagnosis
  - Formula Delivery Method
  - Income
  - Gender
  - Insurance type
- Ensures medical nutrition equity for all

Please contact the offices of Representative McGovern or Representative Winners Boulter to co-sponsor HR 2304, or Senator Casey to become an original sponsor of the Senate bill.
H.R. 4393, Advancing Access to Precision Medicine Act

This legislation would ensure that many children and young adults suffering from an undiagnosed condition will have access to DNA sequencing clinical services that are currently out of reach.

For more information or to connect, please contact Adeola Adeyina, Office of Representative Eric Swalwell (CA) at Adeola.Adeyina@mail.house.gov.

Background
- There are more than 7,000 rare disorders that together affect more than 30 million Americans and their families.
- Approximately 50% of people affected by rare diseases are children, and 30% of those children will not live to see their 5th birthday.
- On average, rare disease patients will see more than 10 specialists and have been misdiagnosed 3 times before receiving an accurate diagnosis.
- This diagnostic odyssey takes an average of 8 years. The current system is an unacceptable reality.

Summary
- H.R. 4393 would encourage coverage of DNA sequencing clinical services through an increase in the Federal Medical Assistance Percentage (FMAP) for state Medicaid plans.
- The bill would also request the National Academy of Medicine to conduct a study on this expanded coverage to understand how such coverage may improve care and reduce health disparities, and how the federal government may reduce barriers to testing. This proposed study would examine potential cost savings resulting from expanded services and bring the nation’s diagnostic infrastructure into the 21st century.
- This legislation would cover “DNA sequencing” services instead of just “whole genome sequencing” (an update from the previous version) which is essential to ensuring appropriate access to care.
- The decision of which diagnostic tool is most appropriate should be made by patients and their physicians; not based on what is or is not covered by Medicaid.
- Diagnostic delays not only costs lives, but often lead to significantly higher treatment costs that would have been unnecessary with a timely diagnosis.

Rare Disease Legislative Advocates (RDLA) is a program of the EveryLife Foundation for Rare Diseases designed 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 policymakers.

Please contact Shannon von Felten (vonfelten@caretheiraccess.com) to learn more about RDLA.
Rare Diseases

In the United States, a disease is considered rare if it affects fewer than 200,000 people. It is estimated that 1 in 10 Americans, and 300 to 400 million people worldwide, are living with a rare disease. For the majority of those individuals, no treatment options are available.\(^1\)\(^2\)

**TODAY**

- 95% of rare diseases lack any FDA-approved treatment.\(^4\)
- Approximately 7,000 rare diseases and disorders have been identified to date.\(^1\)
- 50% of those with a rare disease are children.\(^1\)
- 80% of rare diseases are genetic in origin.\(^1\)

- It takes 18% longer to develop an orphan drug compared to medicines for more common conditions.\(^1\)
- Nearly 1/3 of children with a rare disease die before the age of 5.\(^1\)

**COST**

Each rare disease patient spends an average of $147,000 annually for treatment.\(^8\)

- Only 55% of rare disease caregivers with household incomes under $50,000 are employed, with 42% reporting having only fair or poor physical health themselves.\(^9\)
- 3 out of every 4 rare disease caregivers worry about their family’s ability to pay for care.\(^6\)

Research Delivers Solutions

The international research effort to sequence all human genes, known as the Human Genome Project, led to the identification of the precise genetic cause of many rare diseases. This knowledge has led to breakthroughs in treatment, symptom management, and even cures. For example, a 2016 study used genetic sequencing to provide a clinical diagnosis for 42% of patients with persistent abnormalities in brain motor function whose conditions had previously gone undiagnosed using standard methodologies.\(^4\)\(^5\)

A rare disease known as homozygous familial hypercholesterolemia (HoFH) is a life-threatening condition that prevents the body from removing "bad cholesterol." Individuals with untreated HoFH often die before the age of 30. A new treatment option, Evolocumab, has been shown to reduce levels of "bad cholesterol" among HoFH patients by 60% on average, greatly decreasing mortality risk and improving overall health.\(^9\)

Severe combined immune deficiency (SCID) is a rare and fatal immune disorder that causes death before an infant's second birthday. As a result of research-based newborn screening, children with SCID can be diagnosed and treated within 3 months of birth, a period during which they are still protected by their mother’s immune cells. Cost-benefit analyses estimate that every dollar invested in newborn screening for SCID produces $10 in economic and societal benefits.\(^9\)

Majority Favor Doubling Funding for Medical Research Over the Next Five Years

Do you favor or oppose doubling federal spending on medical research over the next five years?

Source: AARP Research and AARP Public Policy Institute, October 2019
Rare Diseases

Then. Now. Imagine.

THEN
In the early 1980s, 20-25 million people in the U.S. were affected by approximately 5,000 rare diseases, and there were only 10 drugs available for treatment.\(^1\)\(^2\)

NOW
The Orphan Drug Act was passed in 1983, creating the orphan drug designation and providing needed incentives for researchers and manufacturers to develop therapies for rare diseases. Since then, the FDA has approved over 500 orphan drugs. In 2016 alone, 30 rare disease indications were approved and 34 novel treatments for rare diseases were approved – 58% of all 2016 FDA drug approvals.\(^3\)

Imagine
A cure for all rare diseases.

Quest for Diagnosis

5.6 to 7.6:
That's the average number of years it takes to correctly diagnose a rare disease patient in the U.S.\(^4\)

25% of patients with the most “common” rare diseases wait between 6 and 36 years to receive a correct diagnosis, with 40% receiving an incorrect initial diagnosis.\(^5\)

Number of Conditions Tested As Part of Newborn Screening, 2018

Newborn Screening

Newborn screening allows for the detection of numerous rare diseases, such as SCD, phenylketonuria, and cystic fibrosis. Early diagnosis leads to better treatment and care, which can lead to much better health outcomes for these infants.

\(^5\) The Albert and Mary Lasker Foundation is a founding partner in this series of fact sheets. www.laskerfoundation.org
Meetings with Members of Congress on February 27, 2020

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<td>Roberto Sada</td>
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<td>Senator Bob Menendez**</td>
<td>Diane Adamson</td>
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<td>Senator Cory Booker</td>
<td>Juan Gomez</td>
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* My district and also the district from which most of the advocates reside (4 in total)
** Served as Team Lead for meeting

Representative Frank Pallone Jr. is the Chairman of the House Energy and Commerce Committee, which has jurisdiction over issues pertaining to energy, environment, health care, commerce, and telecommunications. His staffer, Roberto Sada, indicated that it may present a conflict if Rep. Pallone were to sign the letter to the FDA, but is otherwise very supportive of the idea of it and all the other asks. Rep. Pallone also led Democratic negotiations in the House on the 21st Century Cures Act signed into law in Dec 2016.

Representative Josh Gottheimer shared with us that his mother passed away 18 months ago from a rare disease (sarcoidosis), thus we have his full support on these causes.

Representative Tom Malinowski also stated his support in person, while his staffer mentioned the same before he joined us mid-meeting.
What Can Members of NNPDF Community Do?

WRITE YOUR MEMBERS OF CONGRESS TO ALSO ASK FOR SUPPORT OF THESE CAUSES

There is strength in numbers! Members of NNPDF can use the RDLA link to easily send a letter to their Senators asking them to support and/or Co-sponsor the Newborn Screening Saves Lives Reauthorization Act (S. 2158): https://www.votervoice.net/mobile/EveryLife/campaigns/62708/respond

EveryLife makes this very easy. Once you put in your address, it automatically finds your Senators and Representative. There is a letter already drafted and has an optional section where you can personalize it if you like.

It is also worthwhile asking them to sign the letter to have the FDA form a Center of Excellence for Rare Diseases and for their Representatives to co-sponsor the HR2501 Medical Nutrition Equity Act and Senators to sponsor and introduce a similar act in the Senate.
Pictures

2020 Rare Disease Legislative Advocates
Margo Frey and Anne O’Connor-Smith, sisters diagnosed with ASMD (Niemann-Pick Disease Type B) at the Research America Evening Reception
The NNPDF Crew at the Research America Evening Reception
Representative Mikie Sherrill’s office
Representative Frank Pallone Jr.'s office
Representative Tom Malinowski’s office
Senator Bob Menendez’s office
Senator Cory Booker's office