Galactosemia Foundation

Summer 2018

GALACTOSEMIA GAZETTE

INSIDE THIS ISSUE:

We welcome feedback on the Galactosemia Gazette. Please reach us anytime at newsletter@galactosemia.org.

Galactosemia Foundation
Linked for Life.

PRESIDENT KELLIE WILCOX'S MESSAGE

Greetings, Galactosemia Community!

The Colorado Galactosemia Foundation Conference is only weeks away and if you are still on the fence about the value of attending...then let me convince you! Starting on Thursday during regular and youth registration there will be many fun activities. We are fortune enough to have the Galactosemia Foundation 2018 app again this year, donated by Double Dutch, for which we are very grateful. On the app we will again have the scavenger hunt and a Rock Quest! During registration come paint and seal your rocks in whatever wonderful design you can come up with, let them dry while you participate in the other activities, then come back and pick them up. Hide them on the main level of the hotel (inside or out), post a picture on the app as a hint to where they are hidden, find another rock and keep it going! Remember to take home your souvenir rocks at the end of the conference! There will be a PRE-hidden prize rock.

Activities on Thursday include a 25 ft. Rock Wall, Mysto the Magician, and in true Climbing to New Heights Conference theme, we will have a Camp Counselor with games for all! During all the activities, researchers Dr. Berry, Dr. Rubio and Dr. Judy will be conducting studies on site and our very own Research committee will be recruiting for the 2018-19 year. This will all be in the same area as registration, Silent Auction, Sales and our vendor booths. Unfortunately, David Levine sold Illinois Nut Company and will not be joining us this year BUT Denver, Colorado is home of VooDoo Donuts which has Vegan donuts and will be the main attraction for our Sweet Treat Thursday! WOW, and that's just the start!

Friday's General Session will be one you don't want to miss, and we have new breakout sessions from speakers and companies we have never heard from before. Friday night, for the second year in a row, is the exciting Talent Together show put on by our very talented kids AND it will be the close of the Silent Auction! Don't miss this!

Saturday, join the general session for important updates and Galactosemia speakers, more breakouts and Saturday evening will be the ever-anticipated Dance with live DJ, photo booth and fun all the way until midnight! The GG and GForce coordinators have some great speakers lined up for these groups in addition to daily trips into the city for some fun! See you all in a few weeks!!

Keynote Speaker: Jamie Siminoff

By Galctosemia Conference Committee

There has been much discussion on the education, research and fun sessions that will form part of this year's conference. We also wanted to take a moment to highlight the keynote speaker. This year we are pleased to have tech CEO Jamie Siminoff joining the Foundation Conference.

Jamie founded Your First Step International, Inc., a company that assisted entrepreneurs in bringing ideas from concept to fruition, while obtaining a Bachelor of Science in Entrepreneurship from Babson College. In 2000, the company pivoted to building and operating global wholesale Voice over IP networks in developing countries.

In 2005 – perplexed by the archaic nature of voicemail – Jamie founded the world's first voicemail-to-text company, PhoneTag. In September of 2009,PhoneTag sold to Ditech Networks. In 2010, Jamie took on email and founded Unsubscribe.com to help email users clean commercial email from their inboxes. In 2011 Unsubscribe.com was sold to personal security company, TrustedID.

Jamie now works as Chief Inventor and CEO at Ring, the world's first battery operated smart doorbell. The company's mission is to reduce crime in neighborhoods by empowering homeowners, and Jamie and his team are working on making communities a safer place. When he's not working on Ring, he enjoys spending time with his wife, eight-year-old son (who has classic Galactosemia), and dogs – Short Rib and Pancake.

Don't Miss the Silent Auction!

Silent Auction

By Kellie Wilcox

This will be the BEST Silent Auction we have had yet! All items will be displayed starting Thursday night at registration with opening bids beginning on Friday morning. Items will be displayed and open for bid all day Friday in the same location. Items will be displayed Friday before the talent show starts; the auction will end during the intermission of the Talent Together Social. You will be able to pick up your items after the Talent show Friday night or on Saturday.

Please stop by and see what we have. THERE IS TRULY SOMETHING FOR EVERYONE!!!!

Everything from a Cape Cod vacation, little girls' jewelry, flip flops, Summer Drink Basket and new electronics.

Text or call for donation drop off—Kellie Wilcox 903.539.8815

We are also having a 50/50 Raffle. Tickets will be available at the sales table and the three winners will be announced Saturday night during the dance! New this year is a WINE PULL of donated wines from local area breweries and wineries!

THE 2018 CONFERENCE APP IS LIVE! CHECK SCHEDULES, POST EVENT PHOTOS, AND MORE!

https://dl.doubledutch.me/download.aspx?appId=f970ae52-c10c-4757-b886-39ec72c91cb1&region=us

TREASURER'S REPORT

Statement of Activity

January 1 - May 20, 2018

	Total
REVENUE	
Child Care Registration	9,005.00
Conference Registration	22,699.00
G-Groups Registration	
AGERS	796.00
G-Force Registration	5,224.00
GG Registration	4,627.00
Total G-Groups Registration	**10,647.00**
Paul P Scholarship	600.00
Sales	33.95
Sales of Product Revenue	899.19
Temporarily Restricted Donations	4,975.00
Unrestricted Donations	64,022.91
Total Revenue	**112,882.05**
COST OF GOODS SOLD	
Cost of Goods Sold	
Child Care Expenses	8,748.00
Conference Entertainment	5,530.00
Freight & delivery - COS	567.19
Insurance-Conference	410.00
Total Cost of Goods Sold	**15,255.19**
Total Cost of Goods Sold	**15,255.19**
GROSS PROFIT	**97,626.86**
EXPENDITURES	
Bank Charges	180.30
PayPal Fees	354.46
Stripe Fees	73.82
Total Bank Charges	**608.58**
Dues & Subscriptions	700.00
Telephone	154.70
Website Hosting	192.00
Total Expenditures	**1,655.28**
NET OPERATING REVENUE	**95,971.58**
OTHER REVENUE	
Interest Earned	5.98
Total Other Revenue	**5.98**
NET OTHER REVENUE	**5.98**
NET REVENUE	**$95,977.56**

Statement of Financial Position

As of May 20, 2018

	Total
ASSETS	
Current Assets	
Bank Accounts	
BUSINESS CHECKING (XXXXXX 3329)	318,870.52
EventBright	2,911.18
PayPal Bank	6,104.95
Research Fund (XXXXXX 6770)	6,268.32
Scholarship Fund (XXXXXX 6788)	3,286.49
Stripe Account	23.97
Total Bank Accounts	**337,465.43**
Other Current Assets	
Undeposited Funds	14,275.78
Total Other Current Assets	**14,275.78**
Total Current Assets	**351,741.21**
TOTAL ASSETS	**$351,741.21**
LIABILITIES AND EQUITY	
Liabilities	
Current Liabilities	
Accounts Payable	
Accounts Payable (A/P)	12,817.00
Total Accounts Payable	**12,817.00**
Total Current Liabilities	**12,817.00**
Total Liabilities	**12,817.00**
Equity	
Retained Earnings	242,946.65
Net Revenue	95,977.56
Total Equity	**338,924.21**
TOTAL LIABILITIES AND EQUITY	**$351,741.21**

Like us on Facebook for information, events, and support!

Galactosemia Foundation
Linked for Life.

Scan this QR code for all the latest information about the Galactosemia Foundation

Applied Theraputics to Attend GF Conference

By: Galactosemia Foundation

Applied Therapeutics is a privately held biotechnology company, focused on developing transformative drugs in areas of high unmet medical need. They are currently collaborating on projects with Emory University. Galactosemia Foundation approved their request to attend and have an information table at the July 2018 conference. They are excited to meet our community and interact with our members. Please see below their press release from Rare Disease Day 2018.

APPLIED THERAPEUTICS ANNOUNCES SUPPORT FOR RARE DISEASE DAY 2018

INITIATES GALACTOSEMIA RESEARCH COLLABORATION WITH EMORY UNIVERSITY SCHOOL OF MEDICINE

NEW YORK, N.Y. – February 20, 2018 - Applied Therapeutics Inc., a privately-held biotechnology company focused on developing transformative drugs in areas of high unmet medical need, today announced its support of Rare Disease Day 2018. Rare Disease Day is an international campaign organized by the European Organisation for Rare Diseases (EURORDIS), which unites the community in an effort to improve the awareness, diagnosis and treatment of rare diseases.

February 28, 2018 marks the 11th International Rare Disease Day. This year's theme, "Research," emphasizes the importance of rare disease research toward the development of diagnostic tools, treatments and cures, as well as improved health and social care for patients and their families. In celebration of this theme, Applied Therapeutics is pleased to announce a research collaboration with Emory University to develop treatments for galactosemia.

Galactosemia is a rare genetic disease that affects the body's ability to metabolize galactose, a sugar produced at low levels in all human cells that is also found at high levels in milk and dairy products. Currently, no treatments exist for galactosemia, which is fatal in infants if undiagnosed; early identification through newborn screening and prompt initiation of a restricted diet can prevent the most severe consequences of the disease. However, despite strict dietary control, patients with galactosemia often suffer from serious long-term consequences of disease. These complications, which frequently include cataracts, cognitive, speech, and behavioral disabilities, neurological and motor problems, and premature ovarian insufficiency in women, significantly diminish patients' health and quality of life.

Applied Therapeutics recognizes the urgent need to develop safe and effective treatments to prevent or reverse consequences of galactosemia, and is committed to supporting research that will drive forward a more precise understanding of the disease's complications and potential therapeutic targets. This effort will benefit from the contributions of scientific and clinical researchers across the globe, including Applied Therapeutics' collaborators at Emory University, who are working to determine the exact causes of long-term galactosemia complications.

"Newborn screening has greatly prevented galactosemia fatalities, but the long-term consequences of disease persist and have a profound effect on patient quality of life," said Shoshana Shendelman, Ph.D., Founder, Chairman and CEO of Applied Therapeutics. "We are proud to announce our research collaboration with Emory, and hope that through this research we can, for the first time, bring therapies to galactosemia patients that will prevent disease complications and improve quality of life."

Cont'd Applied Theraputics

"The long-term complications of galactosemia in children and adults have been recognized for decades, but the mechanism remains unclear," said Judith Fridovich-Keil, Ph.D., Professor of Human Genetics at Emory University School of Medicine. "We believe that by identifying the specific metabolites and pathways responsible for these complications, we may be able to intervene—either by preventing the metabolites from forming, or compensating elsewhere in the pathway. We are excited about our collaboration with Applied Therapeutics and are hopeful that our work together will provide new therapeutic options for patients with galactosemia."

For more information about galactosemia, please visit the Galactosemia Foundation website or the National Organization for Rare Disease (NORD) website.

About Applied Therapeutics Inc.
Applied Therapeutics is a privately held biotechnology company, focused on developing transformative drugs in areas of high unmet medical need – fatal or debilitating diseases for which no therapies are approved. The company applies cutting-edge technology to validated drug targets that have failed to produce meaningful therapies in the past. Applied Therapeutics believes that through innovative science, millions of lives can be saved. For more information, visit www.appliedtherapeutics.com.

About Galactosemia
Galactosemia is a rare genetic metabolic disease that affects the body's ability to convert galactose to glucose. The incidence of galactosemia is estimated to be between 1/30,000 to 1/60,000 in the US, and varies worldwide based on ethnicity.

Newborn screening now identifies almost all infants with classic galactosemia born in the US and many other countries, enabling early dietary intervention to restrict galactose exposure—generally by switching the baby from milk to a soy-based formula. Dietary intervention prevents potentially lethal acute symptoms of the disease, but fails to prevent the many long-term complications that can develop later in childhood and persist through adulthood. The most common complications currently recognized include cataracts, cognitive, behavioral, and speech problems, motor difficulties, and primary or premature ovarian insufficiency in women.

Contact:
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Applied Therapeutics Inc.
212.220.9437
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Presentation, Progression, and Predictors of Ovarian Insufficiency in Classic Galactosemia

Presentation, progression, and predictors of ovarian insufficiency in classic galactosemia

Allison B. Frederick
Alison M. Zinsli
Grace Carlock
Karen Conneely
Judith L. Fridovich-Keil (corresponding author at jfridov@emory.edu, Department of Human Genetics, Emory University School of Medicine, Atlanta, GA 30322)

Published in Journal of Inherited Metabolic Disease (available online 5-2-18)
https://doi.org/10.1007/s10545-018-0177-0

Summary:

Classic galactosemia (CG) is an inherited disorder that affects about 1/50,000 live births in the United States and many other countries. Thanks to neonatal diagnosis and dietary restriction of galactose most affected infants are spared the early and potentially lethal symptoms of disease. However, most patients grow to experience a range of long-term complications that include premature ovarian insufficiency in the majority of girls and young women. Our goal in this study was to define the presentation, progression, and predictors of ovarian insufficiency in a cohort of 102 post-pubertal girls and women with CG. To our knowledge this is the largest cohort studied to date. We found that 68% of the girls and women in our study completed puberty without taking hormone treatment, while 32% completed puberty only after starting hormone replacement therapy (HRT). Of those who completed puberty naturally, less than half were still cycling regularly after 3 years, and less than 15% were still cycling regularly after 10 years. Of factors tested for possible association with spontaneous puberty, only detectable (≥0.04ng/mL) plasma Anti-Müllerian Hormone (AMH) level was significant. These results confirm that detectable plasma AMH is a useful predictor of ovarian function in girls and women with CG.

Fundraising Committee Update

By GF Fundraising Committee

The Galactosemia Foundation depends on donations and support from this conference to fund our yearly goals. We will have many fundraising activities at the conference - starting with the sales table where you can purchase your Galactosemia Foundation branded gear – so make sure to bring your wallet! We will also have a raffle and a silent auction as well. All proceeds go directly to Galactosemia Foundation. I am also working on having some items for auction from a local consignment shop including sports memorabilia and one-of-a-kind items where a portion of each sale goes to the Galactosemia Foundation. We will have our 50/50 raffle. You can also set up an annual family donation. We appreciate your support; with no paid employees and minimal expenses your donations have a large impact.You can support the Galactosemia Foundation all year long by doing the things you do every day, including:

- Make a donation with each search of the internet with Goodsearch.
- Make a donation each time you buy from Amazon with Amazon/Smile.

These can have a huge impact if you get your family, friends and co-workers involved! See me at the conference for more information.

Fundraising update - June 2018

We continue to improve year over year. This year, we had multiple fundraisers eclipse the $10,000 mark and many more eclipse $5,000. I will be unveiling the top fundraisers during my general session update. I will also be talking about fundraising ideas in the Fundraising 101 breakout session with several other families who have been successful at fundraising over the years.

Please come and find out how to get started!

The 2018 "Fore the Cause" charity golf outing wrapped up on May 18 in Chesterfield, Va. Despite a lot of rain, the funds raised were not dampened. I would like to thank the Rodgers, Berling, Stroop and Balch families for making this year's event a success. We raised $28,000 this year and considering the weather that was a miracle.

Galactosemia Activity Book

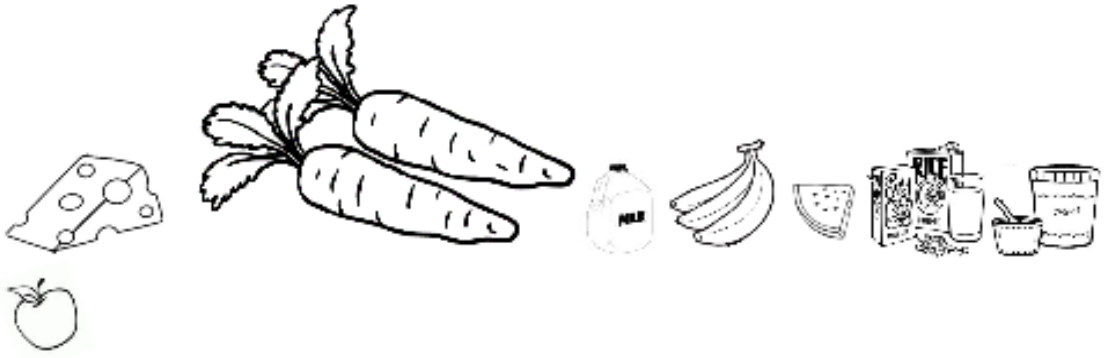
Galactosemia Activity Book

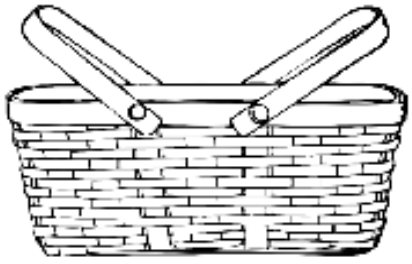
By Nicole Casale

A few months ago, my niece, Cecilia excitedly told me that her K-Kids group (an after-school group where students learn about and do good deeds for others) was going to be selling popcorn to raise money for the Galactosemia Foundation. She asked if I could come and speak to the group about my son Jojo (age 6), and of course galactosemia. I happily obliged, and created a word search and coloring page, so they would have something to do while I spoke. I brought Jojo with me, so they could meet him. He also participated and was really excited to show me how he colored and crossed off items that would "hurt his belly." He has never had the opportunity to do something like this, but I could see how beneficial it was for him. This led me to my next project, a coloring book about galactosemia. Children learn in so many ways, and I believe this could be a great resource for them to learn and teach others. Since this is for the galactosemia community, I would love to get your input and incorporate as much as I can into the book. I asked my son to draw what galactosemia means to him, and I think it would be really neat to see what it means to others. So please get your coloring pencils out and get to work. Send your completed image to nicoleacasale@gmail.com I will add them to the book, and hopefully have it complete for July.

We are packing for our picnic!

Could you help fill the basket by coloring in the "yes" foods? Cross out the "no" foods or foods that you are not sure about.





WANTED: NORD COORDINATOR

By Galactosemia Foundation

In 2016, Galactosemia Foundation was fortunate to be awarded participation in an FDA-NORD program to build patient longitudinal studies. Funds were made available to the patient advocacy organizations, to register members and track their outcomes. Many organizations have used this platform todocument important statistics for their condition (e.g.,time to diagnosis, speech capabilities, motor function) while adhering to the privacy requirements and protocols of an independent review board (IRB). Galactosemia Foundationwas pleased to launch their registry using a basic set of information athttps://galactosemia.iamrare.org/. While this is a good first step, there is much more value that could be extracted from the registry. First, there are additional customizations that could be done to capture more Galactosemia specific information (e.g., information on women's health, menstruation, hormonal therapy; information on Duarte members and their care/health). Additionally, there is work to be done to encourage additional enrollments and survey completions. The study's goal is to reach as many members as possible regardless of income, geography or language.

Galactosemia Foundation is seeking a volunteer study coordinator to lead the effort of improving this high potential board asset. It is an outstanding opportunity to demonstrate performance in a science research setting. The selected candidate would have access to full-time NORD coordinators as well as recurring NORD webinars on how to enhance the study's performance. The selected candidate would predominately work remotely, however at times they would have opportunity to attend NORD face-to-face meetings on the program. Please contact registry@galactosemia.org for more information.

Your Expertise is Needed!

Please join a network of patients and other stakeholders who want to influence health outcomes for individuals with inborn errors of metabolism.

The Inborn Errors of Metabolism Collaborative QUEST project will work to develop research that answers the questions and concerns most important to individuals with SCAD deficiency, 3MCC deficiency, 2MBCD deficiency, DG galactosemia, H-PHE, or partial biotinidase (pBio) deficiency.

Who: Individuals who have been diagnosed with one of the IBEM listed above, or have a family member with one

What: Work side-by-side with clinicians to identify health outcomes that are important to you or your family member

Compensation: You will be paid for your time, and all travel costs.

Join our QUEST for answers

If you have questions please contact Jane Pilditch at jpilditc@mphi.org, or 517-324-8391.

IBEMC Inborn Errors of Metabolism Collaborative Stakeholder Network

QUEST

NORD Coalition Endorsement Letter

ED Note: In 2015 Galactosemia Foundation joined the National Organization for Rare Disorders (NORD), a nonprofit that represents the combined interests of rare and ultra-rare conditions. One area where NORD has helped member organizations tremendously is in seeking federal and state regulations that support the interests of their memberships. Recently, a coalition of conditions formed to support proposed federal legislation for medically necessary nutrition. Recognizing the similarities between our Galactosemia members and theirs, the PKU alliance invited Galactosemia Foundation to join the conversation on this matter. Below please see the coalitions letter to the key senators. GF is working to have our name included as a signatory on this matter.

Senator Bob Casey
393 Russell Senate Office Building
Washington, DC 20510

Sen Grassley
135 Hart Senate Office Building
Washington, DC 20510

Rep. John Delaney
1632 Longworth House Office Building
Washington, DC 20515

Rep. Herrera-Beutler
1107 Longworth House Office Building
Washington, DC 20515

February 23, 2018

Dear Senators Casey and Grassley and Representatives Delaney and Herrera-Beutler:
On behalf of Patients &Providers for Medical Nutrition Equity, a coalition of patient and provider organizations that represent individuals for whom specialized nutritionis medically necessary for treatment of their digestive or inherited metabolic disorder, we write to provide our strong endorsement of the Medical Nutrition Equity Act (S. 1194/H.R. 2587). We applaud your leadership in recognizing the problems that patients with these chronic conditions face, and for introducing legislation that allows these patients and their physicians to pick the treatments that are medically appropriate for them.

For a subset of the patients with the diseases and conditions covered under your legislation, specific formulas and vitaminsessential to the treatment of these conditions offer a more medically appropriate, less risky, and less costly alternative. For example, for many of the digestive diseases covered in your legislation, including Crohn's disease and eosinophilic esophagitis, medical nutritionmay constitute the only safe and effective therapy available to a patient.Alternatively, medical nutrition may be the preferred medical treatment, with fewer risks and side-effects than other therapeutic options.

Additionally, more than four million newborns in the United States receive state-mandated testing for inborn errors of metabolism as recommended by the Secretary of HHS' Recommended Uniform Screening Panel. Each year, approximately 7,200 of these babies are diagnosed with inherited metabolic disorders as a result of this mandated testing. For most of these babies, such as those diagnosed with phenylketonuria (PKU), the use of medical nutrition is not merely an optional, alternative food choice, but a medical necessity.

When diseases of the digestive system or inherited metabolic disorders are left unmanaged or untreated, the medical consequences are often significant, permanent, and costly. The implications of denied or delayed access to medical nutrition in pediatric populations are particularly profound — inadequate growth, abnormal development, cognitive impairment, and behavioral disorders. In severe cases, without medical nutrition, the outcome can be surgery, hospitalizations,intellectual disability, or even death. Children with an unmanaged disease also suffer emotionally and socially.

Unfortunately, patients often find that their insurance provider considers these medical foods and other forms of medical nutrition to be "groceries." Consequently, insurance coverage is denied and patients are forced to seek alternative treatments or go without treatment altogether, resulting in disability. Medical foods and other forms of medical nutrition are not groceries, but rather are a major treatment modality for inherited metabolic disorders and diseases of the digestive system.

Further, without coverage, medical nutrition is unaffordable for many families. For example, some children with Crohn's disease require a pre-digested formula such as Peptamen 1.5, which, at five cans per day, can cost an average of $1,500/month. For many patients and their families, the out-of-pocket costs for specialized formulas are prohibitive, particularly when you consider that these formulas cost less than biologics that are covered for some of these conditions. Biologic therapies are not only costly, but confer medical risks, such as suppression of the immune system which can increase a patient's risk of infection.

When an insurance company does cover a medical formula, it often comes with the stipulation that the formula be administered through the nose or through a tube surgically placed in the patient's gut, both of which carry additional risks. For example, a gastrostomy tube can leak, cause ulcerations, or a patient may experience infection at the insertion site. In severe cases, some patients with these feeding tubes experience a perforation in the intestinal tract.

These types of coverage policies are irrational and interfere with thoughtful medical decision making. The Medical Nutrition Equity Act would ensure coverage parity, providing patients the ability to choose the best treatment option in consultation with their physician. Patients &Providers for Medical Nutrition Equity thanks you for your critical leadership on this important issue. Passage of S.1194/H.R.2587 will have a profound effect on the many patients who require medically necessary nutrition to survive and thrive. We stand ready to work with you to build critical bi-partisan support for this legislation and to move toward passage. Please contact Megan Gordon Don at 202.246.8095 or mgdon@mgdstrategies.com if you have any questions or need more information.

Sincerely,

American Academy of Pediatrics
American College of Gastroenterology
American College of Medical Genetics and Genomics
American Gastroenterological Association
American Partnership for Eosinophilic Disorders
Association for Creatine Deficiencies
Association of Pediatric Gastroenterology and Nutrition Nurses
Children's Hospital at Dartmouth
Children's Hospital Colorado
Children's Medical Nutrition Alliance
Children's MAGIC US
Children's National Health System
Crohn's & Colitis Foundation
Campaign Urging Research for Eosinophilic Disease (CURED)
EveryLife Foundation for Rare Diseases
FOD (Fatty Oxidation Disorders) Family Support Group
Genetic Metabolic Dietitians International (GMDI)
International Foundation for Functional Gastrointestinal Disorders
Maple Syrup Urine Disease Family Support Group
March of Dimes
National Organization for Rare Disorders (NORD)
National PKU Alliance, Inc.
National PKU News
National Urea Cycle Disorders Foundation
Nationwide Children's Hospital
North American Society for Pediatric Gastroenterology, Hepatology
Organic Acidemia Association
Pediatric IBD Foundation
Propionic Acidemia Foundation
Society for Inherited Metabolic Disorders