WWW.MSUD-SUPPORT.ORG

MSUD NEWSLETTER

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MSUD Family Support Group joins the Global Genes® RARE Foundation Alliance

Jordann Coleman, Advocacy Chairperson

Global Genes®, a Rare Disease advocacy organization, has created an alliance of rare disease organizations to harness the power of collective impact. This alliance aims to provide patient organizations with an avenue for sharing best practices and lessons learned to drive better outcomes for the entire rare disease community. The MSUD Family Support Group has joined this alliance, giving us the opportunity to become stronger in a variety of areas including:

- Marketing
- Research
- Networking
- Public Policy
- Family & Patient Support
- Education
- Resources

An immediate benefit of our membership in this organization is the ability to raise money for the MSUD Family Support Group. Global Genes® is hosting their 3rd Annual "Denim Dash" Virtual 5K. This virtual race occurs throughout the month of April and encourages rare disease supporters to run/walk/bike a 5k to raise money to support the rare disease community. New this year is the 50/50 split of net proceeds between Global Genes and Foundation Alliance Members. The MSUD Family Support Group has created a team and we encourage you to join! Help us spread awareness and raise money. Visit **www.crowdrise.com/msud-road-warriors**

With the support of the Global Genes® and the RARE Foundation Alliance, the MSUD Family Support Group looks forward to growing our impact and reach to ultimately improve the lives of people living with MSUD and their families. For more information about this organization, visit their website at **www.globalgenes.org**

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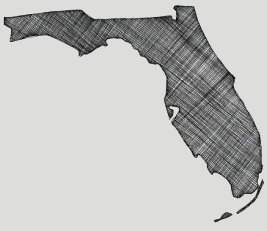

A Message from the Editor

By Karen Dolins

We are publishing our newsletter electronically for the first time!

Many organizations have moved away from print newsletters due to the expense involved in printing and mailing. As mentioned in earlier issues, we have elected to continue with a print newsletter for Summer/Fall while using our eblast list to publish a Winter/Spring newsletter electronically.

We are also using our eblast list to communicate with you on timely issues so you can expect to hear from us more frequently than in the past.

This enewsletter contains research updates, information about our next Symposium in June 2018, and information about rare disease advocacy and how you can help. §

Rare Disease Week 2017

February 27-March 1, 2017

Karen Dolins

I was privileged to represent our group in Washington DC for Rare Disease Week. The week began with a day at the National Institutes of Health (NIH), where we heard from leading scientists about efforts to advance diagnosis and treatment for rare diseases. The following day was an advocacy training session where issues of concern to the rare disease community and ongoing efforts to impact public policy were discussed. Much of the discussion centered on ways in which the new Congress and administration will impact rare disease policy. This was followed by a day on Capitol Hill where we visited the offices of our representatives in Congress and our Senators to ask for their support on key issues. Here is a brief summary of the events of this inspiring and empowering week.

10th International Rare Disease Day at the NIH

The theme for 2017 Rare Disease Day at the NIH was Research. There are 7000 different types of rare diseases, and 80% are genetic. Treatment is available for fewer than 500 of these.

The international rare diseases consortium (www.irdic.org) was established in 2011 with the aim of fostering international cooperation in rare disease research. The goal is to transform rare disease research to improve efficiency and effectiveness by sharing knowledge, data, infrastructure, expertise and viewpoints.

FDA and Rare Diseases

The FDA evaluates drugs for rare diseases through its Office of Orphan Products Development. This office has the flexibility to expedite the approval process for drugs for rare diseases when deemed necessary. The Prescription Drug User Fee Act allows private companies to pay a fee to the FDA for expedited review. The speaker noted that well organized groups can help a drug company move along the drug approval process.

The 21st Century Cures Act was signed into law by President Obama on December 13, 2016. It provides incentives for the development of treatments for rare pediatric diseases and increases funding for NIH research. The current administration has not made their perspective on this act clear. It must be reauthorized by September 30, 2017 or it will be dropped.

The FDA is currently without a commissioner as one has not been named by the current administration yet. The concern is that this may hamper initiatives.

ADVOCACY and LEGISLATIVE ISSUES

Everylife Foundation for Rare Diseases (rareadvocates.org)

RDLA Rare Disease Legislative Advocates

This organization advocates for policy and regulatory reform through partnerships with rare disease organizations. At the state level, they are

working to improve newborn screening by adding diseases to the list of those currently included. (Our organization signed on as a supporter of legislation in Florida – see page 5)

They have a free tool to help individuals and groups with their advocacy efforts, and publish monthly newsletters. Important websites include:

- Rare diseases clinical research networks www.rarediseasesnetwork.org
- Genetic and rare disease information center www.rarediseases.info.nih.gov
- Rare disease registry: www.ncats.nih.gov/grdr

Rare Disease Congressional Caucus

This caucus provides a platform for representatives to discuss issues of importance to the rare disease community. They also hold quarterly briefings with patients, advocates, and providers. Everyone is encouraged to ask their legislators to join. We can be effective by using our personal stories.

It is important to lobby so our voice is heard. Items of current interest include:

OPEN Act: This piece was dropped from the 21st Century Cures Act. It provides incentives for the development of orphan products. While passed in the House, it did not reach the Senate and has been resubmitted. New legislation must be passed by Congress and signed by the president by July 30 or the bill will die. **Patient organizations must sign on.**

HEALTH CARE REFORM

Affordable Care Act (ACA/Obamacare): This was discussed from both the Democrat and Republican perspectives. The Democrat perspective is that any changes must protect those with preexisting illnesses and maintain the regulation against annual and lifetime caps for insurance reimbursement. It was noted that we now have the lowest rate of uninsured people/families in history and that health care costs, while still increasing, are doing so at the lowest level of increase in years. The ACA has also increased treatment for opioid addiction through Medicaid expansion. The Republican perspective notes the need to stabilize the individual market place, and proposes tax credits which will be based on age, not income. Participants asked how people will be able to afford the level of insurance they need with tax credits based on age rather than income (as opposed to subsidies, families must be in a high enough income bracket to have a tax bill high enough to support the credits).

American Academy of Pediatrics: Supports the ACA and keeping Medicaid and Children's Health Insurance Program (CHIP) strong. It was noted that the rate of coverage for kids is now 95%, and 36% of children get their coverage through Medicaid and CHIP. 44% of children with rare diseases receive Medicaid. The concern is that block grants and caps will reduce the level of care as states won't be provided with adequate funds according to need. CHIP was created in 1997 to provide insurance to children whose families cannot afford insurance but have incomes too high to qualify for Medicaid. Funding for this program will run out at the end of September. The Academy is concerned that current proposals for health care will cause children's health to suffer.

NORD: Supports the ACA which created out of pocket caps, protecting patients against exorbitant costs and lifetime limits. Before ACA, individuals were put into high risk pools which were problematic for those with rare diseases due to waiting lists, enrollment caps, and significant out of pocket costs. They also oppose any weakening of state Medicaid programs as this will disincentivize states from covering care and orphan drugs.

With the recent withdrawal of the Republican plan to replace the ACA, it is imperative that congress act to strengthen it.

Advocacy: Ask your representative why they did or didn't support legislation important to the rare disease community (ex OPEN Act). Build a relationship. Tell your personal story and be passionate. Use social media and include calls to action.

Always thank them for the work they do. §

Acer Therapeutics: working diligently to advance ACER-001

Robert Steiner, M.D.
Chief Medical Officer
Acer Therapeutics

As discussed at the 2016 MSUD Symposium and in the previous issue of this newsletter, Acer Therapeutics is following up on the research initiated by Dr. Lee on phenylbutyrate and its potential to aid in the control of branched chain amino acid levels in those with MSUD. I was asked to provide a quick update on ACER-001 – the taste-masked, immediate release formulation of sodium phenylbutyrate – that we at Acer are developing for the treatment of MSUD.

We've spent the better part of the past year optimizing the formulation, ensuring that it is completely taste-masked, yet behaves like regular sodium phenylbutyrate once it's in the body. It's really important that we get this right, so that patients taking ACER-001 get the maximum benefit of the drug, while not detecting the horrific taste of phenylbutyrate! While this has been an incredibly challenging project, we believe we now have the best formulation developed to take into the clinic.

Before we begin the phase 2 clinical trial for MSUD, we need to run a quick study in healthy volunteers to make sure the drug is completely taste-masked, safe, and behaves similarly to regular phenylbutyrate in the body. This is a very quick study that should be completed in the fall. Assuming everything goes well with this study, we will begin the MSUD trial shortly afterward.

As a reminder, the MSUD phase 2 trial will enroll approximately 60 patients at 10-15 clinics across the US. Once a patient begins the study, they will be required to maintain a very tightly-controlled diet for about a month. ACER-001 will be added on top of their diet for about two weeks, to determine if ACER-001 + diet can lower levels of leucine by a meaningful amount over diet alone.

We invite you to visit our recently updated website, where you can access patient resources, subscribe to our mailing list to receive program updates and clinical trial information, or ask questions directly to the company. Please visit:

www.acertx.com/contact-acer-therapeutics

We look forward to hearing from you, and keeping you updated on our progress! §

SAVE THE DATE

MSUD SYMPOSIUM 2018
will be held
Thursday June 28th to
Saturday June 30th, 2018
at the
DoubleTree Greentree near Pittsburgh, PA.

More information to follow in the fall 2017 newsletter and on the MSUD website www.msud-support.org.

For questions, contact Sandy Bulcher at sandybulcher@gmail.com or 740-972-5619

NEW RESEARCH -
HOT OFF THE PRESS!

Neurocognitive profiles in MSUD school-age patients

A newly published paper describes a study conducted by Bouchereau and colleagues on neurocognitive development in 21 children with MSUD in France. All of the children had classical MSUD and were diagnosed in the neonatal period. The children underwent neuropsychological testing between the ages of 6 and 12 years.

All but 3 of the children had IQ scores within the normal range. The 3 who scored a low IQ were diagnosed later and had poorer metabolic control. Almost all of the children had attention deficit disorder and hyperactivity. Testing showed impaired mental flexibility and inhibitory control with impulsivity. Verbal abilities were higher than performance abilities for some of the children.

The authors concluded that it is important for children with MSUD to undergo neurological testing so that appropriate interventions can be initiated. §

J Inherit Metab Dis. 2017 Mar 21. doi: 10.1007/s10545-017-0033-7. [Epub ahead of print]

NEWBORN SCREENING LEGISLATION

Our organization joined 108 others in signing a letter in support of expanded newborn screening in Florida. While MSUD is now a part of newborn screening in every state, many other rare diseases are not. This effort, spearheaded by The EveryLife Foundation for Rare Diseases, aims to support legislation which would expanding newborn screening in the state of Florida to include diseases recommended by the state's Genetics and Newborn Screening Advisory Council. §

NUTRICIA Metabolics
Inspiring Futures

Attn: Patients using Maxamaid® and Milupa products

Dear Metabolic Families,

Having improved our line of Metabolic formulas to be in line with the latest nutritional guidelines we will **no longer supply our lines of Maxamaid and Milupa products after June of 2017 or until supplies last**.

At Nutricia North America, we work very hard to ensure metabolic patients receive the best possible nutritional support. The past couple of years have been exciting as we have enhanced our product portfolio to grow with patients from infancy to adulthood. We proudly introduced our Periflex® / Anamix® Early Years line for infants with the added benefits of DHA & ARA and prebiotic fiber to be closer to breast milk than ever before*! In addition, we launched the follow-on formulas Periflex® Junior Plus and Anamix® Next, which contain added DHA & a multi-fiber blend.

Although we are confident that these changes are in the best interest of metabolic patients, we certainly understand the challenges that may come with transitioning to new formula. If you are currently using a Maxamaid or Milupa formula please follow the steps below to start your transition:

1. **Contact your clinic/healthcare professional to find other products within our portfolio and start the transition process. Please do not initiate a transition on your own.**
2. If you have questions regarding insurance reimbursement, contact our Reimbursement Care team Monday – Friday 8:30am – 5:00pm EST at 1-800-605-0410.
3. If you have product related questions or need help finding a supplier for your new formula , contact our Customer Service team Monday – Friday 8:30am – 5:00pm EST at 1-800-605-0410.

Please know our goal is to provide the best nutrition possible, and we are committed to being the best in specialized nutrition for the metabolic community.

Sincerely,

The Nutricia Metabolic Team

** Compared to Nutricia's Periflex Infant and Analog formulas*

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