The HCU *Herald*

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HEREDITARY METABOLIC DISORDERS AND COVID-19 VACCINATION

MetabERN RECOMMENDATIONS

The experts of MetabERN recommend that **IMD patients get vaccinated against COVID-19** when they are offered this opportunity.

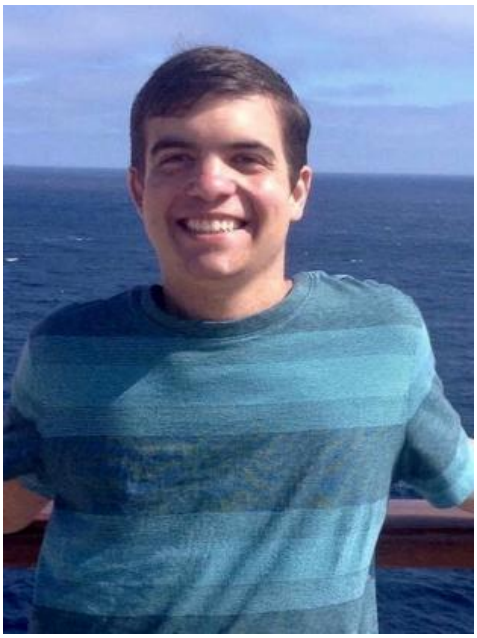
All IMDs should be considered as a high priority for COVID-19 vaccination.
Patients who present a high risk of acute metabolic decompensation, respiratory or cardiac complications, and frequent exacerbation induced by infection, such as those with AOA, PM-MD, C-FAO or LSD, should be vaccinated with the highest priority.

Based on the current evidence, our experts' advice is to **use any vaccine available in the country**, with **no particular contraindications** for any metabolic disease.

AOA: amino and organic acids-related disorders; C-FAO: carbohydrate, fatty acid oxidation and ketone bodies disorders; IMD: inherited metabolic disease; LSD: lysosomal storage disorders; PM-MD: disorder of pyruvate metabolism, Krebs cycle defects, mitochondrial oxidative phosphorylation disorders, disorders of thiamine transport and metabolism.

European Reference Network | MetabERN

HCU HEROES: SHANE, KY AND KADEN



Shane at 29 years old

We became parents for the first time in September of 1991. Our baby Shane was the cutest little boy at 6 lbs 12 oz! The doctors handed him to me and said “you have a healthy boy!” I looked him over and my first thought was how adorable he was. My second thought was “he’s NOT healthy”. To this day I don’t know why I thought that, but that thought stayed with me throughout his entire infancy. He was floppy. Tiny. He still felt like a newborn even at two and three months old. My friends’ babies looked and acted differently than he did. He cried constantly for the first two years of his life. I shared my fears with his doctor. “He’s fine,” one said. “You have new mom jitters” said another. “He has colic” was another comment. He was meeting all of his milestones just a little bit late. Not too far on the outside of what was considered normal for babies of his age, so doctors said he was fine and not to “compare him with other babies”. Yeah, sure!

When Shane was about one year old I began noticing brown spots appearing all over his body. Some were small, like a dime. Some were as big as his entire thigh. All of his fingertips were dark brown just before the nail and his tummy was several shades darker than the rest of his body. My husband is part Hispanic and part Native American. Maybe it's just coming out in his coloring?

He began getting sick. He’d run fevers of 104 to even 106. He was given a spinal tap twice due to his very high fevers but they’d come back clear. No one knew why he got so sick. At this point he’d also fallen off the growth chart for both weight and height. He was perfectly proportioned and didn’t appear to be a failure to thrive. He was just small. So, they said they’d keep an eye on it.

Finally at age three and a half, he was seen by an amazing doctor in Urgent Care for yet another fever. This doctor noticed his dark spots and was very interested in his medical history. He referred us to Dermatology.

The dermatologist we saw diagnosed Shane with Neurofibromatosis. He told us to make a second appointment in six months and don’t “look it up”. The first thing I did was look it up! It scared me to death. I called the doctor from Urgent Care who had referred us, and he brought us right in. He said he didn’t think it was NF. So, he sent us to be seen by Genetics. Finally, things began to happen! The Genetics doctor did a full work up on Shane who was now almost four. He wasn’t 100% sure, but he felt that Shane had a genetic condition called Noonan Syndrome. We read everything we could get our hands on to learn more. We joined a support group and began to meet other people whose children had Noonan Syndrome as well. Finally, things began to make sense! At that time, I had just had my second son, Ky. He was chubby, healthy and met all of his milestones early. It appeared that he didn’t have any of the same issues Shane had. Our focus was on getting Shane early intervention for speech and OT. He seemed very bright and knew all of his colors, letters and sounds and could count to 100 before he was

four years old. We still had the worry that he may also carry the gene for NF, so we stayed in touch with the Neurofibromatosis community as well as Noonan Syndrome as sometimes the two conditions can go hand in hand.

When Ky was 15 months old I had my third baby and only daughter Danica. We were happy and feeling like we were getting a handle on this parenting thing! About six weeks after Danica was born Ky became very sick. He ran a high fever off and on for a few months. No other symptoms except for the fever and he began falling off the growth chart. Our doctor hospitalized him and he was tested for several diseases and disorders. His blood count was abnormal. He showed signs of a possible rheumatoid condition, and scariest of all, they thought he may have Neuroblastoma cancer. We completely freaked out! It was terrifying and we thought we were going to lose our little boy. After being ill for several months and just before our first oncology appointment he began to rebound. His blood tests began coming back normal. The fevers stopped and everything stabilized. It was a crazy situation but by the time he was two years old he was once again fine. At this point, our Genetics doctor began seeing Ky as well. By the time Ky was in kindergarten he was healthy and very smart. He was ahead academically and was doing great, besides being a bit small for his age. Shane, on the other hand, was losing ground academically. He struggled in all areas and had a full IEP by first grade. It went along with the Noonan Syndrome diagnosis. We felt that Ky was in the clear and seemed to be perfectly healthy.

As Shane continued through school he fell further and further behind. He tried harder than anyone I've ever met. We had tears at the table doing homework, yet he kept trying. Finally, we pulled him out of school and put him in a homeschool group where he could continue his education to the best of his ability, at his own pace and with less stress. He loved it! Meanwhile,

Ky was a straight A student, cruising through school with no problems whatsoever except by this time he was quite a bit smaller in stature than his friends. He and his sister Danica went to the same school, shared the same group of friends and both of them were very involved in community theatre, even performing in a Broadway touring company professional production. We had our fourth (surprise) baby in 2003. Our last little boy, Kaden. Around the time Kaden was born, Shane was 12. Our Genetics doctor who had diagnosed him with Noonan syndrome said he was changing his diagnosis. Shane was "outgrowing" some of the symptoms (which doesn't happen). At this point a blood test had finally come out for Noonan Syndrome. Shane tested negative. We were back to square one!

The family all together in (approximately) 2006

Little Kaden was growing very slowly. He was tiny! He struggled to feed and was diagnosed as failure to thrive. He was seen by Genetics as well.

He had been born with several large skin tags on both sides of his face, and one of his ears was deformed by the largest skin tag during fetal development . Genetics thought he had Goldenhar Syndrome. We couldn't understand how all our kids had these unrelated illnesses and genetic conditions. Somehow this had to all be related, but how?

At age 14, we began to notice Shane was dragging his right foot. He began tripping over it. He walked, running his hands along the walls for balance. As a little boy he surfed and played soccer. His preschool teacher had nicknamed him Speedy because he ran everywhere! Now as a teenager he was unable to walk normally and had developed a spastic gait. He also began having significant leg tremors.

During this same time Ky began developing large brown spots on his neck and Kaden had them on his legs. We were sent to do a full work up in Los Angeles. It was another dead end. No answers. No idea what our boys had. No idea why, if they all had the same thing, they each presented so differently.

When Shane turned 18 we gave up. He was sick to death of being poked and prodded. No one could give us answers. At that point, we decided that his quality of life was the most important thing. So, we stopped all testing. Shane got a job and was happy. Ky was doing great in school and was hoping to pursue a career in music. Danica volunteered at a private zoo and wanted to work with animals someday. Kaden was our little water baby who loved to surf! We were content even though we wished we could have answers, we assumed we'd never have them.

When Kaden hit 5th grade he began struggling in math. He was diagnosed with ADHD and put on medication (which he hated and begged us to take him off them). He also was, by far, our smallest child and was put on growth hormones. The summer before 6th grade his pediatrician mentioned that we might want to give Genetics one more try. Lots of new tests had come out over the past few years and we might actually find an answer for the different quirks our kids had. We, reluctantly, agreed to give it one more go. The Genetics doctor chose to do all of the testing on Shane. They did the full genetic diagnostic workup on him. If anything showed up, then they would test Kaden as well, and possibly Ky (even though Ky seemed fine in every way except the brown spots and being a bit shorter than average).

In the spring of 2017, the test came back, and we were shocked that there was an answer. A real answer! Shane had Cobalamin F Disease. What?? Like most people, we had never heard of it, and we had no clue what it meant for Shane or the rest of our family. It was so rare that the doctor asked if my husband and I were related (uh...NO). They tested the rest of the family and both Ky and Kaden also had CblF. (My daughter did not). The most shocking part of the diagnosis was that there was actually medication to treat this disease! It was hard to get help from the Genetics department so I turned to Facebook! I found the MMA Facebook page, and suddenly things began to happen! This wonderful group of people put us in touch with Dr. Venditti at NIH. Dr. Vendetti's team had us come out to Maryland and begin full blown testing. We learned about what the disease was. We found out that our kids had extremely high homocysteine levels, Ky's levels were so high that they said he was at risk of having a stroke. It was at NIH that our kids finally began getting on B12 injections. We were put in touch with a great metabolic specialist here on the West Coast and have now been seeing him for the past four years. The boys (now young men) are healthy, and their homocysteine levels are in normal range.

Shane lost ALL of his brown hyperpigmentation! As he lived 25 years without treatment, he's had the hardest road, however, he was able to find a job where he was very happy for several years through the ARC program. Sadly, that ended due to Covid. Happily, he's now in an amazing

program for adults who are learning to work in the film industry. He loves it and is set to graduate from the program this coming January.

Ky continues to be the least affected. He studied music and musical theatre in college. He's been a performer and singer at theme parks and has taught voice and theatre classes. He hopes someday to move to New York to continue pursuing his dreams. His brown spots are also gone and he continues to be healthy and doing well!

Kaden is now going into 11th Grade. He's had the most miraculous comeback! His health is excellent and has begun growing and is now the tallest of his brothers. He has an IEP but does extremely well in all his school classes and is hoping to study computers in college after high school. He has a great group of friends and was recently voted Prom Prince by his peers at the high school spring dance. He's a strong and dedicated surfer (he's been surfing since he was about three years old) and he's also quite a good drummer and would like to eventually be a studio session drummer.

Kaden at 17 years old

Writing this has reminded me of what a crazy ride it's been trying to find a diagnosis! We honestly never thought we'd be here. It's hard sometimes to wonder what might have been had we only known all of this when they were babies. Especially for Shane, I sometimes feel very sad. I see what the treatment has done for Kaden and wonder if it would have done the same for Shane had he had it as a child. I'll never know. I do know however, that I am a mom to three amazing young men (and one incredible young woman). We are actually very lucky!

Email us your patient story! info@hcunetworkamerica.org

HCU COMMUNITY COOKBOOK: BACK TO SCHOOL IDEAS

For Breakfast:

Banana Bread Waffles

These toaster size waffles are a great breakfast to cook in batches and freeze for those busy school mornings!

Makes 5 Servings | Serving size: 1 waffle
Per serving | Protein: 0.3 g | Calories: 135

Click the picture to get the recipe!

For Lunch:

Roasted Farmers Market Pasta

This delicious pasta is the perfect hearty lunch for your school age kiddo. Keep it warm in a thermos and they'll be all set to go!

Makes 3 Servings | Serving size: 120 g
Per serving | Protein: 2.3 g | Calories: 309

Click the picture to get the recipe!

For Snack:

Brownie Dip

This is the perfect school, or after school snack. It's quick, delicious, and sure to please!

Makes 5 Servings | Serving size: 1 oz
Per serving | Protein: 0.5 g | Calories: 53

Click the picture to get the recipe!

For more ideas, visit the Back to School Section of our Special Edition Topics:
https://hcunetworkamerica.org/special-edition-topics/

THIS WEEK'S MENU

Each day has meals for <10 grams (g) of protein/day, 20-30 g. of protein/day, and 30-40 g. of protein/day.

M
Breakfast: Toast and apples
Lunch: Veggie Wraps
Dinner: Macaroni and Cheese

T
Breakfast: Pancakes
Lunch: Veggie Soup & Salad
Dinner: Spaghetti and "Meatballs"

W
Breakfast: Cinnamon Roll & Yogurt
Lunch: Jackfruit Tacos
Dinner: Pasta Salad

T
Breakfast: Breakfast Hash
Lunch: Fritters / Nuggets & Fruit Salad
Dinner: Macaroni and Cheese

F
Breakfast: Yogurt Parfait
Lunch: Asian Stir Fry Pasta
Dinner: Rice Bowl

Shopping List

Click each day to view the week long menu!

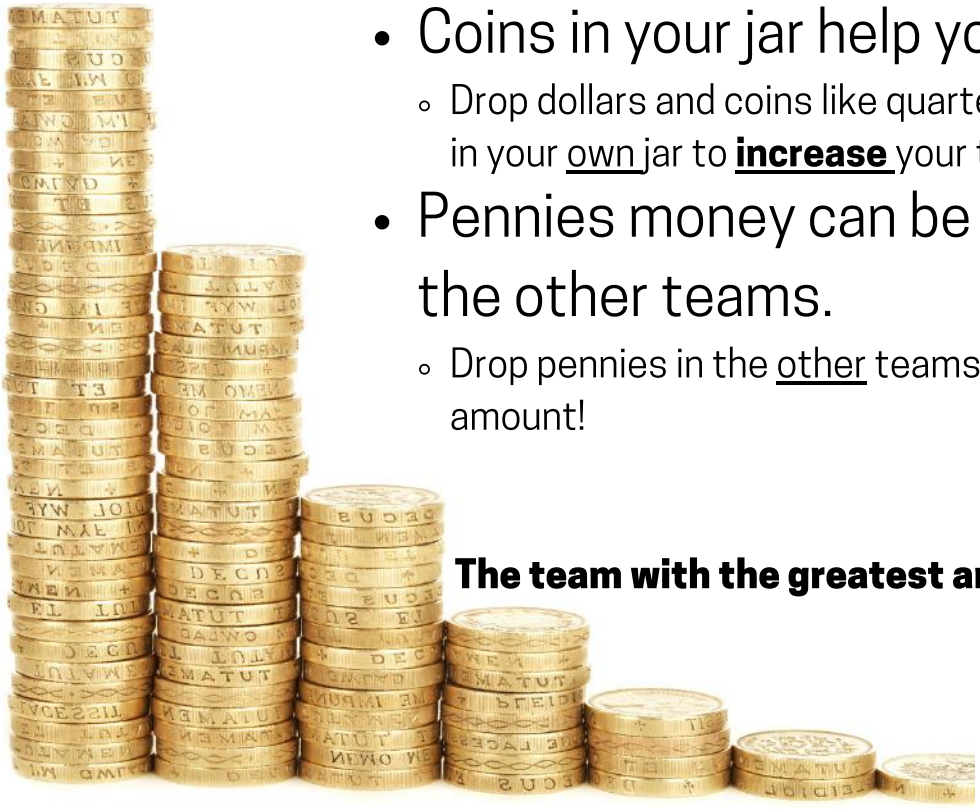
Disclaimer: This meal plan is intended to be a foundation or guide to what meals could look like on a low protein diet. It does not take into account individual caloric, protein and formula requirements, which are all patient-specific. Please consult with your metabolic geneticist and dietitian prior to making any significant dietary changes or following any meal plans of which you are unsure.

BACK TO SCHOOL FUNDRAISER: PENNY WARS

Penny Wars

1. Decide dates for your school wide Change War (work with school administration in advance)
2. Create flyer with dates and rules, distribute to students and teachers
3. Distribute containers to classrooms (or in a common area if you are going to do one per grade).
4. Encourage kids with announcements or charts
5. Count the donations and reward the winning class.

Change Wars Rules



- Coins in your jar help your team win!
  - Drop dollars and coins like quarters, dimes, and nickels in your own jar to **increase** your total amount
- Pennies money can be used against the other teams.
  - Drop pennies in the other teams jars to **decrease** their amount!

The team with the greatest amount of money wins!

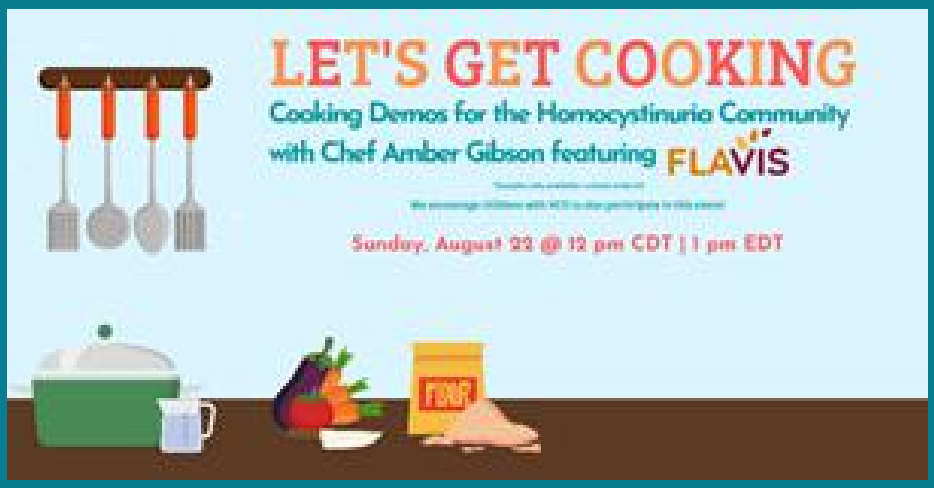
SUPPORT EVENTS

To learn more or register, visit: https://www.eventbrite.com/o/hcu-network-america-30163980100

All good things must come to an end (eventually). The end of monthly Classical and Cobalamin Meetups.

As things have opened back up, we've noticed everyone and everything is getting busier! Over the last year and 2 months we have connected with 98 caregivers, parents, and patients young and old, in 9 countries and 24 states! We have enjoyed getting to know each of you and learning how we can help the community further. While we are sad to stop the meet-ups, we know many of us are happy to start back to some resemblance of "normal". We will be hosting a Classical HCU meet up and Cobalamin meet up in October, as part of HCU Awareness Month and in February, as part of Rare Disease Month. We then hope to meet all of you in person at our 2022 conference! Our door is always open, and we are glad to continue to assist and support in any way possible! Please don't be a stranger.

Sincerely,
Danae' Bartke, Executive Director
HCU Network America



Come cook with us as Chef Amber Gibson guides us through recipes using Flavis low protein products! Free products will be sent to US participants affected by HCU who register by August 11th, 2021!

Those outside of the US or who register after August 11th are welcome to attend too. (Flavis products are equivalent to the Mevalia line).

Fall means back to school! Preschool, K-12, after school care, or College, no need to worry or fret, we are here to support you!

Come chat with other parents and caregivers about this transition and what has worked, hasn't worked and learn what resources HCU Network America has to offer!

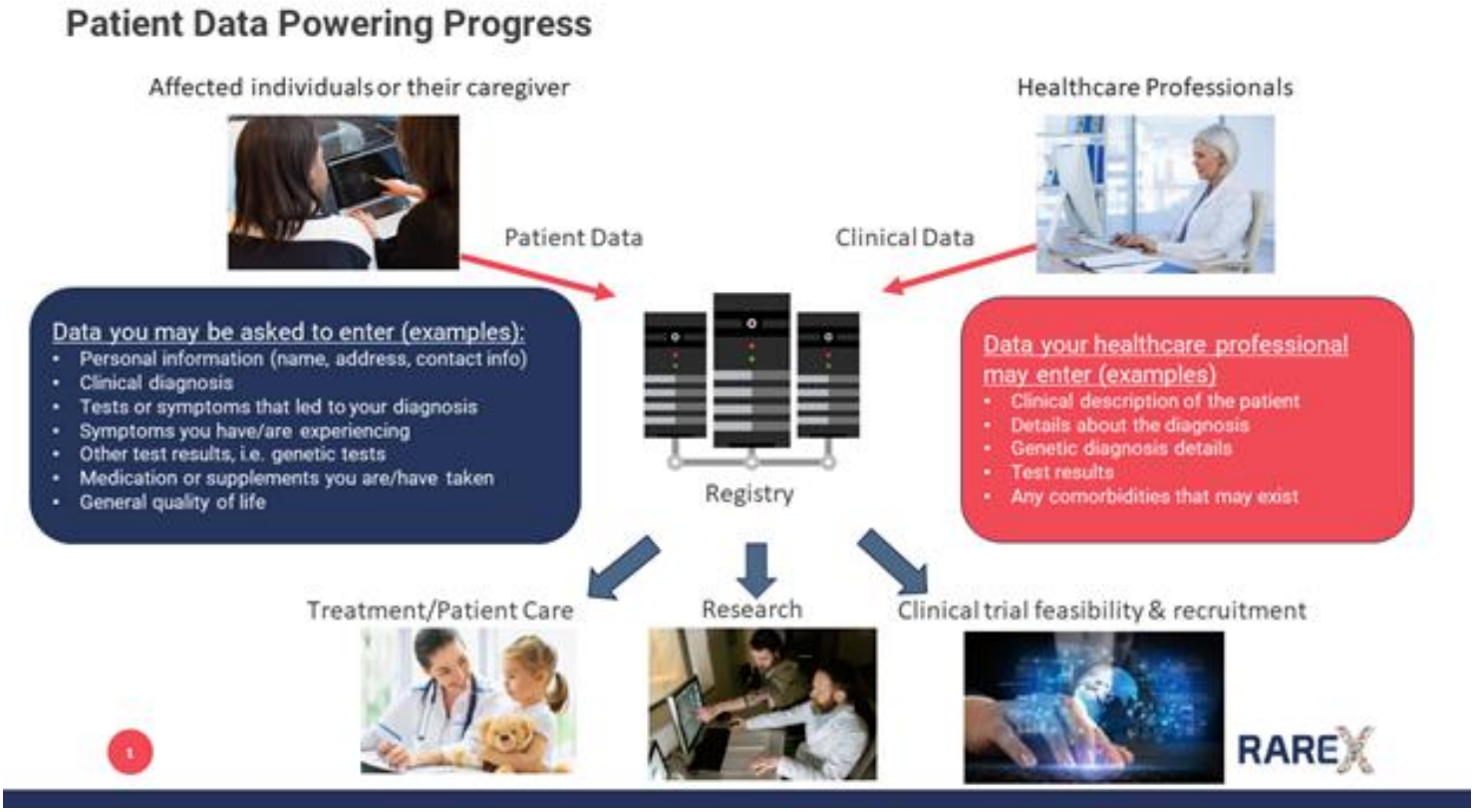
Come join us
Wed., 8/25 @ 5 pm PT | 7 pm CT | 8 pm ET

To learn more or register, visit: https://www.eventbrite.com/o/hcu-network-america-30163980100

MARK YOUR CALENDARS!

Come join HCU Network America, The Organic Acidemia Association, and the Propionic Acidemia Foundation for our first joint conference, June 25-26, 2022 in Bethesda, Maryland. There will be combined keynote speakers, disease specific speakers, breakout sessions, networking sessions and more! Mark your calendar now, registration will open later this year.

Rare – X:
The Importance of Data Collection to the Homocystinuria Community



Homocystinuria is a disease that can affect our community in many different ways and has many diagnostic challenges. To support communities with rare diseases, Patient Advocacy Groups are involved in advancing the development of a data collection or registry, based on patient-reported data, for their disease. The objectives are to increase understanding of their disease, improve patient care and further clinical research. Researchers of all kinds - academic and the biopharmaceutical industry - need open access to such information as they research future medicines and conduct clinical trials. HCU Network America is exploring this opportunity for the homocystinuria community. We are partnering with RARE-X, a non-profit patient advocacy organization with a focus on patient-owned data and open access for researchers. HCU Network America is committed to providing this opportunity for our community as the collection of data (as portrayed in the graphic) will benefit treatment/patient care, research, and clinical trial feasibility and recruitment. More will be shared at the summer mini-series (Roadmap to Innovation for HCU) and in future communications.

Danae' Bartke, Executive Director
HCU Network America

RESEARCH:
SUMMER RESEARCH MINI SERIES

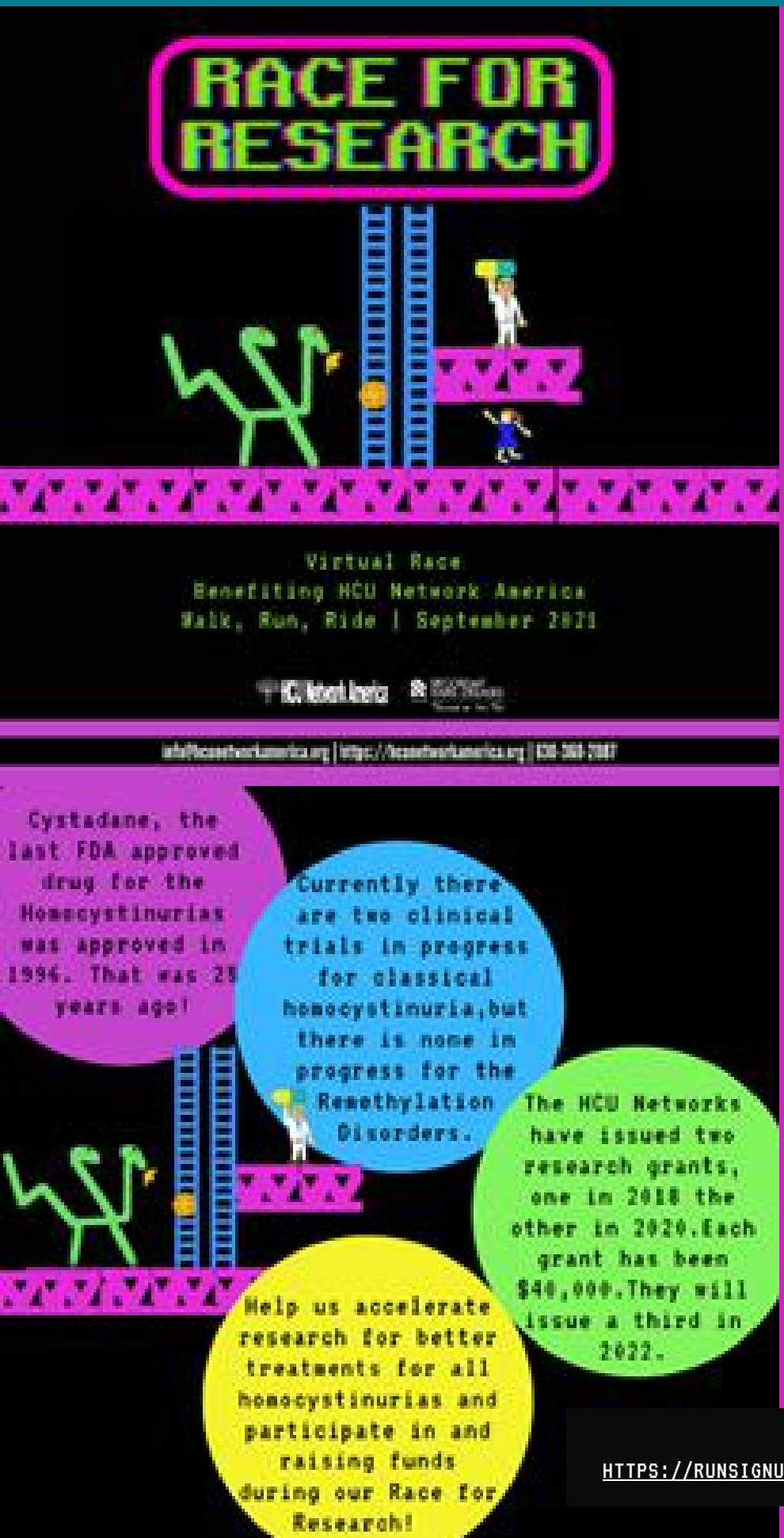
To learn more or register, visit: https://www.eventbrite.com/o/hcu-network-america-30163980100

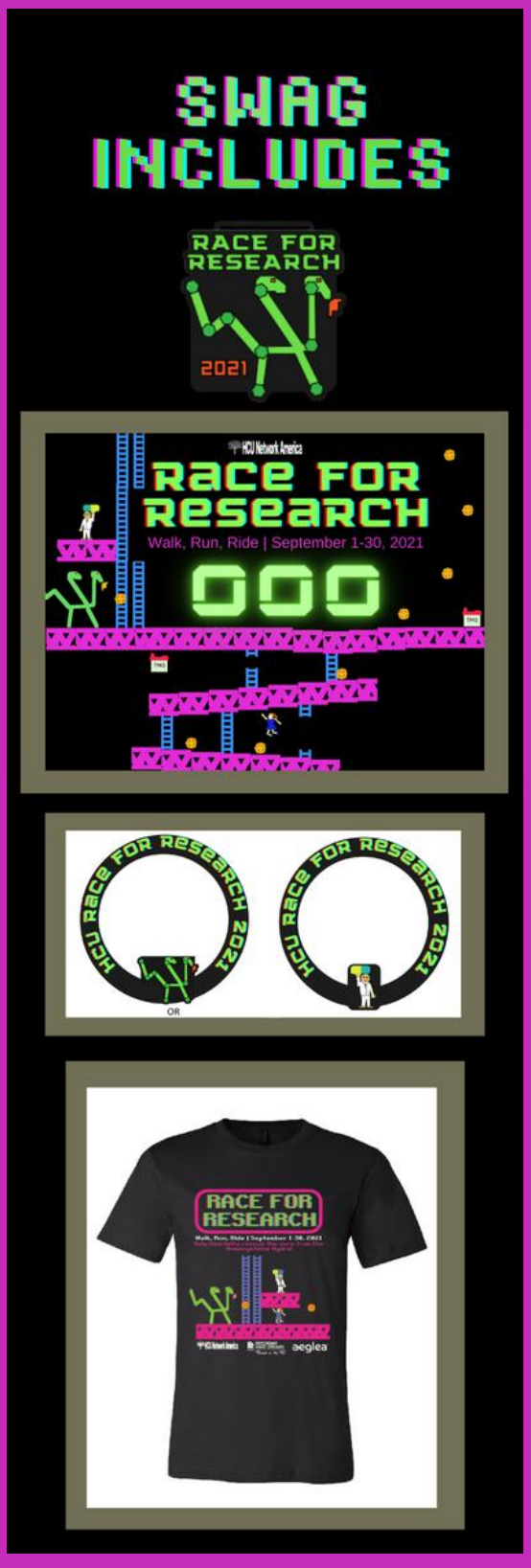
Exciting things are happening for the Homocystinuria Community. Join us for the last two sessions of our Summer Mini Series with these scientific sessions. They are held back-to-back with a brief intermission between for your convenience!

Kick off our Summer Mini Series scientific presentations with Warren Kruger, PhD, of the Fox Chase Cancer Institute Saturday, August 28, 2021 at 8:30 am CT | 9:30 am ET as he provides us with background information and the progress of gene therapy for Classical Homocystinuria. This webinar is designed for anyone else interested in Classical Homocystinuria (Cystathionine Beta-Synthase).

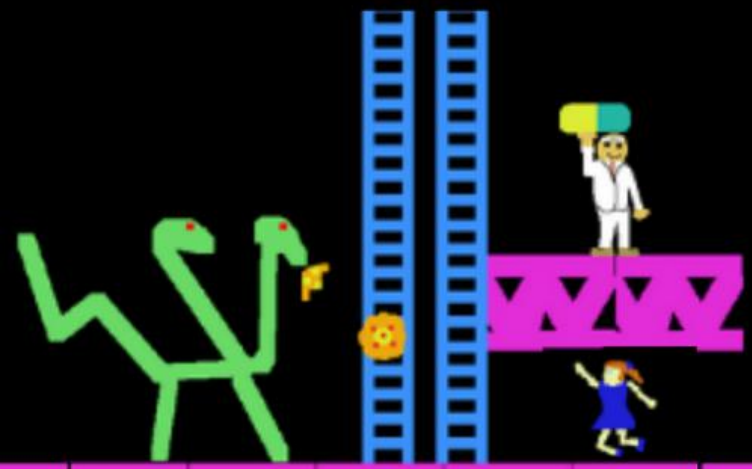
Continue with us after our intermission to learn about patient registries, natural history studies and clinical trials all for HCU! Hear the most up-to-date information about what is being done to accelerate research and bring new therapies to market!

ACCELERATE THIS RESEARCH: RACE FOR RESEARCH!





REGISTER AT:
HTTPS://RUNSIGNUP.COM/RACE/IL/BATAVIA/HCURACEFORRESEARCH



RACE FOR RESEARCH

HCU Network America | RECORDATI RARE DISEASES GROUP — Focused on the Few™

Virtual Race
Benefiting HCU Network America
Walk, Run, Ride | September 2021

What is a virtual race?

A virtual race is a race that can be walked, ran, or biked from any location you choose. You can participate on the road, on the trail, on the treadmill (or stationary bike), at the gym or on the track (or even at another race). You get to run your own race, at your own pace, and time it yourself. You do not have to complete the miles all at once, in one day, or even a week. You can use the entire month to complete the race.

How do you know how many miles I completed?

- We rely on the honor system. You don't have to use a device to prove your miles.
- If you'd prefer to use an app to track your miles, we recommend Strava. You can join the HCU Network America Club.
- Please use intentional miles - this means no step counting
- Please log all your miles by 11:59 pm ET September, 30, 2021.

How do my miles translate to money raised?

After a racer is registered, they are set up with their own personal donation page. You can direct those who would like to donate to your race link.

Learn more or register at https://runsignup.com/Race/IL/Batavia/HCURaceforResearch

info@hcunetworkamerica.org | https://hcunetworkamerica.org | 630-360-2087

Focused on the Few® ...

At Recordati, we focus on the few - those affected by rare diseases. They are our top priority and at the core of everything we do. Our mission is to reduce the impact of extremely rare and devastating diseases by providing urgently needed therapies. We work side-by-side with rare disease communities to increase awareness, improve diagnosis and expand availability of treatments for people with rare diseases.

We are proud to support the mission and vision of the HCU Network America

RECORDATI RARE DISEASES GROUP
Focused on the Few®

www.recordatirarediseases.com/us
@RecordatiRare
NP-RRD-US-0201

SPEAK UP FOR THE MEDICAL NUTRITION EQUITY ACT

1. **The best way that you can help make sure that the Medical Nutrition Equity Act (MNEA) moves this Congress is by asking contacting your members of Congress NOW.** Please go to nutritionequity.org/contact-congress and use the simple form to send an email to your members of Congress asking that they co-sponsor the bill. It doesn't matter if you have sent an email before. We encourage them to keep asking your members of Congress to sign-on until they do so.
2. **Please also ask contribute stories to the website.** The patient story form is here and the one for medical providers is here. We currently have ZERO or very few stories from a few key states, including Maine, Rhode Island, Mississippi, and several others.
3. **If you have had a meeting with a congressional office in the last few months asking them to co-sponsor the bill, please make sure that you are following up with those offices to see if they have any questions, or what they need to become co-sponsors.** The coalition has created a webpage with a wide-range of resources that should answer most questions that come up, but if you don't see what you need, please let me know.

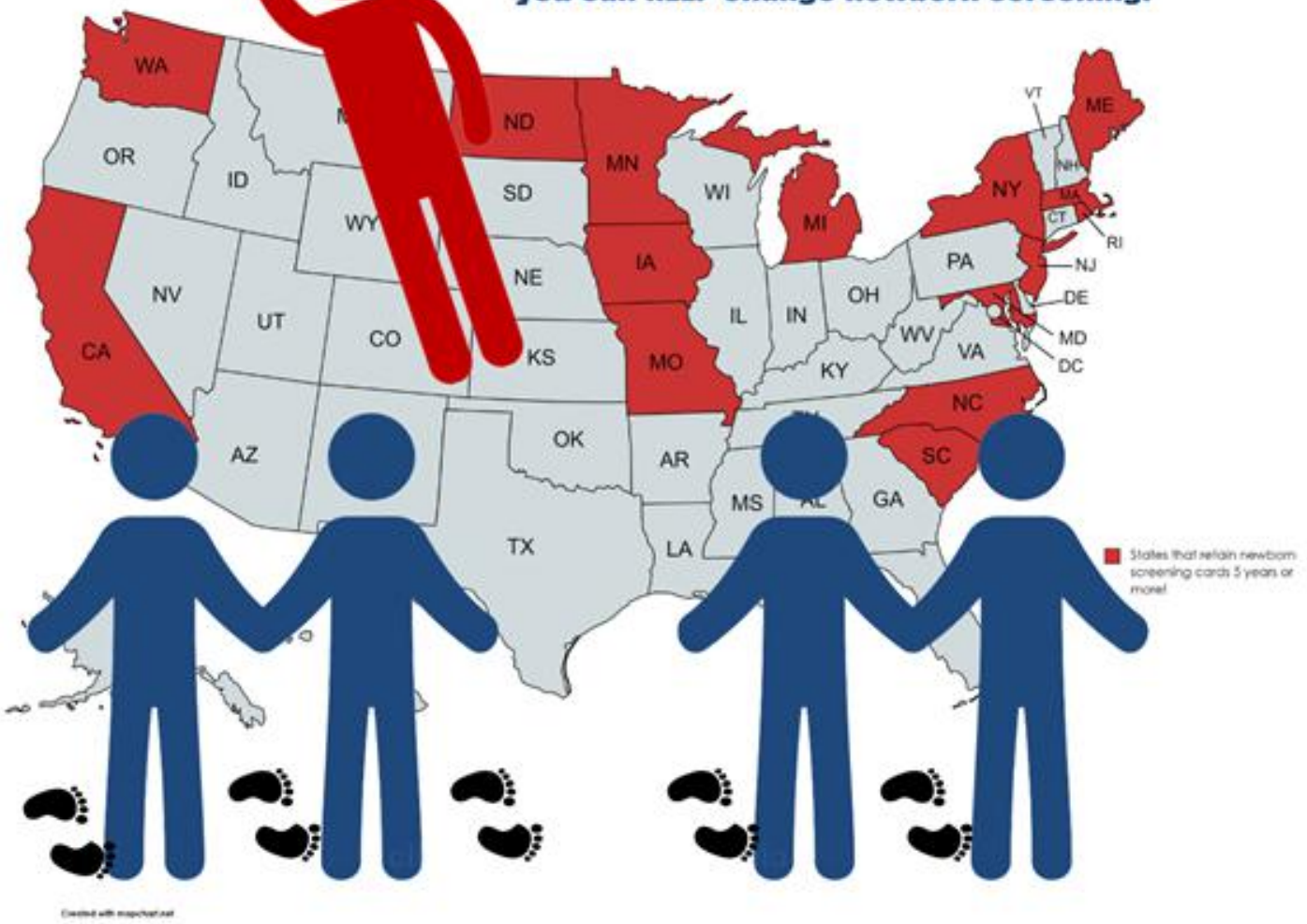
We can get MNEA passed if we all work together to build the support that we need!

The Patients & Providers for Medical Nutrition Equity Coalition

Were you or your child born in one of the highlighted states below?

Were you or your child diagnosed with Classical HCU,Cobalamin with HCU or Severe MTHFR?

If you answered yes, you can HELP change newborn screening!



Emory University is conducting a newborn screening research study on these disorders!

Contact Angela Wittenauer MSN, FNP-C, RN: alwitte@emory.edu | 404-778-8489
Director, Newborn Screening Follow Up Program | Emory Univ. Dept. of Human Genetics

"Regarding speech therapy services, early childhood services, our area really has nothing. **So COVID actually opened up the world a little bit more because now we have access to telehealth services."**

JODI TERVO ROBERTS
MOTHER OF A CHILD WITH HEARING LOSS

The Newborn Screening Family Education Program, a core program of Expecting Health, is excited to share with you the recordings of our recent webinar series, *From Emergency to Everyday: Reflections on a Year of COVID-19 in Newborn Screening*. During these three sessions, we heard from key stakeholders in the newborn screening community including healthcare providers, laboratory programs, and families.

In this series, the newborn screening community came together to discuss what we can "start, stop, and continue" doing to support families through future health emergencies. Together, we brainstormed solutions to challenges individuals and families faced with regard to newborn screening during the COVID-19 pandemic. **Check out what we discussed by watching the webinar recordings on YouTube.**

Provider Reflections on a Year of COVID-19 in Newborn Screening (March 2021)

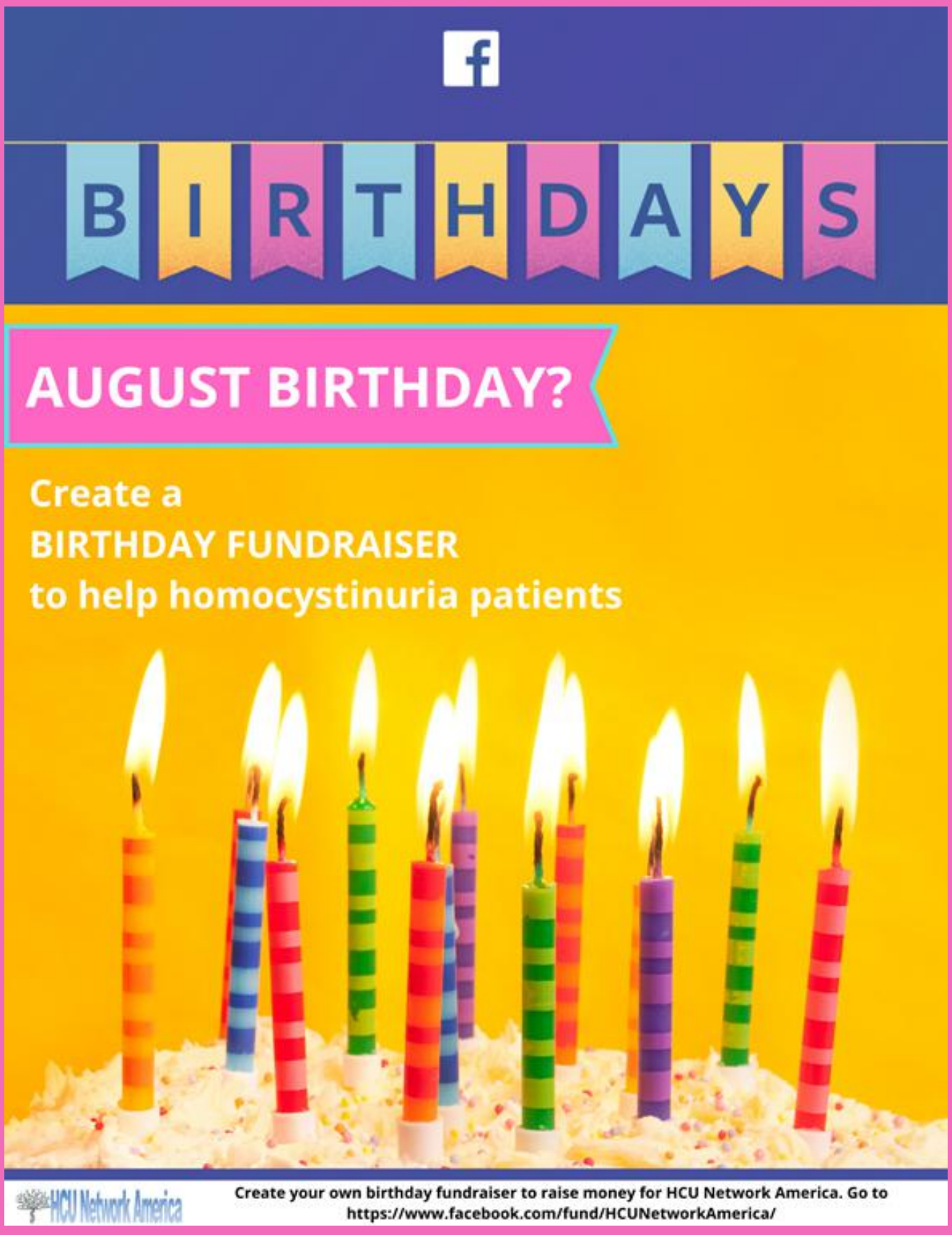
State Newborn Screening Program Reflections on a Year of COVID-19 (April 2021)

Family Reflections on Newborn Screening on a Year of COVID-19 in Newborn Screening (June 2021)

Look out for additional resources coming soon to support and engage families during future health emergencies!

CELEBRATE YOUR BIRTHDAY WITH A FACEBOOK FUNDRAISER!

Did you know that July through September have the overall highest birthrates with August or September typically having the highest number of births?



HCU NETWORK AMERICA

IS LOOKING FOR STATE AMBASSADORS

Looking for active and outgoing members of the HCU community

What does an ambassador do?

Ambassadors...

- *Connect with local HCU families*
- *Share their story*
- *Advocate and raise awareness for HCU*
- *Amplify and support our mission*
- *Help fund-raise*

Get involved today! Contact Danae'
dbartke@hcunetworkamerica.org

BECOME A STATE AMBASSADOR FOR HCU NETWORK AMERICA

HCU Network America