The HCU *Herald*

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HCU HERO: VALERIE FROM ARIZONA

Hello, my name is Valerie and I have Classical Homocystinuria (HCU).

I was fortunate enough to have been born in Arizona. Arizona is one of the few states that tested for HCU 34 years ago. As a result, I was diagnosed at birth with Homocystinuria through the Newborn Screening Program. I feel very blessed to not only have been born in Arizona with early diagnosis, but also to have such strong, dedicated, amazing parents who along with the medical team helped navigate my life with HCU. Receiving the news at the time was heart shattering and nerve wracking to say the least for my parents. The doctor explained I had a rare genetic disorder that can lead to cardiovascular complications, skeletal issues, vision disturbances and developmental and potential mental delays. This would be any parent's nightmare. This was before the internet and little research was known about the condition. My parents spent endless days at the public library scouring articles and trying to learn more only to find a few paragraphs. With the help of the genetics team, my parents had mastered the skill of never making me feel different from others, but always accommodating my needs. Nothing really prepares a parent for weekly blood draws and having to explain diet restrictions to a young child. In addition, there is the constant worry about side effects from this disorder and the potential financial toll this may have in the future. I certainly did not make it easy for my parents in my later years when it came to complying with my diet.

My childhood was no different from any other child without this condition. I participated in sports, went to camps, Girl Scouts, had sleepovers, and finished all my schooling. There were of course differences when social events happened. I ate different and had to take medication as a substitute. I often hid my formula from friends and woke up early at camps so no one would know. I referred to my condition as being a vegetarian because that was easier to explain to young children and even some adults. As I got into my late teens after high school, and started college in my twenties, that is when my self-discipline went astray, and I struggled with diet, depression, and anxiety.

Like many college students I made many memories and threw a lot of parties, went to concerts, and played sports. I had always been a

social person. I love people, and college was fun. I began to not take my formula, which was always hard to drink as it tastes disgusting no matter how you try to mask the taste and smell. I ate whatever I wanted, even with a guilty feeling that I knew it was wrong. Unlike diabetes, you don't feel sick initially. My levels kept climbing, and I knew what I was doing was wrong, but became depressed and threw in the towel with a feeling of hopelessness. Eventually this all came crashing down when my levels reached an all-time high of over 500, and I did eventually feel sick; brain fogged, disoriented, tired, and my vison started to rapidly change.

After seeing 25 ophthalmologists, 4 surgeons from Arizona, Iowa, and Utah with each one knowing my history with HCU, they could not figure out why my vison was declining. Then I met Dr Richardson. He was able to explain my lens had dislocated. He never made me feel scared and has helped me tremendously. This was my biggest fear with HCU. It was also my wake-up call to be better.

After 5 years of working on myself and getting back to caring about my levels, they are finally at a good point. Although I still struggle daily to maintain compliance with diet and medication, and deal with depression, I still consider myself blessed. At 34 years of age, I have completed college: I will be starting my own family, and I have a support system. It can always be worse, and it may get worse with time, but in this moment, I am doing better.

In writing this I hope my story can help anyone who is struggling with diet and that I can be an example that no matter how off diet you get you can always change to be better. Eventually it catches up to you, and if you feel like throwing in the towel, I have been there and I'm always open to lending an ear. My advice is to find support from people living with the condition, find a formula that works for you, be your own advocate, and take one step at a time. HCU does not define who I am as an individual, but it is a part of my life.

Travere Therapeutics Announces Positive Topline Results from the Ongoing Phase 1/2 COMPOSE Study of Pegtibatinase in Classical Homocystinuria

December 15, Travere Therapeutics, Inc. announced positive topline results from the ongoing Phase 1/2 COMPOSE Study of pegtibatinase, an investigational enzyme replacement therapy being developed for the treatment of classical homocystinuria (HCU). For HCU patients in the highest dose cohort to date (evaluating 1.5mg/kg of pegtibatinase twice weekly given subcutaneously), treatment with pegtibatinase resulted in rapid and sustained reductions in total homocysteine (tHcy) through 12 weeks of treatment, including a 55% mean relative reduction in tHcy from baseline as well as maintenance of tHcy below a clinically meaningful threshold of 100 µmol. To date in the study, pegtibatinase has been generally well-tolerated. Travere now intends to begin discussions with regulatory agencies on the design of the Phase III study that will be required before marketing approval.

We'd like to recognize the Hempling Foundation for Homocystinuria Research, which was started by HCUNA's President, Margie McGlynn, in honor of her sisters, for the investment in the original proof-of-concept study, as well as, the late Dr. Jan Kraus and Tomas Majtan who discovered the product and conducted the preclinical research. Without them, we would not be at this point where we are hopeful that a new treatment will be available in a few years to help patients better control HCU and improve their quality of life.

To read the full press release, please visit: https://ir.travere.com/news-releases/news-release-details/travere-therapeutics-announces-positive-topline-results-ongoing

Travere is still recruiting patients into the Phase I/II study to gather additional data that will help with the design of Phase III. See ad below if interested

Compose

COMPOSE Study

A study that looks at how safe pegtibatinase is and how well it works in people with classical homocystinuria (HCU)

NOW ENROLLING

Travere Therapeutics has initiated a first-in-human study of pegtibatinase, a new, investigational human enzyme therapy that targets the underlying enzyme deficiency that causes HCU.

The goal of this study is to learn how safe and effective pegtibatinase is and how well it works in people with HCU at different dosage levels.

Approximately 32 subjects will participate in sites in the US. The study will include three key stages (screening, treatment, and extension) and will last approximately 150 weeks.

You (or your child) may be eligible to participate in the COMPOSE Study if you:

- **Have been diagnosed with HCU**
- **Are 12–65 years of age**

You (or your child) will need to meet all other study criteria to take part in the COMPOSE Study.

For additional information on criteria for eligibility, please go to: www.clinicaltrials.gov/show/NCT03406611

Payment for time and travel may be available to subjects who participate in this study.
To inquire about participation in the study, **please contact**:

HCUConnect@labcorp.com

TRAVERE™ THERAPEUTICS

RESEARCH OPPORTUNITIES

We will be launching in soon - sign up to get an email when we launch!
https://hcunetworkamerica.org/homocystinuria-data-collection-program-email-contact/

To learn more visit:
https://hcunetworkamerica.org/homocystinurias-data-collection-program

express™ has been updated to express™ plus+

Vitaflo
Enhancing Lives Together
A Nestlé Health Science Company

Now available!

Announcing... express plus is now available for Homocystinuria (HCU)

Changes from express to express plus include:

express™ plus+

A new name

A new look

And... **because we always try to bring you the latest in nutrition science...**

- Updated nutrition profile
- Increased vitamin D
- Addition of DHA

A new formula prescription will be needed going from express to express plus.

For more information, including nutrition details:

HCU express plus product details

Click to request a sample!

Yours in good health,

The Vitaflo North America Team

Vitaflo
Enhancing Lives Together
A Nestlé Health Science Company

CONTACT
Vitaflo™ USA, LLC
1007 US Highway 202/206
Building JR-2
Bridgewater, NJ 08807
Order Line: 888-848-2356
Fax: 631-693-2002

Email: vitafloNAM@vitafloUSA.com
Web: www.VitafloUSA.com

Please follow the link below to fill out the form to get in touch with a Vitaflo representative.

LEARN MORE

January 2022, HCU and You: Ask Methia

Dear Methia

The Dreaded Formula Change

I have been prescribed the same medical food/formula for the past five years. It's now a huge part of my daily routine, and it's one of the few formulas I've been able to take consistently. Recently, I learned that the manufacturer is changing the ingredients, and I'm panicking! What if it tastes different and I don't like the new flavor or consistency? I want to go into this change with an open mind – do you have any suggestions on how to make the transition?

Sincerely,
A Friend in a Formula Frenzy

Dear *A Friend in a Formula Frenzy,*

This isn't the first time a manufacturer has changed a well-loved formula, and it won't be the last. Research and Development professionals are constantly looking to re-vamp their products to make sure that they are up-to-date with nutrition guidelines. They are also trying to improve the flavor profile of their products, but this comes with the risk that the change will no longer be preferred by loyal consumers. I absolutely understand that this is a stressful time. Here are some suggestions on how to adjust and stay on diet.

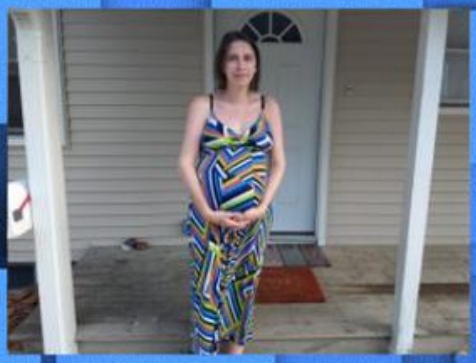
- Just try the new formulation, as it is. There's a chance that you will taste little to no difference in the product. If that's the case, fantastic!
- Titrate "old" formula with "new" formula. If you notice that the new formulation tastes different in a way that you're not sure you could drink long-term, try adding a few ounces of it to your old formula. Start with 25% (for example, adding two ounces of the new formula to six ounces of old formula). Gradually increase the amount of the new formula in proportion to the old formula so that you adjust to the taste over time.
- Add flavorings. Popular ways to improve the flavor of formula include Mio Drops, Crystal Light, and fruit juices. This is also a chance to try new ways to drink your formula, and pack in extra nutrition. For example, a berry-flavored formula with extra acai juice might taste great in a fruit smoothie!
- Ask to try samples of other formulas. If the new formulation is a no-go, use this as an opportunity to try new products. The best person to talk to about what would be a good fit for you is your dietitian. Your dietitian will recommend products to try that will meet your nutritional requirements (everyone's are different!) and are the best for your lifestyle.
- Think about using a combination of medical foods to meet your needs. There's a chance that the new formulation will work for you, but perhaps not every day ("flavor fatigue" is real!). There's also a chance that your ability to take a certain necessary volume of formula will change. That said, combining the new formula with a product such as Easy Tabs or a quick ready-to-drink product might work better for you

Your metabolic clinic is always there to help you find a solution to hiccups in your nutrition prescription and formula routine. Make sure you stay connected with them and communicate about the things that aren't working (and the things that do work, so that other people can benefit from your creativity!) throughout this time.

Sincerely, Methia

CLASSICAL HCU AND COBALAMIN DISORDERS
SHARE YOUR STORY

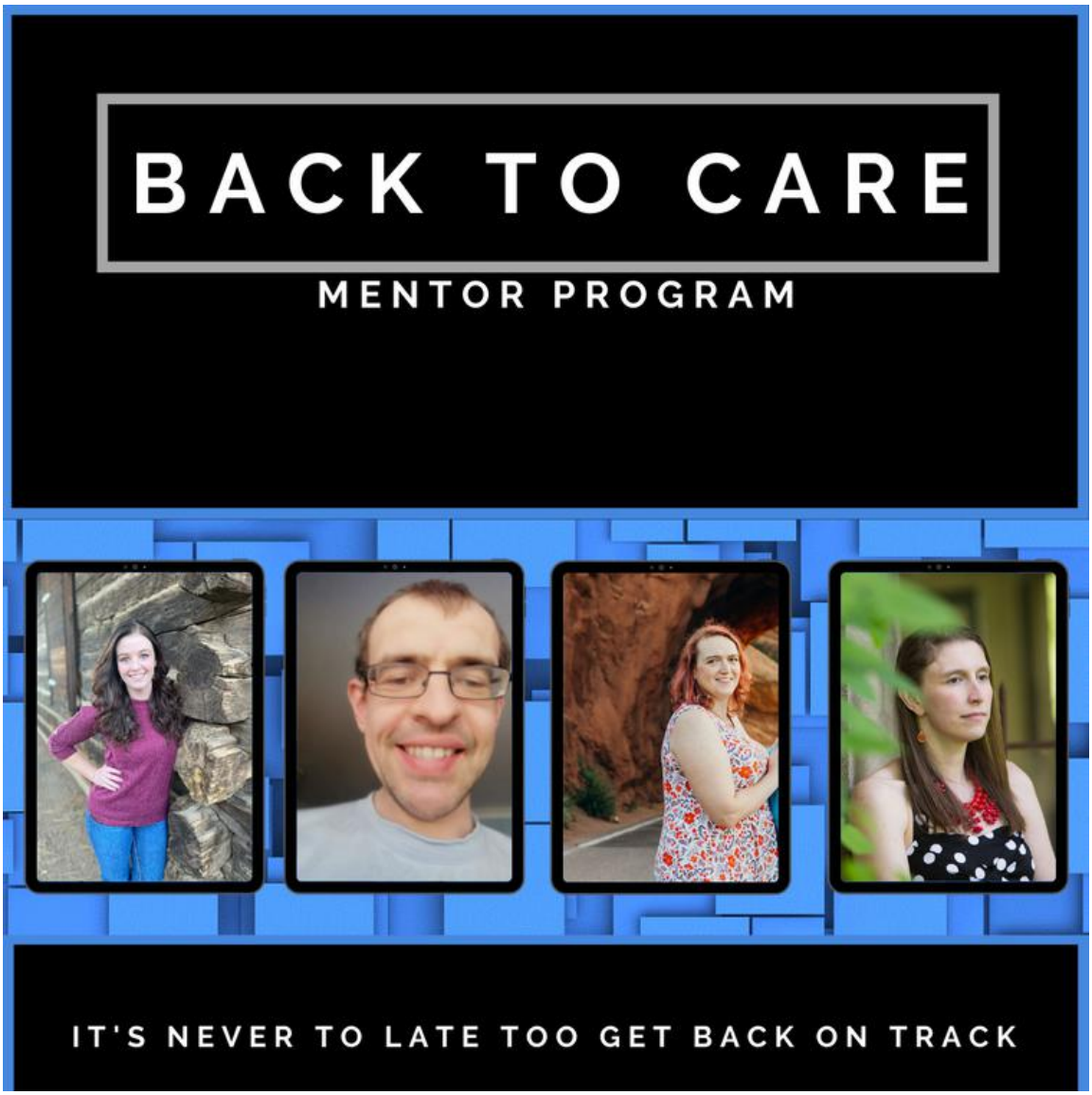
FOR THE MONTH OF JANUARY, SHARE YOUR STORY ON NUTRITIONEQUITY.ORG AND WIN A $100 GIFT CARD



If passed, the Medical Nutrition Equity Act would require insurance companies to cover medical formula, low protein medical food, and hydroxocobalamin. Add your voice to those fighting for medical nutrition coverage. Submit your story: https://nutritionequity.org/share-your-story/

We will select one classical Homocystinuria patient story and one cobalamin patient story from those submitted in January! After your story is live on the Nutritionequity.org site – share it with us to be entered.

NEW RESOURCES



Back to Care Mentor Program

For HCU Network America, 2021 was all about Back to Care. We know it's easy to stray away, but as patients age, they realize the importance of keeping in contact with their clinic, following the diet, and keeping themselves all around healthy. This past year we took steps to help patients get back on track and back to care. In the Spring, we introduced the program via our Back to Care webpage, then in the summer we published our Back to Care Guide. We closed the year (but not in time for the December HCU Herald) with the launch of our Back to Care Mentor Program, and already have our first official patient enrolled.

If you are interested in becoming a mentor, or enrolling in the Back to Care Mentor Program, please visit: https://hcunetworkamerica.org/back-to-care-mentor-program/

NEW RESOURCES

While we didn't officially launch our Back to Care Mentor Program until December, we have been quietly planning and speaking with patients who were, or are, in the process of coming back to care. These patients helped mold our back to care program – one of those such patients was Ashley.

In the past year Ashley has worked hard to transform his diet, and we are proud to say Ashley is successfully back on track. Because of the awesome job he has done at adapting to this, Ashley was interviewed by the HC&U Podcast team. So, get inspired and listen to his experience on Back to Care. Listen here: https://www.stitcher.com/show/hcu/episode/4-interview-with-ashley-longthorn-87448223

NEW RESOURCES

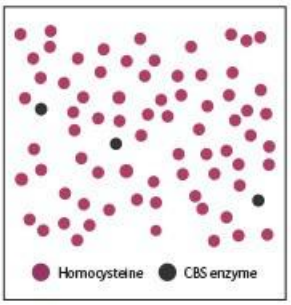
Check out this amazing new resource from Recordati Rare Diseases! Click here, or the infographic to see the full infographic on living with **Classical Homocystinuria, Also known as CBS deficiency**

CLASSICAL HOMOCYSTINURIA

Also known as CBS deficiency

What is it?

A rare inherited disorder caused by a deficiency of an enzyme called cystathionine beta-synthase (CBS).



Not enough CBS enzyme activity

The CBS enzyme helps the body metabolize, or break down, homocysteine. If there is not enough CBS enzyme activity, homocysteine builds up in blood and urine, resulting in **homocystinuria (HCU)**. This can cause serious health problems.

Possible signs and symptoms

- Severe nearsightedness
- Dislocation of the lens of the eye
- Learning problems and developmental delays
- Behavior problems
- Long limbs
- Curved spine
- Weak or brittle bones
- Blood clots
- Strokes

Affected children may be slow to crawl, sit up, walk, and talk. Early intervention programs may help children develop skills.

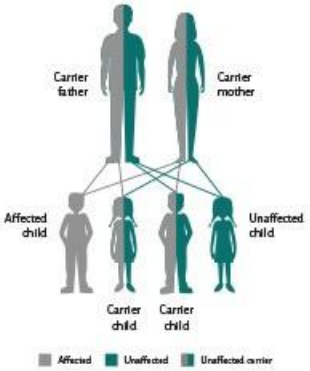
The effects on the body

HIGH BLOOD HOMOCYSTEINE LEVELS can harm the body, mainly in four areas:

brain eyes bones blood

It begins with mom and dad

A child with classical HCU has inherited two *CBS* genes with mutations – one from each parent. People with one unaffected gene and one gene with a mutation are known as carriers. They do not have symptoms. When each parent is a carrier of a *CBS* gene with a mutation, each child in the family has a 25% chance of inheriting classical HCU.



Spicy "Tuna" with Teriyaki Sauce

Yields 1 serving | Serving size: 1 recipe | Protein per serving: 0.8 g | Calories per serving: 67

Ingredients:

- 98 g Small Cubed Watermelon
- 28 g Teriyaki Sauce (see below)
- 1 tsp Minced Ginger
- 1/2 tsp Siracha Sauce, optional

Homemade Teriyaki Sauce

- ½ c. coconut aminos
- 2 Tbsp orange juice
- 2 tsp grated ginger
- ½ tsp fresh, chopped green onion
- 2 tsp brown sugar
- ¼ tsp cornstarch mixed with 1 tsp water

Tuna Directions:

1. Combine all ingredients above in an airtight container and toss to well coat the watermelon. Seal the container and place in fridge to marinate for at least 30 minutes.
2. Once marinating time is done, turn oven on to 350 degrees to preheat. Line a small baking sheet with foil and spray with nonstick spray. Scoop out the watermelon and place on prepared baking sheet. Toss the rest of the marinade.
3. Bake for 15-20 minutes, stirring the watermelon halfway in between. You want the watermelon to begin to look like raw tuna. Remove from oven. This is now ready to use for so many dishes! Can be used warm or cold. Make a poke bowl with rice and vegetables of choice. Drizzle with sriracha mayo for an extra kick. Make sushi rolls, tacos, sashimi, etc.

Teriyaki Sauce Directions:

1. In a small saucepan, combine the first five ingredients. We will add the cornstarch mixture later.
2. Heat the saucepan over medium to medium low heat, stirring frequently, until sauce begins to simmer. Now add the cornstarch mixture and continue to stir, bringing back to a simmer. Continue to simmer until sauce is thick enough to coat the back of a spoon. Remove from heat. Sauce will thicken a little more as it cools. Use right away as a stir fry sauce or as a marinade. Or you can keep in a small glass jar until ready to use.

Pineapple Kiss

This is a quick sweet sauce I came up with to use up some pineapple. If you have the tolerance, you can add a little toasted coconut for nice flavor and texture. This is great with the Cook for love pound cake, ice cream, or alone.

Yields 7.26 serving | Serving size: 1/4 cup | Protein per serving: 0.7 g | Calories per serving: 130

Ingredients:

- 2 TBSP Butter
- 1/4 c Packed Brown Sugar
- 1/2 tsp Rum Flavoring
- 1/2 tsp Vanilla Extract
- 2 c Pineapple chunks
- 1 c Coconut Milk, canned
- 2 tsp Cornstarch, mixed with 2 tsp water to make slurry
- 1/4 tsp Salt, Table

Directions:

1. In a medium skillet over medium heat melt the 2 tablespoons of butter. Add the brown sugar and stir. Heat until smooth. Add the rum and vanilla and stir. Next, add the pineapple to the skillet and stir to coat with sugar mixture. Continue to stir over medium heat until the pineapple gets a little caramelized.
2. Pour in the coconut milk and bring to a simmer. Stir in the prepared cornstarch slurry, stirring constantly until thickened enough to coat the back of a spoon. Remove from heat and stir in the salt. Serve while warm or you can chill and serve cold.

2021 $25K MATCH CHALLENGE HIGHLIGHTS

FABULOUS HCU AWARENESS FUNDRAISERS

Our annual appeal started in October with HCU Awareness month. We had a good start to the month with several fundraisers hosted on behalf of HCU Network America. In October we raised $5,100 thanks to the 6 families who set up fundraisers on behalf of the organization and to the many others for their individual donations.

In November we had another wonderful opportunity to raise awareness and funds for HCU Network America with Thanksgiving and Giving Tuesday. We also sent an email with an appeal letter that those in the community could share with their family, friends, co-workers and others. By the first part of December, we were at more than half way to our goal. Thank you to those who set up #GivingTuesday fundraisers.

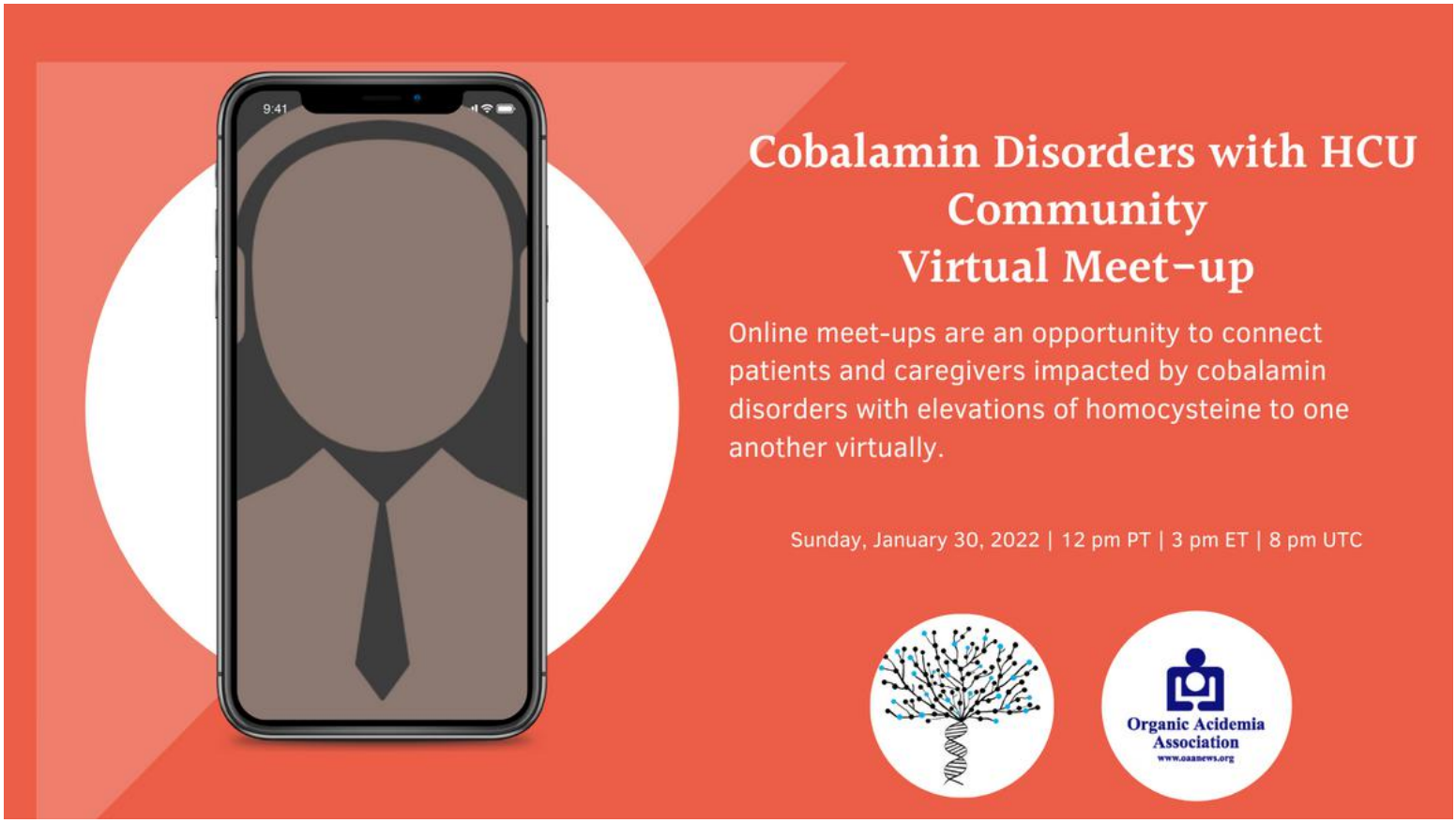
By mid-December, as our annual appeal was about to come to a close, we sent out a few more emails recapping our accomplishments of the past year. Because of our successful campaign, we were able to accomplish a lot in 2021! With these emails we were able to take in **$27,241** to continue our mission in 2022, and we will receive a match for the $25,000 from our anonymous donors. We are very thankful for the generous donations from our supporters! This was our fourth year that we not only met our goal, but surpassed it! We couldn't have had a successful 2021 without you and know this will be true for 2022 as well!

Giving Tuesday Fundraisers

UPCOMING EVENTS

The Rare Disease Legislative Advocates (RDLA), a program of the Everylife Foundation has announced the schedule for Virtual Rare Disease Week on Capitol Hill. The week brings together rare disease community members from across the country to be educated on federal legislative issues, meet other advocates, and share their unique stories with legislators.

No prior experience is necessary. Registration for this event and all RDLA events are free for all rare disease advocates. To learn more and register, visit https://everylifefoundation.org/rare-advocates/rare-disease-week/



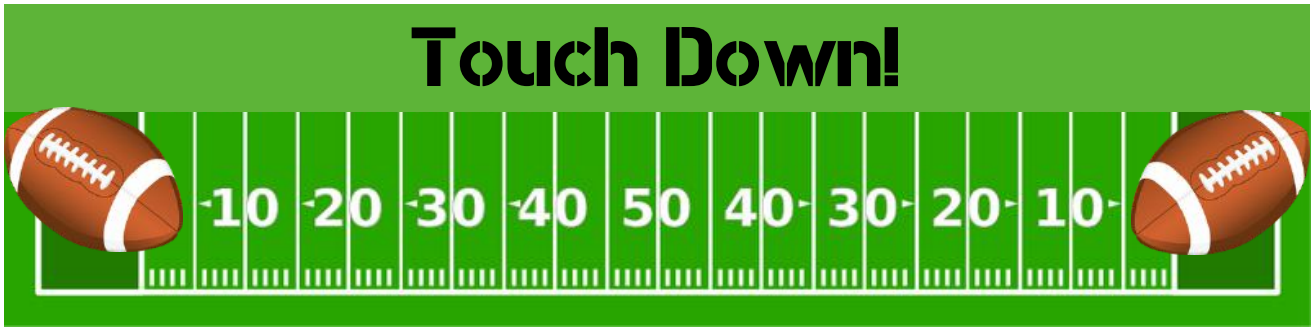
Register for all our virtual meet-ups at: https://www.eventbrite.com/o/hcu-network-america-30163980100

UPCOMING EVENTS

Register for all our virtual meet-ups at: https://www.eventbrite.com/o/hcu-network-america-30163980100

Learn more and register at: https:/hcunetworkamerica.org/2022-conference

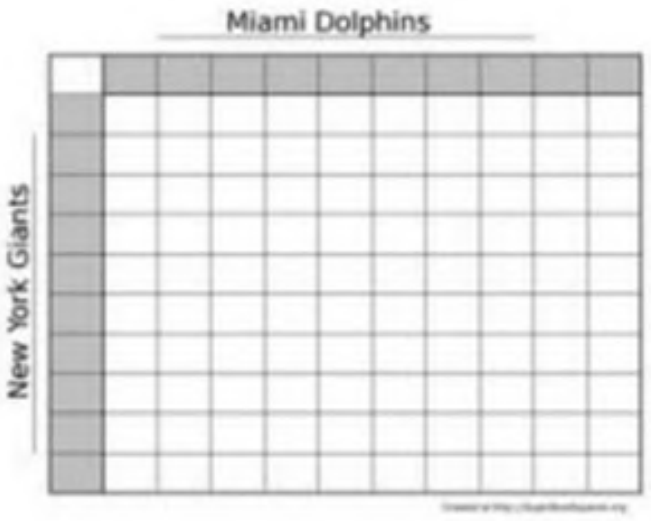
JANUARY FUNDRAISING EVENT: SUPER BOWL SQUARES



Big sporting events are a great way to not just show your team spirit, but they are a fantastic way to **raise funds for your charity of choice - HCU Network America!**

How it works:

The cost is $5 or $10 donation per square. The purchaser writes his or her name in the suqare, and then the seller will randomly pick numbers 0-9 from each team in the game, and assign that number to a particular row or column (the grey shaded columns). These numbers represent the last numbers in the score of each team at the end of the game. In other words, if the final score is Giants 17 - Dolphins 14, then the winning square is the one with a Giants number 7 and a Dolphins number of 4. Since no one knows what numbers each square will represent, the odds are the same for everyone. So good luck and have fun!



Winner Breakdown:

If you sell 100 squares (you must sell all squares to host the fundraiser) at $10 each, the funds raised are $1,000. You can chose to split the pot evenly and the winner will get $500 or get a large item donated for the winner and keep all funds raised toward your fundraising effort. (You can also decide to split the prize money and give some at half time or the end of each quarter.

For further instructions and templates:
https://www.printyourbrackets.com/nflweekly100squares.html

For online tools, check out:
http://footballsquaresonline.com
https://www.runyourpool.com/nfl-football-pools.cfm

NEED HELP HOSTING A FUNDRAISER? EMAIL US - INFO@HCUNETWORKAMERICA.ORG

WHAT'S YOUR STORY?

More than 30 million Americans are living with one or more rare diseases.
Each one of us has a story. We want to hear yours.

Today, the EveryLife Foundation for Rare Diseases launched *What's Your Story?*, a new online rare disease story bank for advocates like you to share your experiences on a wide-range of issues impacting the rare disease community.

What's Your Story? presents a series of questions covering topics ranging from difficulty in getting diagnosed to challenges in gaining access to therapies, to facing discrimination in the workplace, and more. You are also invited to upload photos and videos that help illustrate your rare disease journey. Advocates of any age are welcome to submit stories.

Share Your Story With Us

Benefits of sharing your story:

Influence rare disease policy.

Educate policymakers.

Inspire other advocates.

Be entered to win a $1,500 donation to the patient advocacy organization of your choice! Submit your story by March 2nd, the final day of Rare Disease Week on Capitol Hill 2022 for the chance to win one of three donations.

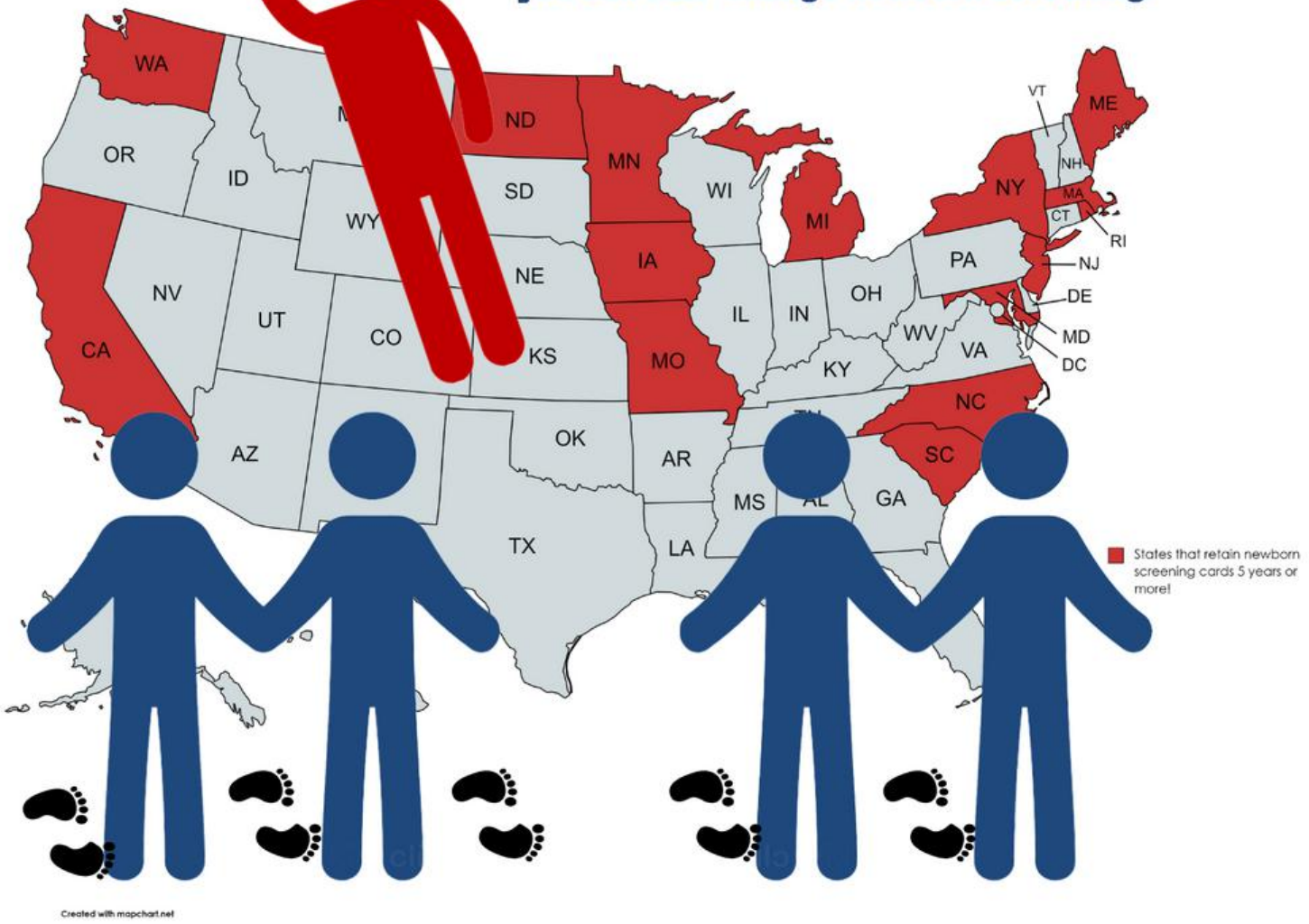
Stories submitted through *What's Your Story?* will help advance rare disease policy priorities through ongoing advocacy efforts to advance the equitable development of and access to lifesaving diagnoses, treatments, and cures. Submissions may be shared during meetings with Members of Congress or with the public via social media, online or in the press.

EVERYLIFE FOUNDATION FOR RARE DISEASES

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

Live better, together!

Contact Register

What is the contact register?

The contact register is a secured private survey that allows you to share information on you or your family member with HCU with us. This includes where you are from, your relationship to homocystinuria, the patient's birthdate, gender, their exact diagnosis (e.g. CBS, cobalamin, or MTHFR), how they were diagnosed, and if the patient was diagnosed through newborn screening. This information is kept confidential and will not be shared unless you give us permission. By registering, you will be able to identify other patients in your state and request their contact information. You will also be able to access information posted over time that can only be shared with the patient community. (For example, we may have webinars that the expert presenter does not want to be publicly available, but is willing to share with the HCU community.)

What will this information be used for?

HCU Network America strives to inform patients and families with resources, create connections, and support advancement of diagnosis and treatment of HCU and related disorders. The information you provide helps us succeed in our mission - plan events, develop resources and educational tools, and ensure everything is being done to support timely and accurate diagnosis from birth. It also allows us to have informed conversations with doctors, pharmaceutical companies, and law makers. Your information helps us understand the landscape better so we can better advocate for you!

How do I participate?

The contact register form takes approximately 3-5 minutes to complete. You can find the form either by visiting our website and clicking on the "Contact Register" tab, or you can fill it out by going directly to: https://hcunetworkamerica.org/contact-register/