HCU Network America

The HCU *Herald*

All things Homocystinuria: patient stories, resources, research, events and more!

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-0285

HCU HERO: DIANA (CBLC) FROM MEXICO

Almost all the people around me, even the people closest to me, don't know that I have a rare disorder. I think it's because they are embarrassed to ask, and they think I'm going to be offended or feel bad. A friend recently told me that her mother asked her what condition I have and she couldn't answer because she didn't know. She explained to me that she had never asked for fear of making me feel uncomfortable. Since I lead such a normal life and I interact like any other person, she didn't feel she had to because I did not need her to help me with anything. Also, because she thought that since we talked about everything except that, that meant I didn't want to talk about it. I told her that it's a topic I never bring up because she never asks me and that I don't just randomly tell people what I have.

DIAGNOSIS

My name is Diana, and I was born in 1998. I'm currently 24 years old, and I was the product of a full-term pregnancy without problems nor complications. Nevertheless, when I was a baby, I didn't crawl or stand up in the crib and that was a cause for concern for my parents. My pediatrician thought that it didn't represent anything important or concerning. When I was about 1 year old the situation had not improved and my parents took me to a traumatologist. They told my parents that there was nothing abnormal and that they were amplifying the situation by worrying so much. They told them that there was nothing wrong with me.

After that, they took me to a neurologist where they did several tests in which all of them came out fine but the doctor noticed that I had low muscle tone, and he recommended physical therapy.

After a year and a half in therapy, the therapist recommended we see a geneticist. At 3 years old, I was diagnosed with Methylmalonic Acidemia and had to treat it with oral vitamin B12, carnitine and a low protein diet. I had slight improvements, but still had delays in my walking and gross motor skills. At 3 years old I was hospitalized for bronchiolitis that ended up in pneumonia. I was in serious condition and my family even thought I wasn't going to make it. One day when my grandmother was taking care of me in the hospital, she called my parents to tell them the doctors said they should come immediately. We all know what that means, so I

can't imagine the immense scare and pain my parents went through, but thank God I'm still here. However, I still have sequels, so I have to take care of myself.

When I was 4 years old, we went to Houston for a second opinion. When I got there, I had normal language development, good social and fine motor skills. I was meeting all my milestones adequately, without any type of mental delays or seizure disorder. I was doing well in school and my parents say I was even doing better than most of my classmates. However, I had delayed gross motor development and decreased reflexes. I couldn't walk on my own or even stand without support. I could sit unassisted in a somewhat tripod position. I couldn't crawl, but when I stood next to furniture or a wall I could walk around the room. In Houston, they told me that I have B12 Disorder and that I should treat it with physical therapy, carnitine and daily injections of hydroxocobalamin, not cyanocobalamin, which is what they gave me in México.

The doctors recommended my diet to be liberalized, and I was now allowed to eat whatever I wanted. This was because my dietary protein wasn't significantly restricted and my methylmalonic acid level had normalized, so they anticipated that I would do well on a regular diet, carnitine, hydroxocobalamin injections and physical therapy. They sent me immediately to a therapist at the hospital to get an evaluation. She said there weren't any solutions for me to be able to walk. I kept going for several years. I continued with the treatment and physical therapy, but as time went by, I stopped walking.

At the age of 9, I went to Cuba for 2 months to receive intensive physical therapy and they gave me another diagnosis called Muscular Dystrophy Type 2. I did make some improvements, but it was probably because I spent almost all day at PT. When I returned to México it was impossible for me to keep up with so many hours of therapy and continue with my normal life as well.

When I was 12, I had surgery for neuromuscular scoliosis due to spinal atrophy, which left me with a huge scar. It may sound like no big deal, but realizing you're going to have a scar all the way down your back for the rest of your life, when you're almost a teen, is not very cool. I spent 2 months in recovery, most of the time in

bed or on a couch. For some reason my legs were injured during the surgery, and I remember I went through a lot of physical pain. Also, my back and other body parts remained numb for months. Eleven years have passed and there were areas where I still didn't have much sensitivity.

As the years went by, I continued with carnitine and physical therapy, but I was getting fewer and fewer injections. I continued with my life and didn't give my health the necessary attention because for me I was "fine" and there was nothing else to do. I thought injections were for energy because most of the time I felt tired, but when I got injected, I felt the same way so I lost the motivation to do it. I really didn't know how important they were and that not having them could affect me.

REEXAMINING MY DIAGNOSIS

It wasn't until last year I reassessed my disease when I met a geneticist, who I now consider my friend. He was the first person with whom I discussed my disease in depth and with whom I even allowed myself to be vulnerable. I'm so grateful he came into my life in such a random and unexpected way because if that had not happened I would probably still not be receiving the proper treatment and something serious could have happened to me. Also, I feel thankful because that situation made me pay attention to my condition and the fact that I'm not okay with it. I told him about my case and he told me how important treatment is and that high homocysteine levels could cause serious consequences like blood clots, heart attacks or strokes; and that high levels of methylmalonic acid can cause kidney problems among other things. To be honest, I was very scared because I'm a very concerned person and in a certain way I felt responsible for my situation.

I began to ask myself a lot of questions. Among them, “What if I had followed the treatment properly? Would I be better? Could I be walking?”. It's worth mentioning the confinement due to the pandemic didn't help my overthinking because I didn't have much opportunity to clear my mind. Even when people started to go out I still didn't, as a precaution due to my history of pneumonia.

I had to collect all my medical records to send them to the doctor and it was very difficult for me to read them because, specifically the ones from Houston, were too explicit. They described everything from my personality and appearance, to the problems of my disease, and I learned many things about myself and my diagnosis that I didn't even know.

Since I had different diagnoses, I researched each of them, and I was so confused because I felt like I didn't fit into any of them. My doctor recommended that I should get a genetic saliva test so I did and we sent it to California.

When it came time to talk about the result, he told me that I was in a safe place to cry, scream or whatever I felt like doing. Then, he told me I have CblC Disorder and explained it to me. I was speechless. He asked me how I felt, and I really didn't know what to think or feel. I just told him I thought it didn't matter. That it sounded the same as what I had heard when I was a little, nothing new. Now that I think about it, maybe I expected a completely different result. Maybe subconsciously I was still clinging to being diagnosed with something different that actually has a cure or at least a treatment with which I could really see big improvements. I'm not really sure what I was expecting, but it certainly wasn't that.

He told me that it was something important because by knowing the type, it would be possible to know

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STATEMENT ON METABOLIC FORMULA SUPPLY ISSUES AS A RESULT OF THE ABBOTT PRODUCT RECALL

At the request of U.S. Food and Drug Administration (FDA), Abbott is releasing limited quantities of metabolic nutrition formulas that were previously on hold following Abbott's recall of some powder formulas from its Sturgis, Michigan facility.

The products have been tested and comply with all product release requirements before distribution. The lots being released were not included in the recall and are safe to consume.

Abbott has limited quantities of available metabolic products and we will be working with you to ensure available product is distributed to the patients with the greatest need. The products being released from hold and available for distribution include: Hominex-1, Hominex-2, Pro-Phree,

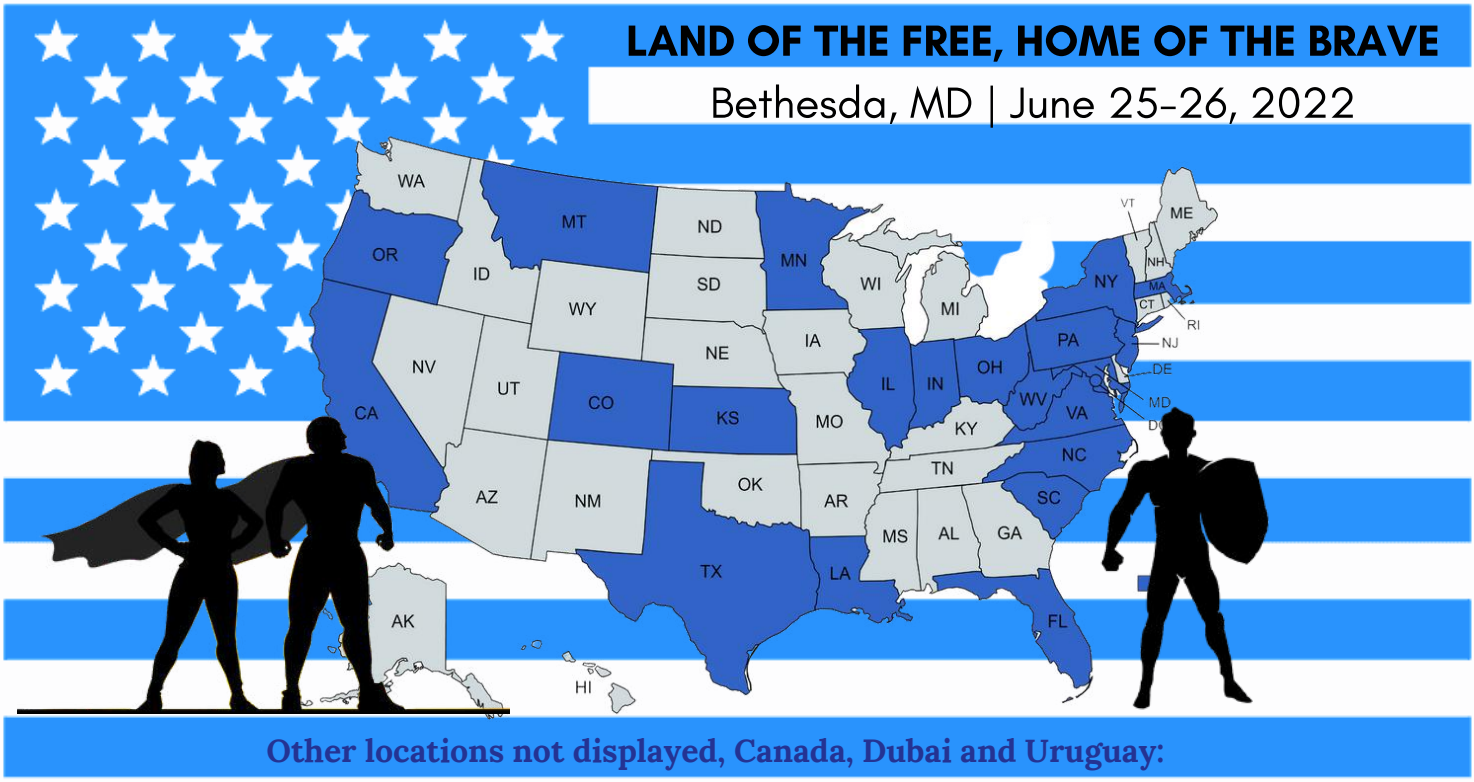
Patients will need a physician/medical professional order to obtain the metabolic product. Abbott is releasing this product at no charge to patients. Abbott is working closely with the U.S. Food and Drug Administration (FDA) to restart operations at the Sturgis, Michigan facility.

Abbott will be reaching out to metabolic healthcare professionals next week with specific instructions on the ordering process. If you have been affected by this recall and are in short supply, please contact your healthcare team to discuss the patient's needs.

Have you been affected by the FDA hold on Abbott products -- either because you can't get your Abbott formula, or another formula is in short supply as others switch from Abbott? The FDA needs to hear your story to understand the impact on our community. Please read our updated statement at metabolicformula.org and share your story to help raise awareness in the media and help the FDA understand the severity of this situation, so they will act to prioritize the clearing of Abbott's metabolic formula products.

Learn more & share your story at MetabolicFormula.org

HCU Heroes Unite!



The countdown is on, one month left to register!
Hurry – help us fill up our map and join in the fun!

Join us in Bethesda, MD for HCU Network America's third homocystinuria conference, Land of the Free, Home of the Brave. Join families from across the country in celebrating our brave HCU Heroes.

Registration runs till Friday, June 3, 2022! | Room rates go up May 27th!
Register aknd book your room at: https://hcunetworkamerica.org/2022-conference

- VENDORS
- NETWORKING WITH PATIENTS AND FAMILIES
- LEARN ABOUT THE BEST PRACTICES IN MANAGEMENT
- HEAR ABOUT CUTTING EDGE RESEARCH FROM RENOWNED EXPERTS
- ENGAGING KIDS' PROGRAM, WITH SELECT ACTIVITIES FOR TEENS

Attention: Volunteers Needed

For HCU Network America, Organic Acidemia Association and Propionic Acidemia Foundations Conference:
Land of the Free, Home of the Brave
(An "educational meeting" for patients and families affected by homocystinuria, a rare genetic disease)

Pooks Hill Bethesda Marriot | 5151 Pooks Hill Rd, Bethesda, MD 20814

We are now seeking volunteers to assist in our children's program:

- Arts & Crafts
- Educational Lessons
- Stories
- Games
- Hands on activities
- And more!!!

Children's program will take place during the main session of the conference.
(Saturday 8:00-5:30 and Sunday from 8:00-12:00)

Please see available slots at: https://signup.com/go/miiZeeB
or
Direct your contact with days and times of availability to:
Danae' Bartke, Executive Director at
info@hcunetworkamerica.org

Dear Families,

On behalf of the HCU Network America, Organic Acidemia Association and Propionic Acidemia Foundation, we offer our deepest condolences on your loss. We would like to honor your loved one during the upcoming 2022 HCUA, OAA & PAF Conference by having a Hero Remembrance photo poster displayed during the conference's dinner banquet.

If you would like to share a picture of your hero, please email Raymonde Degrace, degracemr@gmail.com. Please include your Hero's first name, type of homocystinuria, state and/or country, birthdate and Angel date. Photos must be received by Friday, June 3, 2022.

With Love,
Danae Bartke, HCU Network America
Kathy Stagni, Organic Acidemia Association
Jill Chertow, Propionic Acidemia Foundation

MEET OUR NEWEST MEDICAL ADVISOR

Peter Baker, MD

Dr. Baker is an associate professor of Pediatrics at the University of Colorado School of Medicine. Dr. Baker earned his M.D. at the University of Arizona. He received residency training in pediatrics at Oregon Health and Sciences University in Portland, Oregon. He then went on to pursue fellowship training in medical genetics and clinical biochemical genetics at the University of Colorado. He joined the faculty at the University of Colorado in 2013 and is now the Clinical Medical Director of Clinical Genetics and Metabolism and Director of the Fellowship Training Program in Medical Biochemical Genetics. He is an expert in inherited metabolic diseases and is intimately involved in the treatment of patients with these diseases at Children's Hospital Colorado. His basic and clinic research covers small molecule metabolism in rare and common conditions, and the diagnosis and management of metabolic disorders including phenylketonuria, organic acidemias, urea cycle disorders, fatty acid oxidation disorders, glycogen storage disorders, lysosomal storage disorders, and disorders of cobalamin metabolism.

WILL YOU BE OUR NEXT HCU HERO?

Tell us:

- How you or your child was diagnosed?
- How has HCU affected you, your family and relationships?
- What are some of your successes with HCU
- What are some of your challenges you have faced?
  - How have you overcome them?
- What words of advice would you give to newly diagnosed families?
- For other patient stories, visit: https://hcunetworkamerica.org/patient-stories
- Email your story to: info@hcunetworkamerica.org

RESEARCH OPPORTUNITIES

HOMOCYSTINURIAS
DATA COLLECTION PROGRAM

The Homocystinurias affect multiple systems of the body!

The most common areas of the body effected are:

- The Central Nervous System (brain)
- Ocular (eyes)
- The Cardiovascular System (heart)
- The Skeletal System (bones)

But many patients experience symptoms outside of these areas!

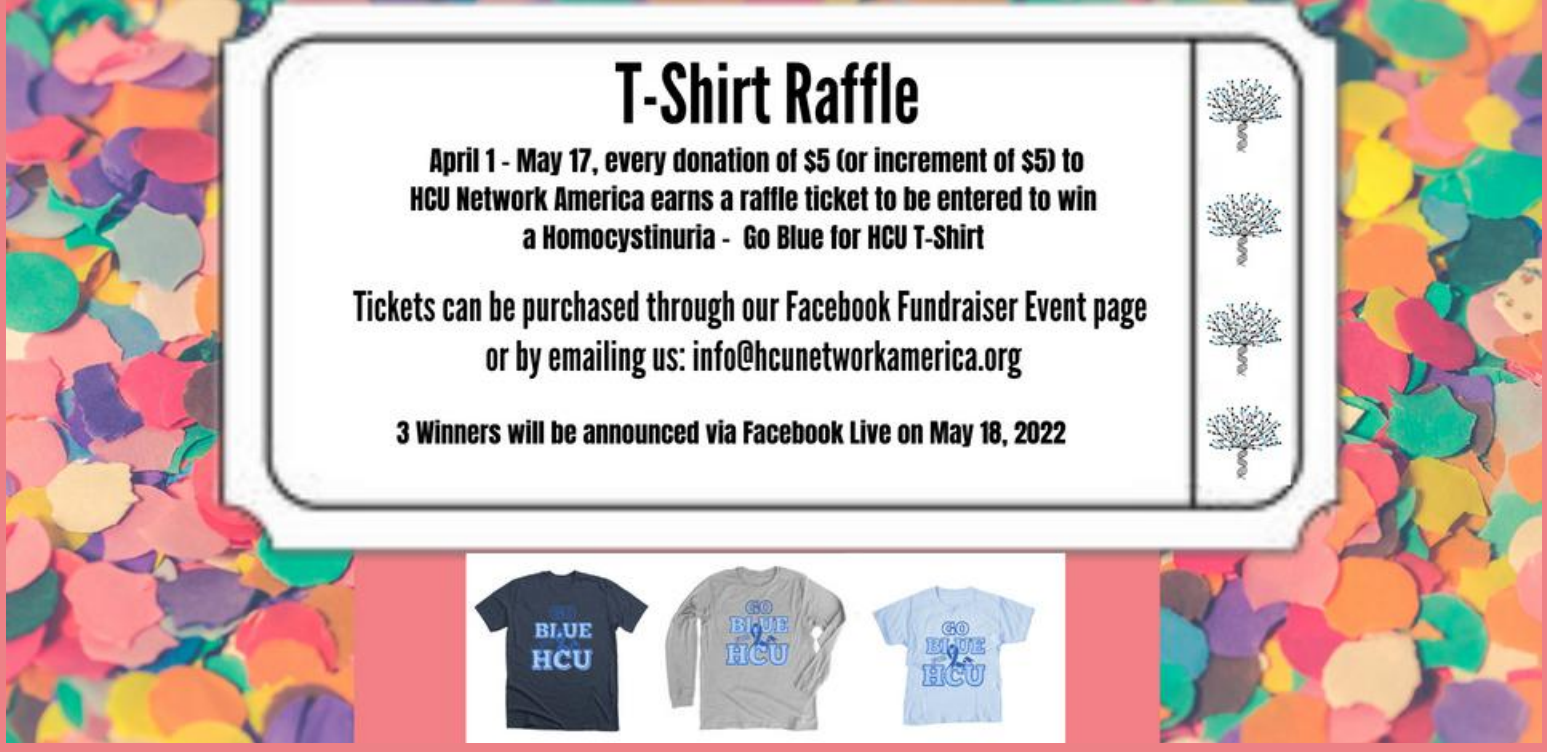
Complete the Health and Development Survey by May 8, 2022 to have your experience be part of the HCU Network America Conference in June!

YOUR QUALITY OF LIFE MATTERS!

Having Homocystinuria takes a toll on our mental and physical health, our relationships, and finances. We need to hear from patients and caregivers to learn how to better support their needs.

Complete the Quality of Life Survey by May 8, 2022 to have your experience be part of the HCU Network America Conference in June!

Share your experience at: homocystinuria.rare-x.org



May 18 is World Homocystinurias Awareness Day!

Get ready, raise awareness and funds for homocystinuria research!

- **You can purchase a raffle ticket: https://www.facebook.com/donate/366933448650459/**
- **Buy a shirt: https://www.bonfire.com/go-blue-for-hcu-2022/**
- **Join the leaderboard and create your shirt here: https://www.bonfire.com/event/go-blue-for-hcu/**

Leaderboard

Rank by

	CAMPAIGN	RAISED	STARTED BY	SOLD	SUPPORTERS
1	Go Blue for Anniston	$150.61	Brittany Hunter	15	10
2	Go Blue For HCU_Team Carson	$34.76	HCU Network America	4	2
3	Go Blue for Classical HCU	$0.00	HCU Network America	1	0

Share with friends and family.

UPCOMING EVENTS

Register here:
https://www.hcunetworkaustralia.org.au/event/navigating-the-homocystinurias-webinar/

Register now at:
https://www.eventbrite.com/o/hcu-network-america-30163980100

SEPTEMBER 1-30, 2022

More swag available for fundraisers!

Per Individual: $30

Per Family (up to 4 – 1 mailing address): $50

Registration opens June 1, 2022

https://runsignup.com/Race/IL/Batavia/HCURaceforResearch

MAY FUNDRAISING REMINDER

Did you submit for your Employers Corporate Matching Gifts Program?

What are Matching Gifts?
Corporate matching gifts are a type of gift giving in which companies financially match donations that their employees make to nonprofit organizations. When an employee makes a donation, they need to request the matching gift from the employer, who then makes their own donation. Companies usually match at a 1:1 ratio, but some will match 1:2 or 2:1.

Why are Corporate Matching Gifts Valuable?
Corporate matching gifts are valuable because they are free money for your nonprofit of choice, HCU Network America! Your donation has double the power without you having to give double the amount.

Does HCU Network America Really Benefit?
If corporate matching gifts weren't valuable, we wouldn't be sending this letter. In 2020 we received approximately $45,000 in corporate matching gifts!

How Does this Benefit my Employer?
Companies of all sizes match donations their employees make to nonprofits because it's an easy way for them to support good work in their communities. Corporate Social Responsibility (CSR), is an important factor in how the public perceives brands and companies these days.

Corporate matching gifts are an efficient and straightforward way for companies to build relationships with charities.

How Do I Find out if my Employer has a Corporate Matching Gifts Program?
Your company website or websites such as doublethedonation.com make it easy for you to search and see if your employer takes part in corporate matching gifts. If you can't find it online, please consult with your employee handbook, HR rep or manager to find out.

How Do I Request my Donation is Matched by my Employer?

1. The donor completes their donation
2. The donor submits matching gift request
3. Company reviews donation and nonprofit eligibility and reaches out to nonprofit
4. Nonprofit verifies the donation was made
5. If eligible, the nonprofit will receive the matching gifts request!

Top Matching Gift Companies

Company	Match Ratio
General Electric	1:1
BP (British Petroleum)	1:1
Gap Corporation	1:1
ExxonMobil	3:1
CarMax	1:1
Johnson & Johnson	2:1
Microsoft	1:1
Pfizer	1:1
Coca-Cola	2:1
Avon	2:1
IBM	1:1
And many, many more!	

Did you know some companies will match donations from their retired employees?

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

APRIL FUNDRAISING REMINDER

Not using Amazon Smile? It's easy, here's how

What is Amazon Smile?

Amazon Smile is a simple and automatic way for you to support HCU Network America every time you shop, at no cost to you. When you shop, you'll find the exact same low prices, vast selection and convenient shopping experience as, with the added bonus that Amazon will donate a portion of the purchase price to us.

How do I set it up?

Simply, go to smile.amazon.com, the first time you go it will ask you to designate an organization. Type in HCU Network America and select us from the list (or go to our direct link: click here). It is important to note that in order for the donations to go to HCU Network America, you MUST check out from this url every time - see best practices below for some pointers on how to do this.

Best practices for using Amazon Smile

Now that your account is set up to use Amazon Smile, it is important to note that Amazon only makes donations to HCU Network America when you checkout from your cart from this url. This is the only way HCU Network America gets any donations from Amazon Smile. Since this is the case here are some best practices to help you make the most of your Amazon Shopping.

Desktop Users:

If you do your Amazon shopping from you desktop/laptop then you can simply bookmark/favorite this url and do your shopping from this web page

Mobile Users:

Most Amazon shoppers use the app on their mobile or tablet. If you are using your smartphone, be sure to download the Amazon Smile App and follow the directions.

What if I'm already set up and would like to switch to HCU Network America?

1. From your desktop, simply select "Your Account" from the navigation at the top of any page
2. Then select the option to "Change your Charity". From your mobile browser, select "Change your Charity" from the options at the bottom of the page.
3. Type "HCU Network America" in the search bar and search for the charity.
4. Select HCU Network America charity to update your account

more specifically how to treat it. He gave me time to research the disorder and asked me to write down any questions I had. So I did and as I read, I realized that again I didn't fit many of the descriptions.

It was hard to read, especially the ugly things. In fact, I wasn't even sure if I wanted to know the answers. Something that worried me was the progressivity of the condition being mentioned. I was afraid of getting worse. It was also mentioned that early detection prevents complications and increases survival, which worried me because although I got diagnosed when little, I later didn't follow the treatment as I should have. The word "survival" traumatized me too much. I was like, am I going to die soon? Something that also worried me was retinal problems that were related to blindness. I just thought, I can't deal with another problem.

The geneticist told me to incorporate betaine and folic acid into my treatment and to start consulting a clinical nutritionist who would give me a semi-vegan diet due to my problem with metabolizing proteins. Although I had been left on a free diet as a child, it was probably because at that time I was in a developmental stage. I'm currently following a diet,which is more of a lifestyle, with low consumption of protein, dairy, and gluten. In general, a cleaner diet without processed food or added sugars. This is complicated for me because I used to eat that every day, and also because once I commit to something I fulfill it, so especially the first few months I followed it by the book. The first month that I followed the food plan, I thought that there wouldn't be much difference in my blood tests because it had been a very short time compared to my whole life but to my surprise, my tests improved a lot even though I still hadn't added the new medications. I also need to be well hydrated, which I probably won't be as good as I should because I don't drink much fluids. I just don't feel the need to. I think I got my body used to that so I don't go to the restroom so often, due to how difficult it is for me because of my lack of mobility.

On to other things, I had to go to a cardiologist and a retina specialist for the problems I mentioned before. In my visit to the cardiologist, everything went well, but I confess that I postponed the appointment with the ophthalmologist because I was really nervous about what he could tell me. Thankfully it went great and everything is fine with my retina.

Overall, I am much more aware of my health now. I have blood tests done every 2-3 months, when I had not been checked for years.

DEEP TALK

When reading and talking with the geneticist, I realized that, within what is possible, I'm doing well compared to other people who have this diagnosis (to date I haven't met anyone who has the same diagnosis with my characteristics). That discussion turned into a deep one that, by the way, I hope I didn't traumatize him by telling him so many things and letting off steam. I remember that at one point I told him that for most of my life I asked God for a miracle to happen until one day I got tired of asking, and I was like enough, I better learn to live with this because that's how it's going to be forever.

I also told him that several times people had said things to me like "I admire you" or "You are so strong" and that I subconsciously became like that. I became what people wanted me to be, or at least I pretended to. I didn't even allow myself to feel what I really felt and believed that I had no right to complain because of how blessed and privileged I am. Or for the simple fact that I wasn't "as bad" as others. It's horrible to think this way because there will always be someone in a more privileged or less privileged situation than you, and that doesn't mean that the way you feel has less value. By not taking

this into account, I tried to be positive or just not to think about it. It wasn't until a year ago that I started seeing a psychologist that I understood that whatever I feel is valid and I don't have to invalidate my feelings nor let others make me feel that way. Okay, yes, I'm better than others, but still not walking isn't cool, and it isn't something to be minimized.

When I was telling the geneticist that I didn't understand why people admire me, he interrupted me and told me reasons why I should be admired and that meant a lot to me. I think that was what I needed, someone to explain it, someone who didn't just say “I admire you”. This is because by listening to that while feeling I didn't deserve to be admired, I imagined a thousand reasons why they could admire me, and not knowing what exactly they were referring to affected me because I felt the need to try to fit in with all those reasons.

I have always presented myself as a strong person, happy despite her condition, who isn't affected by her illness and lives her day-to-day life like everyone else. As I mentioned before, I never allowed myself to feel bad about my situation and every time a negative thought came to my mind I just thought how I am lucky for the fact that "I'm not so bad", for being alive or I just thought that I could be worse so I had no right to complain. I know that the intention of people who told me I was strong and that they admired me was good, but when I heard it, I put pressure on myself so that people really had something to admire. I also thought that in that case, they should admire my parents and all their efforts. I never really understood why I should be admired since that's how I got to live, the disease chose me, I didn't choose it and if I could change the situation I would.

I'm sorry for not being that disabled person who romanticizes her disability, and who says she wouldn't change a thing in her life because that made her strong. I totally would. I mean, why wouldn't I want to have spared myself a lot of hardships that I've been through, I’m going through, and will go through? Why wouldn't I want to have the same skills as most other people?

Either way, why want to change things that can't be changed? Everything happens for a reason and if things hadn't turned out in this exact way; I probably wouldn't have met many of the people who have a very important place in my life. I wouldn't be who I am, nor would I value things as much as I do now. I also believe that each one of us is in this world for different reasons. Personally, I don't think we should limit ourselves to thinking that we have a single specific purpose in life, since I think that we're the masters of our own destiny, or at least that's what I believe in order to keep moving forward.

COMPENSATORY CONDUCT

All my life I have had a hard time saying “no”. I hate arguments and so often I find myself apologizing for everything, even though sometimes I don't even have to. It's very difficult for me to set limits. I'm very selfless. Most of the time I find myself giving in to everything because I always try to please others and, although it's difficult for me to accept it, I care a lot about what people think of me. With my psychologist I learned that these compensatory behaviors are very common in people who have a disability since we feel that we have to make up for our lack of abilities by trying to be as noble as possible. I think that's why it's so hard for me to accept that I'm valued by others.

EMOTIONAL REVOLUTION

While I spoke for several hours with the geneticist, being vulnerable was something very new for me. Normally, others approach me and share very personal things, so personal that sometimes I don't

even get why they tell me because, every so often, we aren't that close. I guess some how generate trust. Don't misunderstand me, I love to listen and try to help others, it's just that usually it happens the other way around. Perhaps I allowed myself to be vulnerable with my geneticist because he was a new person in my life, who didn't live in the same place as me, who didn't know me. So, in a way he made me feel like I was in a "safe place" as he mentioned it, a place where I wasn't going to be judged and where he wasn't going to go tattle to anyone about what I said.

Habitually, I would have immediately told my closest friends everything that had happened because those who know me know that I love to talk, but for the first time I wasn't ready to tell. Although my friends were aware of what had happened, I told them that I needed some time to process everything. That day, at night time, I couldn't stop crying. I probably sound like I am exaggerating because right now that I'm writing this that's how I feel, but I felt bad about a lot of things.

I felt bad for not allowing myself to show that I'm not okay because being with the geneticist I didn't shed a single tear. I felt hypocritical for being so good at hiding what I really felt, for not allowing myself to show those feelings to others and for not letting them see me as “weak”. All because I don’t want to disappoint those who “admire” me. Obviously, all this is subconscious, it's not like being there with him I said "Diana don't cry", it just didn't happen.

I felt really bad emotionally. It was many years of avoiding the subject of my illness. I even expressed it to my mom and she didn't understand. She told me that I had been like this all my life and even when I mentioned that I needed to talk to the psychologist about it, she was caught off guard as she didn't understand why it was so urgent. I felt alone, although I know I wasn't, but I knew that no one could fully understand me because they weren't experiencing what I was experiencing.

I was worried because I felt all these responsibilities and concerns that I hadn't considered before were falling on me at the same time. This is because besides the medical issue, and the confinement due to the pandemic, I think I had an adult life crisis because I was graduating soon.

I was angry because at just 23 years old, I was feeling and learning all about my condition and also because I feel dependent on my parents and their schedules. One of my biggest dreams is to visit many parts of the world and, as privileged as it sounds, I watched everyone on social media traveling while I couldn't buy a ticket and go visit one of my best friends who is studying abroad despite having the money I earned from my work to do so. Watching people going out more and more really aggrievated me because I was still locked in my house. I didn't feel free. In fact, due to my new diet, I wasn't even able to make a decision on what I wanted to eat that day.

I felt angry for wanting to improve myself so much and not being able to continue studying or working abroad, something I always wanted to do, because my dad's job is here and leaving is not an option. If I had to sum it up in one word, I was scared. Scared of uncertainty of the future knowing I have it harder than people around me and of the fact that I still haven't figured out what I'll do once I don't have my parents anymore. I still feel it, the fear has not gone away, but I’ve figured that what's important is to not let it stop you as it did with me at that time, but rather use that fear as a boost to move forward.

BREAKDOWNS

I've had 3 ugly breakdowns in my life. One was when I returned to the subject of my health after a long time. I spent hours crying, I hyperventilated and couldn't stop doing it. It was so bad that even my mom wanted to take me to a psychiatrist because she thought I was depressed and even I thought so. Later I talked about it with my psychologist, and she made me notice that many things had happened to me since I started my re-diagnosis process and that the emotional revolution I was going through was normal. I was only grieving my illness. She even pointed out how in our first session I told her "I have this disease but I'm not here to talk about it, I'm fine with it", when clearly, I wasn't. She told me that from that moment she knew I wasn't okay but she wasn't going to tell me on the first appointment because that could cause me to stop going. I got too frustrated because my mom told me that I wasn't doing any better and even I noticed it too, but in therapy I learned that the healing process is not linear, that some days you can be fine and others not and that's okay. I learned that it's normal to feel bad and that if you don't let yourself feel what you're feeling in the moment, later it's going to be worse.

Another one was when I was very young. I was in elementary school and had some friends but suddenly, they stopped hanging out with me. At that time, I didn't understand why, so I felt very bad and abandoned. I didn't understand what I was doing wrong or why things had changed out of nowhere. Now I understand it was because I was very intense because of my need to have people in my life in order to feel safe, loved and have a sense of belonging. It was difficult because overnight I lost friends that I had had since I was little, but once I got into middle school, I made new friends who, to this day, still are my good friends. That's why, I repeat, everything happens for a reason.

My other breakdown was in high school, when my friends were hanging out at clubs every weekend and I watched how their friendship grew because of it. Anyway, my friends told me all about what happened, I knew all the high school gossip and of course, I always gave them advice. To this day, I continue to do so, and I actually don't consider myself to be bad at it despite the lack of experience I have. Besides, as they say... “Coaches don't play.”, but of course it's not the same as living it, which brings me to the other reason for an emotional bump in my life... It was the time when my friends were starting to have boyfriends, and I felt what they call "fear of missing out". At that time, I was talking to a guy who was my friend. Suddenly I felt there was something more going on because of how I saw boys flirting with my friends and also because of what they told me about the situation, nevertheless, I refused to believe it. I didn't think anyone would notice me that way. In fact, any time I thought it was like that, I felt crazy and that everything was in my head. I think it was my way of protecting myself and, to be honest, I still doubt it. I didn't know whether to include this part, but I consider it important because it's part of the insecurities my condition has brought with it.

MY CONCERN ABOUT SCHOOL

In the process of going to the psychologist, I wondered why all my life I was so worried about school if my parents never demanded anything of me. I was an honor roll student. I even won a scholarship to stay in my school's high school. I also received a scholarship at the high school where I studied and from which I graduated with honors and at college where I graduated with cum laude. I share all of this because I worked hard for it but now, I also think I did it out of a need to prove to myself that I could. There are so many aspects in my life that I have no control over, that I felt my studies were something that I did,have control over, so I put all my effort into doing my best. Now I know that I have nothing to prove to myself or to anyone, although sometimes I have a hard time remembering it. My condition also impacted my career choice. I knew I wanted something related to the arts, but I discarded some options because of how complicated it could get if physical abilities were implied. I decided on graphic design and thought it was perfect because you can get the job done with a computer.

FAMILY

I feel like I never quite fit in with "normal" people because I couldn't do everything they did. In physical therapy I felt like I belonged even less because the kids were so affected that I couldn't even carry on a conversation with them. Something that really helped me and is a key factor in me becoming the person I am today is my family, grandparents, uncles and cousins, have always treated me just like a member of the family which I am, but you know what I mean. They never treated me any different and that helped me a lot so that I developed in a "normal" way and wasn't withdrawn, which I really appreciate because I never felt out of place.

I WOULD LIKE TO PASS UNNOTICED

Something that I have always wanted is to be able to go unnoticed and not be so easy to identify but my mobility scooter makes it impossible. Also, because I would like people to describe me for who I am, not as "Diana, the one on the mobility scooter". I would like people to see me as they see anyone else because, although people closest to me do, others see me differently. It's something I've felt all my life. Strangely, this mostly happens to me in México. In other places they don't even notice me and I love that, so I guess something must change in Mexican culture.

I recently took a selfie with someone who told me we should take a full body shot instead to "truly show who I am". I understand what he meant, and even though his intention wasn't to make me feel bad, I don't agree. My disability may be part of my life, but it doesn't define me. I'm much more than that. I have been reading different opinions of disabled people. Some believe that it does define them and others don't. I think that either way of thinking should be respected. I believe that my disability is just a part of me and does not define me as a whole.

CHALLENGE

One of the challenges I've faced and for which I feel very proud though it may sound very silly to others, is having entered the high school I entered instead of staying in my safe zone by continuing studying at my school. I have to admit that my mom played a big role in this decision for which I'm very grateful. It was a huge change for me, since I had been surrounded by only girls who grew up with me since kindergarten. So, for them, my disability wasn't an issue, I was just someone else. When I entered high school, I was nervous because it was much bigger than my school. I didn't know anyone and people would no longer hang out with me "out of habit." That was the moment I proved to myself that attitude and personality is everything. I made friends with people who were willing to meet me without any kind of prejudice and

who today are the most important people to me. That has been the best decision I've made in my life because, if I had not experienced those 3 years, I would be a totally different person. I changed so much. I became more confident and with a little less fear.

ADVICE TO NEWLY DIAGNOSED FAMILIES

If I could give newly diagnosed families one piece of advice, it would be to find a doctor who doesn't make you feel hyped or crazy. Stop googling about the diagnosis because you never really know how the disease will behave, also because there are always exceptions; just look at me.

I know your children need more attention or time, but otherwise treat them like you would want to be treated. Stop seeing them as little angels because they are not; they are people like everyone else. Don't forget about yourselves either, you deserve to be happy too. Your child can perceive all your emotions so, the more positive, the better.

If possible, take your child to a psychologist to accompany them on this path because it is not easy and mental health is everything. If you want, you can also go, because just like they have a hard time, so do you. Besides, it can help you understand them in a better way.

It's okay to feel sad, angry, frustrated, lost and afraid or whatever you're feeling but never feel guilty because it's nobody's fault. That's just how genetics work and although it sucks, that's how things should have happened. You must remember that everything happens for a reason, and you don't always have to know that reason.

Do everything possible for your child to ensure they have a normal, balanced life. Don't overwhelm them with medical things, because in the end, what good will it do if they doesn't enjoy life? Allow yourself to be helped by those who love you and lean on them, but know that no matter how much they want to; they will never fully understand you. Which leads me to the next piece of advice... look for families who have a situation similar to yours, so that you don't feel alone and misunderstood.

Finally, while your child is young, probably the only thing they need to be told is that everything will be fine, even if you don't know it for sure. Always be open to communication, but as they grow, make a plan with them to not feel so overwhelmed by the future. Take one day at a time.

NOWADAYS

During my last breakdown looking for answers I came across HCU Network America. Now, here I am writing my story to try to help others. They put me in contact with the US National Institute of Health, who invited me to be part of a study. They aren't looking for a cure, so I know that I probably won't see myself as benefiting by the study, or maybe, I just want to think that so I don't create expectations and thus not have disappointments. Nevertheless, I would love to contribute in some way so that people who have my diagnosis can have a better quality of life in the future.

I'm currently following a proper diet, receiving medical treatment and physical therapy. I haven't gotten my blood test results back to normal, but I'm continuing on this journey of self-care and hopefully soon I'll get them back down to a lower level.

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/

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