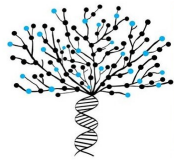


# HCU Herald

**Presented by**



**HCU Network America**

**Connecting for a Cure.**

**There have been a lot of things happening for the HCU community & for HCUNA.  
We strive to keep you informed and connected.**

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- Provides a mixture of amino acids, carbohydrate, vitamins and minerals to support daily nutritional needs
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## HCU cooler15®

- Suitable from 3 years of age
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# Heroes of HCU:

## Ellie from Pennsylvania



Ellie was diagnosed with HCU through newborn screening in April 2018. She had a repeat test done when she was a week old, and was then diagnosed at 13 days old. Her level was over 200, which led her genetic doctor to admit her into the NICU for 6 days. Since she was discharged, she has routine weekly blood draws and we are in close contact with her dietitian and genetic team.

HCU has affected the way our family thinks about food. Although she's not eating solid food yet, I am constantly researching food substitutes and recipes that she can eat along with us. My family has dramatically cut meat from our diet to ease the transition when Ellie starts eating solid food. We have another relative who has been diagnosed with HCU, but never realized the effort it takes to stay within protein goals.

In Ellie's short HCU journey, she has had several accomplishments. The doctors/nutritionist have helped to bring her levels to a safe range by limiting protein intake and giving her betaine, both common in HCU treatment. Weekly blood draws help us monitor maintain her levels. At first, the blood draws were traumatic; it would take two people and several pokes to get a good sample. After about a month she began to get used to the blood draws and even smiles while a sample is taken. This is a huge success and led us to having a positive outlook on her treatment.

Since Ellie was born, she has not been a big eater. The nurses in the NICU even made comments about tube feeding her if she didn't increase her intake. This makes reaching her daily calorie and fluid goals difficult. To overcome this obstacle, we give her less volume per feed and leave room for extra Holminex-1 at night to reach her fluid goals. Some days are better than others, but we make sure to keep close contact with her dietitians.

Ellie's future is very bright. We are lucky that she was diagnosed at birth and could start treatment very early. Although her diet restrictions aren't ideal, we have found several alternatives and live in a time where those food options are easily accessible. Advice I'd give to parents who are just starting their journey is to reach out to people who are affected by HCU. Connecting with people and reading others' stories gives us a peace of mind. We do have days that we are heartbroken for Ellie, but remember she wouldn't be her without HCU and we wouldn't change her for anything.

# HCU and You: *Ask Methia*

Dear Methia,

## How Do I Get Myself Back On Track?

It's been about 2 years since I have been to genetics clinic, and I stopped my low protein diet earlier this year. The diet was so hard to follow with my busy schedule, and has been even harder to follow through the holidays. The truth is, I feel perfectly fine, but I know that I am putting myself at risk for complications if I don't get back on track. Where do I start?

Sincerely,  
*Off The Wagon*

---

Dear *Off The Wagon*,

Low protein diets are really, really hard! So hard, in fact, that occasional slip-ups are almost inevitable. Sometimes one indiscretion can lead to longer periods of non-compliance, and I applaud you for recognizing that getting back on track is incredibly important for your health. Although you may feel completely fine, high homocysteine levels resulting from too much protein are linked to lens dislocation, blood clots, and other clinical complications. Here are some helpful tips to help you get back on diet:

**Schedule an appointment!** Your metabolic clinic is one of your main resources. You are certainly not the first person to ever go off diet, and your team will be well equipped to get you back on track in an effective, safe way.

**Be open and honest with your geneticist and dietitian about your intake.** Having to admit that you've been non-compliant is difficult, especially when telling the people who want you to succeed. Your dietitian will ask you for diet records, and it will be really tempting to minimize the extent of your dietary indiscretions. Remember, though, that your team is using the details of where you are *currently* to calculate numbers and set goals for where you should be for optimal control. Manipulating your recall will only negatively impact your efforts!

**Make small changes first.** Most likely, going off diet started with "just one" non-compliant meal or food item. The same can be said about the road back to compliance – start with one meal! Going immediately back on diet full-swing sets you up to feel over-restricted and may result in regression. Set a couple of small goals for yourself, and try to make one of those goals one that is achievable before your clinic visit. For example, if you are not using any low protein medical foods, make a commitment to substitute one of your food staples for their low protein counterpart, such as bread or pasta.

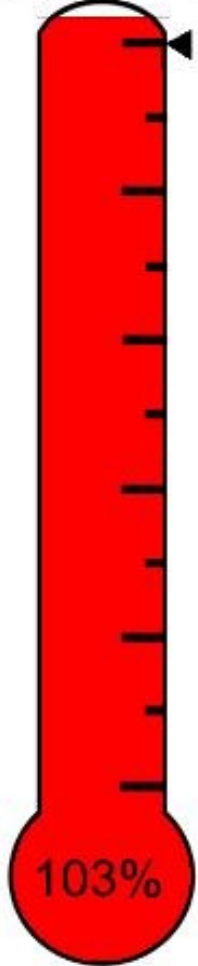
I wish you the best of luck, and commend you for making your health a priority in 2019!

Sincerely,  
*Methia*

# 2018 Annual Appeal Highlights

Thank you to everyone who donated!

\$20,000



\$20,683

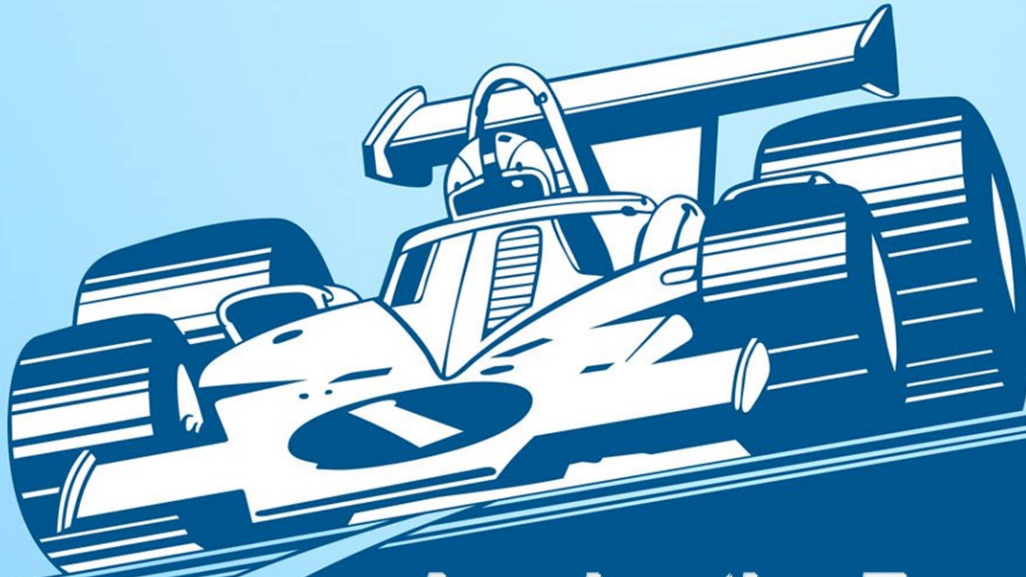
Our annual appeal started in October with a record breaking HCU Awareness Month. In October we raised \$12,866 thanks to the 13 families who set up fundraisers on behalf of the organization and to the many others for their individual donations.

In November we had another wonderful opportunity to raise awareness and funds for HCU Network America through Giving Tuesday and Thanksgiving. 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 end of November we were at more than half way of our goal. Thank you to those who set up #GivingTuesday fundraisers.

In December of 2018, as our annual appeal was about to come to a close, we sent an email recapping our accomplishments of the past year. Because of the communities' generous contributions in 2017, we were able to accomplish a lot this past year. With this email sent in late 2018 we were able to take in \$3952 to continue our mission in 2019!

We are very thankful for our generous donations from our supporters! This was our first year that we not only met our goal, but surpassed it! We couldn't have had a successful 2018 without you and know this will be true for 2019 as well!

# HCU Network America Conference



## *Accelerating Towards a Cure*

*Save the Date ~ 2nd Homocystinuria Conference  
October 19 & 20, 2019 | Indianapolis, Indiana*

*Details to follow at [HCUNetworkAmerica.org](http://HCUNetworkAmerica.org)*

HCU Network America will be holding its second patient/family-expert meeting October 19 and 20, 2019 at the Holiday Inn Indianapolis Airport. It will be a weekend full of friends new and old, networking, keynote speakers, breakout sessions, panels and a reception.

We listened to your feedback from last year and as a result this one is very close to the Indianapolis airport. In addition, each room has a refrigerator, microwave and individual cup coffee maker. The hotel has a 24 hour free shuttle that runs to and from the airport.

Discounted rooms will be available, so while we encourage you to save-the-date on your calendar, don't book your hotel rooms just yet. We will have a direct reservation link on our conference webpage.

Register your expression of interest to attend the meeting: [Click Here](#)



# OT-58, Enzyme Replacement Therapy Clinical Trial Recruitment

Orphan Technologies has initiated a first in human (Phase 1) clinical trial of OT-58, an enzyme replacement therapy that addresses the underlying enzyme deficit for patients living with classical homocystinuria. The goal of this trial is to evaluate the safety and efficacy of OT-58 in patients with classical homocystinuria and identify the appropriate dose. Patients between the ages of 12 and 65 years of age with classical homocystinuria may be eligible to join. For additional information on criteria for eligibility, please go to:

<https://clinicaltrials.gov/ct2/show/NCT03406611?cond=Homocystinuria&rank=1>

There are four sites in the US currently participating in the trial:

- Children's Hospital of Philadelphia – open to patient enrollment
- Boston Children's Hospital – open to patient enrollment
- Indiana University – open to patient enrollment
- Children's Hospital Colorado - open to patient enrollment

Payment for time and travel may be available to patients who participate in this trial.

To inquire about participation into the trial, please email: [info@orphantechnologies.com](mailto:info@orphantechnologies.com)



**OT-58**

Enzyme Replacement Therapy

# 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 to. By registering, you will be also be able to identify other affected patients in your state and request their contact information, and you will 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 and provide resources for patients and families, 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/>



# Formula Survey



Love your formula, hate your formula? Let us know exactly what helps you or stops you from taking it!

This short 5 minute survey will be used to work with formula manufacturers to develop and improve on current formulas' texture, smell and taste as well as develop formulas that better fits your life style.

[Take the Survey Now](#)

**We'd like to thank the following content contributors:**

**Editor in Chief: Danae' Bartke**

**Heroes of HCU: Ellie from Pennsylvania**

**HCU and You: *Ask Methia*: Angela Pipitone**

[\*\*Click to donate directly\*\*](#)