HCU Herald

Presented by HCU Network America

Connecting for a Cure.

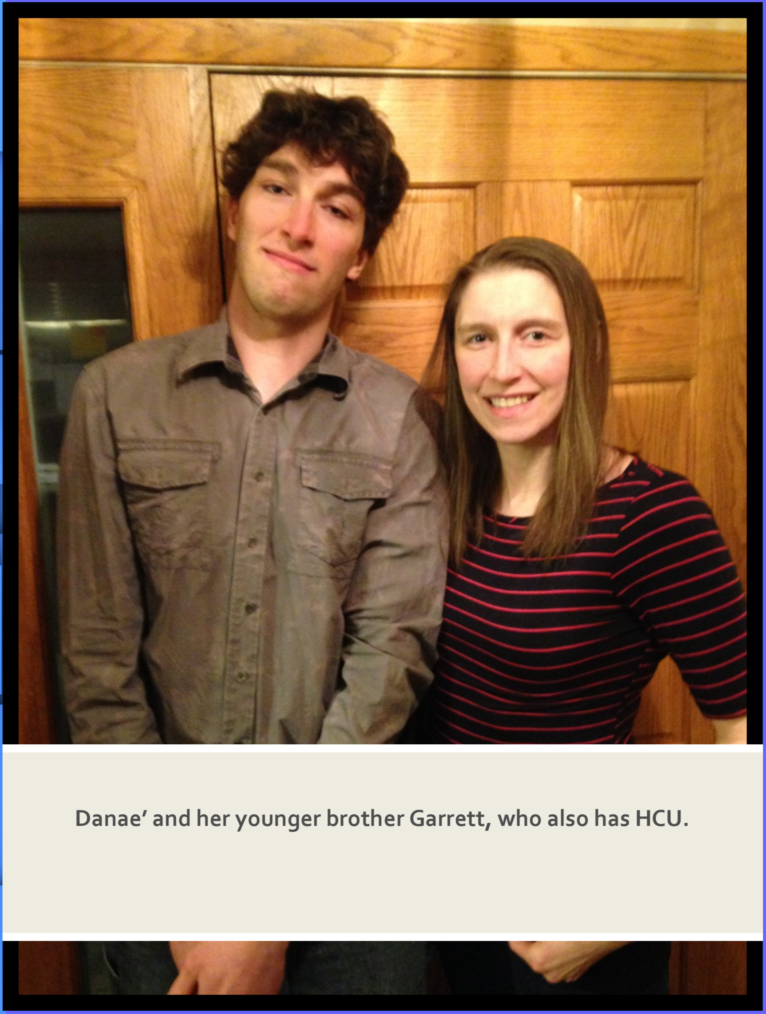
There have been a lot of things happening for the HCU community & for HCUNA.

We strive to keep you informed and connected.

Inside this issue:

Heroes of HCU

Danae' from Illinois



Danae' and her younger brother Garrett, who also has HCU.

My name is Danae' Bartke and I was diagnosed with homocystinuria in 1995 at the age of 10. We discovered the diagnoses because my younger brother had bumped his head on a table in school and then complained that he could not see. After a week or so, my mom took him into an eye doctor who referred us to the Wheaton Eye Clinic. At the Wheaton Eye Clinic they discovered that his lenses had detached from his retina and popped through his pupil. He then had immediate eye surgery to remove the lenses in both of his eyes. The doctor, at the time, told us that it could only be one of two conditions that had caused the detachment. Garrett was disqualified from one because he was not old enough, so it left us with only one possibility: homocystinuria. The eye doctor at the Wheaton Eye Clinic then got in touch with Dr. Paul Wong and told him of the case. Immediately, Garrett was tested and it came back positive. The next step was to get myself and my six siblings tested. Out of rest of us, I was the only one who also tested back positive for homocystinuria.

After the diagnoses Dr. Wong took us through a couple of treatments to see which ones we had responded too. The first one was just a regiment of B6. From that, we discovered we did not have the kind of homocystinuria that was responsive to B6. He then started us on B6, B12, baby aspirin, folic acid, Hominex-2 and a low-protein diet. After that we were on track; at least for a bit. I can't say that I was a very sweet child. My brother and I gave my mother a very difficult time. We hated the food, hated our Hominex, and we did not really follow the diet like we were supposed to. Our father had died a year prior to our diagnoses, so our mom did not have the time or energy to battle us every step of the way. Eventually, I did adhere to taking the Hominex and attempted to follow the diet. I wasn't the best patient, but I did make an attempt.

In 2009, I had a really big fork thrown in my road. As a result of not following the diet as well as I should've, I developed a blood clot in my wrist. Dr. Wong advised me to go immediately into the Emergency Room at Rush and he would have someone there to meet me. After a week in the hospital I was released with a new lease on life. I realized how lucky I was and that if I was going to have a healthy, productive and long life, that I was going to have to take my diet seriously.

About two weeks after being released from the hospital I received a letter from the PKU Organization of Illinois inviting me to a low-protein cooking class. I had neither heard of the PKU Organization of Illinois nor did I know there were any other disorders out there that had to follow a low-protein diet. I was so excited! After the cooking class, we then went to their annual meeting where we met Malathy from Taste Connections. From her, we found out about the first national conference for homocystinuria. We went to that in March 2011. In the course of a couple of years we went from having no community to a community that was just so helpful and kind.

Heroes of HCU, cont.

2011 Denver Meeting

In 2014 I joined the PKU Organization of Illinois board. In 2014 and 2015 I was in charge of the PKU Press. The PKU Press is PKU Organization of Illinois' newsletter. It lists the events for the Spring and Fall. It also has special interest pieces, tips, recipes and much more. In 2016 I became the President of the PKU Organization of Illinois. As President, I oversaw the Board of Directors and various committees. I put myself on every committee to get a solid understanding of each function. My term ended in December 2017, but left me with a lifetime of experience and knowledge.

In 2016, I traveled to Prague for the first Methylation Defects Patient-Expert Meeting. There I was able to meet with world-renowned researchers in the Methylation Defects field, along with other patient organizations and families. It became very apparent that we needed an organization in the US for the Methylation Defects. Upon arriving back, Margie McGlynn and I began work to start up what is now HCU Network America!

2016 Patient-Expert Meeting, Prague, Czech Republic

In March 2017 I was hired as the executive director of HCU Network America. This opportunity has allowed me to connect at a much deeper level to the patient community. It has also given me the chance to help organize and create resources that not only help patients, but help clinics provide better care. Last but not least, it has given me the ability to delve into some of the more complicated ongoing issues in the community such as formula choices, treatment methods and newborn screening,. I look forward to serving the community for many years to come!

HCU and You: *Ask Methia*

DEAR Methia,

How Can I Balance the Low-Protein Diet, Formula and Calories?

Balancing calories, natural protein, and my medical food every day is really hard. At the end of the day, I find that I am always over or above one of these things. Sometimes, I'm not sure if the values I'm calculating are even correct, as I'm finding that I have to guess about the protein or methionine content of many foods. I know that it is possible to balance all three, but I need some extra resources. Do you have any suggestions?

Love,
Meal Planning Maniac

Dear *Meal Planning Maniac,*

Meal planning is definitely a challenge, but you are right – it is definitely possible to meet your caloric and protein needs on a daily basis with the right amount of resources and practice. Here are some tips for successful meal planning that will eventually become quick and easy!

Use books, telephone apps and databases to keep a running list of nutrition information. With advanced research and technology, the information is out there and ready for you to access! For example, the USDA Nutrient Database is an online resource that provides not only the calorie and protein content of 200,000+ foods, but many foods have also been analyzed to provide their methionine content as well. Additionally, many companies have created databases that you can access on a phone app to keep a running tally of your intake for the day (metabolic diet apps are the best choice for this). In order to avoid having to look up the same foods again and again, you can make a "quick list," either in your phone or a Word document, so that inserting these foods into another meal plan becomes easier.

Make a number of "sample menus." When you have some free time, use your quick lists, low-protein medical foods and formula data to make 3-5 sample menus that fit your caloric and protein requirements for the day. These menus can be evolved as your tastes change or your desire for more variety increases, and it will soon become easy to "switch out" one food for another.

Work with your metabolic clinic and other patients! Your geneticist and metabolic dietitian will be more than happy to help you troubleshoot any meal planning roadblocks you might have. They are also available to help you navigate the best telephone apps and databases. Your team can also connect you to other patients with HCU, with whom you can collaborate ideas, talk about what works and what doesn't, and to support one another in successfully following your diets.

Remember, the time you spend today will be time that you save tomorrow. This will get easier with practice!

Sincerely,
Methia

SIMD Recap

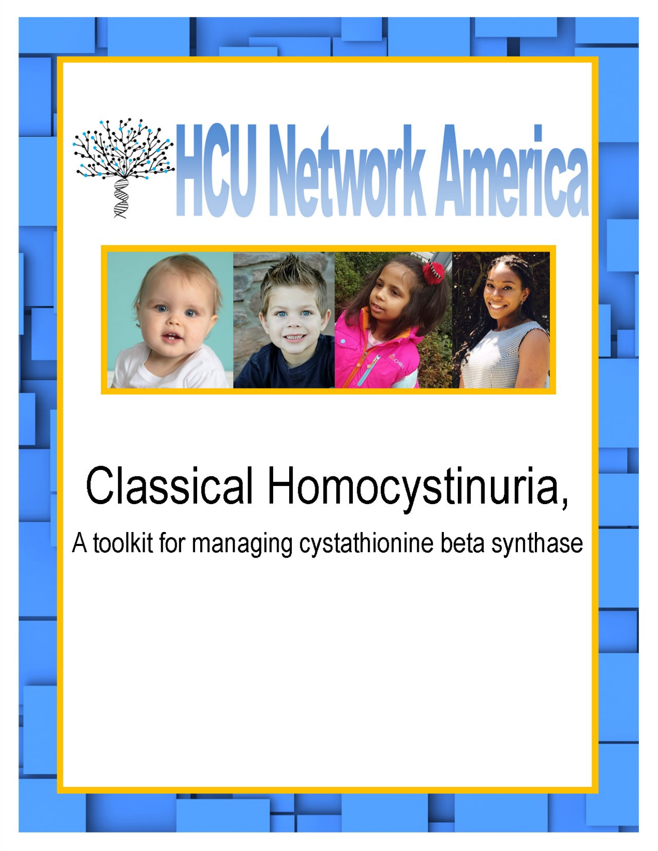
HCU Network America participated in the Society for Inherited Metabolic Disorders 2018 conference in San Diego California, in mid March.

The 4 day conference brings together patient organizations, medical professionals, researchers and pharmaceutical companies that all have a vested interest in metabolic disorders. The four days are comprised of poster sessions, keynote speakers, and panels with lots of time to network. The topics that were included were the political landscape and advocacy for metabolic diseases, pricing and economics related to the treatment of Inborn Errors of the Metabolism, newborn screening, genetic based therapies, other advances in research for specific metabolic disorders, dietary treatment guidelines, unknown and challenging metabolic cases and more.

During coffee breaks and receptions, the exhibits (this includes the booths like HCU Network America) were open to those in attendance. With each break, we saw a lot of interest in our organization and the resources and materials that we had available to both clinics and patients. At the conference we were able to debut our newly published New Patient Toolkit , as well as share our infographic, resource list and the Guidelines for Diagnosis and Management of CBS Deficiency. Doctors were thrilled to know that there was finally a group for their HCU patients!

Despite a heavy push in the past 6 months to connect with clinics, many of those who stopped by were not aware of our organization. This just impresses upon us how important it is that we continue to network, reach out and continue our clinic introductions and have patients connect us as well. While we are a US organization, we also do realize the impact we have on the global homocystinuria community. We had medical professionals from other countries, like Turkey, Qatar, and Ireland stop by and say how they wish there was something like this for their patients in their countries. We hope we not only helped provide Information and resources to those who stopped by, but also were a source of inspiration and hope!

New Patient Toolkit



HCU Network America

Classical Homocystinuria,

A toolkit for managing cystathionine beta synthase

For the past year, HCU Network America has been working hard to develop their first toolkit in a series. The first toolkits now available via our website, is Classical Homocystinuria: A toolkit for managing cystathionine beta- synthase. The toolkit was put together by executive director, Danae' Bartke, doctors Kimberly Chapman, MD, PhD and Harvey Levy, MD, and dietitians, Angela Pipetone, RD, LDN and Krista Viau PhD, RD.

The toolkit is designed for patients and caregivers regardless of age of the patient at the time of diagnosis. It's current version has 6 sections and with plans to expand it further. The first section focuses on what homocystinuria is, followed by: a HCU Nutritional Guide, HCU and Pregnancy, Tips and Tricks, Food and Formula Resources and a section on Medications, labs and doctors. We do have plans in the near future to add an educators guide, babysitters guide and Natural disaster checklist.

Later this year we will begin our toolkit on the cobalamin-related remethylation disorders. Due to the complex nature and variety within the cobalamin-related disorders, we may decide to break these into more than one toolkit. If you are a cobalamin patient or caregiver and would like to provide insight into areas that would be helpful to cover, please email us directly at HCUNetworkAmerica@gmail.com

To view the toolkit, please visit https://hcunetworkamerica.org/wp-content/uploads/2018/03/CBSToolkit.pdf

If you have questions or comments about the toolkit, please email us directly at HCUNetworkAmerica@gmail.com

Contact Register

Did you know that just under one year ago we launched our 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. In order to succeed in our mission the input you provide, helps us plan events, develop resources and educational tools, and to help better to ensure everything is being done to guarantee 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/

Upcoming Events

Taking the Lead for HCU

Save the Date ~ April 21 & 22, 2018

The first HCU Network America Conference

Westford, Massachusetts

Conference Stats

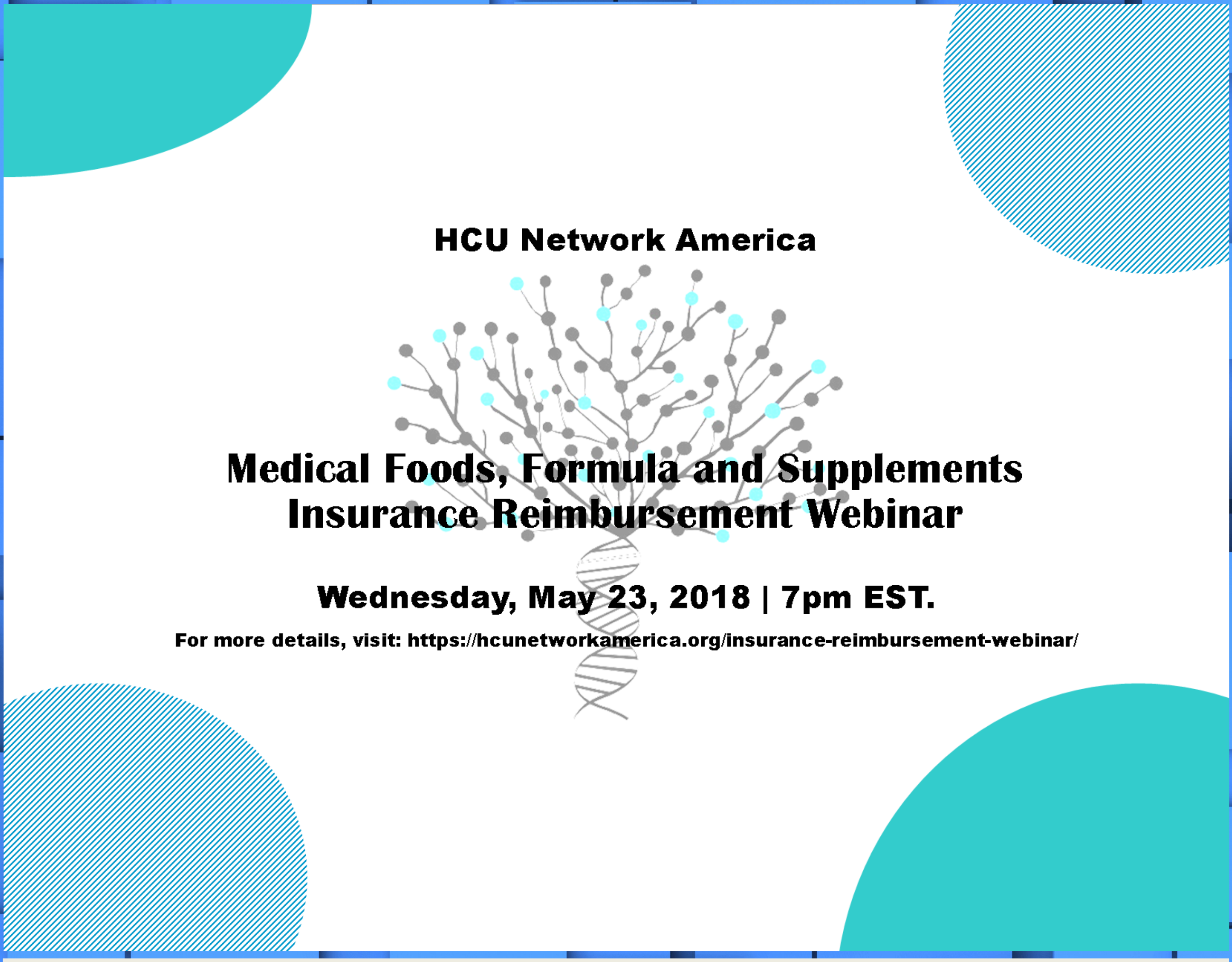
113 Registrants to date!
34 Health Care Professionals and Industry Reps
50 Parents, Spouses, and Relatives
15 Adult Patients
7 Children Patients
5 Children

7 Vendors Booths:

Cambrooke Therapeutics, NECPAD, Nutricia North America, Orphan Technologies, Recordati Rare Diseases, Taste Connections, and Vitaflo USA

Haven't registered yet? Registration closes April 13th. Register today

Upcoming Events, Cont.



HCU Network America

Medical Foods, Formula and Supplements Insurance Reimbursement Webinar

Wednesday, May 23, 2018 | 7pm EST.

For more details, visit: https://hcunetworkamerica.org/insurance-reimbursement-webinar/

During this webinar, Raenette will review information such as these coverage statistics:

- 50% of states have some type of insurance mandate.
    - Of those states, 84 % have some type of mandate on both food and formula coverage.
    - The other 16% percent of those states, do have mandates only on formula coverage.

*Some states may have programs that cover food or formula directly that are not included in these statistics.

Learn how you can get your food, formula and supplements covered by attending our FREE online webinar with Raenette Franco of Compassion Works Medical, LLC. Raenette is a Certified Biller Coder Specialist (CBCS), with over 17 years of experience helping patients get coverage on food, formula, and supplements they need!

To attend this webinar on May 23 at 7 PM EST, please register at:

https://hcunetworkamerica.org/insurance-reimbursement-webinar/

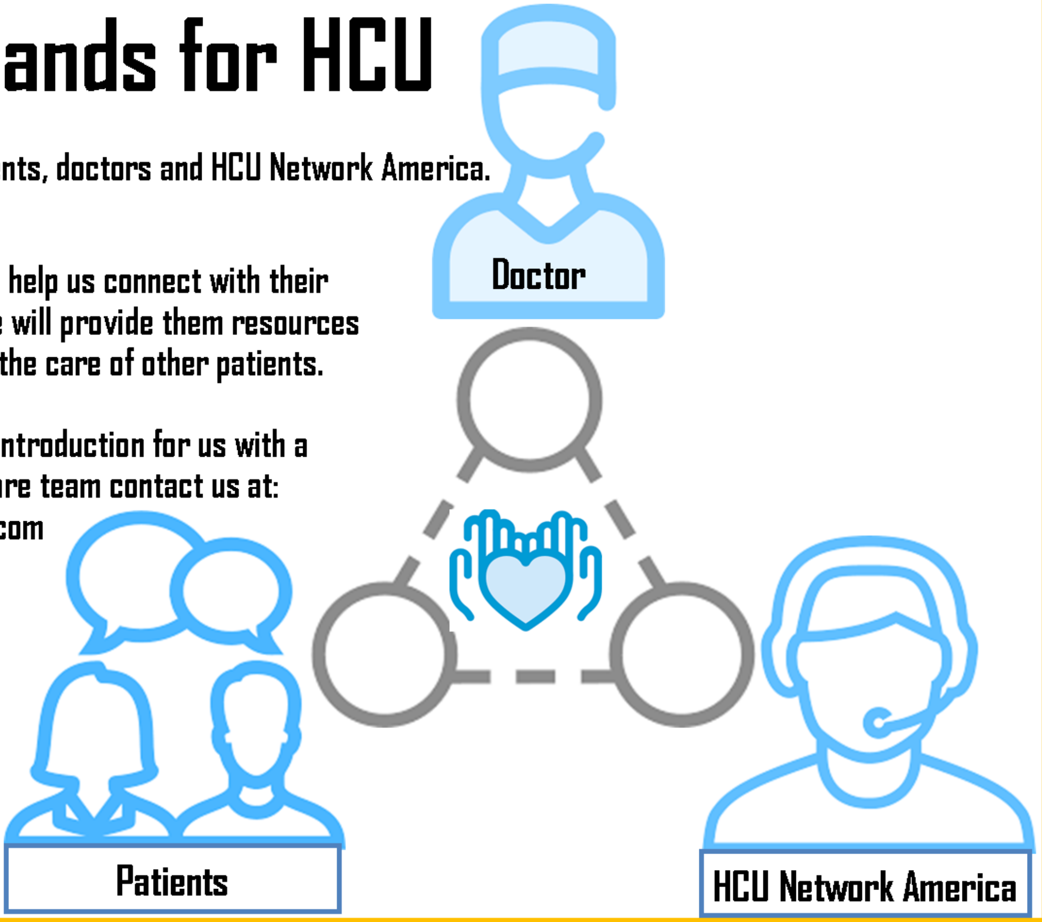
Ways to Get Involved

Helping Hands for HCU

A collaborative effort of patients, doctors and HCU Network America.

We are asking for patients to help us connect with their metabolic team. In return, we will provide them resources to help better your care and the care of other patients.

If you are willing to make an introduction for us with a member of your metabolic care team contact us at: HCUNetworkAmerica@gmail.com



Natural History Study

Current sites include: Boston, Philadelphia & Atlanta.

Joining the Natural History Study allows researchers to find out more about Homocystinuria and issues that patients face. Natural history studies help drive new therapies and a cure! If you qualify, we highly suggest you participate if there is a center in your area. You do not have to be a patient at one of these clinics to participate.

Amazon Smile

Shop Smile.Amazon.com when purchasing through Amazon and 0.5% will be donated to HCU Network America when you designate us as your charity of choice.

Click here to find out how!

Company Matching

Did you know that many companies big and small may match donations made to HCU Network America?

Click here to find out more

Click to donate directly

We'd like to thank the following content contributors:

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