
Aurora, Colorado

HCU NETWORK AMERICA

# HCU Network America

**Mission** HCU Network America strives to inform and provide resources for patients and families, create connections, influence state and federal policy, and support advancement of diagnosis and treatment for HCU and related disorders.

**Goals**

- To support research to improve diagnosis and treatment including a cure for the disease
- To provide information and resources to better manage the disease
- To create connections across the community and facilitate sharing of information and best practices through in-person and virtual events and discussions
- To assure all patients are diagnosed as early and efficiently as possible to enable access to care and avoid complications.

HCU Network America is a 501c3 nonprofit organization dedicated to helping patients and their families affected by classical Homocystinuria (HCU), severe Methylenetetrahydrofolate reductase (MTHFR), and cobalamin deficiencies C-X (Cbl). Since 2016, HCU Network America has strived to inform and provide resources for patients and families, create connections, influence state and federal policy, and support the advancement of diagnosis and treatment for the homocystinurias.

June 29, 2024

Dear Members of the Homocystinuria Community,

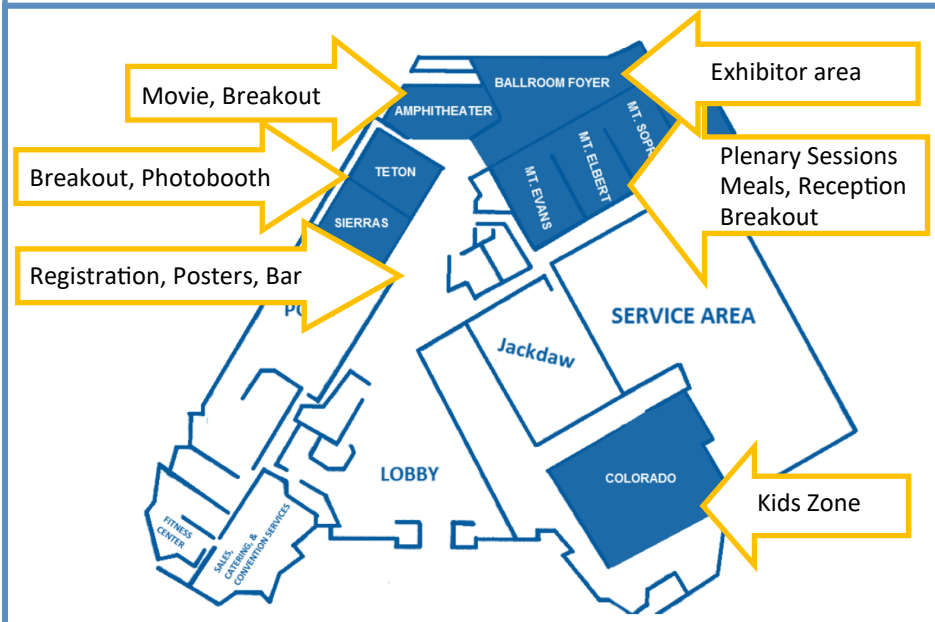
On behalf of the board of HCU Network America, we are elated you are able to join us for Moving Mountains, our fourth Patient/Family Conference, and we hope that you find it very informative and uplifting.

The theme of this meeting, *Moving Mountains*, recognizes the overwhelming feelings that a diagnosis of homocystinuria can bring but also serves as a reminder that together, *we are moving mountains*. From the initial diagnosis, to navigating treatment, and the obstacles that present along the path as patients age, our goal is to help you traverse the uncharted landscape. This meeting is all about providing you, our HCU community, with the knowledge, tools and support you need to successfully take on the challenges that may come you way.

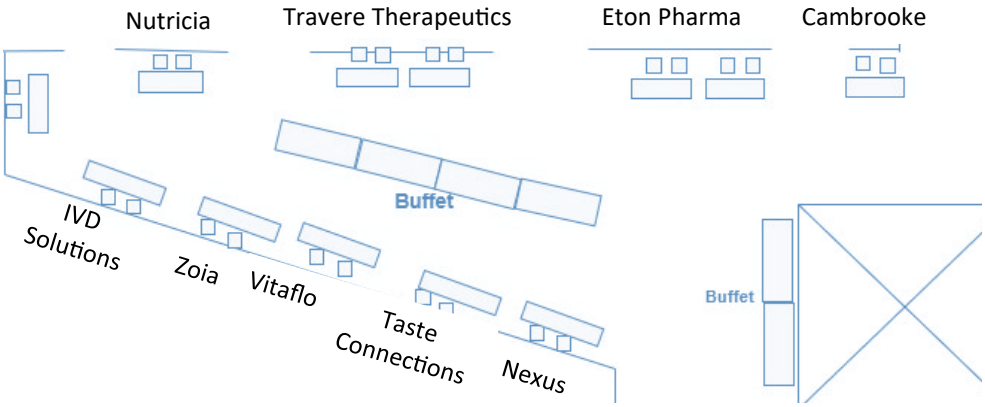
We are proud of what HCU Network America has been able to accomplish with your support, in our eight years of existence. I hope you leave this meeting with even greater hope for the future and the confidence that you or your loved one with HCU will have a long, healthy, and enjoyable life ahead.

Warm Regards,
Danae' Bartke, Executive Director
HCU Network America

# Hotel Floor Plan




## Exhibitor Layout



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# Agenda of Events

Friday, June 28, 2024

01:00 pm - 04:00 pm **Pool time**

04:00 pm - 08:00 pm **Registration**
Location: Ballroom Foyer

04:00 pm - 07:00 pm **Dinner On Your Own**
See folder insert for low-protein options in the area!

06:30 pm **Movie Night!**
Location: Amphitheater

This a family friendly opportunity to meet up and hang out together. We will have free concessions available!

## Saturday: Breakfast

| High Protein | Low Protein | Shared Items |
|---|---|---|
| • Pancakes<br>• Potatoes<br>• Scrambled eggs<br>• Sausage links<br>• Yogurt parfait bar<br>• Granola | • Pancakes w/ syrup (Cookforlove.org)<br>• Scrambled eggs (PKU Perspectives)<br>• Sausage (PKU Perspectives)<br>• So Delicious Coconut Yogurt<br>• Crunchy Granola (Cookforlove.org)<br>• Blueberry scones (Cambrooke) | • Coffee and tea<br>• Orange juice<br>• Seasonal berries<br>• Dried fruits<br>• Honey |

## Saturday: Lunch

| High Protein | Low Protein | Shared Items |
|---|---|---|
| • Smoked brisket<br>• Mac and cheese<br>• Cornbread<br>• Carmel Apple Pie | • Pulled jackfruit sliders<br>• Slider buns (Taste Connections)<br>• Mac and cheese (cookforlove.org)<br>• Katie's cornbread (cookforlove.org)<br>• Brownies (Cambrooke) | • Ice water & tea<br>• Garden salad<br>• Green apple coleslaw<br>• Potato salad<br>• Vegetable medley |

# Agenda of Events

## Saturday, June 29, 2024

07:00 am **Registration, Breakfast, and Exhibits**

08:15 am **Kid Zone Opens** - Kids must be dropped off and picked up by parents.
Location: Colorado
See page **16** for Kids Zone agenda

08:30 am **Welcome and Introduction**
**Danae' Bartke, HCUNA, Executive Director**

09:00 am **Keynote Presenter**
**Presenter: Dr. Albert Freedman**
**Founder, Rare Counseling**
This session will highlight common challenges faced by families affected by rare disease and perspective on how these complicated challenges can be met. Dr. Freedman is a practicing psychologist who specializes in serving patients and families affected by rare disease. His son, Jack, lived to the age of 26 with SMA (spinal muscular atrophy).

10:00 am **Intermission**
Please take this time and visit our conference sponsors!

10:30 am **Panel: Aging with a Homocystinuria**
**Moderator: Danae' Bartke, ED, HCU Network America**
**Panelists:**

- **Adam Settle, Living with Cobalamin C**
- **Alex Zarou Levine, Living with Classical HCU**
- **Annette Settle, Parent of child with Cobalamin C**
- **Barbara Zarou, Parent of child with Classical HCU**

This session will highlight the nuances of what it means to age with HCU navigating what the initial diagnosis and the various transitions of medications, relationships, and independence.

12:00 pm **Lunch | Networking Activity | Exhibits open**

01:30 pm **Session 1 Breakouts**

Location: Sierra

### Supporting Students with HCU through School Accommodations

**Presenter: Stephanie A. Hitti, PhD**

**Neuropsychology Postdoctoral Fellow, Children's Hospital Colorado**

Join Stephanie Hitti, PhD for a discussion about school accommodations for children with HCU. This session will explore the unique challenges faced by students with HCU in the educational setting and provide valuable insights into creating effective support systems. Participants will gain an understanding between the differences between 504 plans and Individualized Education Programs (IEPS) and how these documents can be utilized to ensure that students with HCU receive the necessary accommodations to thrive academically.

Or

Location: Ballroom

### Making Your Move: Strategies for a Great Transition to Adulthood

**Presenter: Laura Pickler, MD, MPH**

**Associate Professor, University of Colorado School of Medicine**

Dr. Laura Pickler will facilitate discussion to help you along your journey from pediatric to adult health care systems. Come ready to learn from each other and get ideas to guide the next step in your life! HCU Network America's excellent transition planning tool will be utilized.

02:30 pm **Intermission**

02:45 pm **Session 2 Breakouts**

Location: Sierra

### Navigating Low Vision Across the Life Span

**Presenter: Kara Hanson, OD, FAAO**

**Director, Low Vision Rehabilitation Service at Anschutz Medical Campus**

**Presenter: Dr. Catherine Smyth**

**Independent Research Consultant, University of Utah**

Low vision habilitation (for congenital vision loss) and rehabilitation (for acquired loss) helps children and adults with visual impairment maximize the way they use their vision. This course will review the possible developmental signs, symptoms and functional implications of vision loss caused by HCU. Educational, non-optical, and optical strategies, skills and resources can be used throughout the life span to improve ability to perform daily activities to increase independence and quality of life.

**Or - Continued on next page**

02:45 pm **Session 2 Breakouts**

Location: Ballroom

**Traveling with Classical HCU**

**Presenter: Danielle Baez, Living with HCU**

**Patient Advocate, World Traveler**

Intimidated by traveling while managing the low protein diet and formula? Worried that all your planning will be for nothing? Join Danielle for a lively discussion as she shares her experiences, best practices and tips for traveling with classical HCU.

03:45 pm **Intermission**

04:00 pm **Session 3 Breakout**

Location: Amphitheater

**Legal Strategies to Protect Loved Ones W/ Special Medical Needs**

**Presenter: James L. Parke, J.D., LL.M.**

**Keele and Parke, LLC**

Medically fragile family members are confronted with a variety of complicated situations. Questions of decision-making, government program eligibility, and payment can be complicated and overwhelming. This presentation will discuss various planning opportunities and potential solutions to help care for individuals with sensitive health needs. This includes a discussion on Supplemental Needs Trusts (also called Special Needs Trusts), ABLE Accounts, standalone irrevocable trusts, (d)(4)(A) trusts, (c)(2)(B)(iv) trusts, 76-270 trusts, Medicaid eligibility, powers of attorney for both healthcare and finances, as well as guardianship issues.

04:00 pm **Exhibits, Cocktails, & Poster Presentations** (Location: Foyer areas)

04:00 pm **Stop by the Photobooth!** (Location: Teton)

06:00 pm **Dinner Reception starts** (see menu below)

06:30 pm **Award Ceremony and Entertainment** (Location: Ballroom)

| High Protein | Low Protein | Shared Items |
|---|---|---|
| • Cavatappi pasta<br>• Linguini pasta<br>• Marinara sauce<br>• Alfredo sauce<br>• Meatballs<br>• Broccoli<br>• Parmesan<br>• Garlic Bread | • Spaghetti (Cambrooke)<br>• Fusilli (Cambrooke)<br>• Marinara<br>• Alfredo (cookforlove.org)<br>• Veggie balls (cookforlove.org)<br>• Zucchini and yellow squash<br>• Assorted cupcakes (Taste Connections) | • Ice water & tea<br>• Garden salad |

# Poster Presentations

Sierra-Teton Foyer

| Poster | Presenter |
|---|---|
| 1 | **HCU Network America Surveys help define what is important to patients**<br>Kimberly Champman, PhD, Children's National |
| 2 | **When to look for homocystinuria: Preliminary results from HCU Network America's Patient surveys**.<br>Kimberly Champman, PhD, Children's National |
| 3 | **Assessing the potential of enzymes as inhaled therapeutics for inborn errors of metabolism**<br>Kristen Skvorak, PhD |
| 4 | **Hydroxocobalamin Dose Intensification Improves Ocular, Neurological, and Biochemical Outcomes in CblC Deficiency**<br>Irini Manoli, MD, NIH |
| 5 | **A Case Series of Pancreatitis in Patients with Poorly Controlled Classic Homocystinuria**<br>Rana Aljaberi, MD, Emory University |
| 6 | **A microbiome-mediated dietary treatment for classical homocystinuria**<br>Joseph Schinaman, PhD, Petri Bio |
| 7 | **Latest Results From the COMPOSE® Phase 1/2 Trial for the Treatment of Classical Homocystinuria (HCU) Using Pegtibatinase, a Novel Investigational Enzyme Replacement Therapy**<br>Jalé Güner, Travere Therapeutics |
| 8 | **Pegtibatinase, an Investigational Enzyme Replacement Therapy, for the Treatment of Classical Homocystinuria (HCU): Design of the HARMONY Phase 3 Study**<br>Sagar Vaidya, MD, PhD, Travere Therapeutics |

# James Weisfeld-Adams

## 1979-2018

### 2024 HCU Hero Award Recipient

Dr. James Weisfeld-Adams was an accomplished physician and researcher who made significant contributions to the field of clinical genetics and metabolism. Born in Hastings, UK, in 1979, he pursued his medical degree at the University of Aberdeen in Scotland. His passion for understanding inborn errors of metabolism led him to complete a combined Pediatric and Medical Genetics residency, followed by a Clinical Biochemical Genetics fellowship at Mount Sinai School of Medicine in New York.

In January 2012, Dr. Weisfeld-Adams joined the faculty at Mount Sinai, where he focused on research related to homocystinuria, cobalamin defects, and urea cycle disorders. He actively participated in various clinical care initiatives, including serving as the local site principal investigator for the national Urea Cycle Disorders Consortium and the European Network and Registry for Homocystinurias and Methylation Defects. His dedication to patient care and academic pursuits earned him respect from colleagues worldwide.

In December 2013, Dr. Weisfeld-Adams moved to Colorado, becoming an Assistant Professor in Pediatrics at the University of Colorado School of Medicine. He continued his work in metabolic disorders, mentoring graduate genetic counseling students, medical genetics residents, and biochemical genetics fellows. Despite health challenges, he remained committed to his academic endeavors, contributing insightful comments to the field up until the last week of his life.

James was known for his compassionate and reflective personality, making him easy to collaborate with and considerate of his colleagues. His love for both Exmoor in England and Scotland remained strong, even as he embraced his new home in the USA. Tragically, his promising academic career was cut short by renal cancer, but his legacy lives on through his contributions to medicine and the lives he touched. He is survived by his wife, Emma, and their two children.

HCU Network America is bestowing this HCU Hero award posthumously to Dr. Weisfeld-Adams in light of the many contributions he made to our community through his research and clinical care and the passion and commitment he demonstrated in add he did for us. May his memory continue to inspire those who follow in his footsteps.

# Agenda of Events

**Sunday, June 30, 2024**

07:00 am **Breakfast and Exhibits Open**

08:15 am **Kid Zone Opens** - Kids must be dropped off and picked up by parents.
Location: Colorado
See page **16** for Kids Zone agenda

08:30 am **Diagnostics and Testing Opening Remarks**
**Danae' Bartke, HCUNA, Executive Director**

08:40 am **The Future Is Not Just New Disorders: Reviewing, Updating and Education on HCU**
**Presenter: Gregory Bonn MT (ASCP)**
**Newborn Screening Mgr., Colorado Depart. of Public Health and Environment**
In 1979 homocystinuria was added to the Colorado Newborn Screening Program panel with four other conditions. At some point in the 1990's homocystinuria and maple syrup urine disease were dropped from the CONBSP panel. The conditions were added back on to the panel in 2006 with the implementation of tandem mass spectrometry (MS). In the fall of 2021, the CONBSP underwent a process review for Classical HCU after a webinar and discussion about state cutoffs. After discussing the status of the CONBSP cutoff with the Colorado MS Scientist it was clear that was significant improvement. The cutoff for methionine was changed from 100 umol/L to 48 umol/L. Additional changes based on initial and second screen algorithms were updated with secondary ratios. These updates allowed for the lowering of the cutoff while limiting the impact on calling parents into the hospital for diagnostic testing. In the spring of 2022 the follow up specialists at Children's Hospital Colorado called to check on methionine results for a missed case in 2020. This child would have been identified with the new cutoff.

08:50 am **Enhancing the Detection of Newborns with Homocystinuria**
**Presenter: Carla Cuthbert, PhD, FACMG**
**Chief, Newborn Screening and Molecular Biology Branch, Division of Laboratory Sciences, Centers for Disease Control and Prevention**
Carla D. Cuthbert, PhD is the Chief of the Newborn Screening and Molecular Biology Branch (NSMBB) in the Division of Laboratory Sciences, National Center for Environmental Health, in the US Centers for Disease Control and Prevention (CDC). She has held this position since December 2009. Under her leadership,

Cont. NSMBB has focused on enhancing laboratory disease detection in newborns by implementing technologies and capabilities to improve test development and translational research, screening test performance and result interpretation, training and technology transfer, and by assisting state programs to expand screening to include new conditions of high priority.

09:00 am **A Point of Care-Device for the Determination of Total Homocysteine (tHCY) the HCY Now**

**Presenter: Robert Harper**

**Founder and CEO, In Vitro Diagnostic Solutions**

The discussion will be center around the progress of a home monitoring system for patients diagnosed with HCU to monitor their total blood homocysteine concentrations at home. In Vitro Diagnostic Solutions has received over 8 million dollars in NIH funding for the development of point-of-care devices for patients born with IEMs. We are currently in clinical studies for PKU at University of Pittsburg Medical Center and Boston's Children's Hospital. As well, we are currently in clinical studies for Glucose-6-Phosphate Dehydrogenase (G6PD) for Oxford MORU Institute, Cambodia and Menzies (Australia) in Bangladesh. We have filed for a Phase I application to the National Institute of Health for the home monitoring of blood homocysteine. We appreciate the support of the HCU Network America.

09:10 am **Update on Classical HCU Research Map**

**Presenter: Margie McGlynn, RPh, HonDr Sci**

**Cofounder and President, HCU Network America**

This presentation will provide an update on therapies in development for Classical HCU

09:25 am **Pegtibatinase, an Investigational Enzyme Replacement Therapy for the Treatment of Classical Homocystinuria due to cystathionine-beta synthase (CBS) -deficiency**

**Presenter: Sagar A. Vaidya, MD, PhD**

**Vice President, Clinical Development at Travere Therapeutics, Inc.**

The presentation will provide an overview of pegtibatinase, an investigational treatment being developed by Travere Therapeutics for classical homocystinuria due to CBS-deficiency, and how pegtibatinase may work in the body to lower homocysteine. The latest results on the safety and effectiveness of pegtibatinase from the COMPOSE Phase 1/2 clinical trial will be shared. Additionally, information about the currently open Phase 3 HARMONY study will be provided.

09:35 am **Crystallography-based fragment screening to develop pharmacological chaperones for classic homocystinuria**

**Presenter: Thomas J. McCorvie, PhD**

**Senior Research Associate, Newcastle University**

Classical homocystinuria (HCU) is associated with mutations of cystathionine beta

Cont. -synthase (CBS), the central enzyme in the transsulfuration pathway. Many of these mutations cause misfolding and aggregation of CBS. It has been suggested that CBS is a tetrameric enzyme however contradicting studies suggest larger oligomers. We have determined the structure of full-length CBS showing that it forms filaments. Efforts are underway to screen for molecules that could be developed into a therapy for HCU.

09:45 am **Novel strategies to improve treatment for HCU without injecting proteins: Let me count the ways**

**Presenter: Ken Maclean, PhD**

**Professor of Pediatrics, University of Colorado School of Medicine, Section of Genetics and Metabolism**

The Maclean Lab currently have four novel therapies for HCU in development. Ken will be describing his lab's progress with these strategies and outlining their advantages in efficacy, cost, simplicity and longevity of action over other therapies.

10:00 am **The Remethylation Landscape**

**Presenter: Brittany Parke,**

**Director of Remethylation Research, Board Member, HCU Network America**

This presentation will provide an update on the Remethylation Research and Strategy Map.

10:15 am **How Biophysics enhances our understanding of CblC Disease, suggesting new therapeutic approaches**

**Presenter: Silvia Vilasi, PhD**

**Researcher at Institute of Biophysics, National Research Council (CNR), Italy**

The presentation will focus on the results obtained from the research project titled 'Identification of Compounds to Rescue MMACHC Functional Deficiency in CblC Disease,' supported by the associations HCU America, Organic Acidemia, and Italian cblC aps. Dr. Vilasi will demonstrate how a biophysical approach facilitates the understanding of the chemico-physical defects induced on the MMACHC protein by cblC pathological mutations. Insights into a therapeutic strategy involving compounds capable of correcting these defects will be provided.

10:25 am **What is the right dose? Hydroxocobalamin dose escalation vs intensification for cobalamin-related disorders.**

**Presenter: Irini Manoli, MD, PhD**

**Clinical Associate Investigator, Senior Research Physician, Metabolic Medicine Branch, NHGRI, NIH**

Join Dr Manoli to discuss the evolving recommendations for the management of cobalamin and remethylation disorders. We will compare the standard dose escalation with the ultra-high dose approach regarding injectable hydroxocobalamin, and what we know about the effect on long-term outcomes for this group of

Cont. disorders. We will discuss the collaborative efforts towards updated treatment guidelines and improved access to concentrated hydroxocobalamin.

10:35 am **Intermission**
Please take this time and visit our conference sponsors!

11:15 am **Registries and Why, As an Organization, We Care**
**Presenter: MD, PhD**
**Attending Geneticist Children's National Rare Disease Institute**
The Homocystinurias Data Collection Program launched in early 2022 is a secure global platform for the HCU community to share their personal experience and help to increase understanding of how HCU affects the body. Dr. Chapman will speak about some early learnings and why you - the expert in your disease – need to participate to help accelerate the development of treatments. Your de-identified data will be available to your community, to your physicians and to researchers worldwide.

12:00 pm **Ask the Expert Panel**
**Moderator: Margie McGlynn, RPh, HonDr Sci**
**Cofounder and President, HCU Network America**
Did something get discussed throughout the conference that you would like clarified? What about something that you are interested in, but wasn't discussed? This is your time to have any remain-ing questions answered by our panel of experts.

12:30 pm **Closing Remarks, then Lunch**

## Sunday: Breakfast

| High Protein | Low Protein | Shared Items |
|---|---|---|
| • Pancakes | • Pancakes w/ syrup (Cookforlove.org) | • Coffee and tea |
| • Potatoes | • Scrambled eggs (PKU Perspectives) | • Orange juice |
| • Scrambled eggs | • Sausage (PKU Perspectives) | • Seasonal berries |
| • Sausage links | • So Delicious Coconut Yogurt | • Dried fruits |
| • Yogurt parfait bar | • Crunchy Granola (Cookforlove.org) | • Honey |
| • Granola | • Blueberry scones (Cambrooke) | |

## Sunday: Lunch

| High Protein | Low Protein | Shared Items |
|---|---|---|
| • Macaroni salad | • Macaroni salad (Promin) | • Ice water & tea |
| • Hamburgers | • Beet burgers (Taste Connections) | • Condiments |
| • Hotdogs | • Carrot dogs (Radical Plants) | • Garden salad |
| • Tater tots | • Yucca taters (Cambrooke) | • Vegetable medley |
| • Cookies | • Cookies (Cambrooke & PKU Perspec) | • Watermelon wedges |

# Kids Zone Agenda

Location: Colorado

| Saturday | Kids Zone | Teen |
|---|---|---|
| 08:15 am | Kids Zone opens | Kids Zone opens |
| 08:30 am | Icebreaker activities | Icebreaker activity |
| 09:00 am | Arts & crafts activity | **Sharing Your Story with Young Adult Rare Representatives (YARR)** |
| 10:00 am | Board games | Board games |
| 10:30 am | Sibling Workshop | Sibling Workshop |
| 12:00 pm | Lunch with parents | Lunch with parents |
| 01:15 pm | Kids Zone resumes | Kids Zone resumes |
| 01:30 pm | Movie: Inside Out | Transition Breakout |
| 03:00 pm | Movement activity | Movement activity |
| 03:30 pm | **Science is Fun with Petri Bio** | **Science is Fun with Petri Bio** |
| 04:30 | Board Games<br>**please pick your child up no later than 5:10* | Board games |

| Sunday | Kids Zone | Teen |
|---|---|---|
| 08:15 am | Kids Zone opens | Kids Zone opens |
| 08:30 am | Outside activities | Wake and Take Coffee Shop<br>*Permission slip required |
| 09:00 | Outside activities | Wake and Take Coffee Shop |
| 10:00 am | **Armed with Umbrellas** | **Armed with Umbrellas** |
| 11:00 am | Free play—choice of arts & crafts or board games | Free play—choice of arts & crafts or board games |
| 12:45 pm | Lunch with Parents | Lunch with Parents |

# Kids Zone Agenda

## Kid & Teen Workshops

### Sharing Your Story With Young Adult Rare Representatives (YARR)

**Target participant ages: 10+**
**Presenter: McKenna Spence-Olson**
**YARR Representative**

Are you open to sharing about your disorder with others? What benefits do you think sharing your story might have? These are questions that will be explored in this engaging workshop! McKenna, a rare disease patient herself, will guide participants through the process of crafting their own 'stories' and participants will leave with an 'elevator pitch' version of their stories to be used for self-advocacy, spreading awareness, and other future advocacy opportunities.

### Armed with Umbrellas

Target participant ages: 3rd grade & up
**Presenter: Christa Green , Educational Therapist & adult living with Classical HCU)**
**Presenter: Becky Ransome Founder of Armed With Umbrellas, LLC**

What does it mean to be 'Armed with Umbrellas'? That's exactly what we will explore in this exciting and engaging workshop! First, we will dive into some self-exploration and get to know ourselves a bit better. What bugs you most about having HCU? How do we manage our feelings and emotions? Participants will be encouraged to consider what strategies they currently utilize for self-regulation, self-care and overall wellness, and will be 'armed' with some additional tools in the toolkit! Finally, we'll discuss how once we know ourselves better, we can help others by bringing them under our 'umbrella'

# Conference Speakers

**Danielle Benton-Baez**
Danielle is an adult thriving with Classical HCU. Through experiences with travel, food, sports, and leisure, coupled with a passion and zest for life, Danielle has created a life in which HCU is not her defining characteristic.

She shares her love for travel with her husband, three children, and dogs, all while managing a low-protein diet and medications. Her adventures have taken her across the globe by land, air, and sea, offering her a rich tapestry of awe-inspiring memories.

Danielle loves discovering new cultures and culinary delights through food tours and exploring remote destinations. She is excited to help other HCU heroes collect life-long experiences through travel and food, one step easier at a time.

**Gregory Bonn MT (ASCP)**
Gregory (Greg) Bonn MT (ASCP) is the program manager for the Colorado Newborn Screening Program with the Department of Health and Environment (CDPHE). He has been with the program for over 12 years initially working as the molecular technologist during the implementation of Severe Combined Immunodeficiency (SCID). In January of 2022 he officially took on the role of program manager. Prior to coming to CDPHE he worked in hospital laboratories, an organ donation laboratory, and a medical device company. Greg is a native of North Dakota and graduated from North Dakota State with B.S. degrees in Microbiology and Clinical Laboratory Science.

**Kimberly Chapman, MD, PhD, FAAP, FABIM**
Kimberly Chapman is an attending physician in Genetics and Metabolism at Children's National and an assistant Professor of Pediatrics and Integrated Systems Biology at George Washington University in Washington DC, USA. She specializes in taking care of individuals with defects of methylation, homocysteine and propionate metabolism.

**Keynote: Albert Freedman, PhD**
Albert Freedman, PhD, is a psychologist in independent practice, serving clients and families nationally. Dr. Freedman provides counseling support for patients and families affected by rare disease. He serves as a consulting psychologist to rare disease advocacy organizations and biopharmaceutical companies globally. Dr. Freedman speaks widely on the topic of mental health and rare disease. Al's son, Jack, lived with Spinal Muscular Atrophy (SMA) for 26 years.

**Kara Hanson, OD, FAAO**
Dr. Hanson provides functional vision evaluations for children, adults, and seniors with visual impairments at UCHealth Sue Anschutz-Rodgers Eye Center, Children's Hospital Colorado and Anchor Center for Blind Children. She teaches ophthalmology and optometry residents in the clinical and didactic settings.

**Robert Harper, Founder and CEO**
**In Vitro Diagnostic Solutions**
Founded in 2017 to address home monitoring for patients with inborn errors of metabolism (IEMs). IVDS focus IEMs for HCU, PKU, CPK and G6PD.

**Stephanie A. Hitti, Ph.D.**
Stephanie Hitti, Ph.D. is a postdoctoral fellow in bilingual/multicultural pediatric neuropsychology at Children's Hospital Colorado. She received her undergraduate degree in Psychology and Child Development at Vanderbilt University and went on to get her Ph.D. in Child Clinical Psychology from Virginia Commonwealth University. She has specialized training in how the brain develops and uses this training to evaluate and help manage children with health conditions that affect the brain. She grew up in Venezuela and has a special interest in bilingual/multicultural populations. As such, many of the evaluations she conducts are in Spanish.

**Shu Li PhD**

Shu holds a Ph.D. in Plant Biology and brings extensive experience from the Agri-food tech and alternative protein sectors. Her journey in science is marked by a commitment to harnessing technology to tackle metabolic disorders and improve overall well-being. Beyond her professional achievements, Dr. Li is a dedicated advocate for diversity and inclusion in science. As a long-term member and past president of the National Graduate Women in Science (GWIS), she works tirelessly to support and empower scientists who face challenges due to gender or underrepresented backgrounds.

**Ken Maclean, PhD**

Dr Ken Maclean is a Professor of Pediatrics at the University of Colorado School of Medicine and holds the Ehst-Hummel-Kaufman Family Endowed chair in Inherited Metabolic Disease. In 1998, Dr Maclean came to Colorado to work on the transcriptional and post-translational regulation of cystathionine beta-synthase (CBS) in the laboratory of Dr Jan Kraus. Since establishing his laboratory in 2002, Dr Maclean's work has centered on using transgenic and knockout/in mouse models to investigate the pathobiology of CBS deficient homocystinuria (HCU), homocysteine remethylation defects, Down Syndrome, propionic acidemia and non-ketotic-hyperglycinemia.

**Irini Manoli, MD, PhD**

Dr Manoli combines work on animal models and clinical studies with the aim to develop new therapies for methylmalonic acidemias and defects of intracellular cobalamin metabolism. She studies the pathophysiology underlying disease manifestations to develop biomarkers and clinical outcome parameters that will enable translating experimental genomic therapies from animal models to the clinic. Her clinical research on medical nutrition therapy in MMA and cobalamin C resulted in the reevaluation of the dietary management of this group of inborn errors of metabolism. She is part of an international collaborative effort to revise the guidelines for the diagnosis and management of cobalamin-related remethylation disorders.

**Thomas J. McCorvie, PhD**

Thomas McCorvie is a senior research associate at Newcastle University, UK. He received a PhD based on his work on the biochemical basis of galactosemia from Queen's University, Belfast. He has had multiple postdoctoral positions at University of Oxford and Imperial College studying human proteins involved in rare diseases using structural biology. Currently his interest is in understanding enzymes involved in glycogen storage disorders, homocystinuria, and vitamin B12 metabolism towards developing novel therapies.

**Margie McGlynn, R. Ph., HonDr Sci**

Margie McGlynn is Cofounder and President of HCU Network America, and President of the Hempling Foundation for Homocystinuria Research, a fund she established to support research on new therapies in honor of her late sisters. Margie actively supports a chair she endowed at University at Buffalo, which focuses on state-of-the-art clinical pharmacy care for patients with rare genetic metabolic disorders. Margie received a BS in Pharmacy, MBA in Marketing and Honorary Doctorate in Sciences from University at Buffalo. She spent 26 years at Merck culminating as President, Global Vaccines and Anti-infectives, and now serves on the boards of Amicus Therapeutics, Novavax and University at Buffalo Foundation.

**Brittany Parke**

Brittany Parke serves on the board for HCU Network America and is the director of Remethylation Research. Brittany is the mother of two boys born with Homocystinuria Cobalamin G. Drew unfortunately was diagnosed only after suffering from seizures and severe brain atrophy. He sadly passed away at the age of three and a half months. Thanks to early screening measures, they knew their son Grayson would also have the same disorder when he was born in 2019. An amazing team from Colorado Children's Hospital helped her family welcome their little boy and began treatment immediately. Grayson is now five years old and thriving. While he has a mild speech delay, he remains non-symptomatic.

**James L. Parke, J.D., LL.M.**

Jim Parke is an experienced attorney, business executive, and board member. His professional passion is helping people find answers to their most pressing problems. He has extensive experience providing legal advice to people with medically fragile family members. His experience helps his clients find real-world solutions to complex family, business, tax, and estate planning issues. Jim is a graduate of Weber State University, Gonzaga University School of Law, and New York University School of Law. Jim and his wife Megan have five exceptional children. Jim is the chairman of the board for Poudre Valley Hospital and Medical Center of the Rockies and serves as the president of the Fort Collins Stake of the Church of Jesus Christ of Latter-day Saints. He is a member of the advisory board for Gonzaga University School of Law and Weber State University. He is also a board member for Verus Bank, as well as several startup companies. Jim is also a supportive uncle to a nephew with Homocystinuria CblG.

**Laura Pickler, MD, MPH**

Early in her medical training, Dr. Laura Pickler became aware that young adults with complex medical needs often experienced suboptimal health and lacked fundamental access to basic primary care services especially as they transitioned to the adult health care system. Over the last 20 years she has worked in diverse ways to improve the health of this patient population. Currently, she is a faculty member at the University of Colorado School of Medicine and uses this role as a platform to improve transition from pediatric to adult health care for young adults with HCU and other genetic conditions.

**Adam Settle**

When Pennsylvania started testing newborns for metabolic diseases. Adam Settle was one of the first to be diagnosed with Cobalamin-C Deficiency. Legally blind, Adam never let his limitations hold him back from accomplishing his dreams. His love for people and his desire to help has allowed him to achieve things others never thought possible. Adam is now an adult who travels the globe independently as a missionary.

**Annette Settle**

Annettte Settle is a wife, mom, and Nona to 9 children and 20 grandchildren. Her youngest, Adam was diagnosed at birth with Cobalamin C deficiency. Being the 8th child, Annette encouraged Adam's siblings to not treat him any differently than any other child. She believes this mindset has allowed Adam to accomplish many things in his life.

**Dr. Catherine Smyth**

Dr. Smyth has provided support for families in early intervention in the area of visual impairment for over thirty years as a teacher of students with visual impairment. Her research interests include concept development and tactual assessments for young children with visual impairment, how vision loss affects the mealtime process, using tele-intervention for EI vision services, and early visual screening for infants.

**McKenna Spence-Olson**

McKenna Spence-Olson has lived in Colorado for most of her life and greatly enjoys experiencing all that the mountains have to offer! McKenna is a two-time recipient of an EveryLife Foundation #RAREis Scholarship and has been a Young Adult Rare Representative (YARR) since 2021. She was diagnosed with Systemic Mastocytosis (SM) when she was 17 and appreciates the opportunities she has had over the last 8 years to share her story about growing into adulthood with a rare disease, both personally through YARR and professionally through her career in speech-language pathology. She is honored to have the opportunity to connect with the HCU community at this year's conference!

**Sagar A. Vaidya, MD, PhD**

Dr. Vaidya is a rare disease drug developer and the Vice President of Clinical Development at Travere Therapeutics since 2019. Dr. Vaidya is responsible for overseeing clinical development of the company's metabolic portfolio of products. Dr. Vaidya previously served in roles in clinical development at Sangamo Therapeutics and BioMarin Pharmaceuticals, and remains deeply committed to developing new treatments for people with rare diseases. Dr. Vaidya completed his Infectious Diseases fellowship at Massachusetts General Hospital, Internal Medicine/ Pediatrics residency at Icahn School of Medicine at Mount Sinai Hospital, and received his M.D. and Ph.D. in Molecular Genetics from UCLA .

**Barbara Zarou**

Barbara is a board-certified OB/GYN and a mother to three sons and two stepsons. Having practiced medicine for 25 years and later becoming involved in education, she currently serves as a medical specialist for the NYC Office of School Health.
Barbara's middle son, Alexander, was born in 1987 with Classical Homocystinuria. Knowing very little about the illness except what she had learned in medical school, Barbara was, of course, devastated.

From an early age, creating music would help Alex get through the difficult years of living with a chronic illness, and he devoted his life to becoming a singer/songwriter (aka alexorangedrink). Barbara recognizes that her other sons also grew up experiencing issues around Alex's illness. Each was impacted differently, but all have become better people because of it. Barbara now wants to share her experiences with other parents to offer reassurance as they navigate raising a child with Homocystinuria.


**Alex Zarou Levine**

Alex Orange Drink is the moniker for Alex Zarou Levine, lead singer & songwriter of The So So Glos. Alex was diagnosed with HCU in 1987 in New York through newborn screening. Along with his band, he co-founded the beloved Brooklyn venues Shea Stadium, and Market Hotel, which served as creative hubs for the New York City D.I.Y. artist community. Alex's music is deeply personal in that it confronts his health issues with a unique and playful vulnerability. Because of this, Alex has spoken and performed internationally at various medical conferences, where he has been embraced by the rare disease community.

# Sponsor Information

We come together every day to help patients, families, and caregivers of all backgrounds as they navigate life with a rare disease. On this path, we know the need for treatment options is urgent – that is why our global team works with the rare disease community to identify, develop, and deliver life-changing therapies. In pursuit of this mission, we continuously seek to understand the diverse perspectives of rare patients and to courageously forge new paths to make a difference in their lives and provide hope – today and tomorrow.

At **Eton**, we search the world over for meaningful therapies that we can bring to patients living with rare diseases.

The **EveryLife Foundation for Rare Diseases** is a 501(c)(3) non-profit, nonpartisan organization dedicated to empowering the rare disease patient community to advocate for impactful, science-driven legislation and policy that advances the equitable development of and access to lifesaving diagnoses, treatments and cures.

At **Nutricia** we believe in the power of nutrition to make a positive and proven difference to health. Everyday we apply our collective expertise in nutrition to some of the world's biggest health challenges in life in the nutritional management of diseases, disorders and medical conditions. To find out more about our products please visit https://www.nutriciametabolics.com/

**Ajinomoto Cambrooke** is a leader and global innovator in the field of advanced medical nutrition. We are passionate in our pursuit of improving the lives of others by bringing new therapeutic options to those with therapeutic and chronic medical needs. A company with over 100 low protein foods and unique formulas like, Homactin AA Plus Powder 15 an amino acid-based **methionine-free**, nutritionally complete, powder medical food for the dietary management of Homocystinuria and comes in a light, fresh taste in a Lemon Lime flavor and suitable for ages 1 through adulthood and can be ordered at Cambrooke.com.

**IVDS** was founded to offer Point of Care simple to use diagnostics for rare metabolic diseases. IVDS is composed of skilled engineers and scientists for innovative technology development. IVDS's main developmental effort is focused on developing novel rare metabolic disease platforms that utilize a portable handheld meter and mobile application.

Designed with to fit into your lifestyle, **Nexus Medical Nutrition** offers unique and innovative medical formula tablets to make dietary management of homocystinuria a little easier. Nexus is proud to support HCU Network America's 2024 Family Conference. For more information, please visit www.nexusmedicalnutrition.com

**Taste Connections** specializes in low-protein products such as bread mix, multi-baking mix, etc. breads, cookies, and other baked goods that are suitable for metabolic conditions such as Phenylketonuria, Homocystinuria, etc. We are committed to providing best quality low-protein products to our customers at lowest possible price. Visit Tasteconnections.com to view our low protein items and mixes.

**Tome Biosciences, Inc.,** is the programmable genomic integration (PGI) company. PGI technologies allow for the insertion of any genetic sequence of any size at any location in the genome with site-specific precision. PGI is a revolutionary approach for the development of potentially curative cell and integrative gene therapies.

**Vitaflo™** develops innovative specialized medical foods for Inherited Metabolic Diseases. We offer a range of Homocystinuria (HCU) formulas for the dietary management of HCU from childhood through adulthood. Vitaflo products and services offer choice, flexibility and convenience to support a sustainable therapeutic diet. For more information, visit https://www.vitaflousa.com/

**ZOIA Pharma** provides exceptional service to individuals diagnosed with Inborn Errors of Metabolism (IEMs) and other rare metabolic conditions, supplying a broad portfolio of medical foods and nutritional products. We specialize in navigating the complexities of healthcare, ensuring patients gain access to necessary dietary formula and low protein medical foods without the financial strain.

**Abbott** is a global healthcare leader that helps people live more fully at all stages of life. Our portfolio of life-changing technologies spans the spectrum of healthcare, with leading businesses and products in diagnostics, medical devices, nutritionals and branded generic medicines. Our 114,000 colleagues serve people in more than 160 countries. Connect with us at www.abbott.com and on LinkedIn, Facebook, Instagram, X and YouTube.

Now Enrolling

# Acappella

**Sponsor:** Travere Therapeutics
**Study type:** Natural History (no investigational medicine given)
**Study duration:** About 6.5 years
**Goal:** To learn more about classical HCU & the course of the disease

| TO QUALIFY* | AGE OF PARTICIPANTS | DETAILS |
|---|---|---|
| Diagnosis of Homocystinuria due to CBS Deficiency (Classical Homocystinuria) | Currently enrolling 1 to 4 years old | The study will include three key stages (screening, enrollment, and observational follow-up) and will last approximately 6.5 years. |

**Your child will need to meet all other study criteria to take part in the ACAPPELLA Study.*

**Study Locations**
**United States: Colorado, Washington DC, Georgia, Pennsylvania**
**Countries outside of the US: Ireland and Qatar**

150

**The ACAPPELLA Study has already enrolled 100 adults and children over 5 years old,** and is now looking for children aged 1 to 4 years old to take part.

**Approximately 150 people aged between 1 and 65** will participate at sites in the US, Europe and other countries around the world.

**You may be able to receive payment** for time and travel if your child participates in this study.

**Talk to your doctor and family members about your child joining the ACAPPELLA study.**

Sites are open and currently enrolling participants. For the most current information about the ACAPPELLA Study and to see additional eligibility criteria, please visit:

https://www.clinicaltrials.gov/study/NCT02998710

If you have any questions, please email:
**medinfo@travere.com**

For more information, please scan the QR code or visit:
***www.hcuconnection.com***

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TRAVERE THERAPEUTICS