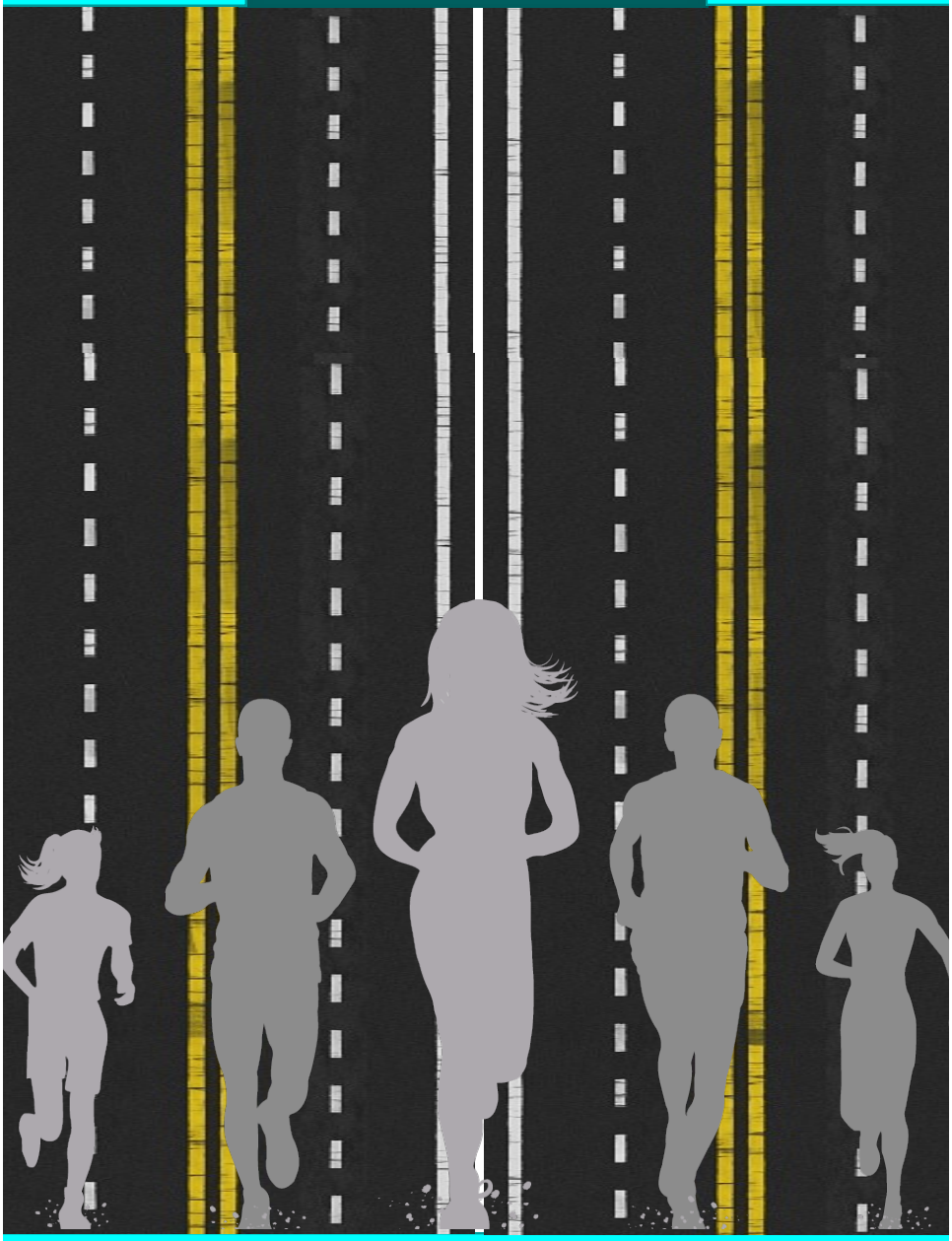
START

Taking the Lead for HCU



IT'S GO TIME

2018 HCU Network America Conference | Westford, MA

April 21, 2018

Dear Members of the Homocystinuria Community,

On behalf of the board of HCU Network America, we are so pleased you are able to join us for this Patient/Family Conference, and we hope that you find it to be very rewarding.

This meeting is all about you. Our goal is to help you as patients or caregivers more easily and successfully manage HCU. We know how devastated you were when you learned you were affected by the disease. It can be very scary. It can be very complicated. It can make you feel socially isolated. But we also know all of that can be overcome if you have the right resources and support system in place to help you.

As many of you know, the first ever national HCU Patient/Family Conference in the US was held in Denver in 2011. Many of us affected by HCU attended this conference, and we vowed that we would build upon the success of that meeting and establish a formal HCU patient support organization that could help patients and families more easily manage the disease. We now have that organization in place at HCU Network America, and we are lucky to have Danae Bartke, a patient with HCU, in place as our Executive Director.

So what do we hope to accomplish with our two-day meeting.? We planned the agenda around what many of you have told us your greatest needs are, and we hope you leave here armed with resources and tools to help you address those needs. But probably the most important take-away for all of you will come from the social interactions - the camaraderie you feel and the new friends you meet; others who are walking in your shoes every day. You will give each other comfort. You will give each other new ideas on what works for you and your family. You will give each other a sense of community and belonging, and hopefully you will never again feel alone in managing this disease.

May you leave this meeting with greater hope for the future and confidence that you or your loved one with HCU will live an easier, happier and more fulfilling life.

Warm Regards,

Margie McGlynn
President, HCU Network America

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HCU Network America Board of Directors & Medical Advisors

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Margie McGlynn, President
Kristin Rapp, Vice President
Danae' Bartke, Executive Director
Kimberly Chapman, MD, PhD
Harvey Levy, MD

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NECPAD

T-Shirts were graciously provided from grant provided by the Hempling Foundation

Conference Menu

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All meals, except for the Cambrooke Reception which is all low-protein, will include both low-protein and high protein creations. Protein content of each item will be designated in the buffet lines.

Breakfast
Saturday, April 21 and Sunday, April 22

High Protein	Low Protein	Shared Items
• Scrambled Eggs	• Eggz*	• Seasonal Fruit
• Waffles/French Toast	• Pancakes**	• Preserves
• Bacon/ Sausage	• Creamy Hot Cereal*	• Coffee and Tea
• Home Fried Potatoes	• Yucca Fries*	• Chilled Juices
• Breakfast breads and pastries	• Breakfast breads and pastries**	

Lunch: Saturday, April 21

High Protein:
- Sicilian Sausage Soup
- Pasta Primavera
- Parmesan Chicken
- Sicilian Pizza
- Caesar Salad
- Cannoli & Tiramisu

Low Protein:
- Minestrone Soup*
- Pasta Primavera*
- Pasta Marinara*
- Garlic Baguette**
- Pizza**
- Garden Salad
- Chocolate Chip Cookies* **&** Brownies**

Lunch: Sunday, April 22

High Protein:
- Chicken tortilla soup
- Beef Fajitas
- Chicken Fajitas
- Flour Tortillas
- Refried beans
- Spanish Rice
- Southwest Salad
- Tres leches cake & Churros

Low Protein:
- Tortilla Soup **
- Veggie Fajitas w/ Portobello mushrooms or CBF Meat*
- Tortillas**
- Mock Refried beans**
- Spanish Rice*
- Baja Salad
- Cookies and cake**

Beverages for both lunches: Coffee, Tea, Soft Drinks and Bottled Water

HCU Network America would like to thank Cambrooke Therapeutics (CBF) and Taste Connections for generously donating their products for meals and snacks for the conference. Food items that use Cambrooke products are denoted with * and Taste Connections are denoted with **

Conference Schedule

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Saturday, April 21

08:00 **Registration opens, Breakfast and Networking**

08:50 **Kid Zone Opens** - Kids must be dropped off and picked up by parents.
Location: Whittier Room
See page 12-13 for Kids Zone agenda and volunteer bios.

09:00 **Introduction and Meeting Expectations**
Presenter: Danae' Bartke, HCUNA Executive Director and Margie McGlynn, HCUNA President

09:30 **CBS Guidelines**
Presenter: Kimberly Chapman, MD, PhD, FAAP, FABIM
Children's National Hospital
Dr. Kimberly Chapman was a member of the E-HOD Guidelines Committee and was responsible for gathering the data from a review of the scientific literature that lead to the Guidelines for Diagnoses and Management of Cystathionine Beta-Synthase Deficiency that were recently published in the Journal of Metabolic Diseases.

10:30 **Intermission**—Please take this time and visit our vendors!

10:50 **Dietary Best Practices**
Presenter: Krista Viau, PhD, RDN CSP
Boston Children's Hospital
Dietary management is the key to success with homocystinuria. Krista Viau will guide us through the rationale for nutrition therapy, review different approaches to diet monitoring, and identify strategies to support success with a low methionine diet.

12:00 **Lunch & Medical Nutrition Reimbursement**
Presenter: Raenette Franco
Compassion Works Medical, LLC
Learn to keep up with insurance terminology and communicating with your health insurance carrier. Helpful tips for challenges with coverage for your medical foods and formula.

01:30 **Transitioning into Adulthood Panel**
Presenter: Next Steps Organization
Moderator Bill Kubicek will lead the panel through a discussion on various situations that our teenage metabolic patients face as they transition to high school and college. Some of the topics will include dating, college life and work. An open Q&A session will take place at the end of the panel.

03:15 **Intermission**—Please take this time and visit our vendors!

3:30 **HCU and the Brain**
Presenter: Benjamin D. Goodlett, PhD
Psychology Postdoctoral Fellow, Boston Children's Hospital
Join Benjamin Goodlett, PhD, for a discussion on the relationships between homocystinuria, emotions, and mental processes. The future of treatment is bright, but homocystinuria may leave individuals with an increased risk for subtle deficits in brain function. This will be an interactive discussion of what is known and not known. Importantly, this will also be a solution focused discussion of practical strategies for daily life.

See Flyer **Cambrooke Tour and Reception**
4 Copeland Drive | Ayer, MA | 01432
Please join us for an evening reception and tour of Cambrooke Therapeutics headquarters.

Sunday, April 22

08:00 **Registration opens, Breakfast and Networking**

08:00 **Orphan Technologies Breakout Session**
Location: Thoreau Room

08:50 **Kid Zone Opens** - Kids must be dropped off and picked up by parents.
Location: Whittier Room

09:00 **Vendor Acknowledgement**

09:15 **Natural History Study**
Presenter: Harvey Levy, MD
Boston Children's Hospital
Boston Children's is currently one of three sites within the US that is participating in the Natural History Study of CBS Deficiency Homocystinuria (CBSDH). Dr. Levy will speak on what the Natural History Study has already revealed and the important of Natural History Studies.

Sound drug development requires a comprehensive understanding of the disease being treated. To design reliable clinical studies and achieve meaningful outcome measures, researchers must apply known etiology and thorough knowledge of the disease's progression.

10:15 **Intermission**—Please take this time and visit our vendors!

10:30 **Research Map & Strategy**
Presenter: Margie McGlynn, HCUNA President
Over the past year, a project was sponsored by HCU Network America and HCU Network Australia to develop a global research map that describes the research underway for new therapies to treat homocystinuria, focusing on HCU caused by CBS Deficiency. A global research strategy was also developed and a Scientific Advisory Board was convened to define the top priorities for funding via a new global grants program. This presentation will summarize the findings from this project and the status of the global grants process.

11:30 **Enzyme Replacement Therapy**
Presenter: Patrick Horn, MD, PhD
Orphan Technologies
Orphan Technologies is dedicated to developing novel therapies to dramatically improve the lives of patients suffering from rare disorders. OT-58, our lead drug development candidate, has been optimized as an enzyme replacement therapy for classical homocystinuria, a genetic disease characterized by cardiovascular, skeletal, neurologic, and ophthalmologic complications. OT-58 is designed to reduce homocysteine levels via a targeted mechanism of action and may have therapeutic applications in other diseases. For more information, please visit www.orphantechnologies.com

12:30 **Lunch & Patient Panel: Keys to Success**
Presenter: Patient-Expert Panel
Our patient-expert panel is comprised of three patients and two experts who we feel are exemplary examples and leaders in the HCU community. Their experience and expertise make these patients and experts an invaluable asset to the HCU community.

01:45 **Moving Forward: Input from the HCU Community**
Led by: Danae' Bartke, HCUNA ED & Kristin Rapp, VP

2:30 **End of Conference—Thank you for Coming!**

Kids Zone Schedule

Saturday, April 21

09:00-09:30 Icebreaker Activities

09:30-10:00 Story about HCU

10:00-10:45 Board Games

10:45-11:45 Flower Pot Craft

11:45-12:00 Restroom Break

12:00-12:45 Lunch

12:45-01:00 Restroom Break

01:00-02:00 Boston Children's Dietitian Program

02:00-2:30 Movement Games/Activities

02:30-4:30 Movie: Inside Out

Sunday, April 22

09:00-09:45 Games and Activities

09:45-11:15 Movie: Trolls

11:15-12:15 Trolls Craft

12:15-12:30 Restroom Break

12:30-01:15 Lunch

01:15-01:30 Restroom Break

1:30-02:30 Create your own slime!

Only food and formula that parents provide will be given to children. All food and formula needs to be labeled with the child's name. If it requires being given at a specific time, please label the time to be given on the packaging.

Please note, there isn't a refrigerator in the Kids Zone room, but we can store things in a small cooler.

Lunch will be eaten in the Kids Zone room, but parents will help their children get their plates. If you have directions on how to handle the food not eaten, please speak with one of the children's program Team Leaders.

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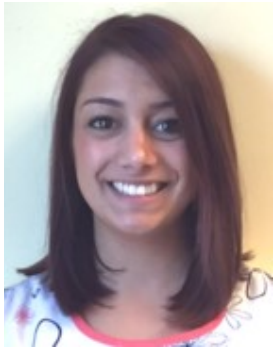
Kids Zone Team Leaders

Jaclyn is currently a nursing student at Mount Wachusett Community college. She will be graduating with her RN degree in June of this year. With her dream of becoming a pediatric nurse she carried out her love of working with children through her volunteering. Jaclyn is dedicated to her healthcare career and currently works at an Urgent Care. In her spare time she enjoys going to the gym, traveling, and participating in events in her community. Jaclyn is excited for the opportunity to be a part of the HCU conference. She looks forward to creating a fun and exciting experience for the children that will be participating in the program!



Laurie works as a medical assistant at Newton Wellesley Family Pediatrics and will be graduating from Mount Wachusett Community College with her RN this June. Laurie has been serving the community in healthcare for about 8 years and especially enjoys working with children. Her hopes are to work in a pediatric emergency department after graduating to share her positivity with the community. Laurie enjoys cooking, running, spending time with family and friends, and volunteering at her local church. Laurie is honored to be a part of the HCU conference and is excited to run a fun and educating children's program.

Mikeala is currently a nursing student at Mount Wachusett Community college, and will be graduating with her RN in June. With over six years of experience as a nanny, Mikeala's love for children has continued through her career in healthcare, as she aspires to be a NICU nurse. In her free time, she enjoys hiking, yoga, and volunteering in the community. Excited for the opportunity to be part of the HCU conference, Mikeala looks forward to running a kids program that will be fun for all ages!

HCU Hero Award

Harvey Levy, MD, Boston Children's

Following graduation from the Medical College of Georgia, Dr. Levy trained in pediatrics in Boston, New York, and Baltimore. Following pediatric training, Dr. Levy then completed a fellowship in metabolic disorders at the Massachusetts General

Hospital (MGH) under Dr. Mary Efron, a pioneer in metabolism. After completing this training, Dr. Levy joined the faculty of the MGH and Harvard and also served as Consultant in Metabolism to the Massachusetts Newborn Screening Program. In 1978, Dr. Levy moved to Boston Children's Hospital to found and serve as Director of the Metabolic Program, now the Biochemical Genetics section of the Division of Genetics.

Dr. Levy developed a specific interest in homocystinuria during his fellowship when he identified the first case diagnosed from newborn screening. Shortly thereafter he and Dr. Harvey Mudd discovered the first example of increased homocysteine due to a non-homocystinuric disorder, now known as cobalamin C defect. This was also the first example of a human disorder of vitamin B12 metabolism. Disorders of sulfur metabolism have continued to be a major interest of Dr. Levy throughout his career. He has authored over 400 medical articles and book chapters on metabolic disorders, including many on the homocystinemias, and has received a number of national and international awards for his achievements.

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Conference Speakers

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, United States. She specializes in taking care of all ages of individuals with defects of methylation, homocysteine and propionate metabolism.

She currently divides her time between clinical responsibilities, clinical research including several patient registries and other clinical trials, and a basic science laboratory which studies the Propionate pathway and Krebs cycle. She firmly believes that clinical care informs bench research and research enhances clinical care. Thus, with collaborators, she is exploring and designing novel therapeutics for a number of the metabolic disorders

Krista Viau, PhD, RD
Krista Viau is a metabolic dietitian at Boston Children's Hospital. She has been working with children and adults with inherited metabolic disorders for 9 years. Krista is a recent addition to the Boston Children's team. She moved with her fiancé after spending 8 years at the University of Utah Metabolic Clinic. Krista completed her Masters and Doctoral degrees at the University of Utah studying cognitive outcomes in PKU and strategies to improve treatment adherence. Her research interests include studying the natural history of rare metabolic disorders and improving nutritional therapies for these conditions

Raenette Franco, CBCS
A native New Yorker, Raenette Franco came to New Jersey in 2001 and landed a career as a Medical Biller Insurance Specialist/ Consultant. Inspired by helping others, Raenette expanded her career as founder of Compassion Works Medical. Raenette received her BA in Business Administration from Columbia University, NY and Certified Biller Coder Specialist (CBCS), NCCA Accreditation, NJ. Her big heart and deep passion is dedicated to helping patients, dietitians, and physicians avoid the difficult tasks of insurance coverage and reimbursement issues for medical foods/ enteral nutrition.

Next Step Organization
Our program for people living with rare genetic disorders helps these individuals develop life skills, find community and become self -advocates so they can move beyond their challenges to become strong, independent, productive adults. The program is co-designed by the young participants, health care professionals and Next Step to create a curriculum that addresses transitional issues and arms young people with tools, information and hope.

Benjamin D. Goodlett, PhD
Benjamin Goodlett earned his doctorate in clinical psychology from Wayne State University, and he is currently a postdoctoral fellow in the Metabolism Clinic, Division of Genetics and Genomics, at Boston Children's Hospital. He provides neuropsychological assessments to children and adolescents with metabolic disorders to monitor cognitive functioning, assist educational planning, and provide concrete recommendations for families. Ben also provides psychotherapy to children and adults impacted by metabolic disorders with a focus on family functioning, adherence to medically necessary behaviors, and finding an identity beyond diagnosis. His passion is helping families balance medical needs with everyday life.

Harvey Levy, MD
Dr. Levy developed a specific interest in Homocystinuria during his fellowship when he identified the first case diagnosed from newborn screening. Shortly thereafter he and Dr. Harvey Mudd discovered the first example of increased homocysteine due to a non-homocystinuric disorder, now known as cobalamin C defect. This was also the first example of a human disorder of vitamin B12 metabolism. Disorders of sulfur metabolism have continued to be a major interest of Dr. Levy throughout his career. He has authored over 400 medical articles and book chapters on metabolic disorders, including many on the homocystinemias, and has received a number of national and international awards for his achievements.

Margie McGlynn, HCUNA President
Margaret (Margie) McGlynn is President of the Board of HCU Network America, a patient advocacy organization she co-founded to provide support for patients and families affected by homocystinuria. She is also President of the Hempling Foundation for Homocystinuria Research, a fund she established to support research on new therapies for HCU in honor of her late sisters, Judy and Susie Hempling.

Patrick Horn, MD, PhD
Patrick Horn recently joined Orphan Therapeutics where he is leading the clinical development of an enzyme replacement therapy for the treatment of Homocystinuria. Pat received both his MD and PhD from the University of Chicago and was a practicing pediatrician for almost 20 years in Chicago before transitioning to drug development. He has held leadership positions at Abbott Laboratories, Dyax, and Tetraphase Pharmaceuticals before joining the clinical team at Orphan Technologies.

Patient–Expert Panel

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Moderator:

Margie McGlynn
Margaret (Margie) McGlynn is President of the Board of HCU Network America, a patient advocacy organization she co-founded to provide support for patients and families affected by homocystinuria. She is also President of the Hempling Foundation for Homocystinuria Research, a fund she established to support research on new therapies for HCU in honor of her late sisters, Judy and Susie Hempling.

Experts:

Kimberly Chapman, MD, PhD
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, United States. She specializes in taking care of all ages of individuals with defects of methylation, homocysteine and propionate metabolism. She is also is one of the authors on the Guidelines for Diagnoses and Management of Cystathionine Beta-Synthase.

Harvey Levy, MD
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Patients:

Pamela Penrose
After growing up with a diagnosis of Marfan's syndrome and a series of health issues including lens dislocation, blood clots and strokes at the age of 54 Pam was diagnosed with homocystinuria. After her diagnosis in 2011, Pam was started on a low protein diet, formula and betaine therapy. She has seen her levels go from the 400s to 20s where they remain. Pam is an exceptional example that the diet can be followed!

Kristin Rapp
Kristin was the first person in the state of Maryland to be diagnosed with HCU following the start of mandatory newborn screening. Because of her early diagnosis and great care from medical professionals and her parents, she has been able to lead a relatively normal life, free of medical issues. Kristin has been a great ambassador for HCU and raised thousands of dollars by running marathons to raise awareness and funds for HCU.

Sarah Sullivan
Sarah Sullivan is the mom to Colbie and Cayle, two patients who despite having newborn screening for HCU were not diagnosed until they were 3 and 1 years old. Their diagnosis came as a shock after Colbie, at the age of 3, suffered a major blood clot to the brain. Stories like the Sullivan's are a reminder of the work we still have to do on newborn screening.

Sponsor Information

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Recordati Rare Diseases is a biopharmaceutical company committed to providing often overlooked orphan therapies to the underserved rare disease communities of the United States. Our experienced team works side-by-side with rare disease communities to increase awareness, improve diagnosis, and expand availability of treatments. For more information, please visit www.recordatirarediseases.com

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Orphan Technologies is committed to reduce the burden of patients suffering from homocystinuria. OT-58, our lead drug development candidate, has been optimized as an enzyme replacement therapy for classical homocystinuria, a genetic disease characterized by cardiovascular, skeletal, neurologic, and ophthalmologic complications. OT-58 is designed to reduce homocysteine levels via a targeted mechanism of action and may have therapeutic applications in other diseases. To learn more, visit www.orphantechnologies.com/ot-58/

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Inspiring Futures

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 more out about our products please visit www.medicalfood.com

CAMBROOKE THERAPEUTICS

Founded in 2000, Cambrooke Therapeutics recognizes the critical nature of managing serious medical disorders through medically based nutrition therapies. For people with serious unmet medical nutrition needs, it is a daily challenge to find flavorful, easy options that can sustain their diets and maintain their nutritional needs. To check out our line of low protein foods and methionine free formula, please visit www.Cambrooke.com

Taste Connections, LLC

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. In the near future, we will be adding several new low-protein products for your convenience.

Visit Tasteconnections.com tview our low protein items and mixes.

Vitaflo®
Innovation in Nutrition

Vitaflo® is at the forefront of developing innovate specialized foods for metabolic disorders, pediatric renal disease, ketogenic diet, and other areas of disease specific nutrition. Our aim is to create nutritional products that combine the best of cutting-edge research with the lifestyle demands of modern living, ensuring the most acceptable products are available for the patient. By constantly evolving to meet patient needs, Vitaflo will continue to develop products that offer patients choice and help support them in complying with restrictive therapeutic diets. For more information, visit www.nestlehealthscience.us/vitaflo-usa

NECPAD

NECPAD was created in the early 1990s to meet the need of several families in the New England area who were starved for information on PKU and other metabolic disorders. At the time, the Internet was not around as a source of information as it is today.

A small support group was started by two parents getting together to talk about their children's conditions, then it became three parents, and then four and so on. These informal get-togethers continued to grow and it didn't take long to realize there was a significant need for an organization to provide support and information to families and individuals with these conditions in New England.

In 1995, the official New England Connection for PKU and Allied Disorders was established and became a 501(c)(3) non-profit organization. And the rest is history... so Welcome to the Family. To find out more information, visit www.necpad.org

Dear Friends,

I would like to thank you again for attending our first conference and for your support in *Taking the Lead for HCU*. As I reflect back on the past year and a half since HCU Network America was founded, I'm blown away at the progress our organization and community has made. In the short time, we have paved the way by:

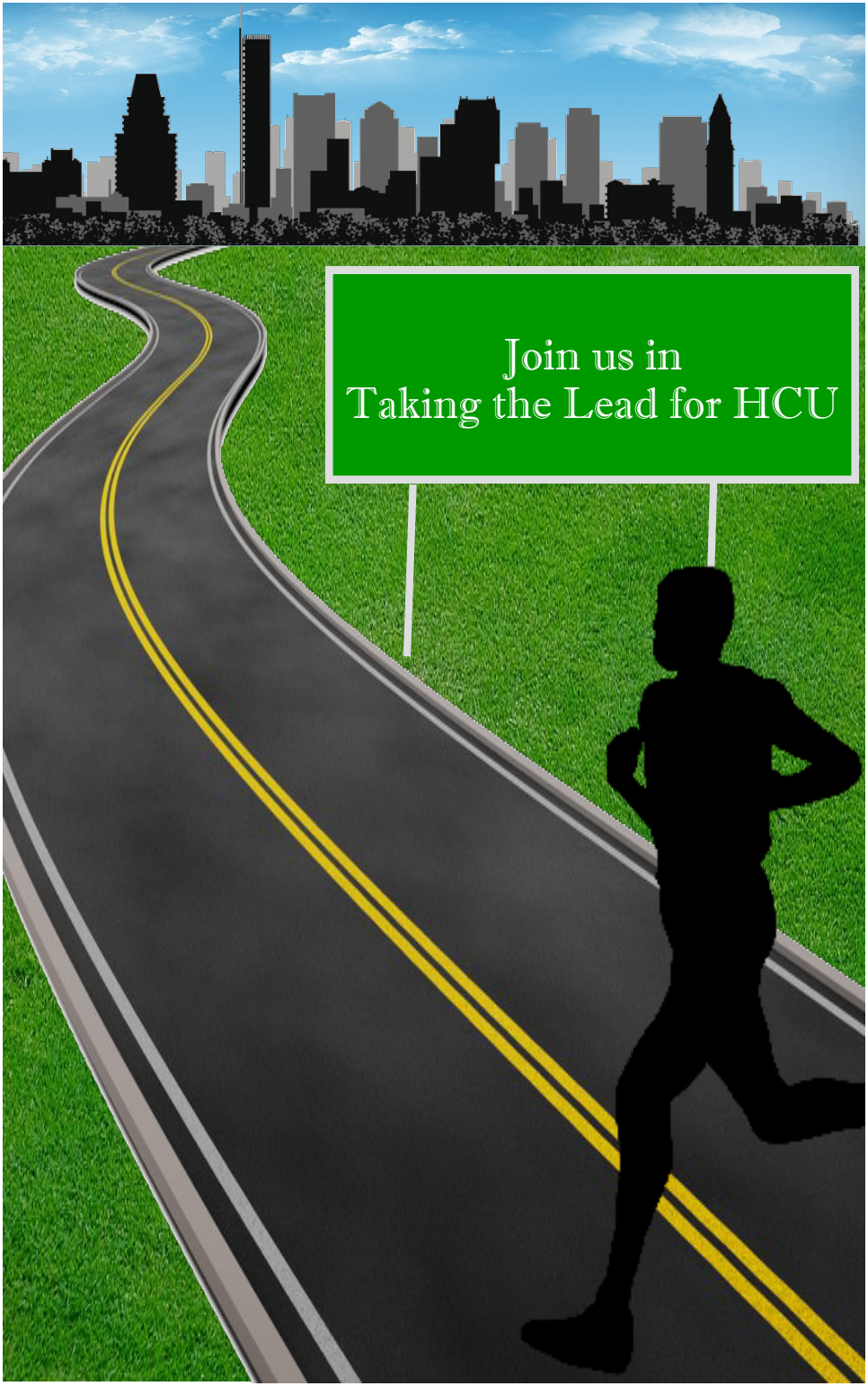
- Developing a robust user friendly website full of resources for patients and clinics. Some of our website resources include: a HCU checklist, new-patient toolkit, find a clinic feature, contact register, food and formula vendor list, patient stories, as well as a rotating news banner.
- Sent introductory clinics packets to over 100 clinics and have had over 70 follow-up conference calls with those clinics.
- Collaborated with industry to find ways to better the HCU Community. This next year you will see resources and surveys that we have worked together on.
- Engaged patients in various ways including our monthly E-Newsletter, social media pages and HCU Awareness Month. In just the past 6 months, we have seen our following more than double!
- Hired a Reimbursement Specialist to assist patients and families one-on-one within low-protein food, formula and supplement coverage.
- Spread awareness for Homocystinuria. In 2017 we attended "Disorder: The Rare Disease Film Festival" in Cambridge, Massachusetts, attended a low protein workshop "Cooking with Cambrooke" that highlighted the similarities between HCU and other metabolic disorders and launched the first HCU Awareness Month.
- Organized our first patient-expert meeting!

Through our efforts, so far we have brought together patients, families, clinics and industry. These are just the initial strides of many to come on our journey. The first few steps have helped define our organization and communities needs for those who can drive new therapies, treatments and a cure. In order to further the path, we need to know we can depend upon each other to continue the pace; this means joining "our team" and helping take the lead for HCU.

"The race does not always go to the swift, but to those who keep running" –Anonymous

With gratitude and hope,

Danae' Bartke, Executive Director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