



2024 CCHS NETWORK FAMILY CONFERENCE

WELCOME

Where: Newport Beach, CA

When: June 26 – 28, 2024

Venue: Renaissance Newport Beach Hotel

This conference offers more than an educational agenda. It celebrates the collective journey that is unique to CCHS families while renewing friendships, celebrating accomplishments and emphasizing the importance of empowerment and advocacy. By blending attendance with clinical specialists and research experts, we have created a space where knowledge is shared, connections are formed and hope is fortified. We also recognize the significance of 2024 for our rare disease community; marking the 35th anniversary of the CCHS Network, our 9th family conference and our 10th International CCHS Day in September.

We are so glad you are here!

A VERY BIG THANK YOU to everyone who has provided support for this conference: Avery BioMedical, KeepMeBreathing, and donations from the CCHS community.

PROGRAM

Day/Time	Speaker	Topic
Wednesday, June 26	Registration & Welcome Reception	
1:00 – 3:00p	Registration	
3:00 – 3:30p	Melinda Riccitelli, President, CCHS Network	Welcome
3:30 – 4:10p	Thomas Keens, M.D., Childrens' Hospital of Los Angeles	Keynote Speaker: From Discovery to Advancements: Tracing the History of Congenital Central Hypoventilation Syndrome
4:15 – 5:00p	CCHS Group Activity: Let the Games Begin	
5:00 – 6:30p	Reception (Finger Foods)	
Thursday, June 27	CCHS Clinic Pathways: Empowering Health and Wellness	
7:30 – 9:00a	Continental Breakfast	
8:30a – 5:00p	Daycare	
9:00 – 9:40a	Gad Vatine, Ph.D., Ben Gurion University of the Negev	CCHS Molecular Research: The Big Picture
9:45 -10:25a	Debra Weese-Mayer, M.D., Lurie Children's Hospital	Natural History Studies and Registries in the Development of Rare Disease Treatments
10:25 – 10:45a	Break	
10:45 – 11:25a	Jack Feldman, Ph.D., UCLA	Every Breath You Take
11:30a – 12:10p	Ajay Kasi, M.D., Childrens' Healthcare of Atlanta	Vents, Stomas & Masks Oh My
12:15 – 1:30p	Lunch Sponsored by Avery Biomedical	
1:30 – 2:10p	Iris Perez, M.D., Childrens' Hospital Los Angeles	CCHS Chronicles: More Than Just Breathing
2:15 – 3:05p	James Oakley, CEO, KMB Xenia Proton de la Chapelle, CEO, AtmosR Tony Martins, COO & President, Avery Biomedical	Cutting-edge Technologies and Emerging Treatments
3:05 – 3:15p	Break	
3:15 – 4:45p	Breakout Sessions	
6:00 – 7:30p	Dinner	
Friday, June 28	CCHS Patient Focus: Living Well, Living Strong	
7:30 – 9:00a	Continental Breakfast	
8:30a – 12:30p	Daycare	
9:00 – 9:40a	Maida Chen, M.D., Seattle Childrens' Hospital	CCHS Survival Guide: Transitioning to Adult Care – The Fun Facts You Need
9:45 – 10:25a	Jeremy Orr, M.D., UCSD Health	Adulting with CCHS: Navigating Your Path Forward
10:25 – 10:40a	Break	
10:40 – 11:20a	Liya Rabkina, MS, CGC, Igenomix	Navigating Fertility in CCHS
11:25a – 12:25p	Individual Patient Stories; Patient Panel	
12:25p	Closing remarks	

SPEAKER BIOS AND TALK SUMMARIES

Dr. Maida Chen, M.D. is Director of the Pediatric Sleep Center at Seattle Children's Hospital. She is also an Associate Professor in the Department of Pediatrics at the University of Washington School of Medicine. She obtained her medical degree at Northwestern University, did her residency in Pediatrics with DR. Weese-Mayer at Rush-Presbyterian-St. Luke's Medical Center, and then completed a Pediatric Pulmonary Fellowship with a special focus on respiratory control and sleep medicine with Dr. Keens at Children's Hospital Los Angeles. Her clinical practice centers on sleep disorders in infants, children, and adolescents. Her research focuses on respiratory control disorders and sleep-disordered breathing in special needs populations, like CCHS. She is a member of the American Academy of Pediatrics, American Thoracic Society, American Academy of Sleep Medicine and Sleep Research Society. Dr. Chen also serves as a board member on the CCHS Network Research Advisory Board, a panel that reviews CCHS research grant applications for funding.

Title: *CCHS Survival Guide: Transitioning to Adult Care, The Fun Facts You Need*

Transitioning care for individuals with CCHS from childhood to adulthood involves coordinated efforts to address changing medical, social, and psychological needs. This includes ensuring continued respiratory support, monitoring for associated health issues, and facilitating independence in managing the condition. The transition process also emphasizes education about the syndrome, fostering self-advocacy, and connecting patients with adult healthcare providers experienced in CCHS. Comprehensive planning and support are crucial to ensure a smooth transition and maintain optimal health outcomes.

Jack Feldman, Ph.D. is a renowned breathing researcher at the University of California, Los Angeles (UCLA). With a focus on unraveling the intricacies of respiratory physiology, Dr. Feldman has become a leading figure in the field of breathing science. His significant research contributions include the discovery and exploration of the Pre-Bötzinger Complex, a critical neural center in the brainstem that plays a pivotal role in respiratory rhythm generation. This groundbreaking discovery has provided crucial insights into the neural mechanisms governing breathing patterns and respiratory control. Driven by a profound dedication to respiratory research, Dr. Feldman continues to advance our understanding of the respiratory system, making lasting contributions to scientific knowledge.

Title: *Every Breath You Take*

Understanding the neural mechanisms underlying dysfunctions of breathing is a prerequisite for developing and optimizing therapies. Our current understanding of the etiology of CCHS followed from identification of the critical sites for generation of breathing movements, particularly as related to generation of breathing rhythm and pattern, and in central chemoreception for carbon dioxide. I will discuss our current knowledge of the basic mechanisms underlying generation breathing pattern, including the discoveries of the past 30 years that underlie our current understanding of CCHS.

Dr. Ajay Kasi, M.D. is a pediatric pulmonologist and assistant professor of pediatrics at Emory University and Children's Healthcare of Atlanta. He completed pediatric residency in Nicklaus Children's Hospital, Miami, FL and pediatric pulmonology fellowship in Children's Hospital Los Angeles. Since starting as a pediatric pulmonologist in Atlanta, Dr. Kasi helped establish the Diaphragm Pacing Program at Children's Healthcare of Atlanta. This unique program is specifically designed for children with CCHS and is one of only a few of its kind offered in the country. His research focuses on ventilatory management strategies in CCHS and cystic fibrosis early lung disease. He has published on clinical aspects of caring for children with CCHS. Dr. Kasi is board certified in pediatrics and pediatric pulmonology and is a member of American Thoracic Society, American Academy of Pediatrics, and American Academy of Sleep Medicine. Dr. Kasi also serves on the CCHS Network Research Advisory Board, a panel that reviews CCHS research grant applications for funding.

Title: *Vents, Stomas and Masks, Oh My!*

In this presentation, different ventilatory options available for children and adults with CCHS – positive pressure ventilation via tracheostomy, mask ventilation, and diaphragm pacing – will be discussed. The pros and cons of each type of ventilatory modality will be reviewed. Pathways for tracheostomy decannulation to mask ventilation and diaphragm pacing and subsequent closure of tracheostomy stoma will be discussed.

Dr. Thomas Keens, M.D. has recently retired after 40 years as a Pediatric Pulmonologist at Children's Hospital Los Angeles (CHLA) and a Professor of Pediatrics at the Keck School of Medicine at the University of Southern California. Dr. Keens is among a handful of experts in the world experienced in treating children with CCHS. He has authored or co-authored well-over 50 articles about CCHS. Dr. Keens research efforts was in the area of Cystic fibrosis (CF), Sudden infant death syndrome (SIDS), CCHS, Pediatric respiratory physiology and home mechanical ventilation. Dr. Keens is Board certified in Pediatrics, Neonatal-Perinatal Medicine, and Pediatric Pulmonology. He is a member of the American Physiological Society; Society for Pediatric Research; American Pediatric Society; American Academy of Pediatrics; American Thoracic Society; American College of Chest Physicians; Sleep Research Society; and the International Society for the Study and Prevention of Infant Deaths. In 2019, Dr. Keens was awarded the most prestigious "Pediatric Founders Award" by the Pediatric Scientific Assembly of the American Thoracic Society for his seminal contributions to the science and practice of pediatric respiratory medicine. Dr. Keens serves as an Advisory Board Member for the CCHS Network One World Registry.

Title: *From Discovery to Advancements: Tracing the History of Congenital Central Hypoventilation Syndrome*

Dr. Keens will trace the evolution of Congenital Central Hypoventilation Syndrome (CCHS) from its earliest observations to current molecular advancements. His presentation aims to deepen understanding, celebrate scientific achievements, and inspire further progress. He examines the initial perplexity and challenges faced by medical professionals, highlights key breakthroughs, and recognizes the contributions of clinicians and researchers. Personal stories of resilience from individuals with CCHS underscore the importance of ongoing research, education, and support networks. Together, we honor the past, embrace the present, and strive for a brighter future for those affected by CCHS.

Tony Martins, with over three decades of experience in the field of breathing pacemaker technology, serves as the Chief Operating Officer and President of Avery Biomedical. Mr. Martin's expertise has been instrumental in the development and advancement of the Avery Diaphragm Pacing System, a pioneering solution in the realm of implanted phrenic nerve and diaphragm stimulation. The Avery Diaphragm Pacing System comprises surgically implanted electrodes and receivers, complemented by an external transmitter and antennas. This innovative system offers a transformative approach to addressing respiratory issues, providing patients with enhanced control over their breathing function. With a track record of more than 2,000 successful patient implants, Avery Biomedical has established itself as a beacon of safety and reliability in the medical device industry.

Title: *Cutting-edge Technologies and Emerging Treatments*

Mr. Martins will discuss the current use of Avery Diaphragm Pacing System in CCHS patients and possible future advancements in the technology.

James Oakley, CEO, KeepMeBreathing a lawyer and astute businessman, shares a common bond with the CCHS community as a devoted parent. Inspired by the challenges faced by his son, Casper, who battles with CCHS, Mr. Oakley decided to channel his energy and expertise into making a tangible difference. In response to the critical need for enhanced ventilatory equipment for CCHS patients, James founded a nonprofit technology company, KeepMeBreathing (KMB). Mr. Oakley's dual role as a dedicated parent and visionary entrepreneur showcases the power of personal experiences in driving positive change within the CCHS community and highlights the potential for collaboration between the business world and healthcare to bring about meaningful advancements for those living with rare respiratory conditions.

Title: *Cutting-edge Technologies and Emerging Treatments*

Mr. Oakley will discuss the critical role of innovative technologies in addressing the unmet medical needs of CCHS patients.

Jeremy Orr, M.D., is a board-certified pulmonary and critical care physician who specializes in hypoventilation disorders, neuromuscular respiratory weakness, chronic lung disease, non-invasive ventilation management and critical care medicine. Dr. Orr's research is focused on sleep apnea, particularly in patients with chronic lung and heart disease. He has published articles in the American Journal of Respiratory and Critical Care Medicine, American Journal of Cardiology, Sleep, The Lancet Respiratory Medicine, Journal of Clinical Sleep Medicine and Annals of the American Thoracic Society.

Title: *Adulting with CCHS: Navigating the Path Forward*

For patients with childhood-onset conditions, reaching adulthood comes with new challenges. I will provide an adult pulmonologist's perspective on the transition process, aspects of adult focused health systems that might differ from pediatrics, and strategies to assure that you get the right care.

Iris Perez, M.D. succeeded Dr. Thomas Keens as Director of the Congenital Central Hypoventilation Syndrome and Diaphragm Pacing Program at Children's Hospital Los Angeles. Dr. Perez is recognized for her exceptional care and expertise in managing patients with Congenital Central Hypoventilation Syndrome (CCHS). Dr. Perez's commitment to the well-being of children affected by CCHS is evident in her specialized approach to diagnosis, treatment, and ongoing care for these individuals. Her dedication to advancing both clinical and research aspects of CCHS has positioned her as a leading figure in the field of pediatric pulmonology, making a significant impact on the lives of patients and contributing to the broader understanding of this rare respiratory disorder. Under her leadership, CHLA continues to provide compassionate and cutting-edge care for children with CCHS.

Title: *CCHS Chronicles: More Than Just Breathing*

Patients with CCHS can have a spectrum of autonomic nervous system dysfunction. This is generally determined by their PHOX2B genotype. This session will review the nonrespiratory manifestations of CCHS and provide an update on their surveillance and management.

Xenia Proton de la Chapelle, CEO, AtmosR, a BioPharma company, shares a profound connection with the CCHS community as a fellow parent of a child with CCHS. In her dual role as a business leader and dedicated parent, Xenia brings a unique perspective to her work, driven by personal experiences with her child, Joany. Under her leadership, AtmosR focuses on advancing biopharmaceutical solutions in CCHS, with a keen and heartfelt understanding of the pressing needs faced by individuals like Joany. Xenia's commitment to both her professional endeavors and the CCHS community exemplifies the potential for meaningful contributions at the intersection of business and healthcare, with a focus on improving the lives of those affected by rare respiratory conditions.

Title: *Cutting-edge Technologies and Emerging Treatments*

The mission of AtmosR is to develop innovative treatments for severe neurological disorders with high unmet medical needs. In her talk, Ms. Proton de la Chapelle will focus on the company's advancements in drug development for Congenital Central Hypoventilation Syndrome (CCHS). She will highlight the progress and challenges in creating effective treatments for this rare and debilitating condition, emphasizing AtmosR's commitment to addressing critical gaps in medical care for CCHS patients.

Liya Rabkina, MS, CGC is a licensed and board-certified genetic counselor at Igenomix USA, a company specializing in reproductive genetics. Ms. Rabkina assists individuals and families in navigating pre-implantation genetic testing (testing embryos for inherited single-gene conditions and chromosomal abnormalities). She is passionate about helping families understand their reproductive genetic options, empowering them with the knowledge to make informed decisions regarding their reproductive choices.

Title: *Navigating Fertility in CCHS:*

Families affected by CCHS often seek guidance on the likelihood of passing the condition to future offspring and the available reproductive options. This talk will offer a brief overview of the inheritance patterns associated with Congenital Central Hypoventilation Syndrome (CCHS). It will explore the probabilities of CCHS recurrence in offspring and outline various genetic testing and family planning strategies that can be employed both pre-conception and prenatally to mitigate these risks.

Dr. Gad Vatine, Ph.D. is a Professor and Researcher in the Physiology and Cell Biology Department and the Regenerative Medicine and Stem Cell (RMSC) Research Center at Ben-Gurion University of the Negev. Dr. Vatine is at the forefront of CCHS cellular and molecular research, and his bioengineered iPSC platforms give real hope in uncovering the intricacies of CCHS. Dr. Vatine received his Ph.D. in 2011 from Tel Aviv University where he studied the mechanisms underlying light-entrainment of the circadian clock using zebrafish as a model. He followed this with a short postdoc at Bar-Ilan University where he established a zebrafish model for the rare psychomotor disability disorder MCT8-deficiency. Next, he joined the laboratory of Dr. Clive Svendsen at the Regenerative Medicine Institute at Cedars-Sinai where he pioneered the concept of disease modeling at blood-brain barrier (BBB) using patient-specific induced pluripotent stem cells (iPSCs). In 2017, he became an assistant professor at The Physiology and Cell Biology Department and the Regenerative Medicine and Stem Cell Research Center at Ben-Gurion University of the Negev. The Vatine lab generates personalized iPSC-based models to study mechanisms underlying various rare neurological disorders, including CCHS, and to test potential treatments in these conditions. He is also the director of the BGU-iPS-core facility. The CCHS Network has wholeheartedly supported the research endeavors led by Dr. Vatine over the past few years, alongside his collaborative efforts with Dr. Ashkenazi.

Title: *CCHS Molecular Research: The Big Picture*

Current molecular research on CCHS primarily focuses on PHOX2B gene and protein research, investigating the molecular mechanisms by which PHOX2b mutations disrupt normal functioning, and examining potential molecular therapies to correct or compensate for the defective PHOX2B function. An overview of these topics will be discussed.

Debra Weese-Mayer, M.D. is the Beatrice Cummings Mayer Professor in Pediatric Autonomic Medicine at Northwestern University Feinberg School of Medicine and Chief of the first Center for Autonomic Medicine in Pediatrics (CAMP) at Lurie Children's Hospital. Dr. Weese-Mayer is recognized internationally for her clinical and research work in the emerging discipline of pediatric autonomic medicine and is a lead translational researcher at Ann & Robert H. Lurie Children's Hospital and the Stanley Manne Children's Research Institute. Dr. Weese-Mayer's notable work includes her involvement in the discovery and understanding of the PHOX2B gene's role in CCHS. As a key figure in unraveling the genetic underpinnings of this rare disorder, she has played a crucial role in advancing diagnostic and therapeutic approaches for patients with CCHS. She has authored more than 250 peer-reviewed articles, reviews, chapters, and abstracts. Her clinical and research programs have grown to include a wide-ranging referral base (48 states; 50 foreign countries). Dr. Weese-Mayer was awarded the March of Dimes Jonas Salk Health Leadership Award in Research for 2005. Dr. Weese-Mayer is board Certified in Neonatal-Perinatal Medicine and General Pediatrics and is a member of the American Autonomic Society Statement on Autonomic Medicine in Pediatrics, American Thoracic Society, CJ Foundation for SIDS Scientific Advisory Board, RADICA-FRE Scientific Advisory Board, Society for Pediatric Research. Dr. Weese-Mayer's commitment to bridging clinical care with groundbreaking research underscores her dedication to improving the lives of individuals affected by rare respiratory conditions at Lurie Children's Hospital and beyond.

Title: *Natural History Studies and Registries in the Development of Rare Disease Treatments*
Congenital central hypoventilation syndrome (CCHS), a severe neurocristopathy caused by PHOX2B gene mutations, leads to profound hypoventilation and autonomic nervous system dysregulation (ANS), necessitating lifelong artificial ventilation. Patients face significant neurocognitive risks due to repetitive hypoxemic and hypercarbic exposure, along with autonomic cardiovascular dysfunction that often necessitates a cardiac pacemaker. Additionally, CCHS is linked to a high risk of Hirschsprung's disease and neural crest tumors, presenting with substantial variability in disease severity and lacking consensus on optimal management. Current treatments are invasive, burdensome, and palliative, with no approved pharmacologic interventions despite promising pre-clinical models and anecdotal off-label drug successes. Comprehensive understanding of disease progression and the impact on quality of life remains limited. To address these gaps, we have assembled a team of leading CCHS experts, international centers of excellence, patient advocacy groups, and industry partners. Together, this team has identified candidate measures that reflect the core CCHS phenotype and is collecting validation data through extensive, longitudinal NH (Natural History) data using rigorously defined protocols, common data elements, and established measures reflecting patient and caregiver voices. By consolidating diverse data sources, expertise, and resources into an adaptable data hub of core measures of CCHS, this collaboration aims to enrich knowledge of CCHS, establish measures correlating with disease severity, and accelerate progress toward approved therapeutics in this underserved population, making this effort both timely and critical.