CAMK2 Therapeutics Network

Community Newsletter

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Latest News

1st CAMK2 Research and Clinical Summit

Collabor(ACTION): Building Bridges for CAMK2

We extend our heartfelt gratitude to the entire CAMK2 Community for the resounding success of our inaugural international scientific summit, held on November 4th and 5th in Washington, D.C. As the parent board of the CAMK2 Therapeutics Network, we were deeply moved by the collective dedication evident throughout the event, aimed at supporting our children.

The summit brought together distinguished CAMK2 thought leaders worldwide, including esteemed basic scientists and committed physicians. This two-day conference focused on knowledge exchange and collaborative planning to address challenges faced by patients with CAMK2 gene disorders. Participants represented diverse global locations such as the Netherlands, Italy, Ireland, and various U.S. states (Massachusetts, California, Texas, and Washington, D.C.). We were honoured to welcome virtual participants from Israel and Portugal, underscoring the global nature of our collaborative efforts.

A big thanks to Gabriela Danelluto, another member of CAMK2 Community, that made all the amazing designs of the banner and folders used and to Lisa Dwyer and King and Spalding Law Firm for the beautiful venue.

Dr. Howard Schulman, PhD, known as the "Father of CAMKII," delivered the opening address, outlining the structure and function of the CAMKII protein and its role in cell signalling. Dr. Schulman co-discovered the kinase now referred to as CAMKII with Nobel Laureate Paul Greegard. At Stanford University School of Medicine, as professor of Neurobiology and of Pharmacology, and chair of the Department of Neurobiology, he has been a key contributor to identifying how various mutations alter CAMKII activity. Even in retirement, he continues to inspire future generations of scientists and remains an influential figure in the world of molecular biology and cellular signalling. His insights set the stage for a series of impactful presentations by key researchers.

Dr. Geeske van Woerden, PhD, leading the basic science branch of the CAMK2 Encore center of expertise at the Erasmus Medical Center in the Netherlands, shared her groundbreaking work on mice born without CAMKII alpha protein. Her findings suggested that reinstating the protein later in life could reverse symptoms, offering hope for children with loss-of-function mutations. In addition to mouse models, her work includes the production of Inducible Pluripotent Stem Cells (iPSCs) to study the effect of CAMK2 mutations at a cellular level.

These iPSC are cells that have been derived from skin or blood cells from kids with CAMK2 genetic mutations. These cells have been reprogrammed back into an embryonic-like state that enables the development of an unlimited source of any type of human cell needed for therapeutic or research purposes. In this case, the iPSCs have been reprogrammed to develop into nerve cells mimicking the cells in the brains of our kids. This is an important first step in identifying how the CAMK2 mutations change nerve cell function in our kids' brains, and can be used to study which medication might help to “normalize” the function of those cells. This work complements Dr. Compagnucci’s work.

Dr. Claudia Compagnucci, PhD from Italy then presented her findings. Her specialty is developing iPSC neuronal cells to study rare genetic diseases associated with neurodevelopmental disorders. She is the first to receive a CAMK2 Seed Grant from Telethon, which was co-sponsored both by UNICI (A CAMK2 parent led foundation in Italy) and by the CAMK2 Therapeutics Network (our own foundation).

Dr. Danielle Veenma, MD PhD, leading the clinical part of the CAMK2 Encore program at Erasmus Medical Center, highlighted the CAMK2 Natural History Study, correlating specific mutations with symptoms. She presented her findings in children with CAMK2B P139L mutation, the most common mutation. We look forward to her upcoming publication, the first publication to define this syndrome. This work is important because we need to understand the symptoms caused by the mutations so we can understand how to target future treatments and cures

Dr. Margaret Stratton, PhD, from the University of Massachusetts Amherst, discussed the molecular structure of CAMKII and how gene mutations affect its activity. She helped us further understand the structure and ways we might try to “normalize” abnormal CAMKII activity with designer molecules (designer drugs).

Dr. Kimberley Goodspeed MD, is new to our team. She is a child neurologist and neurodevelopmental specialist who cares for children and young adults with autism spectrum disorders and intellectual disability at the University of Texas, Southwestern. She has particular expertise in rare genetic developmental disorders in children, including CAMK2 gene related disease. The goal of her talk was to promote a clinical and scientific collaboration with the ENCORE CAMK2 program.

Dr. Bonnie Dwyer MD, a CAMK2 parent and co-founder of the CAMK2 Therapeutics Network, concluded the clinical segment, emphasizing the debilitating symptoms and advocating for empiric treatment.

Dr. Wei-Liang Chen MD, an epileptologist from National Children's Hospital who is currently caring for a child with CAMK2 related disease also joined us. CAMK2 parent board members, Matt Gibbs, Nev Ross, Sarah Pace, Liat Vaknin Nisan, Ines Bastos, and Ian Carroll MD were there as parent advocates and were important contributors to the discussion.

Key collaborations emerged, including: 1. A partnership between Dr. Veenma and Dr. Goodspeed to gather data for the Natural History Study, 2. An ongoing collaboration between Dr. van Woerden and Dr. Stratton to investigate how specific CAMK2 gene mutations impact the activity of the CAMKII protein, 3. A collaboration between Dr. van Woerden and Dr. Compagnucci to deepen our understanding of how altered CAMKII function influences neuronal signalling in iPSCs, and 4. A joint effort involving Dr. van Woerden, Dr. Compagnucci, and Dr. Dwyer to leverage iPSC assays for testing specific drugs through drug repurposing. Remarkably, Dr. Compagnucci submitted a grant proposal, which included this collaborative effort, just four days after the summit.

Join us in applauding this collaborative effort, and consider supporting our cause.

Meet the Team Here

back row left to right: Dr Howard Schulman, Dr. Ian Carroll, Nev Ross, Dr. Geeske Van Woerden, Matt Gibbs. Front row left to right: Dr Claudia Compagnucci, Dr Danielle Veenma, Dr Bonnie Dwyer, Sarah Pace, Dr. Kimberley Goodspeed, and Dr Margaret (Meg) Stratton. Dr. Wei-Liang Chen also joined us in person but is not pictured. We also had Liat Vatkin (Israel), Inês Bastos (Portugal) and Dr Ronan Cohen (Israel) join us virtually.