CAMK2 Therapeutics Network

Community Newsletter

#1 | October 2022

Who are we?

CAMK2 Therapeutics Network is a family-led, scientifically supported international collaboration working to improve the lives of individuals with CAMK2 genetic mutations.The rare gene mutations result in severe neurodevelopmental disorders that affect our loved ones and pose extreme challenges for family life.

We aim to nurture a strong CAMK2 community and, in partnership with our medical and scientific colleagues, to work toward developing near-term treatments by repurposing medications. Our long-term goal is to use precision genetics to develop a cure.

See CAMK2.org. Direct questions to info@camk2.org.

Latest News

We launched our website CAMK2.org (May 2022)!

We created this website to help organize highly complex information about CAMK2 for families as well as for clinicians and researchers. Use as much of this information as you can, but don't be overwhelmed — your understanding of your child's or loved one's condition will be a journey.

As you explore the website, start with **Parents Primer**. This page explains the terms, like CAMK2 gene versus CAMKII protein (yes! We use CAMK2 with the Arabic numeral 2 for the gene, to distinguish it from the protein CAMKII which uses the Roman number II).

Also on the website, we highlight physicians who understand this disease and may be able to help. On the website, see **Meet The Team** under Scientific Advisory. **In the US**, Dr. Brunstrom-Hernandez (pediatric neurology, Texas), Dr. Sherr (pediatric neurology, San Francisco), and Dr. Chang (pediatric psychiatry, San Franciso Bay Area) are available to help. Due to US medical licensing, however, there may be some geographic limitations. **In Europe**, Dr. Danielle Veenma at CAMK2 Encore Center of Expertise and her multi-disciplinary team is also available for virtual or in person consults (camk2disorders@erasmusmc.nl).

We have launched our first fundraising campaign and raised almost $50,000 US dollars (as of 7/31/22)!

We made our first donation!

Jointly with UNICI (a parent led CAMK2 organization in Italy), we have sponsored a seed grant to Fondazione Telethon, a biomedical charity whose mission is to advance science toward the cure for human genetic disease. They have just announced the grant will go to an Italian lab to **speed the development of iPSC cells**. iPSC cells are cell cultures derived from CAMK2 affected individuals. These cultures will allow scientists to study the effect of each mutation on cell function and to screen for medications which may be able to normalize cell function.

We have two new members of our parent board! Please welcome Inês Bastos and Sarah Pace!

Donation Status

Total Raised: **$49,650.18 USD** (as of 7/31/22)

First Goal: **$200,000 USD** to support research for drug repurposing (See Research Initiatives)

Help Us With Our Goal!

Donate Now

Stay Tuned

We will be planning a **virtual forum for families and friends with the parent board of CAMK2 Therapeutics Network.** This Forum will include a small presentation regarding Network goals and will provide an opportunity for questions and answers. This will be scheduled in November 2022.

We are planning the **Fourth Annual International Meeting for CAMK2 in 2023.** This can be attended virtually or in person and is designed for family, friends, doctors and scientists. Formal presentations will be made.

Current Research Initiatives

The **current research goals of CAMK2 Therapeutic Network** are to:

1. Work toward finding current FDA approved drugs to lessen symptoms our children are experiencing (termed drug repurposing), and
2. Work toward genetic therapies both to lessen symptoms and with a goal toward cure.

Primary goal: Support the science of drug repurposing for CAMK2 disorders such that clinical trials are available 2 years after funding. Our estimate is that we need $150,000 to $200,000 US dollars to support this effort. Without our funding, the investigations cannot be done although the expertise is available.

The science of drug repurposing will involve 1. Understanding our children's symptoms and which ones we most need to improve (likely seizure, agitation, constipation), 2. Cell culture with iPSC (back engineered cells derived from the blood of affected persons) or other cells to better understand the cellular effect of different CAMK2 mutations, and 3. Possibly animal models to assess effects of different CAMK2 mutations with regard to biologic processes, such as seizures, agitation, and bowel motility. Once we have cell models and/or animal models, we can start screening medications that are already FDA approved to see which ones may be able to normalize function. Once we have this information, clinical trials can be designed. With drug repurposing, drugs found to help one child with a CAMK2 gain of function mutation will likely be generalizable to help all kids with CAMK2A, B, G, and D gain of function mutations. Likewise, medications to help a child with one CAMK2 loss of function mutation will likely be generalizable to help all kids with CAMK2A, B G. and D loss of function mutations

Secondary goal: Fund science working toward gene therapies. With regard to current technology, ASO (anti sense oligonucleotide therapy) is likely the best approach. This will likely atke 3-5 years after funding is initiated before clinical trials can be initiated. We are exploring multiple options to fund this initiative.

Did you know?

Dr. Danielle Veenma at CAMK2 Encore Center of Expertise is doing a **Natural History Study**. She is collecting information about children affected with CAMK2 gene disorders so in the future we can **help families better understand what to expect when diagnosed with specific mutations.** Further, the information is important so that we can identify which symptoms our kids suffer are most important to treat. This will help us design research studies for drug repurposing. **If you are interested in being a part of the Natural History Study, please reach out to camk2disorders@erasmusmc.nl.**

Dr. Geeske van Woerden at CAMK2 Encore Center of Expertise is studying cells back engineered from blood samples from our children in order to understand whether specific mutations cause CAMKII protein to be overactive (gain of function) or underactive (loss of function). **These designations will help us better understand which child may benefit from what medication.**

Money donated to CAMK2 Therapeutics Network **will speed up current research** so that we can get answers to help our children faster.

CAMK2 Therapeutics Network is getting organized to strengthen our community! **If you are interested in having your child highlighted in this newsletter (created for CAMK2 friends and families only), please reach out to us at info@camk2.org.**