CAMK2 Therapeutics Network

Community Newsletter

#3| July 2023

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

A big Thank You to the Gonzales family!

On June 24, they had a fundraiser at a local bowling alley. Their efforts attracted not one, but two TV news outlets. The Gonzales family was featured in two local news segments. They were able to raise over $6000! Our disease is getting noticed and we are getting put on the map!

Behind the Scenes

- The parent board have met with Dr. Claudia Compagnucci who was awarded the Telethon Seed Grant in Italy to study CAMK2 (funded by our CAMK2 Therapeutics Network together with UNICI) She has successfully established cell lines termed induced pleuripotent stem cells (iPSC) with two different CAMK2B mutations. These are cell lines in which the mutations can be studied in vitro (in a petri dish) in order to better understand how each mutation affects CAMK2B activity and associated calcium signaling. Once fully characterized, these cell lines will be used for "drug repurposing." That is, medications that are already FDA approved can be tested on the cell lines to see which ones best "normalize" CAMK2B activity. This is the first step to determining which medications may be most effective. We hope this initial data will lead to formal drug trials.
- We have new collaborators from the Sheba Medical Center in Israel. Physician-scientists there will be working on a formal "N of 1" study. An "N of 1 study" is a clinical trial in which a single patient is the entire trial. It is a single case study that formally tries medications one by one and formally measures benefits and side effects of each medicaton.
- A clinical trial is being initiated at Stanford University to study the effectiveness of intranasal sumatriptan (a safe migraine medication) for treatment of inconsolable crying/agitation in children with CAMK2 related disease. The rationale is that episodes of inconsolable crying/agitation (also called colic) in infants has been linked to migraine. Further, at least one person with CAMK2 related disease often reports headache with during episodes of inconsolable crying/agitation/aggression and these symptoms are significantly improved with sumatriptan. The trial is still being written and must pass through an Ethic Review Board. They likely will not be recruiting patients until early 2024. Stay tuned.
- Our Network family board has met with leaders of other rare disease communities. A big Thank You to co-founder and father, Michael Tranfagilia MD, of the FRAXA Research Foundation which supports research for Fragile X. MIke shared his 20 years of experience running this foundation and fundraising.

Donation Status

Total Raised: **$69,498 USD** as of 30/4/23

First Goal: **$150,000 USD** to support research for drug repurposing

Help Us With Our Goal!

Donate Now

Upcoming Events

9th September
Virtual Community Meeting

Speakers will include CAMK2 researchers
12am Boston | 5pm Dublin | 6pm Roterdam | 9am (San Francisco)

Stay tuned to FB group for link.

WHO ARE WE? | Stories From CAMK2 Families

Alana, 21 years old, female, CAMK2g c.889C>T, p.Arg 297Trp, London, UK.

Alana is myself and Roy's second child. When Alana was born she was a lovely, relaxed and alert baby. Very happy to communicate and we had no concerns at all. But at 6 months she suddenly stopped developing. We did not realise at the time that she had developed epilepsy. The seizures were tiny and not obvious but we later found they were almost constant. We had concerns but the registrar paediatrician could not find anything wrong and just gave us a follow up appointment. Alana was finally diagnosed with epilepsy at 13 months. But what drug was to work? The AED's that made her worse or had no effect were Carbamazepine, Clonazepam and Sodium Valproate. Great Ormond Street recommended Topiramate and that transformed Alana. She began developing again after the Topiramate took effect at 20 months. In later years we changed to Levetiracetam, without any side effects. **Amazingly she has grown out of her epilepsy and has taken no AED's since the age of 16.** She currently is only on medication to control her menstruation and trialling a low dose antibiotic to reduce the instance of UTIs. But she has never been able to catch up and is diagnosed with Autism and Severe Intellectual Impairment.

We were always told that Alana's differences were most likely to be caused by a de nova genetic misspelling and, thanks to the 100,000K Genome Project **she was finally diagnosed with CAMK2g, as causal, in January this year.** We have always found that more information helps us understand Alana so much better. The latest paper from Erasmus about CAMK2g is particularly illuminating. They could be talking about Alana. **I have always described Alana as remembering everything but somehow she doesn't learn.** This seems a contradiction but she has a very good memory of people and events (CAMK2G not needed in the hippocampus) and is able to recite books from memory. But she definitely does not have any intrinsic learning (CAMK2G is needed in the amygdala) and her motor control is poor.

Alana is a lovely girl and happily oblivious to her condition. She is well known in our neighbourhood and many will stop to say hello to her. She requires 24/7 care but the challenges of looking after her have changed over the years. One thing I have learnt **is you need to keep a good sense of humour.** Alana used to be very movement and tactile seeking. We would sit either side of her when in a pub or restaurant otherwise she would be constantly wandering. I was told she would eventually slow down and amazingly she has. She is still movement seeking but will now stay put in one place.

She has learnt to keep her hands out of other people's dinners (carrots and Yorkshire puddings were her favourites). She has stopped stroking the faces of men with stubble (sorry to the lady whose legs she stroked). And the movement seeking has translated into the things she enjoys. **She loves to swing (very high!), to ride her trike, to swim and enjoy wind on her face, to dance and to listen to music. She loves a party, especially the music and dance.**

The main challenges now are toileting and anxiety. Alana has always suffered with anxiety around specific sounds. In the past it has been rain. Currently it is coughing. But she also becomes anxious around situations where she doesn't know what is happening or what she is expected to do next. She attends an autism specific school and the strategies that they use definitely help her. And, despite me thinking one of my greatest achievements was toilet training her at age 4 she has learnt to use toileting as a means of communicating. This has been a serious challenge but we are back on track at home and when out with me and I am determined to get her on track elsewhere.

I am excited to have found the CAMK2 Therapeutics Network and excited about the research that is happening at Erasmus. I hope there will be therapy available for Alana in the future.

We aim to show all the positive and strong stories of CAMK2 families, to share information and collect all brave narratives out there. Let's bring the faces and smiles behind the mutation numbers, bringing us closer together as a community. If you want to be in our next story, please send us an email to: info@camk2.org.