A LETTER FROM OUR FOUNDERS

Nikki Stusick + Turner Binkley
Founders of The TBCK Foundation

Support the TBCK Foundation
tbckfoundation.org/give

Dear TBCK Community,

It seems like not so long ago that I held my son Theo in a NICU hospital room and looked at him wondering what life was going to be like. Flashforward to almost seven years later, I don't think I could have predicted any of it accurately. Our life has been made extraordinary because of our Theo. I write to you today with the biggest sense of gratitude and appreciation to this incredible community.

It is hard to believe that it has been about 5 years since our family and close friends helped us begin The TBCK Foundation. Many of you have been with us since the beginning. What began as another warrior mom, Lindsey McDonel and I starting an online support group for affected TBCK families expanded into an international patient-led collaborative research network funding over $180,000 in research.

What began as a handful of parents looking for others to relate to, connect with, and lean on has grown to a community of 250 parents, grandparents, clinicians and caring family members in our TBCK support group.

What began as an effort to bring families together and fund research became a community of advocates coming together both at in-person conferences and several virtual gatherings. Together, we spread disability awareness and raised funds both for TBCK research and an all-abilities playground through Race for Rare Kids.

What began as an identified 35 patients grew to 100+ in more than 20 countries. We've become a landing place for families after diagnosis, a safe haven to share experiences, learn from one another, and to feel less alone. We take so much pride in this community and what we have been able to build together over the course of these last four years. It's changed my life and connected me with some of the most inspiring and compassionate people I know.

In science, specifically genetics, it is said that sometimes what we can initially identify, we can't initially interpret. In other words, science can find something but we don't often know what it means right away.

Our family became a living example of that when we found out in March 2022 that our son Theo was diagnosed with a different rare disease called CAMK2D. In 2019, his Whole Exome Report had identified this de novo mutation of CAMK2D, but it was only years later that science caught up to confirm this mutation as pathogenic or disease-causing. It is best understood now that Theo was misdiagnosed with TBCK Syndrome; science knows so much more now. It's rare after Whole Exome Sequencing for this to occur. From this, two key takeaways worth sharing are that 1.) It is important for your geneticist to revisit results, and 2.) If, under the smallest chances, you find yourself with a new diagnosis like Theo, know that you will always be part of the TBCK family regardless.

Finding this out was disorienting. However, fortunately for Theo's medical health, our current monitoring more or less continues on the same course, as like TBCK Syndrome, CAMK2D has no intervention, treatment, or cure. We mostly traded in the same problems associated with TBCK for CAMK2D. It has been a tumultuous time, but we continue to learn more about his new disease, and thankfully there is emerging science that is promising within the CAMK2 community. Theo really is our multi-colored zebra.

Theo has always been our North Star and why we started this work, so naturally our advocacy must evolve along with our rare journey. For me that means a restructuring at The TBCK Foundation, where I will remain the Board of Directors President and step aside from any day-to-day operations.

In the past year we have been hard at work building the future of The TBCK Foundation and ensuring its continuation. With the support of a grant through the Chan Zuckerberg Initiative's Rare As One Network, we have been able to bring on a small, but mighty team to continue the momentum of our work. Together, we are pushing The TBCK Foundation into our next chapter.

Our commitment to this community and to all the TBCK Warriors we've come to know, support, and stand along with on this unpredictable journey has always been centered when it comes to future planning for The TBCK Foundation. (This is my plug for any of you interested in volunteering to get in touch. We want to make sure your voices are heard and centered.)

There is no adequate way to say thank you to the community of families, researchers, and clinicians who continue to walk with us on this journey. The connection and meaning you have given our lives continue on, regardless of diagnosis. I have so much gratitude for so many.

Thank you to our Board of Directors and parent volunteers over the years that have helped plan conferences, donate their time and resources, and support the growth and health of our community. All of you have helped get the foundation where it is today.

A special thank you to every single medical professional, researcher, doctor, medical staff, and our Scientific Advisory Board for all of your efforts to improve the lives of all of those impacted by TBCK Syndrome and in our case, all those with other rare diseases too. Thank you for all you do.

Thank you to the donors, sponsors, and volunteers who have generously supported our events and operations since we began. I think of your generosity often.

Of course, to all the parents and kids, thank you for showing me what hope looks like when I see your children's triumphs, what strength looks like on the darkest days, and what love looks like when it's overcoming the impossible everyday. Thank you for trusting me with your stories over the years to share; I can honestly say that has been my very favorite part of this entire journey. I know that you are just doing what needs to be done, but my goodness you really each are so inspiring. I treasure your friendships, and honestly no one does it better than you all.

To my family and friends who made sacrifices to allow the foundation to grow, volunteered in all of the ways, championed this cause in the most generous of ways, and most importantly showed up and loved Theo through all of this, a thank you will never be big enough for all you've done. We love you all.

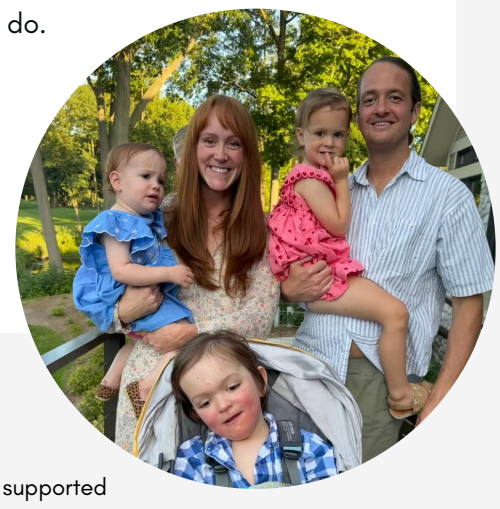
To all of you who have touched our lives in big ways and small ways, because as you know the small ways end up being the big ways too...thank you.

As my dad would say, always remember, never give up. We still have lots of work to do.

In curing rare diseases, even new diagnoses,

Nikki Stusick

Nikki Stusick
Co-founder of The TBCK Foundation



Coming Winter 2024, visit the podcast Storycollider.org where Nikki records her family's story covering Theo's misdiagnosis and new diagnosis.

TBCK Awareness Day coming up on February 1st! **tbckfoundation.org/give**

Stay tuned for upcoming details on the Kaleidoscope Playground at Kiwanis Park which Race for Rare Kids supported the last two years. Follow us @kaleidoscopeplayground on Instagram.