THE TBCK FOUNDATION

Diagnosed Families Resource Guide

A Letter to Families

Dear Families,

We are so glad you are here. Let us begin by saying, that we understand how completely overwhelming it is to receive a diagnosis of a rare disease like TBCK Syndrome. Everyone starts at the point you are now, and although each journey will take different turns, we are all connected. We've found that there is nothing more empowering than a group of compassionate parents and caregivers advocating for their children and leaving no stone unturned to seek answers.

That is why The TBCK Foundation began and why it exists: to push for answers through research, to share our stories, to demystify disability, and rare disease. It's about cheering on each other's kids, leaning on each other when needed, and fighting everyday to give our loved ones the best lives possible.

In this guide, you will find information we think may help as you begin learning about TBCK Syndrome and finding resources to support your family. On our website, we have a page especially for newly diagnosed families, where we suggest a few things that may help you as you're beginning your TBCK journey.

Everyone's journey with diagnosis and treatment is unique, but please know that you are not alone. Staying connected is critical, so we encourage you to follow us on social media, volunteer, and network with other families.

We are so glad you are here, welcome to our TBCK Family.
In finding hope,
Our TBCK Foundation Team

THE TBCK FOUNDATION

About Us

What Is The TBCK Foundation?

The TBCK Foundation is dedicated to serving families impacted by TBCK Syndrome through accelerating patient-led research, advocacy, education, and a dynamic system of support.

The TBCK Foundation is a 501(c)(3) non-profit charity. Our mission has two pillars: **scientific research** and **family connection + support**. Our first focus is research, and we maximize our fundraising and advocacy efforts to drive research towards interventions, treatments, and hopefully even a cure for TBCK Syndrome.

The TBCK Foundation is also a landing place and a community for anyone on this journey with TBCK Syndrome. Perhaps you've found your way to this page because someone you love has been diagnosed with TBCK Syndrome.

How Can You Get Connected?

- TBCK Parent Facebook Support Group
- Follow @tbckfoundation on Instagram, Facebook, + Twitter
- Reach out to our TBCK Foundation Team

We love hearing from you!
info@tbckfoundation.org





Our Global Community

WORLD MAP OF PEOPLE WITH TBCK SYNDROME

100+ DIAGNOSED PATIENTS IN MORE THAN 20 COUNTRIES

Australia
Austria
Belarus
Belgium
Brazil
Canada
Denmark
England
Germany
India
Italy
Latvia
Mexico
New Zealand
Norway
Puerto Rico
Russia
Spain
Sweden
The Netherlands
Turkey
Ukraine
United States
Wales

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Our community continues to grow globally as more patients are diagnosed every year.

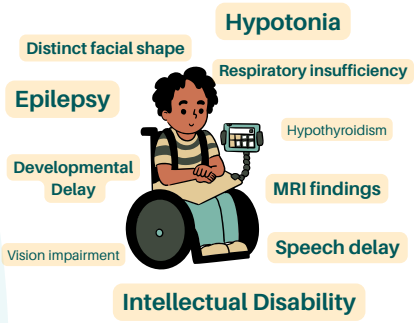
Let us know where you are!

About TBCK Syndrome

What is TBCK Syndrome?

TBCK Syndrome or TBCK-related ID syndrome (named after the gene) is a rare neurogenetic disorder/disease. The disease impacts brain development and causes intellectual disability/delay. TBCK Syndrome is an autosomal recessive condition. The symptoms and conditions of TBCK Syndrome vary depending on the person and specific variant.

TBCK stands for TBC1-domain-containing kinase. TBCK Syndrome is associated with slowed mTOR functioning (a biological pathway regulating cell metabolism, growth, proliferation, and survival).



Prevalence, Diagnosis, and Impact of TBCK Syndrome

More than 100 TBCK warriors have been identified across the globe. Cases are diagnosed with Whole Exome Sequencing. There is a range of genetic variants that have been recorded, with the full extent of symptoms for the disease being variable. Patients have balletic variants in *TBCK*, which can be missed by Sanger Sequencing and exome sequencing may miss small exon-level deletions. It is recommended that both sequencing and deletion/duplication analysis be performed. Neurophysiological studies show axonal motor neuropathy/neuronopathy in severely affected patients. Notably, a founder variant has been identified in the Caribbean region ("Boricua mutation" p.R126X) causing with severe disease, also known as TBCK-encephaloneuropathy.

Treatment for TBCK Syndrome

There is no known cure or treatment at this time. Therapies are highly recommended as well as evaluations for feeding, seizures, and respiratory insufficiency. Research focused on patient outcomes is ongoing. Seizures should be monitored. There is no recommended antiepileptic, with some patients responding well to levetiracetam. Caution is advised for Valproate, bisphosphonate infusion, and the ketogenic diet based on adverse outcomes. Patients can sometimes begin with provoked seizures (fever/illness) and then can progress to epilepsy. Patients should closely monitor aerial blood gases (ABGs) when patients are sick to avoid progression to seizures.

For Diagnosed Families

You are not alone. There are researchers, families, and advocates that are dedicated to improving the lives of those impacted by TBCK Syndrome. Scientists and medical professionals (with the help of our families) have identified these first steps:

- Sleep study (to check for sleep apnea and respiratory muscle weakness especially at night)
- EEG (to check for seizures, they can be subclinical and only detected on EEG; (see above on seizure treatment)
- Cholesterol and thyroid hormone levels monitoring
- Liver levels monitoring
- Swallow study (to evaluate for possible aspiration)
- Connect RARE-X

Questions?

Emily Durham, PhD
Research Engagement Director
emily@tbckfoundation.org

Health Impacts

Did you know, that no two people diagnosed with TBCK Syndrome present the same? Even siblings with the exact same DNA variants have been found to have a range of symptoms.

Here is an overview of symptoms and health impacts, be aware that every patient presents a bit differently and there is a spectrum.

TBCK Syndrome

OVERVIEW, SYMPTOMS + HEALTH IMPACTS

TBCK SYNDROME IS A VERY RARE NEUROGENETIC DISEASE THAT WAS FIRST DESCRIBED IN MEDICAL LITERATURE IN 2016. MOST OF THE PATIENT POPULATION IS CHILDREN + YOUNG ADULTS.

100+
ESTIMATED NUMBER OF PATIENTS DIAGNOSED GLOBALLY

MOST PATIENTS DIAGNOSED IN **FIRST DECADE** OF LIFE

24
AGE OF OLDEST KNOWN PATIENT

BRAIN + EPILEPSY

Seizures are a common symptom, which families often note as the number one health concern. Neuro-degeneration is possible in some patients.

COMMUNICATION

Most patients have speech delay and/or require support for communication. Many utilize augmentative and alternative communication (AAC) devices and methods.

INTELLECTUAL DISABILITY + GLOBAL DEVELOPMENTAL DELAY

Most diagnosed patients require assistance with activities of daily living as well as motor, cognitive, speech/language, and personal/social skills.

AUTOSOMAL RECESSIVE DISEASE

AUTISM

Families often seek support for non-verbal, sensory and communication.

PHYSICAL DISABILITY + HYPOTONIA

Most patients have severe hypotonia. Low muscle tone contributes to musculoskeletal issues (osteoporosis + scoliosis)

ENDOCRINE SYSTEM

Patients often have metabolic abnormaliies and thyroid dysfunction

DIAGNOSTIC CHALLENGES

Diagnosis barriers include lack of familiarity of the disease and access to necessary testing.

DIVERSE POPULATION

- Patient population includes 23 ethnicities, most patients identify as a person of color
- High variability in variants means most mutations are unique to an individual
- Founders mutation identified in the Caribbean: Boricua Mutation

GASTRO-INTESTINAL (GI)

GI issues are common G-tubes are very common for patients due to feeding and dysphagia.

RESPIRATORY SYSTEM

Due to hypotonia (low tone) basic respiratory illnesses can be severe. Some patients require a tracheostomy.

VISION IMPAIRMENT

Majority of patients have vision impairment. Many have strabismus.

CRANIOFACIAL DYSMORPHIA

Aberrant cranial form can be found in most patients.

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First Steps: What We Recommend

Every patient and family will move through this process of understanding a diagnosis differently. Here are specific steps you can take to help make a plan.

- **CONNECT WITH OTHER FAMILIES:** Join our TBCK Parent Facebook Group to get connected to other TBCK families and share experiences. Caregivers, close family members, and clinicians are welcome.
- **TELL US ABOUT YOUR FAMILY:** Complete our TBCK Community Registry so that we can build a rich network of information to better understand the disease and help stay connected.
- **SUPPORT RESEARCH:** Participate in our Patient Registry so we can move research forward through understanding more about TBCK Syndrome. Visit: tbckfoundation.org/rarex
- **SUPPORT RESEARCH:** Participate in the Natural History Study in order to help guide research and learn more about TBCK. Find the survey here: tbckfoundation.org/research
- **CONTACT YOUR DOCTOR:** Many families get connected with the TBCK researchers at Children's Hospital of Philadelphia (CHOP.). They will help answer questions and will give insight on your child's specific variant. Email: tbckresearch@chop.edu
- **FIND SPECIALISTS:** Coordinate with your primary care doctor and request referrals to specialists that help monitor the various body system impacts of TBCK Syndrome. (See the following page for Recommended Medical Monitoring)

Remember, it's one day at a time and do what you can, when you can. You are already doing an incredible job.

Medical Monitoring

Recommended Medical Monitoring

Most families are connected to medical intervention that leads up to genetic testing for TBCK Syndrome. Scientists and medical professionals (with the help of our families) have identified the following medical evaluations that are appropriate and helpful:

- Blood work: liver levels monitoring + cholesterol levels monitoring- if cholesterol levels are elevated, check TSH thyroid level there is sometimes been a correlation notedSleep Medicine: sleep study (to check for sleep apnea)
- Neurologist: EEG and seizure monitoring
- Endocrinologist: Monitor thyroid levels
- Orthopedic: Monitoring for hypotonia and fractures
- Ophthalmology: vision exam for vision-related impairments
- Pulmonology: monitoring for respiratory-related illness protocol
- Speech therapist: Swallow study (to evaluate for possible aspiration)
- Physical therapy assessment
- Occupational therapy assessment
- Vision therapy assessment

Please note: These are recommendations, please be sure to consult directly with your health provider to determine the most accurate plan of care.

Other Insights From Families:

-"Follow online marketplaces for equipment."
-"You know your child best, trust your gut."
-"No one is going to advocate for your kid like you are."
"Search in the Facebook support group for ideas and experiences, many times someone has gone through exactly what you have."

Getting Involved

TBCK Syndrome Awareness Day: February 1st

- Give $21 on 02/01 campaign has raised over $18,000 in 2023!
- Families share photos of their TBCK Warriors and supporters all over the world!
- Families and supporters utilize Facebook fundraisers to support TBCK-related research.

Ask a Scientist

- Every month we host a live call with our Research Director
- Ask all of your scientific TBCK questions in real time
- Look for dates on our Facebook page @tbckfoundation

Monthly Parent Chats

- Every month we host a parent support call
- Allows families to connect with each other
- Look for dates on our TBCK Parent Group page

Annual TBCK Conference

- Every year we host either an in-person or virtual conference for families, clinicians, and researchers
- Connects TBCK Families in person from all over the world
- Researchers share updates and answer questions
- Opportunities to learn about and participate in studies

Host a fundraiser for The TBCK Foundation

- Check out our Fundraiser Toolkit to learn how you can advocate and support TBCK research wherever you are located!

Parent Leadership Opportunities

- *We are always looking for volunteers*!
- *Reach out to us and ask about ways you can help: info@tbckfoundation.org*

Shop TBCK Gear

- We have an online store through Bonfire.com where proceeds benefit The TBCK Foundation

Sign up for newsletters

- We have quarterly newsletters to give updates and share information about our incredible community.

Additional Resources

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Visit our Resource Center:

We have many resources covering the wide needs of families.

tbckfoundation.org/resourcecenter

-Financial resources
-Governmental supports
-Caregiver resources
-Epilepsy management resources
-Travel resources
-Equipment resources
-Family support resources

We'd love to hear from you!
Please reach out to us anytime:
info@tbckfoundation.org

Follow us:
@tbckfoundation