Connect with us!
Social Media:

We also have a private Facebook group (for families and individuals with TBRS). Please reach out below to join.

Newsletter: tbrsyndrome.org/newsletter
Website: tbrsyndrome.org
Email: jill@tbrsyndrome.org

Make a Difference in our Community!

Participate in TBRS Research
Patient Registry: tbrsregistry.iamrare.org

Biorepository & Clinical Research ID: Contact Research Coordinator, Kit Church at kit@tbrsyndrome.org

Join our Collaborative Research Network
Contact Research Coordinator, Kit Church at kit@tbrsyndrome.org

Contribute to the Community
Volunteer: contact Board President, Kacee Richter at kacee@tbrsyndrome.org

Make a Difference in our Community!

Participate in TBRS Research
Patient Registry: tbrsregistry.iamrare.org

Biorepository & Clinical Research ID: Contact Research Coordinator, Kit Church at kit@tbrsyndrome.org

Join our Collaborative Research Network
Contact Research Coordinator, Kit Church at kit@tbrsyndrome.org

Contribute to the Community
Volunteer: contact Board President, Kacee Richter at kacee@tbrsyndrome.org

Donate: tbrscommunity-bloom.kindful.com

The TBRS Community aims to support all families affected by Tatton Brown Rahman Syndrome and advance research toward interventions.

Learn more about TBRS, connect with families, and join our research network at tbrsyndrome.org.

We are STRONGER TOGETHER.
About Tatton Brown Rahman Syndrome (TBRS)

TBRS, also called DNMT3A Overgrowth Syndrome, is a rare genetic disease caused by variants in the DNMT3A gene. About 300 people globally have been diagnosed with TBRS to date.

Individuals with TBRS have overgrowth—typically, tall stature, increased weight, and large head circumference (also known as macrocephaly)—mild to severe intellectual disability, and subtle but distinctive facial characteristics. There are a variety of other symptoms that are also associated with TBRS, such as low muscle tone, behavioral and psychiatric disorders, cardiac defects, increased cancer risk, joint hypermobility, scoliosis, seizures, and autism, but not all individuals have every clinical finding reported, and the syndrome varies considerably in its severity.

Diagnosis and Treatment

A diagnosis of TBRS is determined by the presence of a variant in DNMT3A along with the cardinal features of TBRS. If you have received a “variant of uncertain significance,” please reach out to the TBRS Community—we can help clarify diagnosis.

There is currently no cure for TBRS and treatments focus on managing each clinical finding. Doctors familiar with TBRS recommend patients see the following specialists:

- Physical, speech, occupational, and behavioral therapists
- Geneticist and genetic counselor
- Cardiologist, neurologist, hematologist/oncologist, orthopedic physician, or psychiatrist if related issues arise

Join our Patient Registry! tbrsregistry.iamrare.org

This is a powerful opportunity for individuals with TBRS to contribute directly to research that will enhance our understanding of TBRS, thus accelerating the development of new diagnostic and treatment options.

About the TBRS Community

The TBRS Community was formed in 2017 by Jill Kiernan, whose daughter Aevary was one of the first individuals diagnosed with TBRS in 2014. We have since grown into a global community with over 1,500 members, including patients and their loved ones, educators, therapists, clinicians, and researchers.

Community Support

Our programming for families and individuals with TBRS seeks to support the social, emotional, and educational needs of our community. We organize virtual hangouts for teens and young adults with TBRS, support groups for caregivers, and regional in-person meet ups for families to connect with each other and with local doctors and scientists.

Our Education & Outreach Series provides critical information on pressing topics such as cancer risk and financial planning.

Since 2018, the TBRS Community’s annual Family Conference has gathered dozens of members for socializing and information sessions with clinicians and researchers. We are excited to be hosting our 2023 Family Conference at Morgan’s Wonderland in San Antonio, Texas, October 12–15. The 2023 TBRS Community Summit will combine our Family Conference and Collaborative Research Network Conference for the first time!

Research Acceleration

The TBRS Community is the organizing hub for research on TBRS. We maintain a Collaborative Research Network representing 200+ experts, host regular Research Roundtables, and sponsor several ongoing research projects. These include:

- TBRS patient registry (tbrsregistry.iamrare.org)
- Biorepository of patient samples and iPSC lines
- Funded projects on neurodevelopment, metabolism, and biomarker development