



TBRS
community










New Family
Welcome
Packet




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tip: **Download or print off the fact sheets and share with your doctors, therapists, and teachers!**

Donate

Welcome. We are so happy you found us!

Dear New Member of the TBRS Community,

Welcome to a place of hope, understanding, and relentless pursuit of knowledge. As a TBRS mom and as the Executive Director of the TBRS Community, I extend a heartfelt welcome to you and your family. You are now part of a unique family, one that understands the challenges and triumphs of living with Tatton Brown Rahman Syndrome (TBRS). Our mission is clear and steadfast: to support everyone affected by TBRS and to advance research toward interventions.

In the TBRS Community, you will find more than just support; you will discover a family you never knew you had. We recognize that living with a rare disease can be a solitary and daunting journey. That's why we strive to make our community a sanctuary where isolation turns into connection, and fear transforms into hope. Here, you are not alone. You are part of a family that celebrates each other's strengths and provides comfort during challenging times.

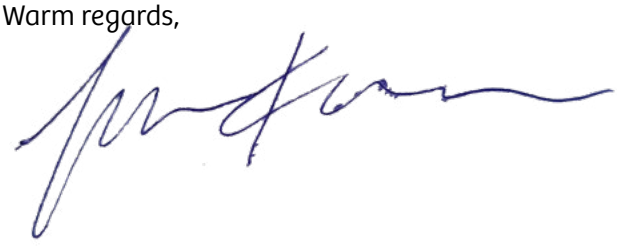
Our passion for supporting families is matched by our dedication to raising awareness and conducting impactful research. The TBRS Collaborative Research Network stands as a testament to this commitment. With over 200 researchers and clinicians, this network is a beacon of hope, driving forward our understanding of TBRS and related conditions. Every program, every research initiative we undertake is patient-focused, echoing the priorities and needs of our community members like you.

In our mission to advance research and interventions, we also encompass Heyn Sproul Jackson Syndrome (HESJAS) and other DNMT3A related disorders within our research program. We believe that by broadening our scope, we can accelerate our understanding and discovery of solutions that benefit our entire community. Together, we are stronger and more capable of unraveling the complexities of these conditions.

As you navigate through our program guide, you will see the breadth of what the TBRS Community has to offer. From support groups to educational resources, every aspect of our community is designed with you in mind. We encourage you to reach out, ask questions, and share your journey with us. Together, we are stronger, wiser, and capable of incredible feats.

Welcome to your new home, the TBRS Community. Here, you will find more than just friends – you will find a family that stands with you every step of the way.

Warm regards,



Our Vision

We envision a world where people affected by rare diseases, like TBRS, have a place to go for answers, support, treatments, and cures. It's as simple as that. Nobody should be left out when it comes to access to health and happiness.

The TBRS Community is turning this vision into reality for our families, while blazing a trail for all those impacted by rare diseases.

Jill Kiernan
Executive Director & **Founder**
jill@tbrsyndrome.org

New Family *Checklist*

- ☐ Join our private **Facebook Group** to meet other families, share resources, and ask questions.
  - ☐ As part of our regional coordination program, we have subgroups within the larger group, to meet the needs of non-English speakers and to provide more individualized support to different regions around the world.
- ☐ Subscribe to our quarterly **newsletter** to get the latest news and events in the Community.
- ☐ Check out our **YouTube channel** to take a peek into the lives of some of our diagnosed families. You can also view the latest presentations from our clinicians and researchers.
- ☐ Attend a **New Family Virtual Meet-Up** to connect with other TBRS families and learn about what the TBRS Community has to offer. View our calendar for info on the next Meet-Up!
- ☐ **Participate in TBRS Research!** There are a lot of opportunities, and we encourage you to start with Count Me In for TBRS and the Patient Registry.
  - ☐ Count Me In for TBRS - Contact Registry (page 10)
  - ☐ Enroll in the TBRS Community Patient Registry (page 10)
  - ☐ Patient Priority Survey (page 11)
  - ☐ TBRS Community Biorepository (page 11)
  - ☐ Clinical Research ID (page 11)
  - ☐ Citizen Health (page 12)
  - ☐ Brain Gene Registry (page 12)
- ☐ **Introduce yourself** to the Board of Directors and our Executive Director.
- ☐ Check out **events** and **news updates** on our website.
- ☐ **Get involved** with TBRS Community and volunteer.

tip: **Request a 1:1 meeting with Executive Director, Jill Kiernan, to introduce yourself, ask questions, and get to know the community.**

SCHEDULE HERE

We also have a private Facebook group (for families and individuals with TBRS).
https://www.facebook.com/groups/705487016188994

Newsletter: tbrsyndrome.org/newsletter

Website: tbrsyndrome.org

Email: info@tbrsyndrome.org

Tatton Brown Rahman Syndrome
Fact Sheet

Tatton Brown Rahman Syndrome (TBRS), also referred to as DNMT3A Overgrowth Syndrome, is a rare genetic disease caused by variants in the DNMT3A gene. There is a wide range of medical issues associated with TBRS but most individuals have a few unifying characteristics:

- **Overgrowth:** TBRS causes children to grow rapidly, and adults with the syndrome are often above average in height and weight. Head circumference is also often increased in size (this is referred to by doctors as macrocephaly).
- **Intellectual Disability:** Some individuals have severe cognitive impairment, while others have a mild disability.
- **Facial Features:** Horizontal eyebrows, large front teeth, and narrow eye openings are subtle yet distinctive characteristics of people with TBRS.

TBRS was first described in 2014, and although doctors continue to identify more individuals affected by the syndrome each year, it remains extremely rare, with around 350 people diagnosed as of 2024. Physicians are still learning about the full spectrum of conditions associated with TBRS. In addition to having the three characteristic features of overgrowth, intellectual disability, and facial features, some individuals additionally present with **autism, joint hyper-mobility, low muscle tone, scoliosis, seizures, behavioral and mental health disorders, heart defects, and blood disorders.**

Click or scan here to read more about our surveillance recommendations

There is no cure for TBRS. The needs of individuals with TBRS vary greatly—some are able to live independently with minimal aid, while others require lifelong intensive support and medical care. Because of this variability, it is important to gather a thorough assessment of each person's specific needs and for **families to participate in the TBRS Community Patient Registry.**

Doctors familiar with TBRS recommend meeting with the following clinicians:

- **Cardiologist:** To screen for heart defects and get a baseline echo-cardiogram
- **Physical therapist, speech therapist, occupational therapist:** For young children, states' Early Intervention programs can screen for eligibility. Older children may access these services through the school district, and adults, through their medical insurance or local adult services programs.
- **Geneticist and genetic counselor:** To coordinate care and screenings, and keep families updated on new developments with the diagnosis
- **A neurologist, hematologist, orthopedic physician, psychiatrist, or behavioral therapist** should be consulted if specific issues arise.

Leukemia Connection

DNMT3A mutations, when acquired in blood cells later on in life (also called somatic mutations), are known to drive the development of leukemia in people without TBRS. For this reason, researchers are investigating whether TBRS is linked with an increased risk for leukemia. There is evidence to suggest TBRS mutations are associated with a higher risk of acute myeloid leukemia, but studies are ongoing to confirm the size of this risk and it is not observed to be a common occurrence. There is no evidence yet that TBRS causes additional cancers, though there have been cases of TBRS patients with different cancers. If someone with TBRS shows possible signs of leukemia, such as easy, unexplained bruising or fatigue, consult a physician for testing.

The Gene

The protein produced by the DNMT3A gene is involved in a process called DNA methylation, which helps cells determine which genes are turned on or off. Although all cases of TBRS involve mutations or deletions in DNMT3A, the location of the mutation in the gene is not the same for all individuals, with most patients having a unique mutation or deletion of their own.

It appears that most mutations arise from spontaneous changes in the gene (called de novo mutations), rather than mutations inherited from the person's parents. Some individuals may have inherited the DNMT3A variant from a parent who has TBRS or does not have TBRS. In the latter case, this is because the mutation can sometimes occur later in development and therefore be present in only certain types of cells (this is called mosaicism). Individuals with mosaicism could have a mutation in DNMT3A in sperm or eggs, also called the germline, and therefore possibly pass it down to their children.

Mutations in DNMT3A that cause TBRS are heterozygous, meaning they are only on one of the two copies of the gene that each person has. Because each parent provides one copy of a gene, someone with TBRS has a 50 percent chance of having a child with the genetic mutation. Genetic counseling is important to help clarify the inheritance pattern of TBRS.

Resources

Website: www.tbrsyndrome.org **Email:** info@tbrsyndrome.org

Public Facebook page: https://www.facebook.com/dnmt3aovergrowthsyndrome/

Private Facebook group for families and providers:
https://www.facebook.com/groups/705487016188994/

Registry: https://tbrsregistry.iamrare.org/

Ostrowski PJ, Tatton-Brown K. Tatton-Brown-Rahman Syndrome. 2022 Jun 30. In: Adam MP, Everman DB, Mirzaa GM, et al., editors. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2022. Available from: https://www.ncbi.nlm.nih.gov/books/NBK581652/

Guide to Programs:

Education and Outreach

New Family Meet-ups

The TBRS Community hosts quarterly virtual meet-ups for newly diagnosed and existing members of the community. This is an informal gathering to answer questions, talk about current TBRS-related research, review what the TBRS Community has to offer, and simply get to know each other.

- We would also like to focus on patient priorities and what YOU would like to see from our team in terms of programming, research, outreach, and support.
- We want to hear about your needs and how we can best serve you as part of our community!

Meetings with the TBRS Team

We can answer questions, point you to helpful resources, discuss TBRS research, and more. We love to get to know members of our community. https://calendly.com/jill-tbrs

Regional Coordination

Our Regional Coordination Program is recruiting leaders from communities and countries around the world to help provide local resources and support for TBRS families. We are stronger together!

Advocacy

The TBRS Community participates in multiple rare disease advocacy opportunities, including the Rare Disease Diversity Council, Rare Disease Week on Capital Hill, and more

Education & Outreach Series

The TBRS Community Education and Outreach Series are virtual meetings focused on different topics related to TBRS. Some of these meetings are recorded and can be found on the TBRS Community YouTube Channel.

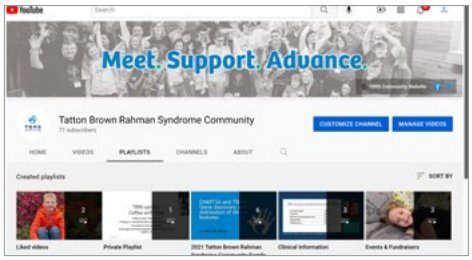
YouTube Channel

The TBRS Community has our own YouTube Channel! This is a great resource to learn more about TBRS by watching previous conferences and presentations from researchers.

- You can meet some of our featured families and share the videos with others who want to learn more about this rare disease.
- The channel is also a wonderful way to inform your doctors, specialists, therapists and teachers who work with your family and to spread awareness.

tip: **Did you know that you can sync the TBRS Community Google calendar to your online calendar so you dont miss any events?!**

Need help with this? Reach out to Kit: kit@tbrsyndrome.org



Check out our YouTube Channel for past recordings of the Family and CRN Conferences and to hear stories of some of our families!

Guide to Programs:

Family Support

Private Facebook Group

The Facebook group is a private and open forum for families and diagnosed individuals to connect with others. It includes updates on research, events, and news from families. Subgroups are available for international patients in a variety of languages and for caregiver-specific support.

Parent/Caregiver Virtual Support Group and Dads' Support Group

The TBRS Community offers monthly virtual Support Groups for Parents/Caregivers and for Dads. Attendees from around the world can participate. We create a safe place to share thoughts, feelings, and concerns without judgment. The meeting seems to take a different shape every time we gather—sometimes we focus on sharing resources or experiences, and we always seek to make everyone feel comfortable and supported.

Social Group for Diagnosed Individuals

Each month a wonderful group of individuals who are diagnosed with TBRS gather for our Social Group. We have a blast with activities that are requested by our group members. You may find us doing anything from dancing and joke telling to karaoke and rock painting. The group is ever evolving and always a fun time. There is no pressure to chat if you are feeling shy, and you don't have to participate in the activity if you would rather just talk.

Regional Meet-ups

The TBRS Community is encouraging and helping to plan regional in-person meet ups in 2024 around the world. Please reach out if you are interested in organizing a meet-up near you.

TBRS Cares

The TBRS Community sends care packages out to families in crisis.

For more information please visit our **website**, the **Facebook Group**, or email **info@tbrsyndrome.org** for more information.

"As I sat with my son to help him interact with the group and the trivia, his face was glowing. He communicated few words, but that hour will stay with me."

- Parent Zoe Wisnoski on a social group zoom

Guide to Programs:

Research Participation

The TBRS Community sponsors the full spectrum of research projects to advance our understanding of the syndrome and to accelerate the path to treatments. Our Community and Research Engagement Manager Kit Church can answer questions and help you keep track of your progress along the way (kit@tbrsyndrome.org). We recommend starting with Count Me In for TBRS and the Patient Registry. Your participation is crucial to supporting people with TBRS!

Count Me In for TBRS

This is a list of contact info for patients and caregivers in our community. We do not share personal information without your consent, and this is an internal document used by TBRS Community staff to know how many individuals are in our community and how to reach them.

- You can choose to be contacted about studies and provide location data so we know where our patients are.
- This is important so we can best advocate for our globally community!

Patient Registry

The TBRS Community has developed a patient registry in partnership with the National Organization for Rare Disorders (NORD). The registry is a series of surveys that ask about the quality of life, medical history, and development of people diagnosed with **TBRS, Heyn Sproul Jackson Syndrome (HESJAS), and other DNMT3A-related disorders.**

- It is important that all families contribute to the registry as it is an invaluable tool to help us further our understanding of TBRS and advance research so we can move toward identifying treatments, improving medical care, and formulating educational, social, and daily living supports.
- This secure, anonymized database helps clinicians and scientists fully understand TBRS and identify opportunities for research.

"Because of the contributions from TBRS families to our research projects, we know so much more about this syndrome."

Shown here, a family member and researcher collaborating together at the TBRS Summit to enhance their studies based on our contributions.

Guide to Programs:

Research Participation

Patient Priority Survey

The Patient Priority Survey asks about research priorities from patients' and caregivers' perspectives, so that we can fund research that matches your priorities and encourage a patient-driven research agenda in our Collaborative Research Network. This survey was created by the Overgrowth Syndromes Alliance (OSA), and organized by the TBRS Community and Malan Syndrome Foundation with the goal of finding treatments for overgrowth intellectual disability disorders.

- The survey can be completed by patients or caregivers. Contact us with accessibility needs.
- We have zoom meetings with patients who need assistance filling out the survey if needed.
- This is important so we make sure that TBRS research is benefiting patients in the most important ways!

TBRS Community Biorepository

People affected by TBRS and their family members can donate blood, urine, skin cells, and other tissues. These can be used by researchers to understanding the science behind TBRS and to accelerate the development of treatments.

- Researchers apply and go through an advisory board to make sure they are researching TBRS and are going to return results to families.
- This is important so that we can get research moving quicker on TBRS!

Clinical Research ID (CRID)

The Clinical Research ID (or CRID) is a unique number that is used to connect patient samples to other research! The patient / caregiver can create this ID without using any personal info, and then it connects these samples in a de-identified way!

- This way, scientists will be able to see Registry data and have a sample for the same patient without knowing any identifiable information, offering researchers a very powerful and informative data set to accelerate research.
- The CRID is required for the Biorepository, but it is optional for the Patient Registry.

"Families have the best knowledge of TBRS - it's so important we share our experiences with doctors and researchers so they can get us the answers we need."

Biorepository collections from the TBRS Summit were critical to our researchers' findings

Guide to Programs:

Research Participation

Citizen Health

Citizen Health is a program that allows patients and/or caregivers to have access to ALL MEDICAL RECORDS! When you sign up with Citizen Health, you give them permission to go to all of your previous hospital systems and clinics to collect this information in one easy place! Many parents have also said that they end up getting back documentation that they have never seen before!

- Additionally, with Citizen Health you are able to choose to share anonymized versions of your medical documents with research! (including our patient registry!)
- This is important because it gives researchers a ton of anonymized information to learn from.

Brain Gene Registry

The Brain Gene Registry is similar to our Patient Registry, but it looks at MANY neurodevelopmental (brain) disorders at the same time to find similarities in symptoms and treatment opportunities. This is run by many academic orgs across the US, including Washington University and Childrens Hospital of Philadelphia (to name a few).

- This can help us find insights between disorders and potential similarities that can be used in clinical trials in the future!

? For questions or more information about or research programs, please contact our Contact Community and Research Engagement Manager, Kit Church: kit@tbrsyndrome.org

Importance of Participating in Research

Participating in research means **real results** for individuals with TBRS! TBRS patients participating in research have already helped scientists to:

- Better understand the features and clinical findings of TBRS
- Estimate the risk of cancer in TBRS
- Make it easier to diagnose patients with TBRS
- Develop research models from patient samples
- Begin learning how TBRS affects the brain
- Create screening guidelines for management

We can also answer patient questions using data from the Patient Registry! From this information, we have learned that cardiac issues, aortic root dilation, seizures, vision and hearing problems, and mental health problems are more common than previously reported.

Guide to Programs:

TBRS Summit

In 2023 we held the TBRS Summit (a combination of the family conference and our scientific conference) using hybrid options to promote a global attendance while providing the in-person connection that so many families love.

2023 Summit

The highlight of the year was the first in-person TBRS Summit since 2019, uniting 187 patients and family members with 27 scientists. The Summit was translated into 5 languages so 18 non-English speaking families and scientists could participate. This event was a melting pot of ideas, experiences, and hope, making strides in bridging the gap between families and researchers.

Future Summits

In-person Summits will be held every other year. We will alternate in-person and virtual summits every other year. Additionally, we will plan meet ups around the world, because nothing can replace getting together in person.






> **"We went eager to meet people who could relate to us and we could learn from. The doctors, geneticists, TBRS staff, volunteers, and information knocked it out of the park."**
>
> - Katie Brennan, Parent and TBRS Summit attendee

Guide to Programs:

Collaborative Research Network

The TBRS Community Collaborative Research Network (CRN) is at the forefront of transforming research and treatment for TBRS. With over 200 clinicians and researchers united in their mission, the network harnesses a patient-centered research agenda to make strides in understanding and treating TBRS. By utilizing research tools provided by the TBRS Community, the CRN accelerates research and ensures that each project aligns with our comprehensive research roadmap. Witnessing the collaboration and dedication of the CRN is truly inspiring, as it propels partnerships to create impactful solutions for those affected by TBRS.

Research Roundtables

The TBRS Community began holding Research Roundtables for scientists in 2023 to ensure that we know about all up-to-date research efforts and can direct this information to our patient community. We also use this platform to inform our research community on patient priorities and TBRS Community activities and initiatives, and form working groups on specific topics like developing diagnostic, surveillance, and treatment guidelines.

These Roundtables provide a collegial space for researchers to discuss unpublished research, share resources, and collaborate.

Meet our Scientific and Medical Advisory Committees

Our Scientific and Medical Advisory Committees provide expert leadership on research efforts and clinical care.











Harrison Gabel Washington University in St. Louis

William Gibson, University of British Columbia

Timothy Ley, Washington University in St. Louis

Joseph Malak, Bambini Pediatrics

Rachel Rau, Baylor College of Medicine

Marwan Shinawi, Washington University in St. Louis

Kate Tatton-Brown, St. George's University Hospital

Ayala Tovy, Novartis/Baylor College of Medicine

Serge McGraw, University of Montreal

Rosanna Weksberg, University of Toronto

Additional members:

- Kit Church
- Kerry Grens
- Jill Kiernan
- Keren Shtiegman

Get *Involved!*

Volunteer Teams

Make an Impact! Volunteer efforts are the driving force behind the TBRS Community. Each contribution, big or small, accelerates our progress towards supporting those affected by TBRS. Whether it's fundraising, planning events, advocating, or something else, your unique skills can make a real difference. Join us and help us move faster towards a brighter future for the TBRS community. The TBRS Community has four Teams and volunteers have many opportunities to contribute based on their skills and interests! For more information, view our volunteer team opportunities here.

Fundraising and Development Team

The Fundraising and Development Team is responsible for coordinating fundraising events, identifying grant and sponsorship opportunities, and designing fundraising campaigns. Available positions include: Thank You Caller, Thank You Letter Writer, Family Fundraiser Coordinator, Grant Prospector, Grant Writer, Sponsor Seeker, and Campaign Writer.

Regional Coordination Team

The Regional Coordination Team is responsible for providing resources and TBRS Community information to diagnosed individuals and families in their region, as well as welcoming new members and planning local events. Available positions include: Regional Coordinator, Meet-Up Host, Caring Ambassador, and Social / Support Group Facilitator.

Social Media Team

The Social Media Team is responsible for creating TBRS content, developing posts, and identifying photos for media use to promote engagement and awareness of TBRS and the TBRS Community. Available positions include: Photo / Video Content Scout, Reel Maker, Content Writer, and Storyteller.

Advocacy and Policy Team

The Advocacy and Policy Team is responsible for attending advocacy events, sharing stories with lawmakers and media, advising on DEI efforts, and hosting events to raise awareness of TBRS. Available positions include: Advocate, Media Contact Specialist, DEI Advisor, and Awareness Event Host.

> **"Volunteering for the Community has been incredibly rewarding. I get to donate my time to a wonderful organization while making a direct impact on advancing research and programs for TBRS that will ultimately benefit my daughter."**
>
> - Erin Rooker, TBRS Marketing Director and Volunteer of the Year, 2023

About the *Community*

Board of Directors

The TBRS Community Board of Directors is passionately committed to finding effective treatments and individualized support for those affected by TBRS. Their actions are driven by a common goal: to ensure that every person impacted by TBRS experiences the best possible health and happiness. They all volunteer their time to help make our Community a success!

Kacee Richter, President

Kerry Grens, Vice President

Jen Isaacs, Secretary

Erin Rooker, Marketing Director

Tom Watson, Treasurer

pictured: Kerry Grens, Kacee Richter, Erin Rooker, Jill Kiernan, Jen Isaacs

Community Staff

Led by Executive Director and Founder, Jill Kiernan, our TBRS Community is supported by these three amazing staff. We are so thankful for all of their hardwork and dedication.

Eric Diehl, PhD
Science Director
eric@tbrsyndrome.org

As our Science Director, Eric surveys the TBRS research landscape to identify opportunities for treatment and gaps in research, communicates research findings in an easily digestible format to the Community, staff, and committees, and develops and maintains a Research Roadmap to guide research based on patient priorities.

Kit Church, MPH
Community and Research Engagement Manager
kit@tbrsyndrome.org

Our Community and Research Engagement Manager, Kit, engages with our Collaborative Research Network and identifies new researchers working on DNMT3A, organizes our annual research meeting and scientific programming, develops resources for families and clinicians on TBRS, and advances research in line with our families' top priorities.

Chelsea Spence
Development Director
chelsea@tbrsyndrome.org

Our Development Director, Chelsea, is responsible for creating a strategic development plan for the organization, advancing donor communications and outreach, assisting with grant prospection, and diversifying funding sources and streams to support the TBRS Community's programs and missions.