

Tatton Brown Rahman Syndrome (TBRs)

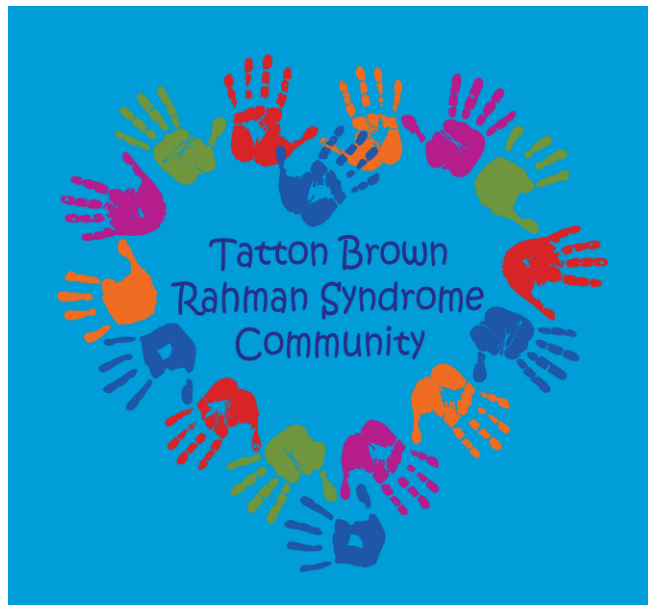
Tatton Brown Rahman Syndrome or DNMT3A Overgrowth Syndrome, is caused by a range of different mutations within the DNMT3A gene. TBRs was first recognized and described in March 2014 through the Childhood Overgrowth Study based at the Institute of Cancer Research, London, UK.

Associated Symptoms

Tall stature, macrocephaly (increased head circumference), variable cognitive disabilities (mild to severe), autism spectrum disorder, joint hypermobility, developmental delay, low muscle tone, low-set heavy horizontal eyebrows (more apparent with age), seizures, cardiac issues, obesity, behavioral / psychiatric issues, scoliosis, others still unknown.

TBRs Community Mission

The Tatton Brown Rahman Syndrome Community supports and educates individuals with TBRs, their families, friends, service providers, and those still seeking a diagnosis.



TBRs COMMUNITY

122 HUNNS LAKE ROAD
STANDFORDVILLE, NY USA 12581

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TBRS Community

TBRS Community was established as a nonprofit October 6, 2017. Dedicated to education and outreach of this new and rare syndrome. TBRS is most commonly de novo (present for the first time) but can occasionally be inherited from a parent. Fundraising is key to developing educational material, creating a global medical registry, conducting the annual TBRS conference, funding grants to further research, planning events to bring awareness to this syndrome, providing care packages for individuals recovering from serious illness. Also sponsor programs to support inclusion for people with other disabilities. TBRS Community has four committees to assist in these efforts. TBRS Community is 100% volunteer run with no paid employees. 501 c3 nonprofit status.

Specialists to Visit

It is important to ensure all possible associated symptoms are addressed by the proper physician. Dr. Tatton-Brown recommends seeing a geneticist, cardiologist (echocardiogram/EKG), neurologist (EEG), physiotherapist (for hypermobility) as well as an orthopedic surgeon/spine specialist (scoliosis) and psychiatric (when behavioral/mental health concerns are present).



Therapies to Consider

Contact your local school district to learn about available services. A specialized class/program can benefit development. Speech, occupational and physical therapies might be necessary depending on need. Applied Behavioral Analysis (ABA) or other evidence-based therapies can assist with autism spectrum disorder.



Conferences

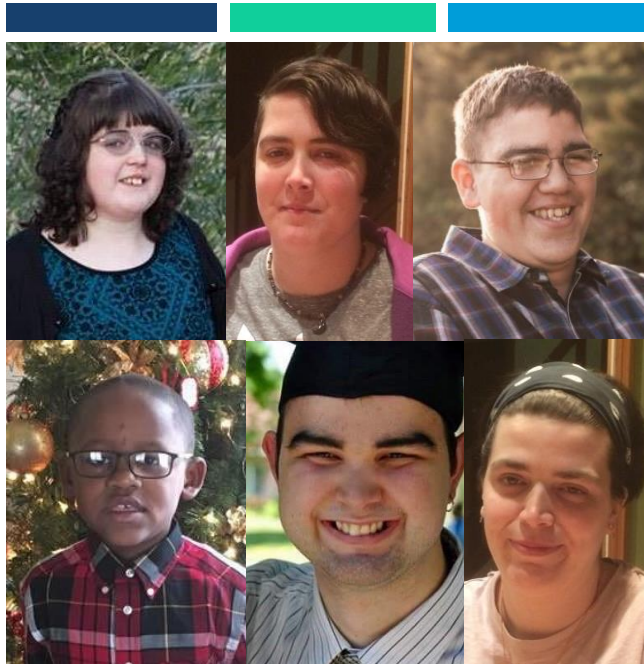
First Annual TBRS Community Conference was held September 2018 in New York, USA. The Child Growth Foundation (UK) holds an annual conference for multiple syndromes, including TBRS. Dr. Tatton-Brown attends and presents at both.





Best Learning Practices

All people learn differently, but many people with TBRS report similar learning styles. It may be important to allow a lot of opportunity and repetition to practice new skills, to encourage social interaction and participation in extra-curricular groups, and to encourage peer mentorship opportunities. In addition, it may be helpful to provide visual cues (visual models, visual schedule, visual timers, picture exchange cards, etc.) in conjunction with any verbal information or instruction. It may be necessary to keep verbal instructions more concise, and to gain joint attention before providing information. Multi-sensory learning modalities are also beneficial.



Medical Advisory Board

Dr. Kate Tatton-Brown: Geneticist working in London, UK. She has been investigating conditions associated with increased growth and a learning disability since 2001 and has published widely in this area.

Dr. Marwin Shinawi: Professor of Pediatrics at Washington University School of Medicine. He is Board certified in Clinical Genetics and Medical Biochemical Genetics.

Dr. Timothy Ley: Professor of Medicine and Genetics at Washington University. Serves as Director of Stem Cell Biology Section in the Department of Medicine.

Dr. Chloe Lane: Post-doctoral research associate in the Sheffield Autism Research Lab at the University of Sheffield.

MANY FAMILIES REPORT

Tendency to over-eat, high pain tolerance, sensory processing disorder, orthodontic concerns (large teeth, overcrowding, high arched palate, extra set of teeth), orthopedic issues, delayed self-care, trouble sleeping, susceptibility to infections, regression, learning pattern that involves inconsistent retention of skills, vision and hearing concerns, large and flat feet with widely spaced toes, difficulty speaking, hyper-flexibility



PERSONAL BIOS



Damion

DOB: September 13, 1998

Located: Netherland

Symptoms: Overgrowth (height and macrocephaly), cognitive disability, signs of ADHD, symptoms of autism, straight eye brows, epilepsy, cryptorchidism, congenital heart defect, AML leukemia

Maverick

DOB: March 17, 2014

Located: USA

Symptoms: Overgrowth (weight, macrocephaly and height), mild scoliosis, developmental delay, autism spectrum disorder



Astrid

DOB: November 12, 2007

Located: Denmark

Symptoms: Overgrowth (height), cognitive disability, ADD, autism spectrum disorder, heart defect, central sleep apnea

Eve

DOB: August 26, 1999

Located: Canada

Symptoms: Overgrowth (height), atrial septal defect, kyphosis, cognitive disability, polycystic ovary syndrome, dyspraxia, slight enlargement of liver and spleen



Henry

DOB: January 13, 2010

Located: USA

Symptoms: Cognitive disability, heart defects, epilepsy, central sleep apnea, heart defect, cryptorchidism, umbilical hernia, hydrocephalus, dysphagia, Chiari Malformation



Alex

DOB: April 15, 2017

Located: Canada

Symptoms: Overgrowth (weight, macrocephaly and height), hypotonia, cognitive disability, developmental delay, central sleep apnea, hypoventilation

Ryker

DOB: September 3, 2013

Located: Canada

Symptoms: Overgrowth (weight, macrocephaly and height), developmental delay, hypermobility, autism spectrum disorder, cognitive disability, keratosis pilaris rubra



Addisyn

DOB: February 14, 2016

Located: USA

Symptoms: Overgrowth (height), cognitive disability, ADHD, developmental delay, small hole in heart, low muscle tone



Ayden

DOB: May 18, 2006

Located: USA

Symptoms: Overgrowth (weight, macrocephaly and height), developmental delay, hypotonia, central sleep apnea, Chiari malformation, autism spectrum disorder, heart defect



Aevary

DOB: November 12, 2003

Located: USA

Symptoms: Overgrowth (weight, height, macrocephaly), autism spectrum disorder, Chiari malformation, scoliosis, development delay, cognitive disability



Stryker

DOB: May 28, 2015

Located: USA

Symptoms: Overgrowth (height, macrocephaly), febrile seizures, hypotonia, cognitive disabilities, central sleep apnea, umbilical hernia, Chiari malformation, apraxia, low muscle tone

Joshua

DOB:

Located: USA

Symptoms: Overgrowth (height, weight, macrocephaly), developmental delay, ADHD, low muscle tone, mitral valve prolapse, super ventricular tachycardia, central sleep apnea



Chrissie

DOB: July 21, 1986

Located: UK

Symptoms: Overgrowth (height), autism spectrum disorder, pervasive development disorder, polycystic ovary syndrome, non-epileptic seizures, hypermobility, osteoporosis

Clara

DOB: November 21, 2014

Located: USA

Symptoms: Overgrowth (height, weight, macrocephaly), developmental delay, low muscle tone, febrile seizure

