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.
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.

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.
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.
Maverick
DOB: March 17, 2014
Located: USA
Symptoms: Overgrowth (weight, macrocephaly and height), mild scoliosis, developmental delay, autism spectrum disorder

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

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

Astrid
DOB: November 12, 2007
Located: Denmark
Symptoms: Overgrowth (height), cognitive disability, ADD, autism spectrum disorder, heart defect, central sleep apnea

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, developmental 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