Tatton Brown Rahman Syndrome
Surveillance Guidelines

Management & Surveillance of TBRS

The TBRS Community has compiled this document to share information about the clinical findings and potential manifestations of Tatton Brown Rahman Syndrome (TBRS). This summary is based on published recommendations (including the *GeneReview* by Dr. Tatton-Brown and colleagues), more recent peer-reviewed literature, and data collected through the TBRS and *DNMT3A* Patient Registry.

The purpose of this document is to support informed discussions between families and their clinical teams. It is not intended to serve as prescriptive medical guidance, but rather to highlight the importance of a multi-systemic, multi-disciplinary approach to care in TBRS and to share emerging knowledge with the medical community.

Initial Management Recommendations (immediately following TBRS diagnosis)

System	Evaluation	More information
Constitutional	Measurement of weight, length/height, and head circumference	Assess for macrosomia.[1]
Development	Developmental assessment	Assess motor, adaptive, cognitive, and speech/ language evaluation. Evaluate for early intervention / special education.[1]
Psychiatric / Behavioral	Neuropsychiatric evaluation	(For individuals over 12 months) screening for behavior concerns, including sleep disturbances, ADHD, anxiety, and traits suggestive of ASD.[1]
Neurologic	Neurologic evaluation	Consider brain MRI (if clinical symptoms) and consider EEG (if seizures are a concern)[1,2,3,4,5]
Musculoskeletal	Orthopedics / physical medicine and rehab / Physical and Occupational Therapy evaluations	Assessment of gross and fine motor skills, joint hypermobility, kyphoscoliosis, mobility, ADL, need for adaptive devices, and need for physical/occupational therapy.[1,5]
Cardiovascular	Baseline echocardiogram	Assess for structural heart defects and aortic dilatation.[1,5,7,8,9]
Respiratory	Polysomnography	Assess for sleep apnea (if suggested by clinical symptoms).[1,5]
Genitourinary and Endocrinology	Exam for cryptorchidism in males	Consider assessment for vesicoureteral reflux in those with a history of recurrent urinary tract infections.[1,5,10]

Hematologic / Lymphatic	Consider a complete blood count (CBC) with differential	Inform patients/families of the potential risk of hematologic malignancy, with an emphasis on symptom awareness. A low threshold should be adopted for investigation for malignancy (in case of symptoms).[1]
Genetic counseling	By genetics professionals	Inform affected persons and their families of the nature, inheritance, and implications of TBRS to facilitate medical and personal decision making.[1]
Family support and resources		Assess the need for community support or online resources, social work involvement for parental support, and home nursing referral.[1]

Ongoing Surveillance Recommendations

System	Evaluation	Frequency
Development	Monitor developmental progress and educational needs.	Every visit [1,11]
Psychiatric / Behavioral	Behavior assessment for anxiety, attention, and aggressive or self-injurious behavior.	Every visit [1,11]
Neurologic	Monitor for seizures. Assess for new manifestations such as seizures and changes in tone.	Every visit [1,2,3,4,5,11]
Musculoskeletal	Physical medicine, occupational/physical therapy assessment of mobility, kyphoscoliosis, and pain.	Every visit [1,5,11]
Respiratory	Assess for signs and symptoms of sleep apnea, infections and other sleep and respiratory conditions.	Every visit [1,5,11]
Endocrinology	Assess for changes in hormones and thyroid function.	Every visit [5,10]
Hematologic / Lymphatic	Assess for signs and symptoms of blood malignancy.	Every visit [1]
Family / Community	Assess family need for social work support and care coordination.	Every visit [1]
Cardiovascular	Echocardiogram to assess aortic root indices.	Ongoing surveillance to assess the size of the aortic root, cardiac function, and structural conditions. [5,7,8,9,11]

If you have any questions about these recommendations, please reach out to our Community and Research Engagement Manager at kit@tbrsyndrome.org.

Disclaimer: The TBRS Community provides this information as an educational resource for families and healthcare professionals. This document does not replace individualized medical evaluation, clinical judgment, or treatment decisions made by qualified healthcare providers. Surveillance and management plans should always be determined by clinicians in consultation with the patient and family.

Citations

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