SATB2-Associated Syndrome

Other names: ***Glass syndrome, 2q33.1 deletion/microdeletion/mutation***

FOR MEDICAL PROFESSIONALS & CLINICIANS

Features

- Significant neurodevelopmental disorders in all affected individuals, which may include: infantile hypotonia and feeding difficulties, global developmental delay including severe speech delay (speech apraxia, commonly absence of speech), gross and fine motor delays (developmental dyspraxia), cognitive delay.
- Behavioral issues: autistic tendencies, hyperactivity, sleep disturbances, aggressiveness, frustration due to lack of communication.
- Palatal anomalies: cleft palate, bifid uvula, or high-arched palate.
- Dental anomalies: prominent upper incisors, other anomalies.

S *evere speech and language anomalies*
A *bnormalities of the palate*
T *eeth anomalies*
B *ehavioral issues with or without bone or brain anomalies*
2 *onset before age 2*

System	Recommended Initial Evaluations and Treatments
Genetic	**Initial:** *SATB2* sequencing with deletion/duplication analysis/array CGH. **Treatment:** Provide genetic counseling.
Neurological	**Initial:** • Consider brain MRI and EEG at baseline if seizures present. • Physical therapy evaluation. • Occupational therapy evaluation. • Consider rehabilitation referral. **Treatment:** • Treat seizures if present, neurosurgery referral if enlarged ventricles present. • Physical and occupational therapies. • Orthotics or mechanical aids.
Psychological & Psychiatric	**Initial:** Developmental evaluation, neuropsychological evaluation. **Treatment:** Treat behavioral issues if needed.
Speech & Language	**Initial:** Speech & language evaluation. **Treatment:** • Intensive speech and language therapy with frequent, highly structured sessions aimed at speech apraxia. • Augmentative and alternative communication devices.
Craniofacial	**Initial:** Evaluate for cleft palate/submucous cleft palate. **Treatment:** Cleft palate/submucous cleft palate repair.
Gastrointestinal	**Initial:** Assess feeding. **Treatment:** Special nipples/bottle for cleft palate, feeding education.
Musculoskeletal	**Initial:** • Consider bone mineralization evaluation (bone density), from age 5 or sooner if indicated (fractures). • Consider referral to orthopedics. **Treatment:** Optimize bone mineralization as needed.
Dental	**Initial:** Dental evaluation. **Treatment:** Dental/orthodontic management, consider referral to specialized center.
Ophthalmology	**Initial:** Baseline ophthalmology exam. **Treatment:** Refractive errors correction/strabismus surgery.

Diagnosis

Established in a proband by detection of one of the following:

- heterozygous intragenic *SATB2* pathogenic variant.
- heterozygous non-recurrent deletion at 2q33.1 that includes *SATB2*.
- intragenic deletion or duplication of *SATB2* detectable by chromosomal microarray analysis (CMA).
- chromosomal translocation with a 2q33.1 breakpoint that disrupts *SATB2*. Molecular genetic testing approaches can include a combination of CMA, a multi-gene panel, comprehensive genome sequencing, and exome array.

Resources

For additional medical and scientific information, as well as registry information, please visit **www.satb2gene.com.**

For more information about the SATB2 Gene Foundation, please visit **www.satb2gene.org.**

Closed Facebook group for families to connect, search for **"SATB2 Syndrome (2q33.1)".**

Additional Resources:

SATB2-Associated Syndrome - GeneReviews®:
www.ncbi.nlm.nih.gov/books/NBK458647

Natural history of *SATB2*-associated syndrome:
www.ncbi.nlm.nih.gov/pubmed/29436146

sat b2 gene foundation

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