

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.

Severe speech and language anomalies
Abnormalities of the palate
Tooth anomalies
Behavioral issues with or without bone or brain anomalies
2 onset before age 2

System	Recommended Initial Evaluations and Treatments
Genetic	Initial: <i>SATB2</i> sequencing with deletion/duplication analysis/array CGH. Treatment: Provide genetic counseling.
Neurological	Initial: <ul style="list-style-type: none">• Consider brain MRI and EEG at baseline if seizures present.• Physical therapy evaluation.• Occupational therapy evaluation.• Consider rehabilitation referral. Treatment: <ul style="list-style-type: none">• 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: <ul style="list-style-type: none">• 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: <ul style="list-style-type: none">• 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

