SATB2-Associated Syndrome

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

FOR PATIENTS & FAMILIES

Features

Individuals with this condition are generally very kind and happy, with the most beautiful smiles you may have ever seen. However, this syndrome significantly affects many areas of development, including speech (absent speech in many or significantly delayed/affected speech), cognition (intellectual disability), fine motor skills, and gross motor skills. They often have palatal abnormalities, including cleft palate or high arched palate, and dental issues, including large teeth. They may also have seizures, sleep difficulties, and growth delays. As the children get older, significant behavioral issues can develop, as well as other medical concerns, such as low bone density.

- **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 (to be shared with your medical team)
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 (broken bones). • 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

Genetic tests detect alterations of the *SATB2* gene. Alterations of the *SATB2* gene may include misspellings (mutations), missing pieces (deletions), or extra pieces (duplications). The SATB2 protein plays an important role in brain and facial development. Most alterations of the *SATB2* gene are new in that individual (known as de novo), and the chance of it happening again is low. Your geneticist can provide more information about this.

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.**

To connect with other families, search for the closed Facebook group **"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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