

Dental Findings for SATB2 Associated Syndrome

People with SATB2 may have dental abnormalities.

These can include:



- Larger than average teeth
- Increased Drooling (called Sialorrhea)
- Delays in when their teeth come through
- Grinding or gnashing of teeth (known as Bruxism)
- Missing second Premolars
- Lower tooth root development in permanent molars

Since most of the people identified with SATB2 are either children or adolescents, there is little knowledge for dental development and maintenance into adulthood

When Visiting a Dentist

Studies have found that people with SATB2 may experience stress, fear, anxiety, irritability, and be uncooperative during a dental appointment.

Unfamiliarity with the environment and the length of the appointment were found to be stressful to the patient. Seeing the same dentist in the same office or rooms may be helpful in building familiarity and comfort.

Families reported children with SATB2 having a higher pain tolerance. For example parents said their child had been injured in the past and not shown signs of discomfort. Carers may consider informing dental staff of this prior to dental procedures.

Recommendations

Carers for people with SATB2 may consider seeking dental care from a medical team experienced with complex behaviours, medical histories and "craniofacial abnormalities" (aka differences in head and facial bones such as cleft lip).



These types of services may be available at hospitals or universities



Explaining SATB2 Project is the collaborative work of Rachel Muir MC-GENCOUN MHGSA and Dalal Baumgartner, SATB2 Connect.

Reference Thomason A, Pankey E, Nutt B, Caffrey AR, Zarate YA. Speech, language, and feeding phenotypes of SATB2-associated syndrome. Clin Genet. 2019 Dec;96(6):485-492. doi: 10.1111/cge.13619. Epub 2019 Aug 12. PMID: 31392730.

Speech and Language with SATB2 Associated Syndrome

Verbal communication in people with SATB2 is quite limited. A 2019 study found that 72% of participants with SATB2-Associated Syndrome were mostly non-verbal. The study investigated speech and language in 61 individuals with SATB2-Associated Syndrome. Looking at vocabulary at different ages.

Kids under 3 years old spoke an average of 2.5 words

Kids aged 3-6 years old spoke an average of 6.2 words

Kids aged 6-12 years old spoke an average of 13.9 words

Young people aged 12-29 years old spoke an average of 10.2 words

Recommendations

- Doctors and Careers may consider Non-verbal IQ testing to help with education planning
- "Augmentative and Alternative Communication" (AAC) should be considered to help people with SATB2 express themselves
- At least 90-120 minutes of intensive speech therapy per week..

What is ACC?

AAC are the ways a person communicates besides talking.

AAC may involve physical aids or tools that help support a person who has difficulties communicating with speech.



Information for Clinicians:

For Speech: Should use a motor-planning approach, and not wait until the child has enough verbal agility to confirm an apraxia diagnosis with formal testing. Rapid Syllable Transitions treatment and the Nuffield Dyspraxia Programme-3 showed comparable gains in word accuracy after 1 month treatment delivered in intensive sessions 4 days a week.

For Hypernasality: If Hypernasality is observed a referral to an otolaryngologist for velopharyngeal insufficiency assessment



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Feeding and Eating Challenges with SATB2 Associated Syndrome

Majority of individuals with SATB2 Associated Syndrome experience feeding difficulties during eating or drinking



These difficulties can include problems with sucking, swallowing, and chewing. Some individuals with SAS may also experience issues such:

- Excessive drooling
- Oropharyngeal dysphagia (choking/gagging, overstuffing/eating),
- Acid reflux
- Aspiration (accidentally inhaling food or liquid into their lungs) .

Recommendations

- Caregivers may consider feeding therapy to help address challenges with eating.
- Caregivers should monitor for signs of "aspiration" (This is when something enters the airway or lungs by accident) during feeding.
- If a SATB2 individual has many challenges during swallowing, a "formal swallow study" may be needed to see if there are any strategies to help protect the individuals respiratory health.

It is important to note that the specific feeding-related issues can vary from person to person, depending on the severity of the syndrome and other factors.

Families and carers may benefit from addressing feeding difficulties through a multidisciplinary approach involving healthcare professionals such as speech-language pathologists, occupational therapists, and dieticians.



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Sleep Challenges with SATB2 Associated Syndrome

Individuals with SATB2-Associated Syndrome may face challenges with Sleep.

A paper published in 2020 found that half of the participants aged 5-15 years old with SAS had at least 1 sleep disorder.

Sleep Challenges

The sleep challenges faced by individuals with SAS may include:



- Difficulties falling and staying asleep
- Sleep breathing disorders (Sleep apnoea)
- Sleep-wake transition disorders (difficulty when waking up)
- Excessive sweating while sleeping (Hyperhidrosis)

Recommendations



- If caregivers suspect sleep issues they may consider asking for an overnight ECG evaluation, also known as a Sleep ECG
- Doctors may consider a low-dose melatonin supplement to assist with mild to moderate sleep issues
- Other medications that have been successfully used for individuals with SAS facing sleep challenges include Alpha agonist medications such as “Guanfacine” or “Clonidine” which can be helpful for both sleep initiation problems and daytime hyperactivity. Low doses of the antidepressant “Trazodone” and antiepileptic medication “Gabapentin” have also been successful in some individuals.



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Reference Cotton AP, Gokarakonda S, Caffrey AR, Zarate YA, Kumar N. Behavioral phenotype and sleep problems in SATB2-associated syndrome. *Dev Med Child Neurol.* 2020 Jul;62(7):827-832. doi: 10.1111/dmcn.14330. Epub 2019 Aug 16. PMID: 31420882.