

Health care professionals,

Arkansas Children's Hospital has been fortunate to host two International *SATB2*-associated syndrome (SAS) conferences for children and their families, clinicians, and researchers. Individuals participating in the SAS Multidisciplinary Clinic were seen by various professionals from multiple departments including Arkansas Children's Genetics, ENT, Plastics, Dental, Orthodontics, Neuropsychology, Speech Pathology, and Audiology. Thirty-seven individuals and their families participated in the first conference in 2017. Thirty-three individuals and their families participated in the second conference in 2018.

Our team of speech-language pathologists gathered data about children and young adults who have SAS through subtests of standardized tests, formal screening tools, rating scales, clinical observations, and caregiver report. Our findings yielded the following information:

- The majority of individuals (72%) were primarily non-verbal communicators. For the purposes of our research, we defined primarily non-verbal communicators as those who reportedly use less than 10 spoken words in a day and use other means of communication more often than spoken language.
- Children with SAS used the following as their reported primary communication modality in order of frequency: gestures (38%), verbal communication (23%), Augmentative and Alternative Communication (AAC) device (13%), gestures combined with signs (11%), gestures combined with AAC device (8%), gestures combined with signs and AAC device (5%), and non-word vocalizations (2%).
- Auditory comprehension is significantly impacted in children who have SAS. In one standardized measure of receptive vocabulary skills, the majority of children who have SAS scored in the severely delayed range, with a few children showing moderate or mild range delays. Most children with SAS exhibited single word receptive vocabulary skills of a child younger than 3-years of age. Receptive vocabulary appears to gradually improve across childhood for most individuals who have SAS. The gap between the receptive vocabulary skills of children who have SAS and typically-developing children increases with age.
- Expressive language skills, especially spoken language is also a marked area of concern for individuals who have SAS. The average number of words of expressive vocabulary among all children studied in this cohort was 28.6, although that figure includes three children with markedly higher spoken language abilities than other children with SAS. The average number of words reported in the individual's expressive vocabulary, (minus the three outlier individuals with expressive lexicons over 200 and 500 words) was 2.5 words for children under 3-years of age, 6.2 words for children 3-years 1-month through 6-years months, 13.9 words for children 6-years 1-month through 12-years, and 10.2 words for individuals 12-years 1-month through 29-years of age. By caregiver report, 16% of individuals with SAS use no words at all. Spoken vocabulary growth does not appear to necessarily improve as a function of age; nor do all children

with SAS eventually develop spoken language skills. The average reported utterance length for children who were considered primarily verbal communicators was 2.39 words, though that utterance length drops to 1.67 words per utterance without a high-functioning outlier individual.

- Communicating with unfamiliar partners is a problem for most children with SAS. Using a rating scale that considers a person's overall abilities using any communication method, most individuals exhibited either inconsistent or effective sending and receiving of messages with familiar partners.
- Essentially all children with SAS who had enough verbal ability to participate in formal screening or criterion referenced measures displayed characteristics consistent with childhood apraxia of speech.
- Children with SAS are at higher risk than the general population to have hypernasal resonance. Some of children with hypernasal resonance had no history of cleft palate; thus, the problem could be attributed to apraxic palatal movement in some cases.
- Feeding problems, specifically difficulty with chewing, overstuffing mouths, and pharyngeal dysphagia were common in children with SAS. Pharyngeal dysphagia was reported to have resolved in most individuals. Sialorrhea is also a common problem but reportedly resolves in the majority of individuals by 4-years of age.

There is obvious individual variability among children who have SAS. Based on our findings from the 61 different children we were able to assess, **our team recommends the following considerations** for clinicians and educational team regarding feeding and communication:

- At least 90-120 minutes of therapy is warranted per week but individuals often benefit from even more time to address all domains impacted by SAS, though many children may need more treatment time.
- Receptive vocabulary skills should be continually targeted for children who have SAS, as these skills appear to have potential for continued growth throughout childhood.
- Individuals with SAS should be considered for Augmentative and Alternative Communication (which includes signing, picture-exchange systems, low tech picture boards, and high tech voice-generating devices) to enhance their expressive abilities as an ongoing part of treatment. AAC use should be targeted with a progressively growing circle communication partners.
- Non-verbal IQ testing should be considered for educational planning purposes, as verbal abilities are generally a weakness for children who have SAS.
- Given our findings to date, apraxia of speech can essentially be assumed to present if a child with SAS develops verbal speaking abilities. Children with enough verbal speaking ability to complete testing should be assessed for apraxia of speech. Clinicians working with a child with SAS who is developing verbal abilities should target speech production with a treatment approach to address childhood apraxia of speech.

- Monitor resonance for hypernasality; refer to ENT for nasendoscopy to rule out velopharyngeal insufficiency if hypernasality perceived. Hypernasal resonance could be due to a history of apraxia of speech, cleft palate, or a combination of both.
- Consider feeding therapy for children who have trouble chewing and pocketing food.
- Monitor for signs of aspiration, and consider a formal swallow study if pharyngeal dysphagia is suspected.
- Consider an ENT referral for sialorrhea (drooling) if persistent after age 4-years of age.
- The incidence of autism is higher in children with SAS than in the general population. Signs of autism such as reduced joint attention, poor eye-contact, and self-stimulating behaviors should not be assumed to be part of the speech presentation of SAS, rather a potential co-occurring autism spectrum disorder that should concurrently be addressed in treatment.

We are happy to provide further clarification and consult as needed regarding recommendations for your child's plan of care.

Sincerely,



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