

NOVEMBER 2021

**SATB2-ASSOCIATED SYNDROME**

# **SPEECH AND LANGUAGE SURVEY**

sat  
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# SPEECH, LANGUAGE, AND SAS

In order to support families with a SATB2-Associated Syndrome (SAS) diagnosis, in June 2021 an independent study supported by the SATB2 Gene Foundation was launched in order to better understand the relationship between a diagnosis of SAS and interventions targeted at speech and language. With only approximately 500 known cases in the world, SAS is incredibly rare. Particularly for families new to the diagnosis, it can be very challenging to understand how to best support someone with the condition and where to focus energy.

The results of the survey cannot provide families with a roadmap of what to do, but it can provide insight into what others have done. It can be reassuring to know the paths of others and where success has been found. The results of the survey demonstrate a great diversity across individuals with SAS in terms of expressive communication and primary mode of communication. The results also show that most individuals with SAS are multi-modal communicators (using many different ways to communicate).

## SURVEY CONTEXT

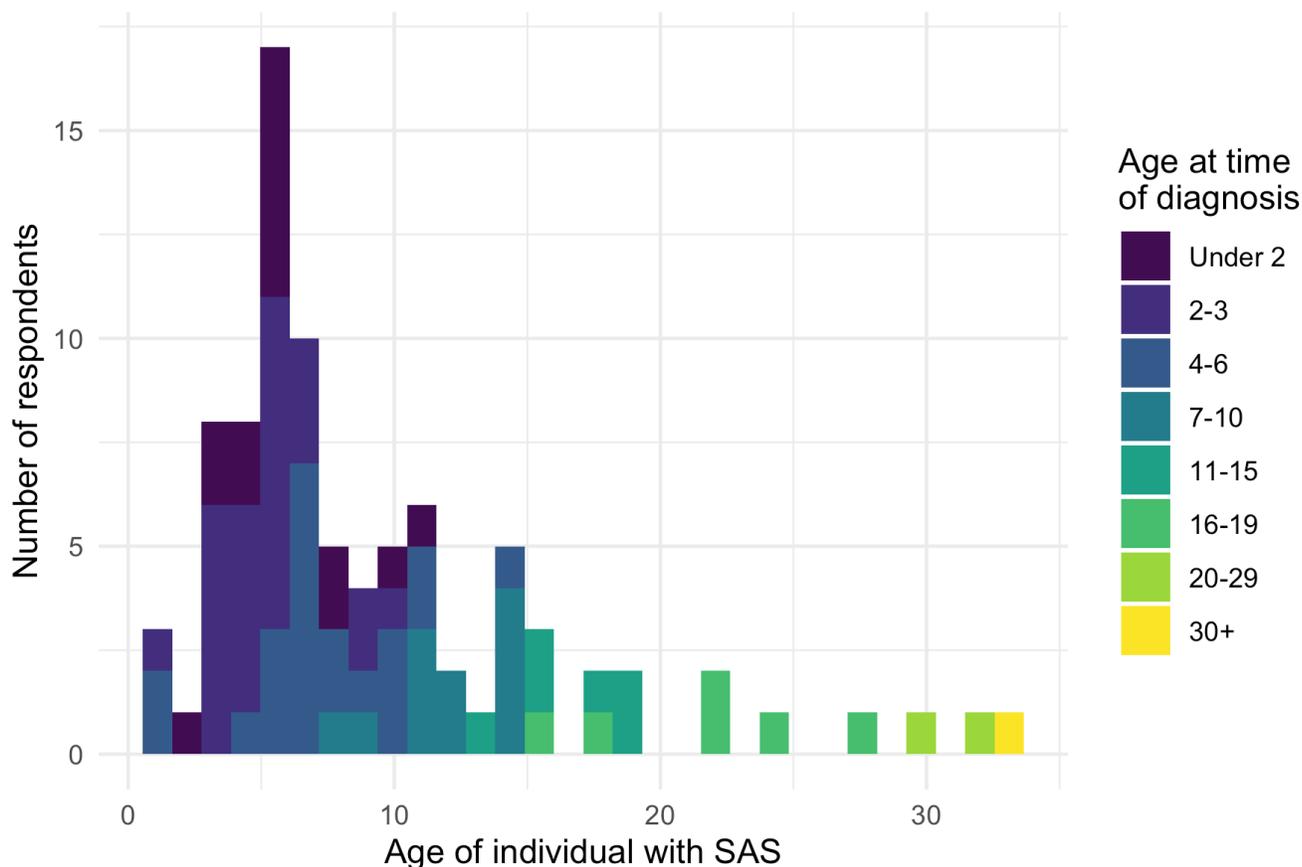
The survey was first shared on June 7, 2021 and remained available for respondents until June 30, 2021. It was written in English and google translated into 5 languages (Dutch, French, German, Portuguese, and Spanish). 89 people completed the survey and 50 began but did not complete the survey. Several people disclosed that they had started the survey and when they could not return to it, restarted the survey a second time in order to complete it. Therefore, any incomplete surveys have not been included in the results below.

We report all results without modifying any answers, which leads to a small number of likely mistakes. For example, in the graph below, several respondents gave an age at time of diagnosis that was older than the individual's current age (there are two individuals who are 1, but list 4-6 as the age at time of diagnosis). These seem to be cases where a small number of respondents misunderstood the questions, which may be attributable to mistranslations.

## PROFILE OF INDIVIDUALS

Survey responses represented individuals whose age ranged from under 2 to 30+. However, the majority of individuals represented (63%) were under 10 years of age. Further, most individuals received a diagnosis before the age of 6 (70%). The identification of SAS through genetic testing is relatively recent, meaning that there is presently a close link between current age and age at time of diagnosis. Future iterations of this survey will likely find greater variation here. As the known population with SAS grows, it may be possible to start mapping outcomes correlated with age of diagnosis.

43% of respondents reside in the United States, with 8% coming from France, 6% coming from Australia, Canada, Spain and the UK, and 4 or fewer responses from remaining countries.<sup>1</sup> 62% of respondents indicated that the individual with SAS for whom they were

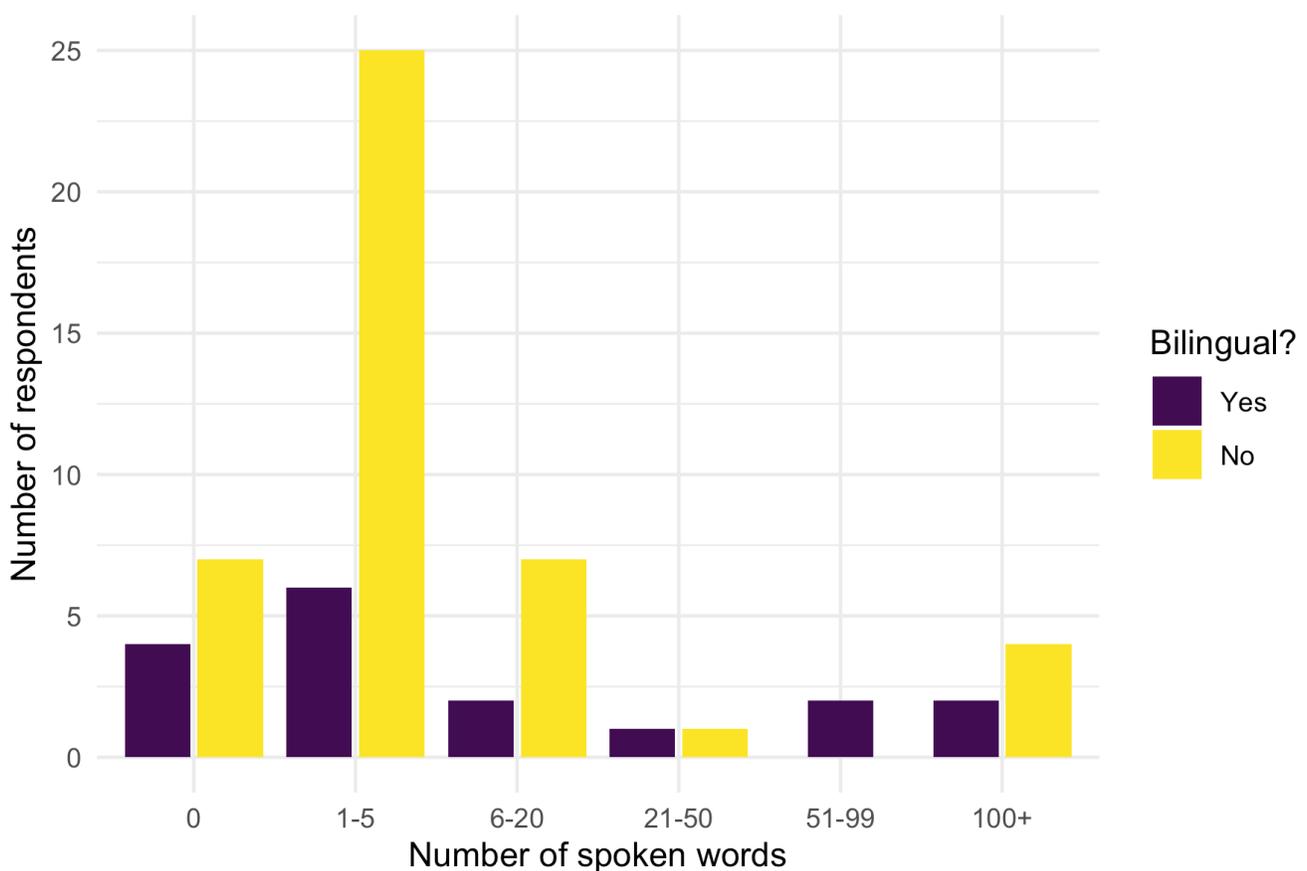


<sup>1</sup> Brazil, Sweden, Austria, Belgium, Denmark, Ecuador, Netherlands, Germany, Lebanon, New Zealand, Poland, Serbia, Slovenia, and Turkey were all represented.

responding spoke English (with French being the second most common language with 7% of respondents).

## BILINGUALISM

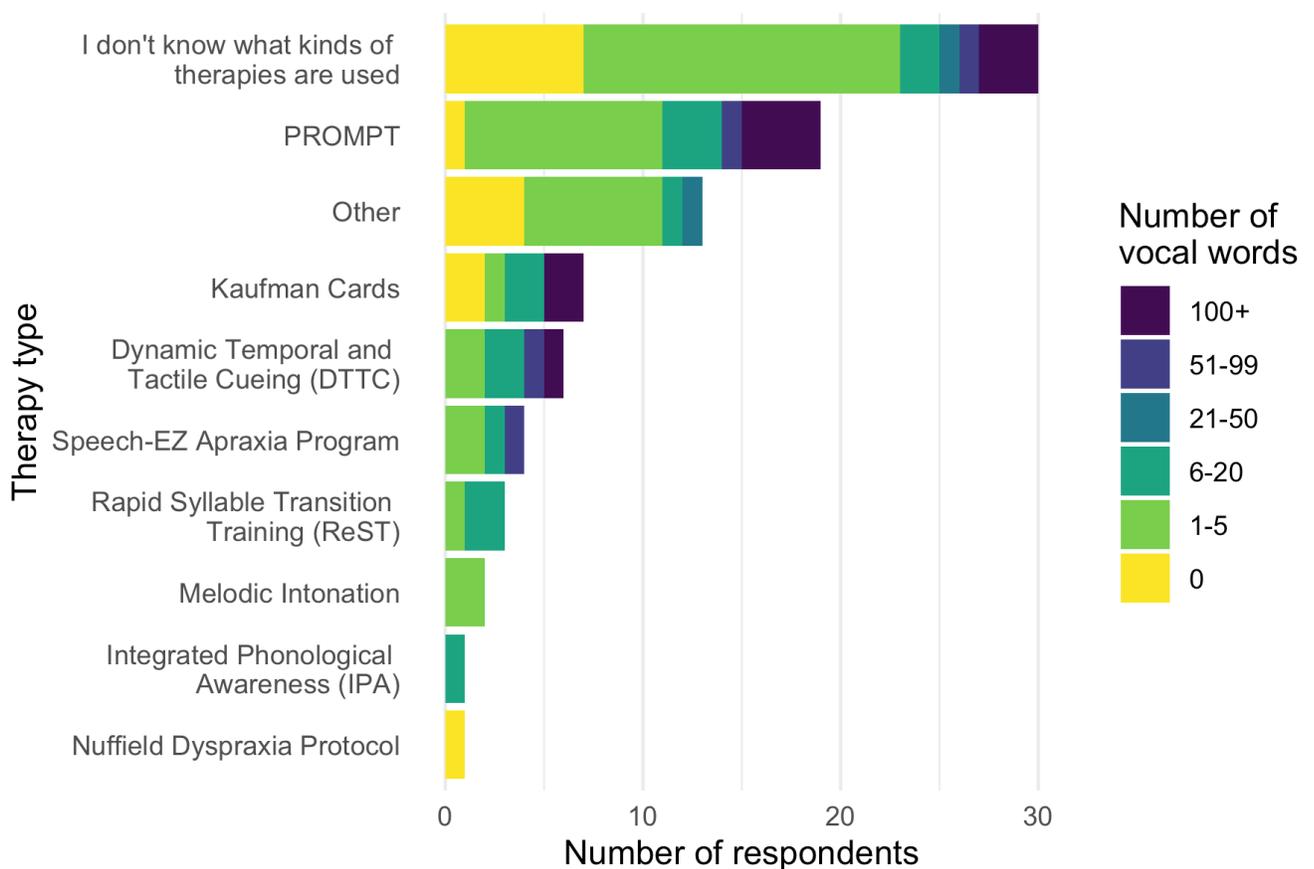
25% of respondents indicated that their dependent with SAS was bilingual. There doesn't appear to be a relationship between the number of spoken or expressed (in AAC) words that an individual has and whether or not they are bilingual. This is true even if we exclude individuals who wrote in an answer of sign language or Makaton as their second language. Second languages are diverse and often from different language families than the individual's primary language. Some respondents included sign languages as a second language, however many respondents who later responded that the individual with SAS communicated in sign language did not list their dependent as bilingual.



## SAS AND VERBAL SPEECH

### TYPES OF SPEECH THERAPY FOR CHILDHOOD APRAXIA OF SPEECH

The limited research on treatments for Childhood Apraxia of Speech (CAS), including for those with SAS, has emphasized the importance of motor-planning based approaches. The survey allowed respondents to select multiple therapies in response to the question of which speech therapy programs are used.



PROMPT was the most common therapy program reported to be used, which is perhaps tied to previous advice on treatment for SAS. The second most reported category was other, which appears linked to alternative therapies used outside English-speaking

countries.<sup>2</sup> Perhaps most interesting was the number of parents or caregivers who were unaware of the type of therapy used by their speech therapist. This may mean that there is room for clearer direction or advice to advocate for therapies specifically targeting childhood apraxia of speech for individuals with SAS.

## **TIME SPENT**

Families reported variable amounts of time spent on speech therapy both with a speech therapist and with others. The majority of respondents spend more than two hours per

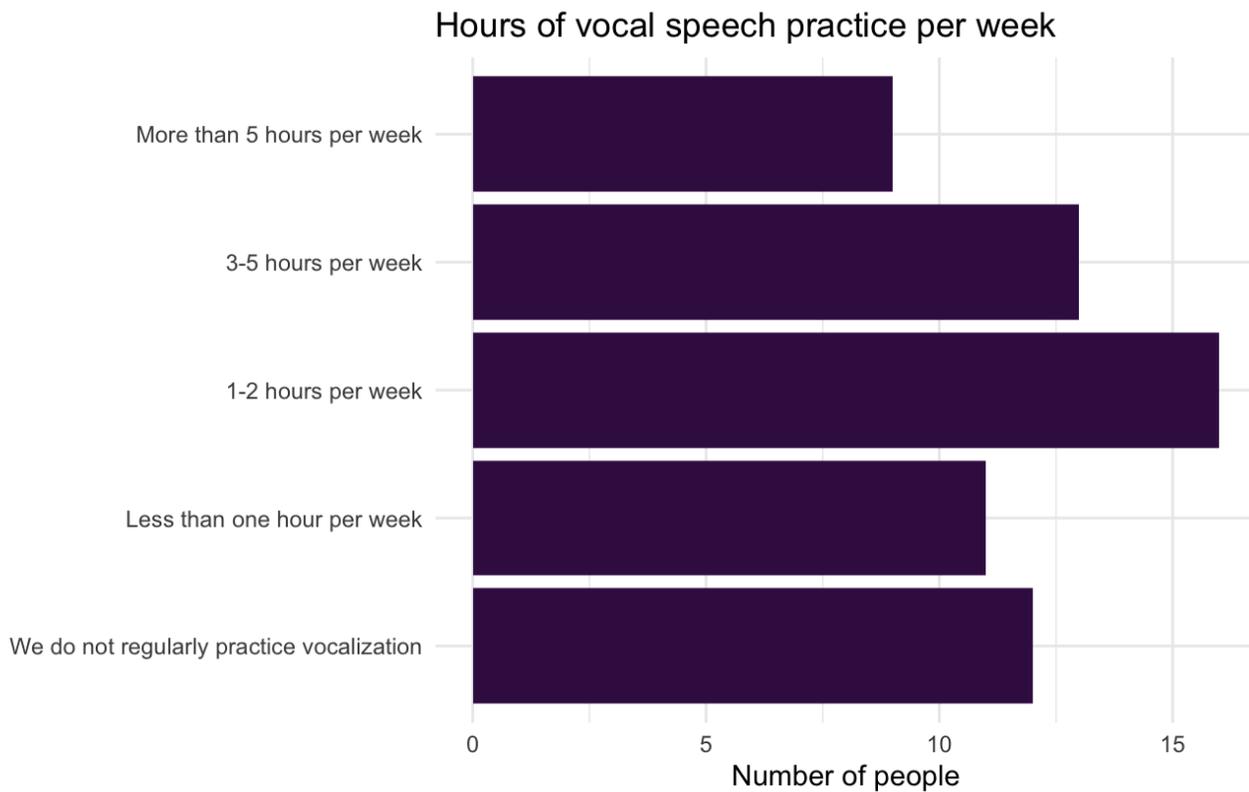


month with a speech language pathologist, with over 1/3 of respondents spending between 3-4 hours per month.

In contrast to time with a speech therapist, where most individuals spent 3-8 hours per month, there was much greater variability in the amount of time spent on practicing

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<sup>2</sup> The "other" category allowed respondents to add in their own therapy options, which included: Bilder, Laut Gesten, SMOG (spreken met ondersteuning van gebaren), FloorTime, oral motor and PECs cards.



speech outside of therapy. There are almost as many families who spend more than five hours a week practicing as the number of families who spend no time practicing.

In open-ended questions, multiple individuals mentioned the importance of having a speech language pathologist who is good at motivating an individual. Several people also discussed the importance of a therapist who has expertise with Childhood Apraxia of Speech. In a question focused on what parents wish they had done differently, multiple parents mentioned wishing they had spent more time doing speech therapy, with one specifying at least one hour per week. Similarly, multiple parents wished they had begun speech therapy earlier. One respondent mentioned the importance of being engaged during speech therapy as a parent or caregiver, to ensure that any practice done with an SLP could be replicated in a home setting.

## **SUCCESS OF SPEECH THERAPY**

Respondents had different ideas on perceived effectiveness of speech therapy focused on vocal speech (including multiple respondents who found that it had definitely not

worked, those who were unsure or had mixed feelings, and those who felt confident in its effectiveness). Many respondents cautioned that success may be more gradual than caregivers might initially expect. In some cases, a change in speech language pathologist made a difference in perceived effectiveness. Multiple families reported little progress until approximately five years of age, when, in some instances, speech seemed to “turn on.” This sudden progress was attributed to factors such as a speech summer camp, improved focus, increased motivation, and better voice control.

Parents and caregivers gave diverse advice on strategies that have supported speech production, including focusing on regular practice, with a number of strategies suggested. These strategies included having regular daily ‘speech time,’ making sure to use visual and motivating cues or prompts to effectively coax speech, and ensuring that the therapy was face-to-face. Parents noted that speech progress would stop or even decline when practice was missed for even a one-week period.

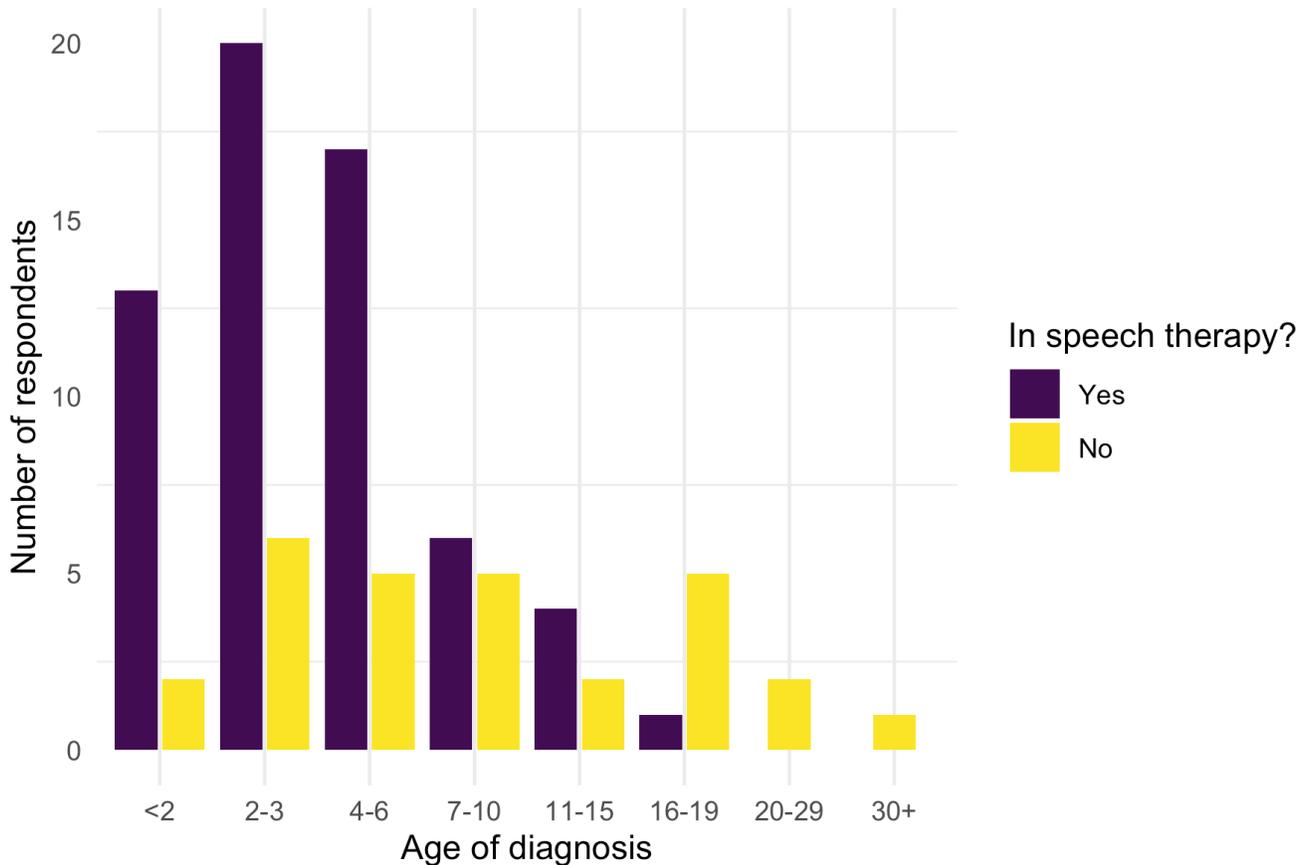
There is no clear relationship between the type of therapy that a family uses and the number of words an individual with SAS says. There seems to be high distribution of success across all therapies that address Childhood Apraxia of Speech.

One respondent mentioned that ‘success’ may not look how you expect. An individual with SAS will not produce sounds like someone without the condition. Expect early speech development to be subtle, and that it may take some focus to recognize.

Some families who had previously concentrated on spoken communication during speech therapy time, have refocused on other domains like Augmentative and Alternative Communication (ACC) or feeding issues due to a lack of success around speech. Another repeated comment was that for individual with focus or behavioural challenges, speech therapy practice is more challenging, and often speech therapy focused on vocal speech was less effective. **Of those who felt that speech therapy is not working, the amount of time dedicated to speech therapy varied dramatically.**

## INDIVIDUALS NOT IN SPEECH THERAPY

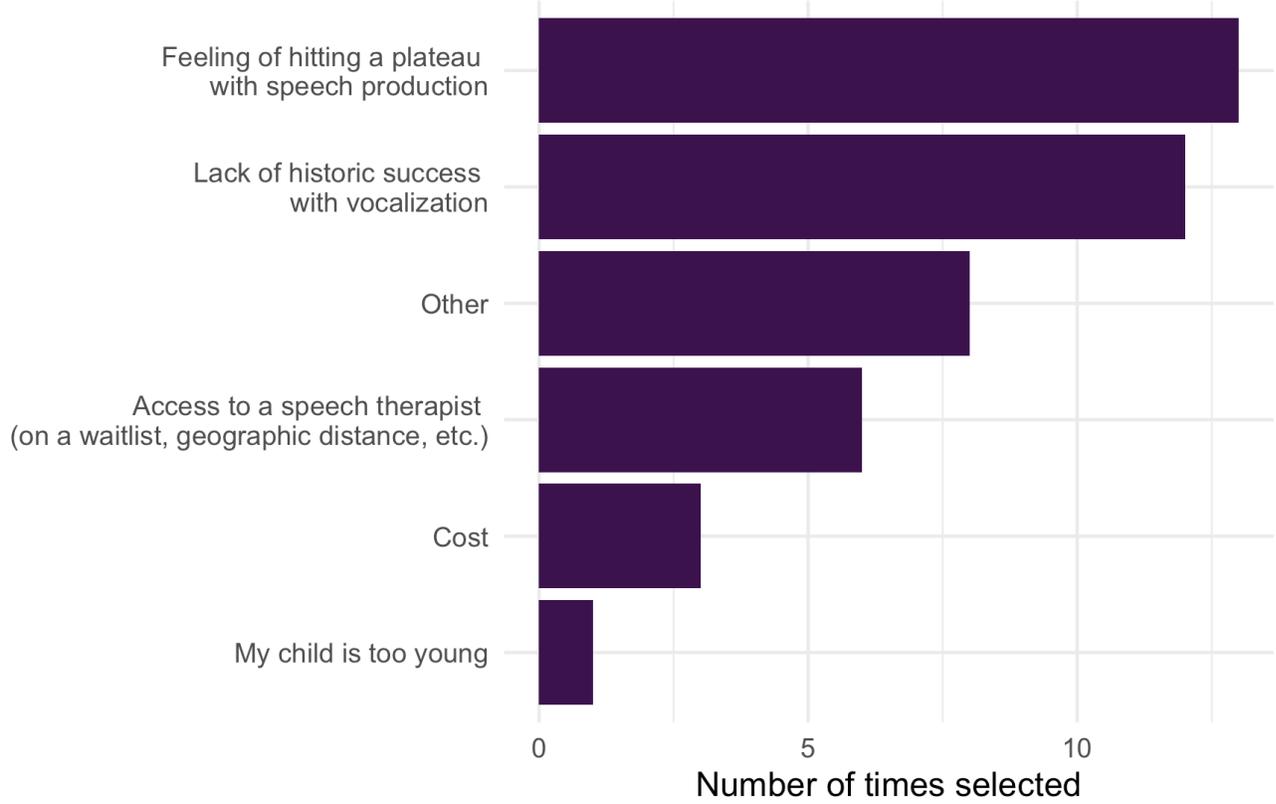
While some respondents continue with speech therapy despite not seeing much success, 31% of families indicated that they are not currently pursuing speech therapy. Individuals who are not currently in speech therapy for voice production were both older on average (14 compared to an average age of 7 for those currently pursuing speech therapy) and also had a later age of diagnosis.



The dominant reasons given for not pursuing speech therapy were a feeling of hitting a plateau or a lack of historic success. Other reasons given included COVID-19 and feeling like time was better spent focusing on communication than speech production.

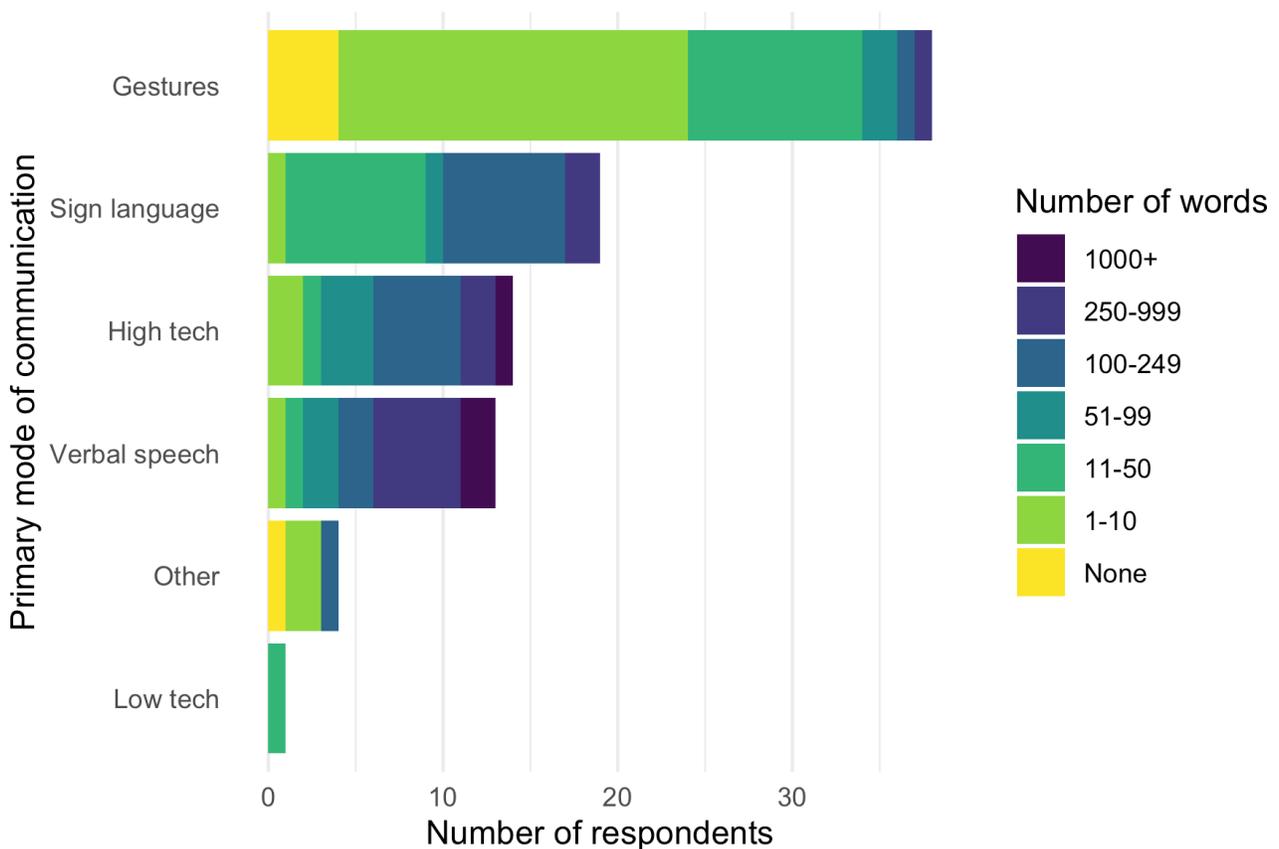
There were multiple respondents who felt that speech development moved hand-in-hand with receptive language development and AAC usage, and that success required emphasis on all three.

### Reasons for not focusing on vocalization



## SAS AND COMMUNICATION

The majority of individuals with SAS do not use spoken language as their primary mode of communication. However, many have robust communication systems and are able to express hundreds, or even thousands, of words. In general, those with the most robust communication are those whose primary mode of communication is sign language, verbal speech, or a high-tech AAC device (defined as a battery-powered device). In contrast, those who use gestures as their primary mode of communication are, in general, able to express far fewer words. The distribution across categories does not change substantially if children under age 3 are removed (with the assumption that they should be expected to have more limited expressive language at that age) and if individuals over age 18 are excluded (on the assumption that diagnosis happened later in life and therefore impacted expressive language abilities).



Respondents who chose "other" as the primary mode of communication gave responses of "screaming" (for a child under the age of 2) and "throat sounds and facial expressions."

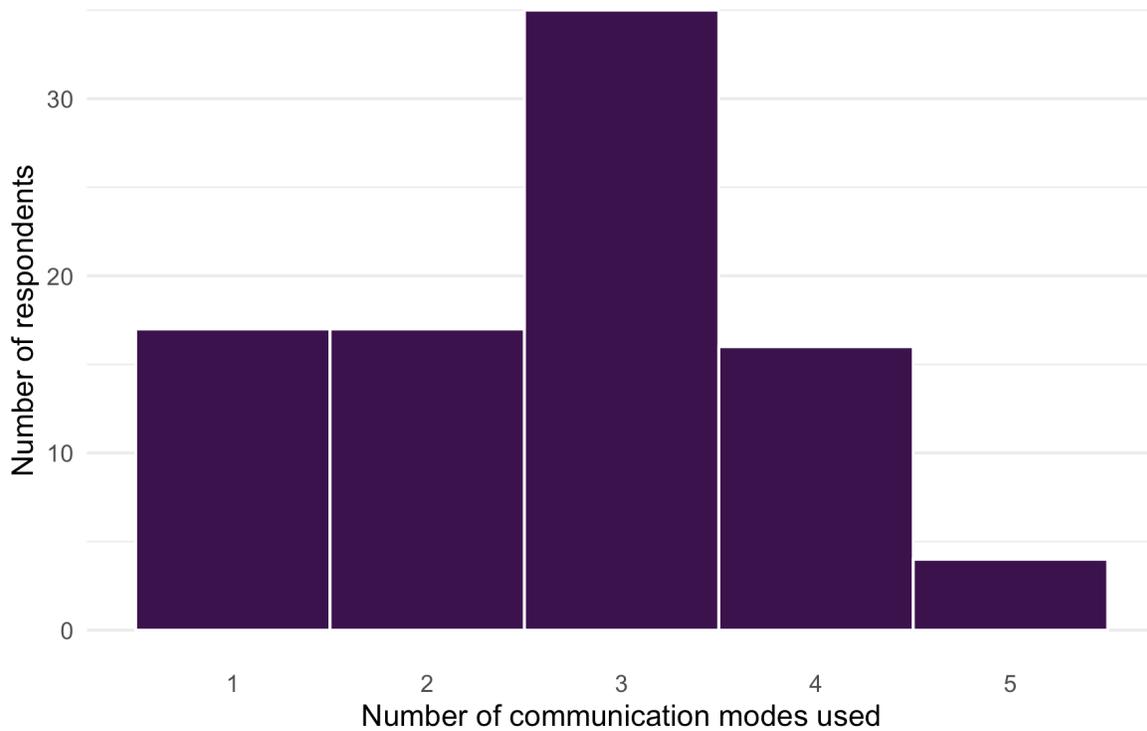
While it may be tempting to assume that families new to the diagnosis should pursue sign language, high tech AAC, or verbal speech in order to increase number of expressed words, this graph cannot be interpreted causally. Some presentations of SAS may both limit expressive language abilities as well as capacity to pursue certain modes of communication.

When asked about what they would do differently if new to the diagnosis today, many parents/caregivers reflected that they wished that they would have started signing or using a communication device earlier on, and recognized the profound positive impacts of an alternative communication outlet for their dependent. Essentially, in the words of one parent, **“I would focus on language as much as on speech.”**

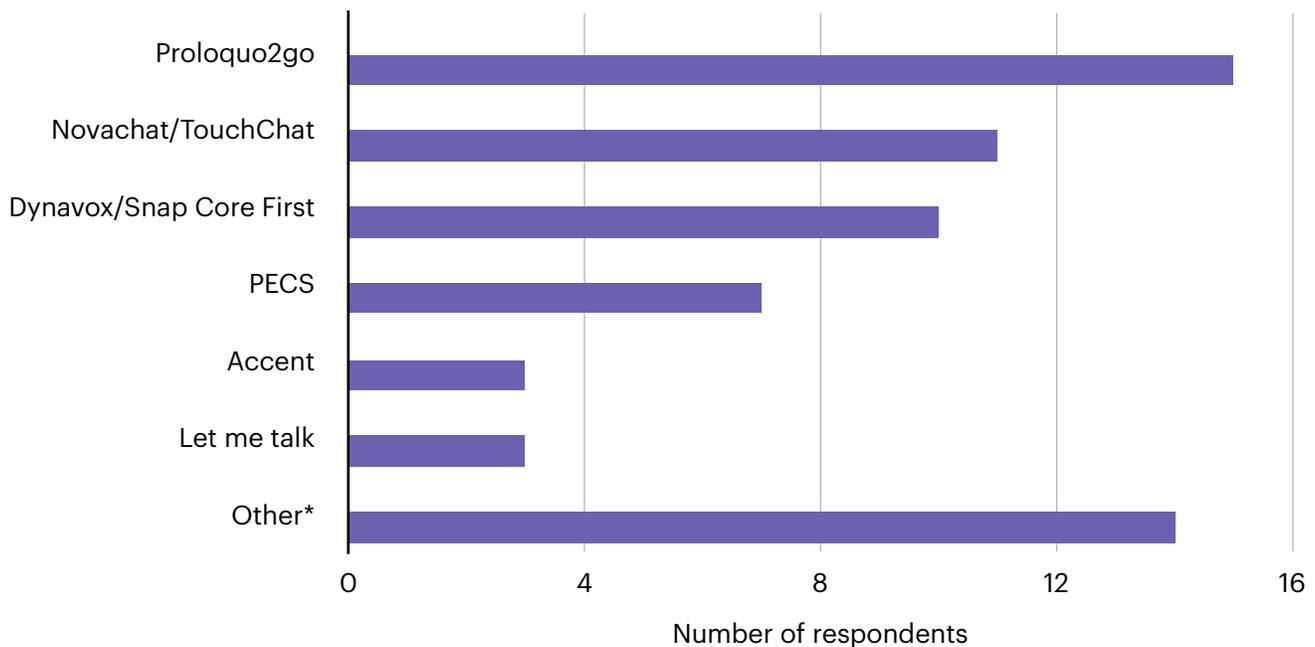
Multiple parents mentioned that sign language came most naturally of all AAC systems to their kids. However, some parents did mention that sign language was not an option because of a dislike of hands being touched, autism, or more severe global apraxia. Many of the families who use sign language still ultimately also pursued other forms of AAC so that there were communication options for when a sign was not known, or with communication partners who did not sign. One limitation of signing that was noted was that because of limited hand dexterity due to motor planning issues, some individuals with SAS had many signs that were not intelligible to new communication partners.

For families who went directly to a high/low-tech AAC device, many trialed multiple options before landing with the system they currently use. Others mentioned being limited by cost or what was available through the school system. Still others felt limited by the maturity level of their dependent and ability to use an AAC system. Lastly, some parents mentioned the challenges of getting some individuals to implement an AAC system as well as the time and effort required for modelling, training communication partners on it, and collaboration required.

Most individuals with SAS are multimodal communicators, with 70 respondents indicating some communication happened through gestures, 53 through sign language or a high-tech AAC device, 18 through a low-tech AAC device (communication equipment without a battery), and 30 indicating communication occurs using spoken language. On average, individuals use 2.7 different modes of communication, with only 17% of respondents relying on a single mode of communication.



There was great diversity across AAC system used, with the most popular systems being Proloquo2go and Novachat/Touchchat. Responses which were selected by two or fewer respondents were combined into an “other” category in the graph below. Answers in the other category included Wego7, Flip N Talk, Metatalk, Auticiel, Coughdrop, Predictable, Nikitalk, Widget Go, AVAZ, Tellus Mind Express, and GoTalk express.



## CONCLUSION

While the number of individuals with SATB2-Associated Syndrome remains small, the communication and language tools that families rely on is rather diverse. There are a couple of take-aways for families that may be of particular interest:

- \* There does not appear to be a relationship between number of words communicated and bilingualism. This is consistent with evidence that bilingualism doesn't impede language acquisition.<sup>3</sup>
- \* PROMPT therapy was the most popular of known speech therapy techniques relied on by families.
- \* By far, the majority of families do not know what kind of speech therapy techniques are being used by speech therapists. This may mean that more work should be explored to support parents in advocating for more evidence-based treatments for Childhood Apraxia of Speech.
- \* There does not appear to be a direct relationship between amount of time practicing spoken communication and number of spoken words.

For any questions or comments regarding this report, please reach out to  
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<sup>3</sup> See, for example: <https://leader.pubs.asha.org/doi/full/10.1044/leader.FTR1.11102006.6>