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Speech, Language, Hearing, and Otopathology Results From the International Smith–Magenis Syndrome Patient Registry

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ABSTRACT

Purpose: Smith–Magenis syndrome (SMS), a rare, genetically linked complex developmental disorder caused by a deletion or mutation within chromosome 17p11.2, is associated with delays in speech-language development, otopathology, and hearing loss, yet previous studies lack comprehensive descriptions of hearing and communication profiles. Here, analyses of patient registry data expand what is known about speech, language, hearing, and otopathology in SMS.
Method: International speech-language and hearing registry survey data for 82 individuals with SMS were analyzed using descriptive and inferential statistics. Hearing loss, history of otitis media and pressure equalization (PE) tubes, communication mode, expressive/receptive language, and vocal quality were analyzed for all subjects and subjects grouped by age. Statistical methods included descriptive statistics and Pearson's chi-square tests of independence to test for differences between age groups for each variable of interest. Association analyses included Pearson's correlations.
Results: Hearing and otological analyses revealed that 35% of subjects had hearing loss, 66% had a history of otitis media, and 62% had received PE tubes. Speech-language analyses revealed that 60% of subjects communicated using speech, 79% began speaking words at/after 24 months of age, 92% combined words at/after 36 months, and 41% used sign language before speech. There was a significant association between the age that first words were spoken and the age that PE tubes were first placed. Communication strengths noted in more than 40% of subjects included social interest, humor, and memory for people, past events, and/or facts.
Conclusions: Significant delays and impairment in speech-language were common, but the majority of those with SMS communicated using speech by age 6 years. Age was a significant factor for some aspects of hearing loss and communication. Neither hearing loss nor otitis media exacerbated language impairment. These results confirm and extend previous findings about the nature of speech, language, hearing, and otopathology in those with SMS.

Smith–Magenis syndrome (SMS) is a rare, genetically linked complex developmental disorder typically caused by a deletion within chromosome 17p11.2 (Smith et al., 1986). The estimated global incidence of SMS is 1:15,000–25,000 births (Elsea & Girirajan, 2008; Greenberg et al., 1991). SMS affects multiple body systems and includes congenital anomalies, intellectual impairment, speech and motor delays, sleep disturbances (for review, c.f. Smith et al., 2022). The behavioral phenotype of SMS includes self-injurious behaviors, aggression, temper tantrums, impulsivity, outbursts, repetitive behaviors, attention deficits, and attention-seeking behaviors (Greenberg et al., 1996; Smith et al., 1986, 1998). Much of the previous research primarily focused on genetics (e.g., Greenberg et al., 1996; Slager et al., 2003; Vlangos et al., 2005), description of the

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general phenotype (i.e., characteristics; e.g., Girirajan et al., 2006; Martin et al., 2006; Smith et al., 1998), and description and treatment of the sleep disturbance (e.g., De Leersnyder et al., 2001; Potocki et al., 2000; Shayota & Elsea, 2019). Research focused on speech-language, and hearing is limited (e.g., Brendal et al., 2017; Hidalgo-De la Guía et al., 2020; Wolters et al., 2009). The current study aimed to address this gap in the literature by focusing on hearing, otopathology, speech, language, and communication profiles of individuals with SMS. Addressing this gap will provide more information about the communication phenotype of SMS, informing future research as well as medical, clinical, and educational practice.

Early SMS research identified conductive and sensorineural hearing loss in 66% of cases (six of nine individuals in this initial study; Smith et al., 1986). Later studies of hearing in SMS reported the prevalence of hearing loss ranging from as low as 48% of cases (Greenberg et al., 1996) to as high as 62%–68% (Edelman et al., 2007; Gamba et al., 2011; Potocki et al., 2003). Conductive hearing loss for younger children with SMS appeared to be related to otitis media (Brendal et al., 2017; Greenberg et al., 1996). Sensorineural loss was more common in older individuals with SMS (Brendal et al., 2017). Sensorineural hearing loss has also been associated with congenital abnormalities in SMS (Greenberg et al., 1996). A more recent longitudinal and cross-sectional study of 133 individuals with SMS investigated the auditory phenotype of this syndrome (Brendal et al., 2017). In this study, longitudinal hearing sensitivity in the worse-hearing ear was evaluated with four or more ear-specific audiograms spanning at least 2 years. The results revealed that hearing loss ranged from mild to severe in 72% of subjects across all age groups. Sensorineural hearing loss occurred most often in subjects in ages 11–49 years. Conductive hearing loss affected approximately 35% of ears and was more prevalent in those ages 1–10 years. Longitudinal analyses revealed fluctuating hearing over time with a tendency for closure of air–bone gaps and progression of the sensory hearing loss component (Brendal et al., 2017).

Many patients with SMS have chronic or recurrent otitis media leading to the possible need for insertion of pressure equalization (PE) tubes (Elsea & Girirajan, 2008). Although otitis media and delayed speech development is widely observed in SMS (Elsea & Girirajan, 2008), it is unknown if or how otopathology (including occurrence of otitis media or insertion of PE tubes) is related to speech-language development. While one previous study reported no differences in spontaneous language performance between typically developing children with and without a history of recurrent otitis media (Casby, 2001), the link between recurrent otitis media and delayed or impaired language development is well established (Lieu et al., 2020). Previous research did not investigate if otitis media in those with SMS exacerbates existing speech-language delays and/or difficulties.

Most individuals with SMS present with mild-to-moderate cognitive disability (Greenberg et al., 1991; Martin et al., 2006). Children with SMS present with delayed speech-language development characterized by greater delays in expressive versus receptive language (Wolters et al., 2009). No previous studies focused on language profiles of adults with SMS. Children with SMS, ages 2–3 years, were found to rarely use spoken words to communicate, and some used nonverbal communication such as gestures and hand signs to communicate their needs (Wolters et al., 2009). The use of sign language (or some form of manual communication) has been noted to potentially decrease frustration and promote more positive communication behaviors, especially when speech is delayed even if hearing is intact (Elsea & Girirajan, 2008; Smith et al., 1998; Wolters et al., 2009). Speech characteristics include over 80% of adults with SMS exhibiting a deep, hoarse vocal quality (Greenberg et al., 1991; Hidalgo-De la Guía et al., 2020). While vocal polyps and nodules have also been reported (Greenberg et al., 1996), it has been argued that high laryngeal tension is most likely the cause of the hoarse vocal quality noted in this population (Hidalgo-De la Guía et al., 2020).

In-depth investigations of language and cognition in SMS compared individuals with the syndrome to age-matched typical controls. These studies revealed impairment in perceptual organization and reasoning, processing speed, working memory, verbal comprehension, vocabulary, and word reasoning (e.g., Greenberg et al., 1996; Osório et al., 2012; Udwin et al., 2001). Individuals with SMS were relatively good at forming verbal concepts, which was interpreted as reflecting relatively unimpaired long-term memory (Osório et al., 2012). This finding is consistent with other previous studies that also reported long-term memory to be a relative strength (Udwin et al., 2001).

Other studies focused on cognition and language found that deficits in receptive and expressive communication appeared to be related to IQ (Madduri et al., 2006; Martin et al., 2006). Significant impairment was found in communication, daily living skills, socialization, and adaptive behavior as measured by the Vineland Adaptive Behavior Scales (VABS; Madduri et al., 2006; Martin et al., 2006). A cross-comparison of the VABS subdomain scales revealed that socialization ability scored higher than communication ability, daily living skills, and adaptive behavior (Madduri et al., 2006; Martin et al., 2006; Udwin et al., 2001; Wolters et al., 2009). Collectively, these studies

indicate that language and communication are impaired in those with SMS with socialization ability perhaps being a relative strength.

While social skills have been reported to be a relative strength (Madduri et al., 2006; Martin et al., 2006; Udwin et al., 2001; Wolters et al., 2009), social interactions tend to be rigid and focused on specific topics with individuals who have SMS being overly demanding of the attention of their communication partners (Dykens & Smith, 1998). Given their difficulty with social skills and rigid focus on topics of interest, one SMS study aimed to determine if individuals with SMS also met the criteria for co-occurring autism spectrum disorder (ASD; Laje et al., 2010). Using different measures of autism symptomatology, different studies reported between 35% and 90% of individuals with SMS having scores in the ASD range (Laje et al., 2010; Nag et al., 2018). In addition to presenting with similar social communication deficits as those with ASD (such as rigid social interaction and focus on topics of interest), SMS is also characterized by sensory processing challenges (Hildenbrand & Smith, 2012), and many (nearly 75%) present with clinical symptoms or signs associated peripheral neuropathy (Greenberg et al., 1996). It is possible that the socialization characteristics combined with sensory processing challenges and clinical symptoms or signs associated with peripheral neuropathy contribute to co-occurring clinical diagnoses of ASD for this population. Beyond these findings, specific similarities and distinctions in social abilities and characteristics for SMS and ASD are unclear.

Aims of the Current Study

While previous research reported limited information about hearing, otopathology, speech, language, and communication in SMS, there is no thorough description of the speech-language and communication phenotype of children and adults with this syndrome. More information is needed about communication in SMS, such as the average age that children with SMS begin speaking and combining words, the percentage of individuals who communicate using speech (note, the term *speech* or *natural speech* is preferred over the outdated terms *verbal communicator* or *verbal* vs. *nonverbal*; please see Biggs et al. [2022] for an example of this more contemporary terminology), sign language (another natural form of language), or augmentative and alternative communication (AAC). Additionally, other than long-term memory and possibly socialization, other communication strengths that may be common in those with SMS have only been speculated. It also remains unknown what percent of those with SMS receive (or received) speech-language services during childhood or if adults with SMS continue to receive speech-language services. Finally, there is limited research focused on the relationship between otopathology and speech-language development in those with SMS. Consequently, educational, clinical, and medical management teams must rely on insufficient information when planning evaluation, treatment, and transition or long-term care plans. This lack of information could lead to unnecessary delays in the provision of effective early evaluation and intervention for hearing, otopathology, and/or speech-language issues.

This study aimed to address some of the existing gaps in the literature by examining the hearing, otopathology, speech, language, and communication characteristics of 82 individuals with SMS using responses from an SMS patient registry questionnaire. In this study, we include analyses on hearing, otologic health, speech-language development, and communication data. We explored age-stratified speech-language and hearing characteristics. As a result, our results provide a more in-depth description of the hearing, otopathology, speech, language, and communication phenotype of children and adults with SMS. Furthermore, while previously published data have not examined how otopathology and speech-language deficits/abilities differ with age for those with SMS, the age-stratified analyses employed here aimed to reveal between-group differences potentially associated with development, maturation, and aging.

The first specific aim of this study was to (a) describe profiles and identify age-related differences for the following: hearing loss, otopathology (history of otitis media and placement of PE tubes), early speech-language development, mode of communication, language abilities, vocal quality, communication strengths, and participation in speech-language services in a large group of individuals with SMS. The second aim was to (b) determine if hearing or otopathology profiles were associated with speech, language, and communication abilities. The third aim was to (c) determine if a hoarse vocal quality was associated with the presence of reflex/gastroesophageal reflux disease (GERD).

Method

Protocol and Patient Registry

Individuals with SMS were identified through the Smith–Magenis syndrome Patient Registry, an initiative of Parents and Researchers Interested in Smith–Magenis syndrome (PRISMS). This registry is housed and managed under a protocol approved by the Baylor College of Medicine (BCM) Institutional Review Board (IRB) with a data use agreement (DUA) for the authors at the

University of Colorado Boulder. Parents/caregivers of those with SMS (all ages) were invited to participate by responding to various questionnaires, and caregiver-provided data were collected using REDCap (Harris et al., 2009). Data for this study were collected from May 2020 through May 2021. Eligibility required that caregiver respondents completed several questionnaires within the registry and submitted genetic reports that confirmed a diagnosis of SMS.

The patient registry included several questionnaires. This study analyzed responses from the "Speech and Language Development in SMS" questionnaire (see the Appendix for full questionnaire). This questionnaire included 155 questions about speech, language, communication (e.g., age first words spoken, age words first combined, use of sign before speech, methods of communication, expressive and receptive language skills, communication strengths), hearing and otologic health (hearing loss, otitis media, and PE tube placement), education (literacy), family members, living situation, school/education, and medical health potentially related to communication status. The questions included yes/no, check all that apply, and open-response questions related to speech, language, communication, reading, writing, hearing, otopathology, and speech-language intervention. All procedures and methods were approved by the IRB at BCM and the University of Colorado Boulder.

Analytical Methods

Many of the results presented here are descriptive. Descriptive data of speech-language, communication, hearing, and otopathology characteristics are provided as mean (standard deviation) for continuous characteristics and number (percent) for categorical characteristics. When relevant, ranges (such as age range) are also presented. Percent occurrence was calculated out of the responses given for a given variable (such as hearing loss type for those with hearing loss), and nonresponses were not included in the denominator for such calculations.

For some analyses, subjects were categorized into seven age groups (ages 3–6 years, 7–10 years, 11–14 years, 15–19 years, 20–29 years, 30–44 years, and 70–80 years). These age groups were used because they align with developmental characteristics and school-age groups and because this grouping resulted in relatively similar group sizes. Developmentally, children without SMS ages 3–6 years are typically in preschool and have basic expressive and receptive language skills. Children in elementary school are typically 7–10 years old, middle school children are 11–14 years old, and high school adolescents are 15–18 or 19 years old. In the United States, individuals may continue to receive special education services through public schools until they are 22 years of age; however, we included individuals ages 20–22 years into the group of young adults ages 20–29 years. The rationale for this grouping was that educational programs for those continuing schooling after age 19 years often focus on transitioning, emphasizing vocational and independent living skills. Also, inclusion of those ages 20–22 years in the high school–age group would have resulted in that being a much larger group than the other groups and the young adult group being much smaller than the other groups. The other adults were broken up into two groups including adults (30–44 years) and older adults (70–80 years). There were no subjects ages 45–69 years in this data set, and there was only one subject in the older adult group.

For categorical variables, chi-square tests of independence were used to examine associations between the seven age groups and each variable of interest (e.g., hearing, otopathology, language). Pearson's correlations were used to assess the association between two continuous characteristics (e.g., age hearing loss first suspected, age of first PE tube placement, age first words were spoken). Analyses were completed using SPSS Version 29 (IBM Corp, 2022). p values $\leq .05$ were considered statistically significant.

Results

Subject Demographics

Parents/guardians/caregivers responded to the speech-language questionnaire for 82 individuals with SMS with confirmed genetic diagnoses of SMS. In this data set, 50% were male and 50% were female, and at the time of participation, the age of subjects with SMS ranged from 36 months to 840 months with a mean age of 204 months (SD = 144 months; see Table 1). According to caregiver responses, 73 (85%) were White, six (7%) were Hispanic, two (2%) were more than one race, and nine (11%) did not provide information about ethnicity. No participants indicated the following ethnicities: Black or African American, Asian, Native Hawaiian or other Pacific Islander, or American Indian/Alaska Native. For location of residence, 73 (89%) reported living in the United States, 58 (70%) reported the individuals with SMS were currently attending school, and 69 (84%) were reportedly living at home with parents or caregivers. Fifty-eight (70%) of subjects were attending school at the time of participation. Of those attending school, 14 (24%) were placed in regular education classrooms, 30 (52%) were placed in special education classrooms, and 14 (24%) were placed in both regular and special education classrooms.

Of the 82 subjects in this study, 72 reportedly have the SMS deletion and 10 have the RAI1 variant. The 10

Table 1. Subject demographics.

Characteristic	(*N* = 82) *M* (*SD*) or *n* (%)
Age (months) at time of study	204 (144)
Age (months) at time of Smith–Magenis syndrome diagnosis	68 (53)
Race & ethnicity	
White	73 (85%)
Black or African American	0 (0%)
Asian	0 (0%)
Native Hawaiian or other Pacific Islander	0 (0%)
American Indian/Alaska Native	0 (0%)
More than one race	2 (2%)
Hispanic	6 (7%)
Not Hispanic	67 (82%)
Did not respond to race/ethnicity question	9 (11%)
Residence & schooling	
Living in the U.S.	73 (89%)
Living at home with parents	69 (84%)
Currently attending school	58 (70%)
Classroom placement[a]	
Regular education classroom	14 (24%)
Special education classroom	30 (52%)
Combination regular and special education	14 (24%)

Note. U.S. = United States.

[a]Classroom placement calculated based on the total number of subjects currently attending school (*n* = 58).

subjects with the RAI1 variant were all between the ages of 7 and 22 years.

Analyses of Hearing Loss and Otologic Health

Descriptive analyses of data related to hearing revealed that 29 (35%) subjects had hearing loss and two (2%) had a diagnosis of auditory processing disorder. Of the 29 individuals with hearing loss, 10 (34%) had a conductive loss, two (7%) had a sensorineural loss, five (17%) had a mixed loss, and 12 (41%) participants responded that they did not know the hearing loss type. Nine respondents did not answer the question, "Does your child have a hearing loss?" and these cases were categorized as not having hearing loss. Of the 29 individuals who were reported to have a hearing loss, three (10%) reportedly used a hearing device. Finally, 34 (41%) subjects reportedly used sign before speech (regardless of hearing status; see Table 2).

In the overall sample, descriptive analyses of otopathology revealed that 54 (66%) subjects had a history of otitis media (middle ear infections) and 51 (62%) had received PE tubes. The average age of first PE tube placement was 24 months (*SD* = 14), ranging from 6 to 72 months. The average number of PE tubes placed was three (*SD* = 3), ranging from one to 18 tubes total. The average age that hearing loss was first suspected was provided by 29 respondents and was 38 months (range: 0–480 months; see Table 3).

Table 2. Hearing characteristics.

Characteristic	*N* = 82 *n* (%)
History of otitis media	54 (66)
History of pressure equalization tube placement	51 (62)
Hearing loss[a]	29 (35)
Conductive	10 (34)
Sensorineural	2 (7)
Mixed	5 (17)
Type of hearing loss unknown by caregiver	12 (41)
Diagnosed with auditory processing disorder	2 (2)
Uses a hearing device (out of 29 with a hearing loss)	3 (10)
Used sign before speech (regardless of hearing status)	34 (41)

[a]There were nine nonresponses for the hearing loss question (i.e., "Does your child have a hearing loss?"), and these nonresponses were categorized as having no hearing loss. Percentages for each of the subtypes of hearing loss and use of a hearing device were calculated out of 29 reported to have hearing loss.

Age Group Analyses: Hearing and Otologic Health

Table 4 shows the distribution of hearing loss (by type) and otopathology (history of otitis media and PE tubes) by age group. There was a significant association between age group and presence of hearing loss (overall; $p = .019$), age group and history of otitis media ($p = .001$), and age group and history of PE tube placement ($p = .001$). The three oldest age groups (i.e., those above age 20 years) had the highest reported percent occurrence of hearing loss. The age group that reported the highest percent occurrence of conductive hearing loss was the youngest group (ages 3–6 years). The age group with the highest occurrence of sensorineural hearing loss was adults ages 20–29 years (15% of 13). The age groups that reported the highest occurrence of otitis media were those ages 70–80 years (100% of 1) and 20–29 years (92% of 13). The age groups with the highest reporting of history of PE tubes were ages 20–29 years (92% of 13) and 11–14 years (79% of 14).

Analyses of Early Speech-Language Development

Descriptive analyses of early speech-language characteristics revealed the average age that words were first spoken was 26 months (range: 11–72 months) and the

Table 3. Early speech-language outcomes and otopathology statistics.

Variable	*n*[a]	*M* (*SD*)	Minimum	Maximum
Age (months) first words spoken	47	26 (15)	11	72
Age (months) words first combined	47	47 (27)	12	108
Age (months) hearing loss first suspected	29	38 (90)	0	480
Age (months) pressure equalization tubes first placed	49	24 (14)	6	72
Number of pressure equalization tubes placed	50	3 (3)	1	18

[a]The number of responses (subjects) for each question is given. Nonresponses are not included in this data set.

average age that words were first combined (i.e., combinations of two or more words) was 47 months (range: 12–108 months; see Table 3). Thirty-seven (79%) subjects spoke first words at or after 24 months, 43 (92%) subjects combined words at or after 36 months, and 34 (41%) subjects used sign to communicate before using speech.

Age Group Analyses: Early Speech-Language Development

There was a significant association between age group and ability to combine words (χ^2 = 17.599, p = .007; see Table 4). The age groups with the highest percentage for combing words were 70–80 years (100% of 1), 7–10 years (71% of 14), and 20–29 years (69% of 13). Percentages for subjects ages 11–14 years and 15–19 years fell between 50% and 57%, and percentages for those ages 3–6 years and 30–44 years fell below 40%.

Analyses of Modes of Communication

Seventy respondents reported the following information about current modes of communication. Speech was the most common mode reported (n = 49, 60% of all subjects). Additionally, 19 (23%) used sign language, eight (10%) used a combination of speech and sign, 21 (26%) used gestures, and eight (10%) used some form of aided AAC to communicate. Aided AAC is a way of communicating using an external communication tool (in this article, aided AAC involved communicating using either a picture communication system or a speech-generating device; as defined by Biggs et al., 2022). Twelve (15%) respondents did not respond to questions about the mode of communication used. For the 70 subjects for whom we have responses, all were reported to use one or more of the following methods to communicate: speech, sign, AAC, gestures, and/or vocalizations (see Table 4).

Age Group Analyses: Modes of Communication

Table 4 shows modes of communication by age group. The association between age group and use of sign was significant (χ^2 = 14.302, p = .026), with subjects ages 3–6 years and ages 30–44 years having the highest percentage using sign (46% of 13 ages 3–6 years and 45% of 11 ages 30–44 years). There was no significant association between age and use of speech, gestures, or other forms of AAC (picture communication system or speech-generating device; see Table 4).

Analyses of Language Abilities

Analyses of reported language abilities indicated that 52 (63%) subjects understand simple directions, 50 (61%) subjects understand complex directions, and 51 (62%) subjects understand simple stories. Additionally, 51 (70%) subjects can answer questions, and 46 (63%) subjects take turns in conversations (see Table 4).

Age Group Analyses: Language Abilities

There were no significant associations between age group and any of the language abilities reported in this data set. The age groups with the highest percentage for understanding complex directions were ages 70–80 years (100% of 1), 7–10 years (71% of 14), and 3–6 years (69% of 13). The age groups with the highest percentage for understanding simple stories were ages 70–80 years (100% of 1), 7–10 years (71% of 14), and 3–6 years and 20–29 years (both 69%). All groups reportedly had greater than 50% who were able to answer questions. Age groups with the highest percentage for taking turns in a conversation were ages 70–80 years (100% of 1), 7–10 years (71% of 14), and 20–29 years (69% of 13; see Table 4).

Analysis of Vocal Quality

Forty (49%) subjects were reported to have a hoarse/gravely vocal quality (see Figure 1).

Age Group Analyses: Vocal Quality

There were at least 43% or more individuals in each age group who reportedly exhibit a hoarse vocal quality (see Figure 1). The age groups with the highest percentage with a hoarse vocal quality were those ages 20–29 years (62% of 13) and 70–80 years (100% of 1). The age groups with the

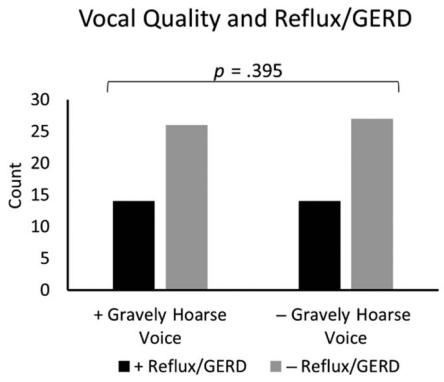
Table 4. Hearing and speech-language characteristics by age group and overall.

Characteristic	3–6 years (*n* = 13)	7–10 years (*n* = 14)	11–14 years (*n* = 14)	15–19 years (*n* = 16)	20–29 years (*n* = 13)	30–44 years (*n* = 11)	70–80 years (*n* = 1)	All subjects (*N* = 82)	*p* values (age-based analyses)
Hearing									
Hearing loss	5 (40%)	4 (29%)	3 (29%)	1 (6%)	8 (62%)	7 (64%)	1 (100%)	29 (35%)	X^2 = 15.193, *p* = .019
Conductive	3 (23%)	2 (14%)	1 (7%)	0 (0%)	2 (15%)	2 (18%)	0 (0%)	10 (12%)	—
Sensorineural	0 (0%)	0 (0%)	0 (0%)	0 (0%)	2 (15%)	0 (0%)	0 (0%)	2 (2%)	—
Mixed	1 (8%)	0 (0%)	0 (0%)	0 (0%)	2 (15%)	(18%)	0 (0%)	5 (6%)	—
Unknown	1 (8%)	2 (14%)	2 (14%)	1 (6%)	2 (15%)	3 (27%)	1 (100%)	12 (15%)	—
Otopathology									
History of middle-ear infections	5 (38%)	11 (79%)	11 (79%)	6 (38%)	12 (92%)	8 (73%)	1 (100%)	54 (66%)	X^2 = 16.867, *p* = .010
Has/had PE tubes	5 (38%)	10 (71%)	11 (79%)	6 (38%)	12 (92%)	7 (64%)	0 (0%)	51 (62%)	X^2 = 17.662, *p* = .007
Current mode of communication									
Sign	6 (46%)	4 (29%)	1 (7%)	0 (0%)	3 (23%)	5 (45%)	0 (0%)	19 (23%)	X^2 = 14.302, *p* = .026
Natural speech	5 (38%)	10 (71%)	7 (57%)	9 (56%)	9 (69%)	5 (64%)	1 (100%)	46 (60%)	ns
Combined speech and sign	2 (23%)	3 (21%)	2 (14%)	0 (0%)	0 (0%)	1 (9%)	0 (0%)	8 (10%)	ns
Gestures	6 (46%)	5 (36%)	2 (14%)	1 (6%)	4 (31%)	3 (30%)	0 (0%)	21 (26%)	ns
AAC system (other than sign/gestures)	3 (23%)	2 (14%)	2 (14%)	1 (6%)	0 (0%)	0 (0%)	0 (0%)	8 (10%)	ns
Language abilities									
Combines words	5 (38%)	10 (71%)	8 (57%)	8 (50%)	9 (69%)	4 (36%)	1 (100%)	44 (54%)	X^2 = 17.599, *p* = .007
Answers questions	7 (54%)	10 (71%)	8 (57%)	9 (56%)	9 (69%)	7 (64%)	1 (100%)	51 (70%)	ns
Takes turns in conversations	6 (46%)	10 (71%)	7 (50%)	9 (56%)	8 (62%)	6 (55%)	0 (0%)	46 (63%)	ns
Follows simple directions	8 (62%)	10 (71%)	8 (57%)	9 (56%)	9 (69%)	7 (64%)	1 (100%)	52 (63%)	ns
Follows complex directions	8 (62%)	10 (71%)	8 (57%)	9 (56%)	8 (62%)	6 (55%)	1 (100%)	50 (61%)	ns
Comprehends simple stories	7 (54%)	10 (71%)	8 (57%)	9 (56%)	9 (69%)	7 (64%)	1 (100%)	51 (62%)	ns
SLT									
Received SLT in the past	8 (62%)	10 (71%)	8 (57%)	9 (56%)	9 (69%)	7 (64%)	1 (100%)	52 (63%)	ns
Receiving SLT at the time of study	6 (46%)	9 (64%)	6 (43%)	6 (38%)	0 (0%)	0 (0%)	0 (0%)	27 (33%)	X^2 = 28.365, *p* = .001

Note. *p* values were calculated using chi-squared tests of independence. When there were limited numbers of subjects (e.g., types of hearing loss), no age-based analyses were conducted (noted in table as "—"). Nonsignificant results are noted as "ns." PE = pressure equalization; AAC = augmentative and alternative communication; SLT = speech-language therapy.

Figure 1. Relationship between gravely hoarse vocal quality and medical diagnosis of reflux/GERD (*p* values from chi-square tests of independence). GERD = gastroesophageal reflux disease.

Age group	Hoarse vocal quality	History of reflex/GERD
3–6 years (*n* = 13)	6 (46%)	6 (46%)
7–10 years (*n* = 14)	7 (50%)	5 (37%)
11–14 years (*n* = 14)	6 (43%)	6 (45%)
15–19 years (*n* = 16)	7 (44%)	5 (31%)
20–29 years (*n* = 13)	8 (62%)	5 (36%)
30–44 years (*n* = 11)	5 (45%)	2 (18%)
70–80 years (*n* = 1)	1 (100%)	0 (0%)
All subjects (*N* = 82)	40 (49%)	29 (35%)



lowest percentage with a hoarse vocal quality were those ages 11–14 years (43% of 114) and 15–19 years (44% of 16). The distribution of individuals who reportedly have a hoarse vocal quality did not significantly differ across age groups.

Relative Communication Strengths

Participants reported that 46 (56%) subjects reportedly had excellent memory for people, events, and facts; 40 (49%) subjects had a great sense of humor; 38 (46%) subjects had a high social interest; and 32 (39%) subjects had strong vocabulary knowledge. The communication skills that had lower percentages as relative strengths included the ability to tell stories (five subjects, 6%), speech/articulation (eight subjects, 10%), problem solving (11 subjects, 13%), ability to follow directions (12 subjects, 15%), ability to answer questions (12 subjects, 15%), and ability to combine words (13 subjects, 16%). Interestingly, while social interest was reported as a communication strength for 38 (46%) subjects, social skills were only reported as a strength for 16 (20%) subjects (see Table 5).

Analysis of Participation in Speech-Language Therapy

Only 52 (63%) participants responded to questions about current and past participation in speech-language therapy (SLT). Responses indicated that 52 (100% of those who responded) subjects had received SLT at some time in the past. Twenty-seven (33% of the overall sample) respondents reported that their child/dependent had received SLT at the time of participation. Individuals that were currently receiving SLT at the time of the study were all younger than 20 years of age. There was a significant effect of age for subjects who were receiving SLT at the time of the study ($\chi^2 = 28.365$, $p = .001$), but no effect of age for having received SLT ever (see Figure 2 and Table 4).

Analyses: Relationship Between Hearing, Otopathology, and Speech-Language Skills

Analyses were conducted to compare the relationship between hearing, otopathology, and early speech. There was a significant correlation between age that first words were spoken and the age that PE tubes were first placed ($p = .028$, $r = .388$; see Figure 3). Additional post hoc analyses revealed no significant associations between hearing loss and other language skills, specifically the ability to answer questions and to understand simple stories.

Analysis of Vocal Quality and Reflux/GERD

Twenty-nine (35%) subjects were reported to have a history of esophageal reflex and/or GERD, but there no association between a hoarse vocal quality and a history of esophageal reflex and/or GERD (see Figure 1).

Discussion

The first aim of this investigation was to describe profiles and identify age-related differences for hearing loss, otopathology (history of otitis media and placement of PE tubes), early speech-language development, mode of communication, language abilities, vocal quality, communication strengths, and participation in speech-language services in a large group of individuals with SMS. We address each of these characteristics in turn, addressing profiles and age-related differences for each.

Profiles and Age-Related Differences for Hearing Loss and Otologic Health

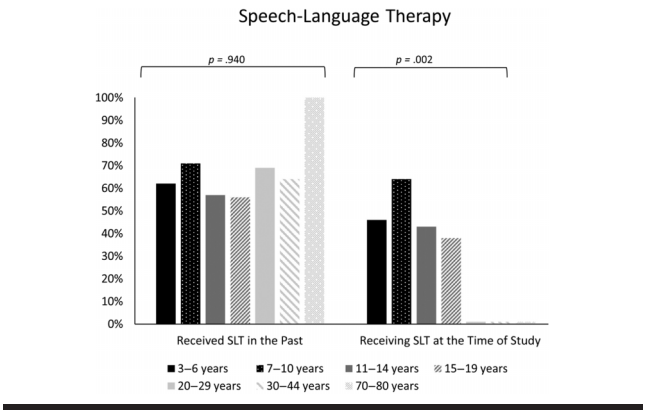
The current study found that 35% of subjects had a reported hearing loss. This is lower than previous reported hearing loss in approximately 48%–72% of SMS cases (e.g., Brendal et al., 2017; Edelman et al., 2007; Gamba et al., 2011; Potocki et al., 2003; Smith et al., 1986). The difference between the current results and previous findings may be due to the nature of data collection. Since the current study involved parent/caregiver responses, it is

Table 5. Communication strengths for each age group.

Identified communication strengths	Age group							
	3–6 years (*n* = 13)	7–10 years (*n* = 14)	11–14 years (*n* = 14)	15–19 years (*n* = 16)	20–29 years (*n* = 13)	30–44 years (*n* = 11)	70–80 years (*n* = 1)	All subjects (*N* = 82)
Memory for people, events, and facts	7 (54%)	8 (57%)	8 (57%)	8 (50%)	9 (69%)	6 (66%)	0 (0%)	46 (56%)
Humor	5 (38%)	8 (57%)	7 (50%)	7 (44%)	6 (46%)	6 (55%)	1 (100%)	40 (49%)
Social interest	5 (38%)	7 (50%)	6 (43%)	7 (44%)	7 (54%)	6 (55%)	0 (0%)	38 (46%)
Vocabulary knowledge	6 (46%)	6 (43%)	5 (36%)	8 (50%)	4 (31%)	3 (27%)	0 (0%)	32 (39%)
Comprehension of stories	3 (23%)	5 (36%)	3 (21%)	4 (25%)	4 (31%)	2 (18%)	0 (0%)	21 (26%)
Reading and/or writing	2 (15%)	6 (43%)	2 (14%)	5 (31%)	4 (31%)	2 (18%)	0 (0%)	21 (26%)
Ability to formulate questions	0 (0%)	6 (43%)	3 (21%)	3 (19%)	5 (38%)	2 (18%)	0 (0%)	19 (23%)
Motivation to be independent	5 (38%)	4 (29%)	1 (7%)	4 (25%)	2 (15%)	3 (27%)	0 (0%)	19 (23%)
Social skills	2 (15%)	4 (29%)	3 (21%)	0 (0%)	4 (31%)	3 (24%)	0 (0%)	16 (20%)
Ability to formulate sentences	0 (0%)	1 (7%)	0 (0%)	4 (25%)	6 (46%)	2 (18%)	0 (0%)	13 (16%)
Ability to answer questions	1 (8%)	0 (0%)	0 (0%)	4 (25%)	6 (46%)	1 (9%)	0 (0%)	12 (15%)
Ability to follow directions	2 (15%)	1 (7%)	1 (7%)	3 (19%)	4 (31%)	1 (9%)	0 (0%)	12 (15%)
Problem solving	2 (15%)	2 (14%)	1 (7%)	3 (19%)	2 (15%)	1 (9%)	0 (0%)	11 (13%)
Speech/articulation	0 (0%)	3 (21%)	0 (0%)	2 (13%)	1 (8%)	2 (18%)	0 (0%)	8 (10%)
Ability to tell stories	0 (0%)	0 (0%)	1 (7%)	1 (6%)	2 (15%)	1 (9%)	0 (0%)	5 (6%)

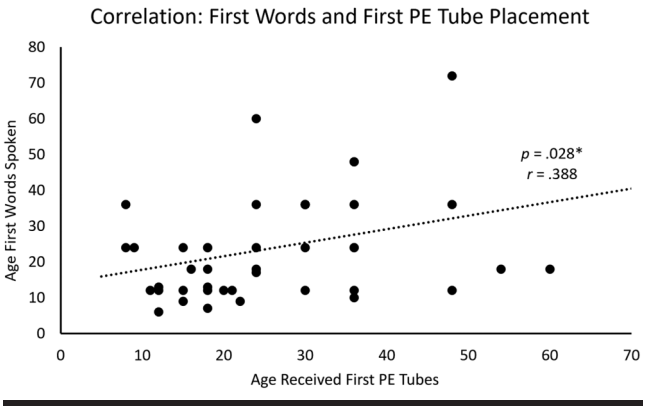
Note. Sorted by highest percentage for all subjects.

Figure 2. Participation in speech-language therapy by age group (*p* values chi-square tests of independence). SLT = speech-language therapy.



possible that some errors were made when reporting no hearing loss for individuals who do in fact have a hearing loss. Since 41% of current respondents indicated the presence of hearing loss but did not know the type of hearing loss, it is possible that other respondents may not have known or did not remember if there was a hearing loss at all. For individuals who have recurrent otitis media, conductive hearing loss may be intermittent and possibly not known of at the time of participation. This may be especially the case for younger children who experience intermittent conductive hearing loss occurring in tandem with acute otitis media. If pure-tone audiometry had been performed here to identify the presence and type of hearing loss, the prevalence may have been higher. While the current study included a large cohort of subjects with SMS compared to many previous studies (e.g., Edelman et al., 2007; Gamba et al., 2011; Potocki et al., 2003), Brendal et al. (2017) directed tested 133 subjects with SMS and obtained pure-tone thresholds (250–8000 Hz). As a result, the results reported by Brendal et al. (2017) are more likely to be a better estimate of hearing loss prevalence in those with SMS than what was found here; however, we would argue that any prevalence related to conductive hearing loss should be interpreted with caution since it can be intermittent when it occurs as a result of otitis media and, as a consequence, may vary from study to study.

Figure 3. Relationship between otopathology and early speech ability (*p* values from Pearson correlation). PE = pressure equalization.



The current results also revealed that 34% of the subjects in this study with hearing loss had a conductive loss, while only 7% had a sensorineural loss and 17% had a mixed loss. Additionally, those ages 3–6 years had the highest occurrence of conductive hearing loss, while those ages 20–29 years had the highest occurrence of sensorineural hearing loss. These results align with Brendal et al. (2017), who found that conductive hearing loss affected 35% of ears mostly in of those ages 1–10 years and sensorineural hearing loss occurred most often in subjects over age 11 years. The current study also reported that 66% of subjects had a history of otitis media, which may be associated with hearing loss in the younger individuals within this data set. The current results are consistent with previous reports that SMS is associated with a high occurrence of otitis media (Elsea & Girirajan, 2008), which is associated with conductive hearing loss.

The current study also reported several results related to otopathology, which no previous SMS studies have specifically done. The current study found a high prevalence of PE tube placement (62%), with tubes being placed between ages 6 and 72 months (average age of 24 months) and individuals having one to 18 tubes placed over time (average of three sets of tubes). These results are consistent with the high percent occurrence of otitis media found here and reported previously (Elsea & Girirajan, 2008). Parents, medical providers, and audiologists should be aware of the elevated risk of otitis media in this population and should know that placement of PE tubes is a common form of management for this population.

Profiles and Age-Related Differences for Early Speech-Language Development

Previous studies reported speech-language delays in those with SMS (e.g., Dykens & Smith, 1998; Greenberg et al., 1996; Smith et al., 1986), and consistent with those reports, the current study found significant delays in the age that words were first spoken and the age that words were first combined. An important novel finding reported here is that 38% of those with SMS ages 3–6 years used speech to communicate (and 46% used sign). For those ages 7–44 years, 56%–71% used speech at the time of the study. Additionally, many subjects used multiple modes of communication, such as sign and speech, or speech, gestures, and AAC (picture communication system or speech-

generating device). Using multiple modes of communication has been shown to improve efficacy of communication for preschool-age children (Jago et al., 1984), individuals with Down syndrome (Powell & Clibbens, 1994), and children with ASD (Brady et al., 2015). This approach also aligns with the intervention methodology known as total communication (see Jago et al., 1984) or the more current approach known as multimodal intervention/therapy (Brady et al., 2015; Pierce et al., 2019). The multimodal intervention described by Brady et al. (2015), which combined speech with AAC to promote vocabulary development in children with ASD, may align with the modalities reported for the individuals with SMS in the current study.

Profiles and Age-Related Differences for Modes of Communication

Analyses of the modality of communication revealed that 65% of the subjects in this study used sign before developing speech and 23% used sign alone or in combination with other communication modes (such as speech) at the time of participation. An interesting finding is that age group had the highest percentage of sign use (46%). For the youngest children in this study, this seems logical since many of them may not yet have speech or may have delayed or limited speech production abilities. The adults ages 30–44 years also had a high percentage of sign use (45%). For these adults, there is no reported use of a picture communication system or speech-generating device, so it is possible that use of sign and speech was relied on more commonly than other AAC methods. Use of sign to support development of communication in children has been reported in previous studies, but this is the first indication of the prevalence of this practice for younger children with SMS. This is an important finding not just to inform parents, educators, and clinicians, but also because the number of individuals who used sign before developing speech (65%) is not the same as those who continued to use sign at the time of the study. Many parents/caregivers (and even educators or clinicians) may be reluctant to use sign (or other forms of alternative communication) for fear that it may exacerbate delays in speech development. This is emphatically not true (Beukelman & Mirenda, 2005; also see Adamson & Dunbar, 1991; Miller et al., 1991; Sedey et al., 1991). The current results along with previous evidence showing that use of sign supports language development can and should be shared with parents, family members, caregivers, and educators to help dispel any inaccurate assumptions. The current results are also consistent with previous recommendations regarding the use of sign language when speech is delayed and to decrease frustration and promote more positive communication behaviors (Elsea & Girirajan, 2008; Smith et al., 1998).

Profiles and Age-Related Differences for Language Abilities

Although children with SMS present with deficits in language, previous studies suggested delays were greater in expressive rather than receptive language (Wolters et al., 2009). The nature of the data in the current study does not lend itself to a direct within-subject comparison between expressive and receptive language skills. Review of the percentage of those in each age group who reportedly demonstrated language skills in the questionnaire revealed slightly higher percentages in all language skills in subjects ages 7–10 years and the single older adult. The age group with the lowest percentage for these language skills was the youngest group, ages 3–6 years. For that group, only 38% reportedly combined words, 46% took turns in a conversation, and 54% could answer questions. Comprehension of directions (simple and complex) and comprehension of stories were reported for 54%–62% of this youngest group. Future research should aim to test expressive and receptive language skills directly, and such investigations should include language sample analyses as well as standardized testing to describe the language profile of children and adults with SMS in greater depth.

The comparison of age groups revealed highly variable percentages for those who could combine words. For example, while 71% of children ages 7–10 years combined words, the percentage for those ages 30–44 years was less than 40%. The current data alone cannot provide a definitive explanation for this finding. It is possible that this difference is due to the variability in language skills in those with SMS. It could also be a sampling effect due to the sample size for each age group. It could also reflect improvements over the years in the efficacy of special education and speech-language intervention. For example, the subjects ages 30 years and older may have had less intense and/or less effective intervention than what is provided to children today. Finally, individuals ages 30 years and older at the time of participation were born as early as 1991, only a few years after SMS was first identified (Smith et al., 1986). Genetic identification of SMS in the 1990s was more difficult than with current genetic testing methods, and many children decades ago may not have received an SMS diagnosis until later childhood or adulthood. As a result, it is possible that education and clinical services were not optimal for the unique needs of those with SMS. Only a direct comparison of adults identified early during childhood versus later could potentially address this question, something that was not done as part of the current study. Additionally, future investigations of language skills in adults with a larger sample size could also confirm or clarify if the 30- to 44-year-old adults in this study were representative of adults in this age range with SMS.

Every age group (other than the one subject in the 70- to 80-year age range) reportedly had 50% of subjects or more that were able to answer questions, take turns in a conversation, and understand simple stories. These are critical skills for social communication, and the higher percentage of those with these abilities aligns with previous reports that socialization was a relative strength compared to other functional skills, such as daily living skills (Madduri et al., 2006; Martin et al., 2006; Udwin et al., 2001; Wolters et al., 2009).

Profiles and Age-Related Differences for Vocal Quality

The current study found that 49% of all SMS individuals in the data set reportedly had a hoarse vocal quality. Previous research indicated that approximately 82% of those with SMS exhibited a hoarse, deep voice in later years (Greenberg et al., 1991). For a hoarse vocal quality, the highest percent for any age group in this data set was for those 20–29 years of age (62%). The only subject in the age range 70–80 years also exhibited a hoarse vocal quality. The current study did not include any adults ages 45–69 years, so the current results cannot confirm or refute the findings reported by Greenberg et al. (1991). Additionally, the current data indicated that for the youngest age children in this study, those ages 3–6 years (46%) presented with a gravely–hoarse voice quality. Furthermore, age-based comparisons were not significant, providing additional evidence that hoarse vocal quality is not something that emerges over time or as the result of aging. The current findings reveal that this characteristic is present in very young children and, thus, is less likely to be the result of vocal polyps, which develop over time, as previously postulated (Greenberg et al., 1991).

Profiles and Age-Related Differences for Participation in SLT

The current results revealed that 63% of those with SMS in this data set received speech-language intervention at some time in the past. Only 33% of individuals with SMS in this study were reported to be receiving speech-language intervention at the time of the study and all of those were ages 3–19 years with no subjects with SMS older than 20 years reported to be currently receiving intervention. For those living in the United States, special education services (including speech-language intervention) are provided through age 22 years if deemed necessary for educational progress. While adults with SMS can continue to receive services after leaving the public school system through outpatient clinics or private practices, there is a financial cost of such services, whereas school-based services are free to parents (again, in the United States). For adults with SMS over age 22 years, the cost of intervention must be covered by insurance or Medicare/Medicaid or paid out of pocket for the individual or his/her/their parents/caregivers. It is possible that the additional cost may be a barrier to speech-language services for those over age 20 years.

In addition to the financial cost, there are other possible reasons why individuals with SMS over 20 years of age in this data set were no longer receiving speech-language intervention. One possibility is that parents/caregivers and previous speech-language pathologists determined continued intervention was not warranted. Perhaps services were discontinued because they had reached a plateau in their progress toward treatment goals. Parents/caregivers may lack knowledge and awareness about what speech-language pathologists can provide to adults who have developmental disabilities and may therefore fail to seek out such services. Another possibility is a lack of local providers with the skills, adequate knowledge, and experience to work with adults who have developmental disabilities. Clinics that serve children with developmental disabilities may provide services to children but not adults. Clinics that specialize in adult services may focus on acquired communication impairments, such as aphasia or dysarthria due to a stroke or brain injury, rather than communication issues associated with a developmental disability. This is, of course, speculation, as the current study did not investigate why subjects over age 20 years were no longer receiving speech-language services; however, the findings here are interesting and motivate the need for further investigation to determine if parents/caregivers are interested in speech-language services for their adult children with SMS and if such adults would benefit from continued services and, if so, determine what barriers may be preventing them from receiving this intervention. Alternatively, if parents/caregivers and speech-language pathologists are determining that services for adults are not necessary, then future research should aim to determine if these decisions are motivated by evidence or if these adults might benefit from continued intervention despite previous termination of treatment.

The data in this study show that adults older than 20 years continue to present with deficits in language and social communication. Based on these findings, we suggest that such adults would likely benefit from intervention, but without knowing why such services are no longer being accessed, it is impossible to address this issue here. We suggest that it is critical for future studies to investigate the reasons for the age-based differences in who is receiving speech-language services. This is an important consideration for adolescents with SMS as they approach adulthood and transition planning is undertaken and for adults who may benefit from intervention but have barriers to accessing such service.

Profiles and Age-Related Differences for Relative Communication Strengths

The current results indicate that long-term memory for people, events, and facts is the most common reported communication strength for individuals with SMS (across all ages), followed by sense of humor, social interest, and vocabulary knowledge. Additionally, while social interest was reported as a strength for 46% of those with SMS in this data set, social skills were reported as a strength for fewer individuals (20% across all ages). The finding that long-term memory was a common strength for those with SMS is consistent with the previous literature (Osório et al., 2012; Udwin et al., 2001). Specifically, individuals with SMS were believed to have relatively unimpaired long-term memory (Osório et al., 2012). The finding that social interest was a commonly reported strength, but social skills were not, results in an interesting dichotomy. Even though individuals with SMS reportedly have good long-term memory, relatively good vocabulary knowledge, and a high social interest, they also (reportedly) lack the social skills needed to establish and maintain social interactions. Additionally, fewer than 20% of those with SMS in this data set reportedly had communication strengths in sentence formulation, problem solving, and the ability to answer questions, following directions, and telling stories. These language skills support social communication, so it should not be surprising that social skills are reportedly an area of deficit in this population despite their strengths. These findings suggest that speech-language intervention should emphasize language skills when improving social skills are identified as an area of need.

Detailed information about the shared and distinct ASD characteristics of those with SMS was not provided by the results of this study. While the current study extended the previous findings regarding socialization, social skills, and social interest, it is unknown what other characteristics of ASD are shared with those who have SMS. Obviously, in those who have SMS, the genetic disorder is the primary diagnosis. It is unclear if those who met the criteria on an ASD rating scale in the previous studies would also receive a clinical diagnosis of ASD. Furthermore, both peripheral neuropathy (Greenberg et al., 1996) and hyperacusis (Brendal et al., 2017) have been reported in SMS and may present as sensory issues. Since sensory processing issues are present in those with ASD, sensory issues in SMS may seems to align with ASD. Behavioral challenges for those with SMS include difficulty with transitions to a new activity or changes in the routine (Smith et al., 1998), a behavioral profile also common in those with ASD. It is important to recognize that despite these similarities, there are also many differences, including the finding here that social interest is very high in those with SMS. Future research is needed to further elucidate the similarities and differences between the behavioral phenotype of those with SMS and those with ASD.

Relationship Between Hearing, Otologic Health, and Speech-Language

The second aim was to determine if hearing or otopathology profiles were associated with speech, language, and communication abilities. We expected to find that the presence of hearing loss and or recurrent otitis media was associated with greater speech-language delays, but the results differed from this expectation. While there was a significant correlation between the age that PE tubes were first placed and the age that first words were spoken, the subjects who received PE tubes earlier also spoke earlier, not later as expected. Although this correlation is significant (p = .028), the correlation coefficient is only .388. Regardless of the strength of this finding, this is an interesting result. This finding may mean that children with recurrent otitis media who are treated early using PE tubes have a slight advantage with early expressive speech skills, perhaps because the occurrence of intermittent conductive hearing loss is reduced. Of course, this study did not collect data to support this specific conclusion, and our hypothesis assumes that otitis media is associated with temporary conductive hearing loss. Additionally, many children with SMS in this study received SLT during childhood (38%–64% of those school aged). It is possible that participation in SLT may have helped identify children who were at greatest risk due to recurrent otitis media and fluctuating hearing status, motivating earlier placement of PE tubes and making speech-language intervention more effective. Direct testing of hearing status and the impact of intermittent conductive hearing loss on early speech-language development in children with SMS is still needed to confirm these speculations.

Relationship Between Vocal Quality and GERD

The third aim was to determine if a hoarse vocal quality was associated with the presence of reflex/GERD. Previous investigation regarding vocal quality in SMS concluded that high laryngeal tension might underlie the characteristic hoarse vocal quality (Hidalgo-De la Guía et al., 2020). It could be that early on, those with SMS do not have vocal polyps but do have laryngeal tension resulting in a hoarse vocal quality. Eventually, this excessive and long-term laryngeal tension/constriction could cause polyps later in life, which would be consistent with Greenberg et al. (1991), who examined children, adults, and older adults. While the questionnaire used in this

study did not include questions about laryngeal tension (nor would this be easy or even possible for respondents to evaluate or recognize), questions about the presence of GERD were included. We compared the presence of a hoarse vocal quality to the diagnosis of GERD, and while both are prevalent in this group of individuals with SMS, there was not a significant association between these factors. The current findings do not conflict with the postulation that high laryngeal tension (as suggested by Hidalgo-De la Guía et al., 2020) may underlie the common hoarse vocal quality of those with SMS, although this cannot be confirmed by the current data. We suggest future studies involve objective measures of vocal quality, direct measures of laryngeal tension, and history of GERD and/or vocal fold pathology (i.e., polyps or nodules) to determine the actual causes that underlie the common hoarse vocal quality of those with SMS.

Limitations

Because this study was based on parent/caregiver responses to questionnaires, accuracy is dependent upon the reliability of participant responses. While it is possible that some caregivers recalled details about their child past hearing and communication abilities with great accuracy or even kept track of such information using a baby book or therapeutic notes, it is also possible that some respondents estimated information or chose not to answer questions if they did not recall the information. As a result, it is important to interpret the results with the understanding that the reliability of responses may be limited.

The validity of the study and results are dependent upon the validity of the questions in the questionnaire. The questionnaire included a broad scope of questions covering many aspects of development and abilities related to speech, language, mode of communication, hearing, otopathology, literacy, and speech-language intervention. Questions were detailed and included yes/no questions, choice-based questions (such as type of hearing loss), and open-ended questions so respondents could provide additional information and individual details. Because of the design and protocols of this study, it was not possible for the authors to conduct any follow-up after respondents submitted their survey responses to obtain additional information about morphosyntax or to clarify unclear or confusing responses.

The current questionnaire did not collect information about comprehension and use of vocabulary, prepositions, pronouns, and grammatical morphemes or other syntactic structures; therefore, the scope of language information gathered was limited. Additionally, there were no questions regarding the nature of word combinations, such as what types of words were combined. Future research should collect data about language ability, perhaps by adding more questions to the language questionnaire focused on comprehension and use of vocabulary and syntax. Alternatively, direct testing that includes standardized measures of language and/or language sample analyses would also provide more information about morphosyntax and address questions about language formulation and use with greater detail.

Future Research

Future research on detailed analyses of communication skills and challenging behaviors in individuals with SMS is of high interest. While this is a rare genetic disorder, this a growing group of individuals with significant needs related to behavior, sleep, hearing, and communication. Clinicians, educators, and medical providers rely on published data to support their evidence-based practices. Without such data, providers are unable to support their clinical decisions using evidence. The questionnaire used in the current study answered many questions but also motivated many potential new research questions. For example, the current study revealed some differences in receptive versus expressive language abilities, but not the nature of language production regarding syntactic complexity or vocabulary use. Additional questions about vocal quality, barriers to receiving speech-language intervention, and pragmatic skills have been motivated by the current results. Future studies should also focus on the relationship between language ability and functional outcomes, including social communication outcomes. Since behavioral issues are so prominent for those with SMS, future studies may also aim to examine individual differences and the relationship between communication ability and the severity of maladaptive behaviors. Additional research is needed that focuses on the efficacy of intervention and education for language and communication in those with SMS, as well as on intervention and management of hearing health and hearing loss in those with SMS. Finally, research examining the communication profiles of individuals with SMS who have a genetic deletion versus the single gene mutation (RAI1 variant) is also needed, as it is unknown if and how speech, language, and communication may or may not differ depending on the genetic nature of this syndrome.

The registry provided a tremendous amount of data for 82 subjects with SMS, making this the largest cohort of subjects with SMS in a study specifically focused on speech, language, communication, hearing, and otopathology to date. While direct testing is important because it increases reliability of the data collected, the type of data collected here provides relevant information about the developmental history, health, current levels of performance, strengths and

needs, and day-to-day functioning of those with SMS. Direct testing of individuals with SMS also poses challenges due to intellectual disability and behavioral challenges (Elsea & Girirajan, 2008; Greenberg et al., 1996; Smith et al., 1998). We suggest that the findings here motivate future studies that combine indirect and direct data collection methods to optimize the number of individuals with SMS tested and ensure an adequate sample size for statistical analyses.

The current study did not include a control group. While milestones, such as the age that children typically begin to speak, can be considered, future investigations may want to consider including a control group for direct comparison to the SMS cohort. An alternative design would be to include two groups of individuals with SMS, including one group with the common SMS deletion (Elsea & Girirajan, 2008) and another with the RAI1 variant version of SMS (Elsea & Girirajan, 2008; Rinaldi et al., 2022). Individuals with the RAI1 variant have been reported to have milder symptomatology (Bi et al., 2004, 2006). The current study included 10 out of 82 subjects with the RAI1 variant. Due to the small number of subjects with RAI1 variant within this large data set and the fact that ages for the RAI1 subjects were limited to 7–22 years, the current study did not include additional analyses focused only on the RAI1 subjects. The correlation between variability in deletion size and communication outcomes remains to be elucidated, although some previous research reported that individuals with the RAI1 variant form of SMS had slightly less impairment in cognition (Bi et al., 2004, 2006; Elsea & Girirajan, 2008). While it is possible that there are also differences in hearing, language, and communication ability based on the nature or size of the deletion/mutation, this remains to be investigated. Such research would be highly informative and would better capture the heterogeneity of SMS symptomatology.

Implications for Clinicians, Educators, and Parents

Parents should seek evaluation of hearing and speech-language as soon as a diagnosis of SMS is confirmed. Compared to children without SMS, children with SMS will begin producing first spoken words later, around age 2 years or even later. In fact, delays in the production of first spoken words may not happen until age 6 years. As previously reported (Elsea & Girirajan, 2008; Smith et al., 1998) and confirmed in the current study, the use of sign language in those with SMS is prevalent and may help provide a means for children to communicate before they are able to use speech. While clinicians know that AAC facilitates language development, several studies have revealed that parents may continue to have the misconception that AAC will replace or hinder their child's spoken language development or that their child will rely on the AAC rather than using their own speech (Donato et al., 2018; Jonsson et al., 2011; Serpentine et al., 2011). Notably, these misconceptions were also found in culturally and linguistically diverse parents (Townsend et al., 2012). Clinicians need to be aware that parents may have misconceptions about AAC and would benefit from education about how the use of sign or other AAC can promote speech and language development rather than hinder it (e.g., Adamson & Dunbar, 1991; Beukelman & Mirenda, 2005; Sedey et al., 1991).

Hearing loss and otopathology are prevalent in this population, but the current data do not show that otitis media or hearing loss exacerbates delays in speech-language development. In past studies with children without SMS who have recurrent otitis media and/or hearing loss, there was evidence of an increased risk of delays or challenges and/or disorders in speech and language (Lieu et al., 2020; Shriberg et al., 2000). Otitis media in typically developing children has also been associated with significant negative effects on attention (Bellussi et al., 2005; Gouma et al., 2011), development of literacy (Bellussi et al., 2005), and anxiety/depression-related disorders (Gouma et al., 2011). Perhaps since most children with SMS are already working with audiologists and speech-language pathologists and many are using sign or other forms of AAC, the possible negative impact of hearing and otopathology on speech-language development is mitigated. In any case, working closely with an audiologist and speech-language pathologist is critically important for those with SMS, especially during early childhood.

Conclusions

Major findings include the observation that those with SMS have a high prevalence of otitis media (66%), PE tube placement (62%), and hearing loss (35%). Findings related to speech-language characteristics included significantly delayed first words (age 26 months), delayed age at which words were first combined (47 months), and a high percentage of those who used sign before speech (53%). Approximately 79% of those with SMS in this study had delayed speech development, and approximately 92% combined words much later than is seen in those without SMS. Importantly, while speech-language delays were common, many subjects in this study (60%) communicated using speech, with 41% reporting the use of sign language before speech developed. The current study extends previous findings by showing that the production of first words and the production of early word combinations are both delayed, establishing that the majority of those with SMS do develop the ability to use speech, and

showing subtle differences in expressive versus receptive language skills. The findings related to otopathology suggest that identification and management of otitis media and hearing loss in those with SMS follow best practices including early identification and treatment. Parents/caregivers and clinical providers should be aware that recurrent otitis media or the presence of hearing loss does not appear to exacerbate speech-language delays in this population. This may be due to the provision of early speech-language services and medical intervention for chronic otitis media (via PE tube placement). Understanding the prevalence and relationship between variables in hearing, otopathology, speech, language, and communication motivates early intervention and informs medical, clinical, and educational providers working with individuals who have SMS. The current results extend previous findings by providing more information about speech, language, communication, and phenotype of SMS. They also motivate future research focused on the speech-language and social communication phenotype of SMS.

Data Availability Statement

The data used in this study were obtained from the SMS Patient Registry housed at BCM. Due to the signed DUA, the authors of this article are not authorized to transmit or share the data obtained from BCM. Researchers interested in obtaining the data set used in this study should follow the procedure used by the authors of the current study. This includes contacting the Elsea Lab at BCM at elsealab-research@bcm.edu with the following information via e-mail: name, title, and affiliation of person requesting the data set, who will have access to the data, which surveys/questionnaires are being requested (the current study used the demographics and the speech, language, and hearing questionnaires), your research questions, a description of how the data will be used, and evidence of current IRB approval. Requests will be reviewed jointly by PRISMS, Inc., and BCM. Reasonable and complete requests will be approved. Researchers will be required to sign a DUA with BCM. Once approved, and the DUA is signed, the de-identified data will be transmitted securely (password protected) through e-mail from the Elsea Lab at BCM directly to the requesting researcher.

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References

Adamson, L. B., & Dunbar, B. (1991). Communication development of young children with tracheostomies. *Augmentative and Alternative Communication, 7*(4), 275–283. https://doi.org/10.1080/07434619112331276013

Bellussi, L., Mandala, M., Passali, F. M., Passali, G. C., Lauriello, M., & Passali, D. (2005). Quality of life and psycho-social development in children with otitis media with effusion. *Acta Otorhinolaryngologica Italica, 25*(6), 359–364.

Beukelman, D., & Mirenda, P. (2005). *Augmentative and alternative communication: Management of severe communication impairments* (3rd ed.). Brookes.

Bi, W., Saifi, G. M., Girirajan, S., Shi, X., Szomju, B., Firth, H., Magenis, R. E., Potocki, L., Elsea, S. H., & Lupski, J. R. (2006). *RAI1* point mutations, CAG repeat variation, and SNP analysis in non-deletion Smith–Magenis syndrome. *American Journal of Medical Genetics Part A, 140*(22), 2454–2463. https://doi.org/10.1002/ajmg.a.31510

Bi, W., Saifi, G. M., Shaw, C. J., Walz, K., Fonseca, P., Wilson, M., Potocki, L., & Lupski, J. R. (2004). Mutations of RAI1, a PHD-containing protein, in nondeletion patients with Smith–Magenis syndrome. *Human Genetics, 115*(6), 515–524. https://doi.org/10.1007/s00439-004-1187-6

Biggs, E. E., Therrien, M. C. S., Snodgrass, M. R., & Douglas, S. N. (2022). Voices from the field: Strategies for effective telepractice for children with autism who use augmentative and alternative communication. *Perspectives of the ASHA Special Interest Groups, 7*(2), 324–337. https://doi.org/10.1044/2021_PERSP-21-00229

Brady, N. C., Storkel, H. L., Bushnell, P., Barker, R. M., Saunders, K., Daniels, D., & Fleming, K. (2015). Investigating a multimodal intervention for children with limited expressive vocabularies associated with autism. *American Journal of Speech-Language Pathology, 24*(3), 438–459. https://doi.org/10.1044/2015_AJSLP-14-0093

Brendal, M. A., King, K. A., Zalewski, C. K., Finucane, B. M., Introne, W., Brewer, C. C., & Smith, A. C. (2017). Auditory phenotype of Smith–Magenis syndrome. *Journal of Speech, Language, and Hearing Research, 60*(4), 1076–1087. https://doi.org/10.1044/2016_JSLHR-H-16-0024

Casby, M. W. (2001). Otitis media and language development: A meta-analysis. *American Journal of Speech-Language Pathology, 10*(1), 65–80. https://doi.org/10.1044/1058-0360(2001/009)

Donato, C., Spencer, E., & Arthur-Kelly, M. (2018). A critical synthesis of barriers and facilitators to the use of AAC by children with autism spectrum disorder and their communication partners. *Augmentative and Alternative Communication, 34*(3), 242–253. https://doi.org/10.1080/07434618.2018.1493141

De Leersnyder, H., de Blois, M. C., Claustrat, B., Romana, S., Albrecht, U., von Kleist-Retzow, J. C., Delobel, B., Viot, B., Lyonnet, S., Vekemans, M., & Munnich, A. (2001). Inversion of the circadian rhythm of melatonin in the Smith–Magenis syndrome. *The Journal of Pediatrics, 139*(1), 111–116. https://doi.org/10.1067/mpd.2001.115018

Dykens, E. M., & Smith, A. C. M. (1998). Distinctiveness and correlates of maladaptive behaviour in children and adolescents with Smith–Magenis syndrome. *Journal of Intellectual Disability Research, 42,* 481–489. https://doi.org/10.1046/j.1365-2788.1998.4260481.x

Edelman, E. A., Girirajan, S., Finucane, B., Patel, P. I., Lupski, J. R., Smith, A. C. M., & Elsea, S. H. (2007). Gender, genotype, and phenotype differences in Smith–Magenis syndrome: A meta-analysis of 105 cases. *Clinical Genetics, 71*(6), 540–550. https://doi.org/10.1111/j.1399-0004.2007.00815.x

Elsea, S. H., & Girirajan, S. (2008). Smith–Magenis syndrome. *European Journal of Human Genetics, 16,* 412–421. https://doi.org/10.1038/sj.ejhg.5202009

Gamba, B. F., Vieira, G. H., Souza, D. H., Monteiro, F. F., Lorenzini, J. J., Carvalho, D. R., & Morreti-Ferreira, D. (2011). Smith–Magenis syndrome: Clinical evaluation in seven Brazilian patients. *Genetics and Molecular Research, 10*(4), 2664–2670. https://doi.org/10.4238/2011.October.31.17

Gouma, P., Mallis, A., Daniilidis, V., Gouveris, H., Armenakis, N., & Naxakis, S. (2011). Behavioral trends in young children with conductive hearing loss: A case–control study. *European Archives of Oto-Rhino-Laryngology, 268*(1), 63–66. https://doi.org/10.1007/s00405-010-1346-4

Girirajan, S., Vlangos, C. N., Szomju, B. B., Edelman, E., Trevors, C. D., Dupuis, L., Nezarati, M., Bunyan, D. J., & Elsea, S. H. (2006). Genotype–phenotype correlation in Smith–Magenis syndrome: Evidence that multiple genes in 17p11.2 contribute to the clinical spectrum. *Genetics in Medicine, 8*(7), 417–427. https://doi.org/10.1097/01.gim.0000228215.32110.89

Greenberg, F., Guzzetta, V., Montes de Oca-Luna, R., Magenis, R. E., Smith, A. C., Richter, S. F., Kondo, I., Dobyns, W. B., Patel, P. I., & Lupski, J. R. (1991). Molecular analysis of the Smith–Magenis syndrome: A possible contiguous-gene syndrome associated with del(17)(p11.2). *American Journal of Human Genetics, 49*(6), 1207–1218.

Greenberg, F., Lewis, R., Potocki, L., Glaze, D., Parke, J., Killian, J., Murphy, M. A., Williamson, D., Brown, F., Dutton, R., McCluggage, C., Friedman, E., Sulek, M., & Lupski, J. R. (1996). Multi-disciplinary clinical study of Smith–Magenis syndrome (deletion 17p11.2). *American Journal of Medical Genetics, 62*(3), 247–254. https://doi.org/10.1002/(SICI)1096-8628(19960329)62:3<247::AID-AJMG9>3.0.CO;2-Q

Harris, P. A., Taylor, R., Thielke, R., Payne, J., Gonzalez, N., & Conde, J. G. (2009). Research electronic data capture (REDCap)—A metadata-driven methodology and workflow process for providing translational research informatics support. *Journal of Biomedical Informatics, 42*(2), 377–381. https://doi.org/10.1016/j.jbi.2008.08.010

Hidalgo-De la Guía, I., Garayzábal-Heinze, E., & Gómez-Vilda, P. (2020). Voice characteristics in Smith–Magenis syndrome: An acoustic study of laryngeal biomechanics. *Language, 5*(3), Article 31. https://doi.org/10.3390/languages5030031

Hildenbrand, H. L., & Smith, A. C. M. (2012). Analysis of the sensory profile in children with Smith–Magenis syndrome. *Physical & Occupational Therapy in Pediatrics, 32*(1), 48–65. https://doi.org/10.3109/01942638.2011.572152

IBM Corp. (2022). *IBM SPSS Statistics for Windows (Version 29.0)* [Computer software].

Jago, J. L., Jago, A. G., & Hart, M. (1984). An evaluation of the total communication approach for teaching language skills to developmentally delayed preschool children. *Education and Training of the Mentally Retarded, 9*(3), 175–182. https://www.jstor.org/stable/23877254

Jonsson, A., Kristoffersson, L., Ferm, U., & Thunberg, G. (2011). The ComAlong communication boards: Parents' use and experiences of aided language stimulation. *Augmentative and Alternative Communication, 27*(2), 103–116. https://doi.org/10.3109/07434618.2011.580780

Laje, G., Morse, R., Richter, W., Ball, J., Pao, M., & Smith, A. C. M. (2010). Autism spectrum features in Smith–Magenis syndrome. *American Journal of Medical Genetics Part C: Seminars in Medical Genetics, 154*(4), 456-462. https://doi.org/10.1002/ajmg.c.30275

Lieu, J. E., Kenna, M., Anne, S., & Davidson, L. (2020). Hearing loss in children: A review. *JAMA, 324*(21), 2195–2205. https://doi.org/10.1001/jama.2020.17647

Madduri, N., Peters, S. U., Voigt, R. G., Llorente, A. M., Lupski, J. R., & Potocki, L. (2006). Cognitive and adaptive behavior profiles in Smith–Magenis syndrome. *Journal of Development & Behavioral Pediatrics, 27*(3), 188–192. https://doi.org/10.1097/00004703-200606000-00002

Martin, S. C., Wolters, P. L., & Smith, A. C. M. (2006). Adaptive and maladaptive behavior in children with Smith–Magenis syndrome. *Journal of Autism and Developmental Disorders, 36*(4), 541–552. https://doi.org/10.1007/s10803-006-0093-2

Miller, J., Sedey, A., Miolo, G., Rosin, M., & Murray-Branch, D. (1991, November). *Spoken and sign vocabulary acquisition in children with Down syndrome* [Poster presentation]. Annual meeting of the American Speech-Language-Hearing Association, Atlanta, GA.

Nag, H. E., Nordgren, A., Anderlid, B. M., & Nærland, T. (2018). Reversed gender ratio of autism spectrum disorder in Smith–Magenis syndrome. *Molecular Autism, 9*(1), 1–9. https://doi.org/10.1186/s13229-017-0184-2

Osório, A., Cruz, R., Sampaio, A., Garayzábal, E., Carracedo, Á., & Fernández-Prieto, M. (2012). Cognitive functioning in children and adults with Smith–Magenis syndrome. *European Journal of Medical Genetics, 55*(6–7), 394–399. https://doi.org/10.1016/j.ejmg.2012.04.001

Pierce, J. E., O'Halloran, R., Togher, L., & Rose, M. L. (2019). What is meant by "multimodal therapy" for aphasia? *American Journal of Speech-Language Pathology, 28*(2), 706–716. https://doi.org/10.1044/2018_AJSLP-18-0157

Potocki, L., Glaze, D., Tan, D. X., Park, S. S., Kashork, C. D., Shaffer, L. G., Reiter, R. J., & Lupski, J. R. (2000). Circadian rhythm abnormalities of melatonin in Smith–Magenis syndrome. *Journal of Medical Genetics, 37*(6), 428–433. https://doi.org/10.1136/jmg.37.6.428

Potocki, L., Shaw, C. J., Stankiewicz, P., & Lupski, J. R. (2003). Variability in clinical phenotype despite common chromosomal deletion in Smith–Magenis syndrome [del(17)(p11.2p11.2)]. *Genetics in Medicine, 5*(6), 430–434. https://doi.org/10.1097/01.GIM.0000095625.14160.AB

Powell, G., & Clibbens, J. (1994). Actions speak louder than words: Signing and speech intelligibility in adults with Down

syndrome. *Down Syndrome Research and Practice, 2*(3), 127–129. https://doi.org/10.3104/reports.43

Rinaldi, B., Villa, R., Sironi, A., Garavelli, L., Finelli, P., & Bedeschi, M. F. (2022). Smith–Magenis syndrome—Clinical review, biological background and related disorders. *Genes, 13*(2), Article 335. https://doi.org/10.3390/genes13020335

Sedey, A., Rosin, M., & Miller, J. (1991, November). *The use of signs among children with Down syndrome* [Poster presentation]. Annual meeting of the American Speech-Language-Hearing Association, Atlanta, GA.

Serpentine, E. C., Tarnai, B., Drager, K. D. R., & Finke, E. H. (2011). Decision making of parents of children with autism spectrum disorder concerning augmentative and alternative communication in Hungary. *Communication Disorders Quarterly, 32*(4), 221–231. https://doi.org/10.1177/1525740109353938

Shayota, B. J., & Elsea, S. H. (2019). Behavior and sleep disturbance in Smith–Magenis syndrome. *Current Opinion in Psychiatry, 32*(2), 73–78. https://doi.org/10.1097/YCO.0000000000000474

Shriberg, L. D., Flipsen, P., Jr., Thielke, H., Kwiatkowski, J., Kertoy, M. K., Katcher, M. L., Nellis, R. A., & Block, M. G. (2000). Risk for speech disorder associated with early recurrent otitis media with effusion: Two retrospective studies. *Journal of Speech, Language, and Hearing Research, 43*(1), 79–99. https://doi.org/10.1044/jslhr.4301.79

Slager, R. E., Newton, T. L., Vlangos, C. N., Finucane, B., & Elsea, S. H. (2003). Mutations in *RAI1* associated with Smith–Magenis syndrome. *Nature Genetics, 33*(4), 466–468. https://doi.org/10.1038/ng1126

Smith, A. C. M., Boyd, K. E., Brennan, C., Charles, J., Elsea, S. H., Finucane, B. M., Foster, R., Gropman, A., Girirajan, S., & Haas-Givler, B. (2022). Smith–Magenis syndrome. In M. P. Adam, H. H. Ardinger, & R. A. Pagon, et al. (Eds.), *GeneReviews®* [Internet]. University of Washington. https://www.ncbi.nlm.nih.gov/books/NBK1310/

Smith, A. C. M., Dykens, E., & Greenberg, F. (1998). Behavioral phenotype of Smith–Magenis syndrome (del 17p11.2). *American Journal of Medical Genetics, 81,* 179–185. https://doi.org/10.1002/(SICI)1096-8628(19980328)81:2<179::AID-AJMG10>3.0.CO;2-E

Smith, A. C. M., McGavran, L., Robinson, J., Waldstein, G., Macfarlane, J., Zonona, J., Reiss, J., Lahr, M., Allen, L., Magenis, E., Opitz, J. M., & Reynolds, J. F. (1986). Interstitial deletion of (17)(p11.2p11.2) in nine patients. *American Journal of Medical Genetics, 24*(3), 393–414. https://doi.org/10.1002/ajmg.1320240303

Townsend, A., Harris, O., & Bland-Stewart, L. (2012). An ethnographic investigation of African American mothers' perceptions of augmentative and alternative communication. *Perspectives on Communication Disorders & Sciences in Culturally & Linguistically Diverse Populations, 19*(3), 84–89. https://doi.org/10.1044/cds19.3.84

Udwin, O., Webber, C., & Horn, I. (2001). Abilities and attainment in Smith–Magenis syndrome. *Developmental Medicine and Child Neurology, 43*(12), 823–828. https://doi.org/10.1017/S0012162201001499

Vlangos, C. N., Wilson, M., Blancato, J., Smith, A. C., & Elsea, S. H. (2005). Diagnostic FISH probes for del(17)(p11.2p11.2) associated with Smith–Magenis syndrome should contain the *RAI1* gene. *American Journal of Medical Genetics Part A, 132A*(3), 278–282. https://doi.org/10.1002/ajmg.a.30461

Wolters, P. L., Gropman, A. L., Martin, S. C., Smith, M. R., Hildenbrand, H. L., Brewer, C. C., & Smith, A. C. (2009). Neurodevelopment of children under 3 years of age with Smith–Magenis syndrome. *Pediatric Neurology, 41*(4), 250–258. https://doi.org/10.1016/j.pediatrneurol.2009.04.015

Speech and Language Development in SMS

Please complete the following survey. This information will help us better understand speech and language deveolpment among individuals with SMS.

Thank you!

Speech and Language Development in SMS	
Does the child with SMS (now or in the past) live with siblings or other children?	◯ Yes ◯ No
If you answered "Yes" to the previous question, please indicate how many other children and if they are older or younger than the child with SMS.	_____
Did your child babble as an infant?	◯ Yes ◯ No ◯ Maybe
Currently, is your child verbal (i.e. does your child communicate using speech)?	◯ Yes ◯ No ◯ Partially ◯ Other
Please explain.	_____
If verbal, at what age did your child begin to talk using single words (specify if age is in years or months)?	_____
If verbal, at what age did your child begin to talk using sentences (specify if years or months)?	_____
If your child is not verbal, how does he/she/they communicate?	_____
If your child (verbal or nonverbal) uses multiple methods of communication, please indicate all of those used regularly:	☐ non-speech vocalizations ☐ speech/speaking/talking ☐ one or more of the following: ASL, SEE, sign language, total communication ☐ picture communication ☐ voice output device ☐ iPad communication system ☐ gestures, pointing, touching, tapping
Does he/she/they communicate (speech, sign, or AAC) in single words or sentences?	◯ single words ◯ sentences

Does your child appear to understand single step directions (such as "turn the page" or "give me the ball")(even if he/she/they doesn't always comply with the direction)?	◯ Yes ◯ No ◯ Maybe
Does your child appear to understand two-step directions (such as "get the book and bring it to me") (again, even if he/she/they doesn't always comply with the direction)?	◯ Yes ◯ No ◯ Maybe
Does your child appear to attend to and understand simple stories (via listening)?	◯ Yes ◯ No ◯ Maybe
Does he/she/they answer questions (about wants, needs, events, feelings, stories)(again, this is about capability, not cooperation)?	◯ Yes ◯ No ◯ Maybe
Does he/she/they participate in conversations (for more than 2 turns - in other words, can comment or ask questions at least twice during a conversation)?	◯ Yes ◯ No ◯ Maybe
For his/her/their age, do you feel conversation skills lag behind typical peers?	◯ Yes ◯ No ◯ Maybe
Has he/she/they EVER received speech-language services?	◯ Yes ◯ No ◯ Maybe
Does he/she/they CURRENTLY receive speech-language services?	◯ Yes ◯ No ◯ Maybe
Does your child attempt to communicate with unfamiliar adults/children?	◯ Yes ◯ No ◯ Maybe
Do unfamiliar adults/children understand her/him/them?	◯ Yes (always or almost always) ◯ No (rarely or never) ◯ About 50% of the time ◯ Sometimes/often (50-80% of the time) ◯ Sometimes/but less often (25-50% of the time) ◯ Other
If other, please explain:	_____
If your child is unable to communicate effectively (or is misunderstood), does he/she/they attempt to communicate another way or with other words/methods?	◯ Yes ◯ No ◯ Sometimes ◯ Other
If other, please explain:	_____

Which of the following are relative strengths (strong compared to his/her/their other skills) in your child's communication profile (may select as many as are relevant)?	☐ speech/articulation ☐ vocabulary knowledge ☐ ability to formulate questions ☐ ability to formulate sentences ☐ ability to answer questions ☐ comprehension/understanding of stories ☐ ability to formulate and tell stories ☐ social skills ☐ social interest (interest in being social) ☐ humor ☐ problem solving ☐ memory for people, past events, and/or facts ☐ ability to follow directions ☐ motivation to be independent ☐ reading and/or writing ☐ other
If other, please specify:	_____
If your child has hearing loss, does he/she/they have a hearing aid or similar device to improve hearing sensitivity?	◯ Yes ◯ No ◯ Other ◯ No hearing loss
Did your child use ASL, sign, or another form of communication as a younger child but now relies mostly on speech?	◯ Yes ◯ No ◯ Other
If other, please specify:	_____
Can your child read?	◯ Yes ◯ No ◯ Reading skills are emerging ◯ Other
If other, please specify:	_____
If your child can read, what can he/she/they read independently? (check all that apply)	☐ words ☐ sentences ☐ cards ☐ simple books ☐ chapter books ☐ newspaper or magazine articles ☐ online content ☐ other
If other, please specify:	_____
Can your child write?	◯ Yes ◯ No ◯ Other

Appendix (p. 4 of 4)

Speech and Language Questionnaire From the SMS Patient Registry

If other, please specify:	_____
If your child can write, what can he/she/they write independently?	☐ his/her/their name ☐ words ☐ sentences ☐ simple stories ☐ cards or notes ☐ complex stories ☐ other
If other, please specify:	_____
Does your child have a gravelly or hoarse voice?	◯ Yes ◯ No ◯ Maybe ◯ Other
If other, please specify:	_____
Is there anything else about your child's speech, language, and communication development (including strengths and/or needs) that you would like to share, add, or clarify?	_____