SMS Patient Registry Results: Speech-Language Development and Hearing Health

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Introduction

Previous SMS research indicates speech delays, communication difficulties, and intellectual impairment (Dykens & Smith, 1998; Elsea & Girirajan, 2008; Girirajan et al., 2006; Greenberg et al., 1996; Smith et al., 1986). Additionally studies have also revealed impairment in working memory, verbal comprehension, vocabulary, and word reasoning with relatively unimpaired long-term memory (Osório et al., 2012; Udwin et al., 2001). Delays in speech-language development are further characterized by greater delays in expressive versus receptive language (Elsea & Girirajan, 2008). Individuals with SMS have been reported to use sign language when speech is delayed to decrease frustration and promote more positive communication behaviors (Elsea & Girirajan, 2008; Smith et al., 1998).

Previous 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; Girirajan et al., 2006; Greenberg et al., 1991, 1996; Potocki et al., 2003). Conductive hearing loss for younger children appeared to be related to otitis media (Brendal et al., 2016; Greenberg, et. al., 1996) and sensorineural loss appeared to be more common in older individuals with SMS (Brendal et al., 2016). Sensorineural hearing loss has also be associated with congenital abnormalities (Greenberg, et. al., 1996). A more recent and large longitudinal study of 133 individuals with SMS investigated the auditory phenotype of this syndrome (Brendal et al., 2016). Hearing loss ranging from mild to severe occurred in 72% of subjects across all age groups. Sensorineural hearing loss occurred most often in participants in the 11–49 year age range and conductive hearing loss affected approximately 35% of ears and was more prevalent in those in the 1–10 year age range (Brendal et al., 2016).

What remains unclear are details about the development of language and communication in those with SMS, such as when children begin speaking, and they begin formulating sentences, and the percent that use sign as a form of communication. It is also unclear what percent of those with SMS receive (or received) speech-language services during childhood or beyond. Further, there is limited research on the relationship between hearing health and speech-language development in those with SMS, such as if otitis media (i.e., middle ear infections) or hearing loss are associated with greater delays in speech-language development. As a result, educational and intervention teams who would be better informed about the impact of hearing

health issues on the development of effective communication skills make decisions despite this gap in the research. This lack of information could cause unnecessary delays in the provision of early evaluation and intervention related to hearing health and speech-language development.

Methods

This study analyzed a subset of data from the Smith-Magenis Syndrome Patient Registry, an initiative of PRISMS and housed/managed by the Baylor College of Medicine. Data for this study included responses about 82 individual with SMS on the Speech and Language Development questionnaire. This questionnaire focused on speech, language, communication, hearing, literacy, and medical health potentially related to communication status. Analyses included descriptive statistics (means, standard deviations, minimum and maximum values, and percentages), comparison of subgroups of subjects based on variables of interest (e.g., with and without hearing loss) utilizing separate analysis of variance testing (ANOVA), and correlation analyses (note: either Pearson, Kolmogorov-Smirnov, or Chi-squared tests were used depending on if variables were continuous and/or categorical).

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% of those with SMS were male. At the time of participation, the individuals with SMS ranged in age from 3-70 years. According to caregiver responses, 83% were white, 5% were more than one race, and 1% 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. Additionally, 82% were not Hispanic, 7% were Hispanic, and 11% did not indicate ethnicity. Eighty-nine percent reported living in the US. Seventy-two percent reported the individuals with SMS were currently attending school and 86% were reportedly living at home with parents or caregivers.

Results

Results related to speech/language (see Tables 1, 2, & 4): 92% used natural speech to communication (i.e., verbal communication), 79% began talking at or after 24 months of age, 92% had delayed language (i.e., combined words to form sentences at or after 36 months of age), and 53% used sign language before speech. The average age that first words were spoken was 26 months (range 11-72 months). The average age of first spoken sentences was 47 months (range 12-108 months).

Results related to hearing health (see Tables 2 & 4): 66% had a history of otitis media (middle ear infections), 62% had at least one set of pressure equalization tubes to prevent additional infections, and 38% had hearing loss.

There were no significant differences for age that first words or first sentences were spoken based on previous history of otitis media or the presence of hearing loss.

Significant correlations (i.e., $p<0.05$) were found between the age that first words were spoken and the age that first sentences were spoken.

Conclusions

While significant delays in speech-language delays were common, the vast majority of those with SMS developed the ability to communicate using natural speech by age 6 years. Neither recurrent otitis media nor the presence of hearing loss exacerbated speech-language delays. The correlation between age that first words and first sentences were spoken is consistent with a language delay/impairment. The correlation between age of pencil holding and age of first sentences suggests a general delay across domains rather than a language specific delay. These results confirm and extend previous findings about the nature of speech, language and hearing health in those with SMS.

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 spoken words later and on average, first spoken words may occur around age 2 years or later. Delays in the production of first spoken words and delays may be significant (not starting to talk until age 6 years). The current results also show that when individual children begin talking later, they are also more likely to begin formulating sentences later.

As previously reported (Elsea & Girirajan, 2008; Smith et al., 1998), use of sign language in those with SMS is prevalent and may help provide a means to communicate before children are using natural speech. Parents, educators, and clinicians should be aware that the use of sign or any alternative form of communication (AAC or Alternative Augmentative Communication) ***will not*** prevent children from starting to produce natural speech. In fact, using sign and/or AAC supports language development and provides the means to communicate and build vocabulary and language skills (Beukelman & Mirenda, 1998; also see Adamson and Dunbar, 1991; Sedey, Rosin, and Miller, 1991; Miller, Sedey, Miolo, Rosin, and Murray-Branch, 1991).

Hearing health (otitis media) and hearing loss is prevalent in this population but the current data does not show that middle ear infections or hearing loss exacerbate delays in speech-language development. In past studies with children who do not have SMS but do have recurrent middle ear infections and/or hearing loss, there is a risk of greater delays or challenges in speech-language (Lieu, Kenna, Anne, & Davidson, 2020). Perhaps since most children with SMS are already working with audiologists and speech-language pathologists, the possible negative impact of these factors 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 childhood.

Table 1. Descriptive statistics related to hearing health and early speech-language characteristics

	n+	Mean (sd)	Minimum	Maximum
Age* first words spoken	47	26 (15)	11	72
Age* sentences first spoken	47	47 (27)	12	108
Age* hearing loss first suspected	29	38 (90)	0	480
Age* PE tubes first placed	49	24 (14)	6	72
Number of PE tubes placed	50	3 (3)	1	18

* Age reported in months

+ Notes: The number of responses (subjects) for each question is given. Nonresponses are not included in this data. Means and ranges were calculated across all ages.

Table 2. Percent occurrence for early speech-language development.

	n+ (number of responses)	Percent responded "yes"
Communicates using natural speech (i.e., verbal communication)	52	92%
Late talker (first words spoken at or after 24 month; only calculated for those who are verbal)	47	79%
Delayed language (first sentences spoken at or after 36 months)	47	92%
Used sign before speech	34	53%

Notes: + Due to individual responses and non-responses to individual questions in the questionnaire, n is reported for each variable and percent reporting “yes” was calculated based on the number of responses for each given question.

Table 3. Hearing Health Statistics

Hearing Health Variables	Percent Occurrence*
History of Otitis Media	66%
History of PE Tube placement	62%
Hearing Loss+	35%
Conductive	34%
Sensorineural	7%
Mixed	17%
Type of hearing loss unknown by caregiver	41%
Diagnosed with Auditory Processing Disorder (APD)	2%
Uses a hearing device (out of 31 with a hearing loss)	13%
Used sign before speech	65%

*Across all ages groups (n=82)

+ Notes: There were nine non-responses for the hearing loss question (i.e., does your child have a hearing loss) and these responses were calculated having no hearing loss. Percent for each of the subtypes of hearing loss was calculated out of twenty-nine reported to have hearing loss.

Table 4. Statistics related to hearing and speech-language for each age group.

	Age Group								Significance	
	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)	F	Effect of age
Hearing										
Has hearing loss	38%	36%	29%	6%	62%	64%	100%	38%	2.598	***p*=0.038**
Hx+ of middle ear infections	38%	79%	79%	38%	92%	73%	100%	72%	3.237	***p*=0.007**
Has/had P.E. tubes	38%	71%	79%	38%	92%	64%	0%	72%	6.351	***p*<0.001**
Mode of Communication										
Currently uses sign	46%	29%	7%	0%	23%	45%	0%	28%	0.687	*p*=0.662
Currently uses natural speech	38%	71%	57%	56%	69%	64%	100%	72%	2.372	***p*=0.045**
Expressive Language										
Speaks in sentences	38%	71%	57%	50%	69%	36%	100%	67%	3.837	***p*=0.004**
Answers questions	62%	71%	57%	56%	77%	91%	100%	80%	3.226	***p*=0.014**
Takes turns in conversations	62%	79%	57%	56%	69%	64%	0%	77%	0.963	*p*=0.467
Receptive Language										
Understands spoken directions	62%	71%	57%	56%	69%	64%	100%	76%	0.275	*p*=0.947
Follows 2-step directions	69%	71%	64%	56%	62%	55%	100%	76%	0.763	*p*=0.605
Understands simple stories	69%	71%	57%	69%	77%	73%	100%	82%	2.215	*p*=0.069
Speech-Language Intervention										
Received SLT* in past	62%	71%	57%	56%	69%	64%	100%	76%	0.275	*p*=0.947
Receiving SLT* at time of study	46%	64%	43%	38%	0%	0%	0%	44%	25.329	***p*<0.001**
Voice										
Hoarse vocal quality	46%	50%	43%	44%	62%	45%	100%	57%	1.935	*p*=0.107

+Hx= history; *SLT=Speech-language therapy.

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