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An investigation of the language and communication characteristics observed in children with Smith-Magenis syndrome

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An Investigation of the Language and Communication Characteristics Observed in Children with
Smith-Magenis Syndrome

by

Anna Hesson

Thesis

Submitted to the Department of Special Education

Eastern Michigan University

in partial fulfillment of the requirements

for the degree of

MASTER OF ARTS

in

Speech Language Pathology

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September 29, 2014

Ypsilanti, Michigan

Dedication

This thesis is dedicated to my parents, David and Myra.

Thank you for your endless love, support, encouragement, and prayers throughout my educational studies. The completion of this milestone in my career as a speech-language pathologist would not have been possible without you.

Acknowledgements

I would like to express my gratitude to my advisor, Dr. Bill Cupples. His continuous support, advice, insightful criticisms, and encouragement aided in the achievement of this thesis in innumerable ways. It is rare for one to find an advisor and future colleague who always is available to listening to the little problems and roadblocks that unavoidably happen in the course of performing research.

The members of my thesis committee, Rhonda Vander Laan Kraai and Pam McClure, have generously given their time and expertise to better my work as a researcher. I thank them for their contribution and their support.

I would also like to express my deep gratitude to the Brehm family. Without their contribution, this thesis would not be possible. Thank you for your passion to inspire students to continue to better the field of education and make a difference in the lives of others.

Thank you to all of the parents who participated in the research, as well as PRISMS and SMS Foundation.

Abstract

This speech-language pathology study explores the communication characteristics exhibited in children with Smith-Magenis Syndrome (SMS), a recently identified genetic disorder arising from the deletion or mutation of part of the 17th chromosome. This qualitative study examines the speech and language needs exhibited by children with SMS through parent surveys and interviews, as well as current speech language pathology intervention strategies being utilized. It also observes the effectiveness and validity of the Language Use Inventory (LUI) as a reliable measure for identifying speech and language delays, which was analyzed in comparison to the MacArthur Bates Communicative Development Inventory (CDI). Results identified commonalities and differences in both language and communication among participants, as well as establishing the LUI as an accurate protocol for determining the speech and language development of children with SMS. Future research should continue to confirm precise language and communication needs of individuals with SMS that will help with the identification, intervention, and overall awareness.

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Definition of Terms

Autism Spectrum Disorder – A developmental disorder that affects children’s ability to communicate and interact socially.

Angelman syndrome – A complex genetic disorder that primarily affects the nervous system.

Cytogenetically – Relating to cytogenetics, which is a branch of biology dealing with heredity and the cellular components, particularly chromosomes.

Brachycephaly – Also known as flat head syndrome. This is a congenital malformation of the skull, which is a result of premature closure of the coronal suture, giving the head a short, broad appearance.

Brachydactyly – An inherited condition that causes shortness of toe and/or finger bones.

Down syndrome – A condition in which a person is born with an extra copy of chromosome 21.

Fluorescence in situ hybridization – A molecular cytogenetic method for identifying chromosomes and parts of chromosomes, deciphering chromosome rearrangements, and locating genes on chromosomes.

Fragile X – A genetic condition involving changes in part of the X chromosome.

Hypernasal – A speech disorder in which air leaks from the nose while speaking due to improper closure of the palate and pharynx.

Hypoplasia – The underdevelopment or incomplete development of a tissue or organ.

Hypotonia – Decreased muscle tone.

Prader-Willi syndrome – A rare, genetic disorder in which seven genes on Chromosome 15 are deleted or unexpressed on the paternal chromosome.

Preschool Language Scale-3 – A widely used standard assessment, which uses two broad subscales, auditory and expressive communication, to assess a variety of language-based skills.

Ra11 gene – Also known as retinoic acid induced 1. It provides instructions for making a protein

that is active in nerve cells in the brain.

Velocardiofacial syndrome – A genetic condition characterized by abnormal pharyngeal arch development that results in defective development of the parathyroid glands, thymus, and conotruncal region of the heart.

Velopharyngeal insufficiency – Inadequate closure of the velopharyngeal port or the soft palate muscle in the mouth, which can affect swallowing and speech.

Chapter 1: Introduction, Problem Statement, and Background

Smith-Magenis syndrome (SMS) is a rare genetic disorder associated with multiple congenital anomalies and cognitive impairments, first identified by Smith et al. in 1982 (Smith et al., 1982; Greenberg, 1991). It is caused by the deletion or mutation of the *RaI1* gene on chromosome 17p11.2. Prevalence for this neurobehavioral disorder is estimated to be one in every 15,000 to 25,000 births, with equal sex ratio across all ethnic groups (Elsea & Girirajan, 2008; Greenberg, 1991). Diagnosis of SMS is confirmed cytogenetically or by fluorescence in situ hybridization, as well as the recognition of the unique phenotype involving physical, developmental, and behavioral aspects (Finucane & Haas-Givler, 2009; Gropman, Duncan, & Smith, 2006). It is likely that the diagnosis of SMS is overlooked or delayed due to the lack of awareness and similarities of other identifying characteristics seen in other genetic disorders (Finucane & Haas-Givler, 2009; Gropman et al., 2006; Wolters et al., 2009). It is not uncommon for individuals to be initially misdiagnosed with autism, Prader- Willi syndrome, Down syndrome, velocardiofacial syndrome, Fragile X, or Angelman syndrome (Elsea & Girirajan, 2008; Gropman et al., 2006; Gropman, & Smith, 2009).

Individuals with SMS display clinically recognizable craniofacial and skeletal features including brachycephaly, midface hypoplasia, broad nasal bridge, flat mid-face, down-turned mouth with cupid's bow, broad hands, inwardly bent fingers, and short stature (Greenberg et al., 1991; Jung et al., 2009; Smith et al., 1986). Chronic ear infections, eye and vision abnormalities, hypotonia, brachydactyly, and cleft lip and/or palate have also been observed (Elsea & Girirajan, 2008; Gropman et al., 2006; Gropman, & Smith, 2009; Smith et al., 1986).

Aggressive and self-injurious behaviors such as hitting and biting oneself, hair pulling, and head banging occur in SMS as in some other syndromes, but skin picking, specifically around the fingernails and fingertips, distinguishes SMS from other syndromes (Finucane,

Dirrigl, & Simon, 2001; Martin, Wolters, & Smith, 2006). Other common and stereotypical behavioral problems include hyperactivity, inattentiveness, teeth grinding, inserting hands and objects into mouth, self-hugging, walking on tip-toes, and sleep disturbances, as well as deficits in all domains of adaptive behavior, including daily living skills and communication (Finucane et al., 2001; Madduri et al., 2006; Udwin, Webber, & Horn, 2001). It has also been noted that the behavior of some children with SMS falls between the mild to moderate classification of autism spectrum disorders (Finucane & Haas-Givler; 2009).

Children with Smith-Magenis syndrome have been identified with having speech and language deficits. However, few reports describe this particular area in detail, and objective data are limited. Wolters et al. (2009) conducted a study specifically focused on the neurodevelopment of children with SMS. Eleven children, 5 to 25 months of age, were evaluated with multidisciplinary assessments. A quick evaluation of the children's receptive and expressive language was measured with the Preschool Language Scale-3. Scores showed that expressive language was more impaired than receptive language, confirming previous informal assessments. Solomon et al. (2002) reported on the oral motor, speech, and voice functions in twenty-seven children with Smith-Magenis syndrome. Oral sensory motor deficits such as labial weakness, drooling, and open mouth posturing were observed in all of the children, as well as decreased range of motion and strength of the tongue. The participants' speech was described as being hypernasal with a hoarse and harsh vocal quality. Babbling, vocalization, and the use of gestures and signs were observed in non-speaking children and velopharyngeal insufficiency and voice impairments in speaking children. Overall, the study's findings reaffirmed functional impairment in swallowing, speech, and voice production. Although the study identified speech and voice issues, Solomon and colleagues research did not investigate other characteristics of speech, language, and communication.

These studies demonstrate that children with Smith-Magenis syndrome have speech and language deficits, but their descriptions of these abilities are cursory. There is no data identifying specific articulation or phonological errors, or a comprehensive description of the language and communication skills that children with SMS may exhibit.

Purpose of the Study

The purpose of this qualitative study was to determine with greater specificity what speech, language, and communication deficits are reported by parents of children with SMS, aged 24-56 months. The qualitative data were obtained from semi-structured interviews of parents conducted over the phone, and parent completion of the Language Use Inventory (LUI) and the MacArthur-Bates Communicative Development Inventory (CDI).

The Language Use Inventory (LUI) was developed to assess a child's language and communication abilities by having parents or caregivers complete a fourteen-page questionnaire (See Appendix C) arranged in four sections. Part 1 solicits parents' impressions of how their child communicates with gestures. Part 2 questions how their child communicates with words. Part 3 elicits parents' impressions of how their child communicates with sentences, including how the child interacts with others, converses, and is able to tell stories and narrate experience. Part 4 asks parents to report on any health issues their child has experienced, including whether hearing loss is an issue, and what other languages, if any, the child may have experienced.

The LUI is a standardized instrument that uses parent report to describe the language development and language use of children 18-47 months old in a range of every day interactions (O'Neill, 2007; Pesco & O'Neill, 2012). O'Neill (2007) conducted a study to investigate internal reliability and test-retest stability for the use of the LUI. One hundred seventy-seven parents participated in the first study and ninety-eight in the second study. Both groups completed the LUI protocol. O'Neill noted that, "Alpha values for the subscales for the LUI were at or above

acceptable levels (.80-.98) and steady growth in children's pragmatic language development was demonstrated. The study of discriminate validity revealed sensitivity and specificity levels over 95%" (p. 214).

Similarly, Pesco and O'Neill (2012) conducted a preliminary study examining the predictive validity of the LUI for accurately identifying later language outcomes in children 18-47 months old. The study consisted of 348 parents. Participants completed the LUI and then the participants' children were given a follow-up assessment of the Diagnostic Evaluation of Language Variation-Norm Referenced (DELV-NR), Clinical Evaluation of Language Fundamentals-Preschool, 2nd Edition (CELF-P2), Children's Communication Checklist-Second Edition, U.S. Edition (CCC-2), and a parent interview. The LUI and assessments scores were then examined for similarities in pragmatics. Significant correlations were found between the LUI Total Standard Score and three other language measures (CCC-2, CELF-P2, DELV-NR) with r-values ranging from .289 to .580, $p < .01$ for the age ranges of 24-41 months.

The Language Use Inventory has been validated through studies conducted by O'Neill (2007) and Pesco & O'Neill (2012). Findings of the studies show that the LUI is reliable for screening and evaluating toddlers and preschool children for early development of language, as well as aspects of pragmatics. It should be noted that independent researchers did not conduct these studies.

The MacArthur-Bates Communicative Development Inventory (CDI) is another standardized instrument that uses parent report to characterize early language skills of children (Dale, 1991) (See Appendix D). Dale (1991) conducted a study to assess the validity of the CDI, which consisted of twenty-four parents with their two-year-old children. The parents completed the CDI, and the Expressive One-Word Picture Vocabulary Test, the Memory for Sentences subtest of the Stanford-Binet Intelligence Scale: Fourth Edition, and selected items of the

Sequenced Inventory of Communicative Development were administered to the children. Results showed concurrent validity correlations in both expressive language and syntax with the parent report ($r = .73-.79$), demonstrating a high validity.

A similar study conducted by Thal et al. (1999) examined the validity of the CDI for children with specific language delay (SLD). There were a total of twenty participants who had children between the ages of 39 and 49-months. Children were previously diagnosed with SLD through the Goldman Fristoe Articulation Test of Articulation (GFTA), Expressive One-Word Picture Vocabulary Test (EOWPVT), the Peabody Picture Vocabulary Test–Revised (PPVT–R), and the Preschool Language Scale–R. Results showed a moderately high to high concurrent validity correlations in both domains, syntax and vocabulary (.67–.86).

The MacArthur-Bates Communicative Development Inventory has been validated through studies conducted by Dale (1991) and Thal et al. (1999). Results demonstrate that the CDI is a valid instrument in assessing children for early development of language, as well as language delays.

Justification and Significance

Currently, no formal research has been conducted specifically to determine the characteristics of speech, language, and communication abilities of children with SMS. Past research has primarily focused on the genetic and cognitive aspects of this recently discovered syndrome, resulting in limited information pertaining to the specific communication, speech, and language needs of individuals with SMS. It is important to establish a more precise definition of speech, language, and communication deficits in individuals with SMS to improve the identification, treatment and educational intervention for these children.

Chapter 2: Research Design and Methodology

Research Questions

This study explored the following questions:

What are the communication and language deficits exhibited by children with Smith-Magenis syndrome? This question establishes the speech and language needs among children with SMS, adding to past and current research, along with increased knowledge of the characteristics seen in individuals with SMS.

Are there any differences in the speech and language characteristics of children with SMS reported in this study? This question establishes if there are any differences in the speech and language needs among children with SMS, adding to past and current research, along with increased knowledge of the characteristics seen in individuals with SMS.

To what extent do the LUI and the MacArthur-Bates CDI describe similar characteristics of speech and language for children with SMS?

What are the general impressions of parents for speech language pathologists working with children with SMS? This final question takes into account parents' perspectives and opinions of the role of speech language pathologists in providing intervention for children with SMS. Gaining insight to parent impressions and possible therapy techniques used by speech language pathologists, will continue to develop the knowledge, planning, and implementation of interventions for these individuals.

Methodology

Participants. Seven families with children with Smith-Magenis syndrome (SMS) participated in the study. Participants were drawn from two sources: membership of the Parents and Researchers Interested in Smith-Magenis Syndrome (PRISMS: U.S. support group) and the National Smith-Magenis Foundation. The support groups agreed to forward letters to the families

of all individuals with children who were known to them. The letters contained information about the study and requested permission for the child's details to be passed on to the researchers. At the time of the study, children ranged in age from 30 to months 56 (Mean age = 42, Median age = 43). Individuals for the sample were as follows: five females and two males; one from Canada, two from the United Kingdom, and four from the United States. All seven children were diagnosed with SMS at birth due to other health factors, such as premature births and cleft palate. It is also noted that all of the participants continue to have significant health issues, such as ear infections, hearing loss, hypotonia, underdevelopment of the trachea, slow weight gain, and gastro esophageal reflux.

Procedure. Data were obtained from parent reports of communication and language abilities of their children with SMS using the MacArthur Communicative Development Inventory: Words and Gestures, and Words and Sentences (CDI-WG & CDI-WS) and the Language Use Inventory (LUI). These were mailed to parents before the informal phone interview. Participants returned the forms by mail or by email. Only the standard written instructions on the form were given.

Before parents of SMS children were allowed to participate, a consent form (see Appendix A) that outlined the purpose and procedures of the study was signed. Once signed and returned, caregivers first completed and returned the LUI and the CDI to solicit specific information about their impressions of their child's speech, language, and communication abilities.

Once completed, a secondary questionnaire (see Appendix B) was administered by the investigator via telephone interview to all of the families who agreed to participate in the first survey. The telephone interview discussed questions from the protocols in greater detail, including level of cognition, developmental level of speech, language, and communication

characteristics, and the preferred means of communication of their children. All interviews were audio recorded for transcription and accurate collection of data.

All participants were randomly assigned an identification number from 1 to 100 from a random table of numbers for the purposes of documentation. Only the randomly assigned code numbers had any indication of the subjects' identities. Consent forms, identifying information, audio recordings, transcriptions, and data were stored in a locked file inside the faculty advisor's office, as well as in a password-protected file on a password-protected computer.

Chapter 3: Results

Qualitative data gathered from participants' interviews were compiled and analyzed. Quantitative data, consisting of raw scores and percentages of each section from the LUI and CDI, were calculated in order to identify reported similarities and differences in speech, language and communication characteristics of the participants. Transcripts of the phone interviews were also analyzed and compared to determine preferred means of communication, duration and types of speech and language therapy received, and parents' reports of the recommendations they have received from their SLP's. The agreements between the CDI and LUI were analyzed to test the validity of the LUI in identifying speech and language delays in children. Results from the study provided specific information pertaining to the strengths and weaknesses of individuals with SMS in the areas of speech, communication, and language, as well as the use of the LUI for accurate measurement of language development.

Common Themes Revealed in the Interviews

Analysis of the phone interviews revealed common themes among participants. Table 1 summarizes the common themes reported by the parents in their interviews.

Table 1

Summary of Interview Themes

Delayed Speech	Adult Interaction	Behavioral Issues
“Her speech is definitely not on par with other children her age.”	“He charms adults.”	“He will cry and fall to the ground... he recently starting throwing things at us and in the air.”
“His speech and language is severely delayed.”	“He is very good at building relationships with adults.”	“She can be aggressive with other children.”
“Her speech is also 8-12 months delayed.”	“She really likes to interact with people.”	“He’s like Mr. Jekyll, Mr. Hyde. He will switch very quickly. Sometimes we don’t even know what triggers it.”
“Her speech is delayed to her peers.”	“He likes to play with adults but not children.”	“Sometimes she lashes out without any prorogation by scratching. She will start hitting her head on the floor or anything.”
“His speech is behind... he just started to speak.”	“She likes to please people.”	“He will beat us up. He doesn’t understand consequences.”
“He doesn’t use words like typically developing children.”	“He wants to play with adults... he would much rather play in the kitchen by himself or with the teacher.”	“He’ll take it out on the dog and me. Most of the time it’s screaming and hurting himself.”
“His speech is behind.”	“She is very good at interacting with people, more so than children her age.”	“She will just drop to the floor and start scuffling and screaming.”

Final Consonant Deletion	Outside Play	Water Play
“She will say part of the word, like more = ‘mo’ and sign it.”	“He loves being outside. That was literally his first word.”	“He loves sensory stuff, especially water. I will fill up the sink and let him go at it...”
“He will say yellow bus, blue car, or little dog, but they don’t sound like word words. He will	“She loves going out in the garden when it is nice.”	“In the bath, she loves to splash and play with her water toys.”

say 'bu ca' for blue car."		
"He just started talking, but will only say the first part of the word."	In the garden, she will kick a ball and go on her swing."	"She loves being in the water."
		"Water play, he loves it."

Recognizes Emotions in Others	Independently Requests Activities	Sign Language
"He internalizes it as if we were talking to him or his brother."	"He will say 'outside' and point towards the outside."	"He prefers natural gestures with some sign language and words."
"When her little brother or if I were to be upset, she would get upset too."	"It's (activities) just available to her, but she will ask for books."	"I would say a combination of words and signs. Most of them are ASL... she doesn't have the skills to do full signs but are similar to the original."
"He will see a face in a book and name it. Also by facial expressions and tone of voice, he can tell that too."	"He can request anything he wants now."	"He uses sentences always. Sometimes he will use a few signs to help clarify a word, but always sentences."
"When you say in a stern voice or tell her off, she knows she's done it wrong."	"She will tell you that she wants to go in the garden and point to it."	"She uses a combination of putting two words together and signing."
"When her sister is crying, she'll sign and pat her on the back."	"He is independent at asking for these things (iPad, train, singing)."	"He doesn't point much, he probably signs something a couple times a day. He prefers to use his speech."
"Somewhat. He will give you a hug if you are upset."	"She will point to them and ask for it."	"Now, more and more she prefers to use her words, but typically does a combination of words and signs."
	"He will either point to what he wants on the communication board or tell us."	"He uses a combination of words and signs."

Speech Therapy Sessions	Therapy Approaches	Parents' Desire to Increase Language
"Now that he is in school, it	"The communication sheets	"I would like him to be able to express himself more through

(therapy) is 2 sessions for 60 minutes.”	have been wonderful.”	speech... we want him to be able to speak more, because he knows more than he can say.”
“Once a month for typically 45 minutes.”	“They’ve given us some sort of laminated picture cards for her.”	“ I would like him to have more speech therapy. I think it’s a matter that everyone around him encourages him.”
“It’s now once a week for 45 minutes because of all of the other therapy.”	“Mixing up the session is really the best for him.”	“Putting more words together to make sentences would be great. It would allow her to explain herself more. It would help us with what upsets or triggers her.”
“She sees the speech and language therapist every 5-6 weeks in her nursery between 1-1.5 hours.”	“The suggestion of using sign language has been a Godsend to use and it’s been the most helpful.”	“I would love for him to speak in clearer sentences or words that others can understand him... I want him to sit in society on his own.”
“He has private speech therapy 2 times a week for 30 minutes each session.”	“Physical touch on his mouth/throat and using sensory stuff... I think it woke him up a little.”	“I would like to see him getting the vowels connected to the consonants and the use of the visual schedules at home.”
“They should be 1 session every 3 months.”	“They are working on /b/ and /p/. Blowing feathers. Working on oral motor exercises with a focus on /b/ and /p/, as well as closure.”	“Just to continue with verbalizing words and start to questions and let us know what’s going on. Just more speech and words.”
“Private therapy once a week for 30 minutes and school therapy twice a week.”		

All parents confirmed cognitive, developmental, and speech/language delays. Of the seven, five (#006, 020, 021, 033, and 095) also reported physical difficulties in fine and gross motor movement. For example, all five participants had difficulties going up and down stairs. Behavioral issues were also noted in all seven participants. Five of the participants’ parents (#020, 021, 033, 043, and 095) specifically stated self-injurious behaviors, a common characteristic exhibited in children with SMS (Finucane, Dirrigl, & Simon, 2001; Martin,

Wolters, & Smith, 2006). The other two (#003 and 006) described tantrums and meltdowns as issues, but did not specify self-injurious behavior. However, all stated that behavioral issues typically arose without cause. Participants were also asked to describe recognition of emotions and feelings. All except one participant (#043) reported that their children were able to recognize emotions in others, as well as internalize the emotions of others.

Of the seven participants, six (#003, 006, 021, 033, 043, and 095) reported that their children independently requested toys or activities through the use of gestures and words, typically in phrases 1-3 words in length. The seventh participant, who was the exception, spoke in full sentences. It was also noted that the six participants also imitated gestures and words when prompted by parents.

Significant differences were also noted through the phone interviews pertaining to the children's speech and language, as well as intervention time and methods used by speech language pathologists. Six of the participants (#006, 020, 021, 033, 043, and 095) gave specific examples of their children's speech showing differences in speech and language errors. Four had final consonant deletion of words, e.g. more = "mo," car = "ca," and toilet = "toile." Two participants reported speech sound substitutions of words, such as Jonny = "Donny" and the = "da." Other reports of speech and language errors were cluster reductions and initial consonant deletions.

All participants stated that their child was receiving speech therapy services, however session time and frequency of sessions were quite variable. Sessions ranged from once every three months to twice a week for thirty minutes to sixty minutes. Therapy methods and interventions were also differentiated, varying between sign language to oral motor exercise to high tech augmentative and alternative devices. It was also noted that the participants were interested in different types of activities; however, all had sensory experiences as a component of

therapy. Four of the seven participants (#020, 021, 033, and 095) reported extensive fascination with water play and outside play.

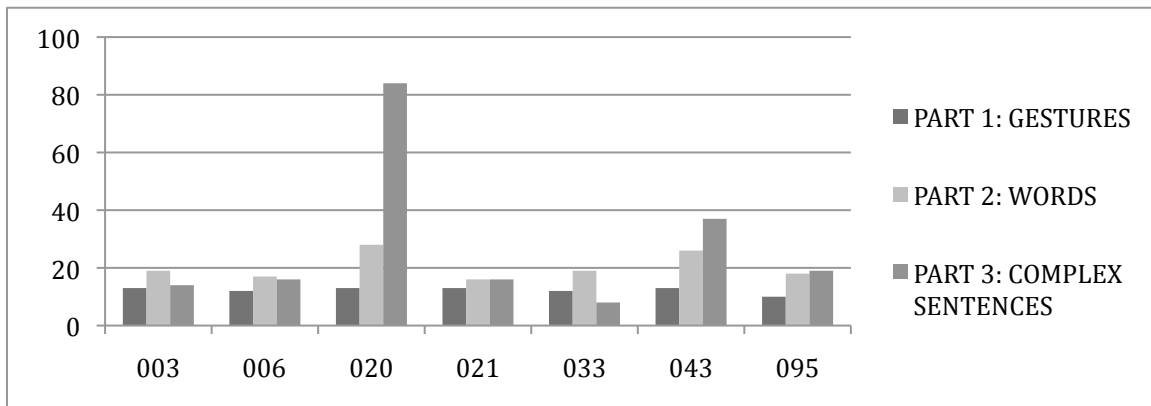
Description of Scores Obtained on the Language Use Inventory

Analysis of the use of gestures, words, and complex sentences through the Language Use Inventory (LUI) showed similarities between participants. Table 2 and Figure 1 present the results obtained.

Table 2
Summary of raw scores from the Language Use Inventory

Participant #	Part 1: Gestures	Part 2: Words	Part 3: Complex Sentences
003	13	19	14
006	12	17	16
020	13	28	84
021	13	16	16
033	12	19	8
043	13	26	37
095	10	18	19

Total raw score: Part 1 = 13; Part 2 = 28; Part 3 = 133



Total raw score: Part 1 = 13; Part 2 = 28; Part 3 = 133

Figure 1. Summary of raw scores from the Language Use Inventory

All children showed commonalities in communication through gestures with raw scores differing by +/-1. Scores ranged from 10 to 13 with a mode of 13. Raw scores in part 2 of the LUI: Communication in Words also showed similarities, however the range of scores varied

more. Five participants (#003, 006, 021, 033, and 095) fell within +/- 3 of the raw score of 18, showing similarities in language use through communication with words. The two outlying scores, 26 and 28, differed by a raw score of +6-10 from the other five participants. Although participants #020 and #043 demonstrated a greater use of words in communication from the other participants, the two scores were close in number (#020 = 28 and # 043 = 26). Complexity of Sentence Length in part 3 of the LUI showed more discrepancy in the raw score range. Scores varied from a raw score of 8 to 84. This suggests that complex language structure is variable between the children with SMS in this sample.

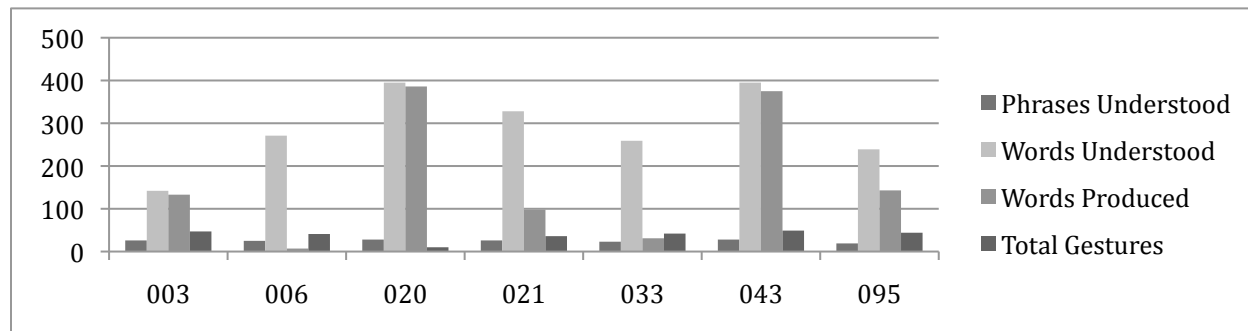
Description of Scores Obtained on the MacArthur Bates CDI: Words and Gestures

Phrases understood by participants through analysis of the MacArthur Bates CDI-WG showed minimal variability from the raw score obtained by all 8 participants. Table 3 and Figure 2 present the results obtained.

Table 3
Summary of raw scores from the MacArthur Bates CDI-WG

Participant #	Phrases Understood	Words Understood	Words Produced	Total Gestures
003	26	142	133	47
006	25	271	107	41
020	28	395	386	41
021	26	328	98	36
033	23	259	31	42
043	28	395	375	49
095	19	239	143	44

Total raw score: PU = 28; WU = 396; WP = 396; TG = 63



Total raw score: PU = 28; WU = 396; WP = 396; TG = 63

Figure 2. Summary of raw scores from the MacArthur Bates CDI-WG

Scores ranged between 19 and 28 phrases understood out of a total of 28. Two participants (#20 and #043), who were also the oldest with ages of 52 and 56 months, scored a perfect score of 28. This may account for the higher understanding of phrases. Of the five participants with lower scores, all had difficulties with some of the command phrases, such as “Hold still.”

Greater discrepancy was noted in the total number of words understood by participants. Scores varied between 142 and 395. The total “number of words understood” were out of 395 words. Once again, the two oldest participants obtained a perfect score.

Data gathered on words produced also showed variability between participants. Scores were dispersed from 91 to 386 out of 396 words, showing no definitive vocabulary size for children with SMS in the specific age range included in this study.

Within the section Total Gestures, there was less variability, with scores falling in the range of 40-56 out of 63. Specifically looking at early gestures, all participants fell between 15-18 out of 18, with two outlying scores of 12 (#006 and 033). In comparing scores in later gestures, scores varied from 21 to 40 out of 45. With the exception of two participants (#020 and 021), who received a score of 21 and 22, the others scored within a small range, with scores of 28, 29, 30, 30, and 32.

Description of Scores Obtained on the MacArthur Bates CDI: Words and Sentences

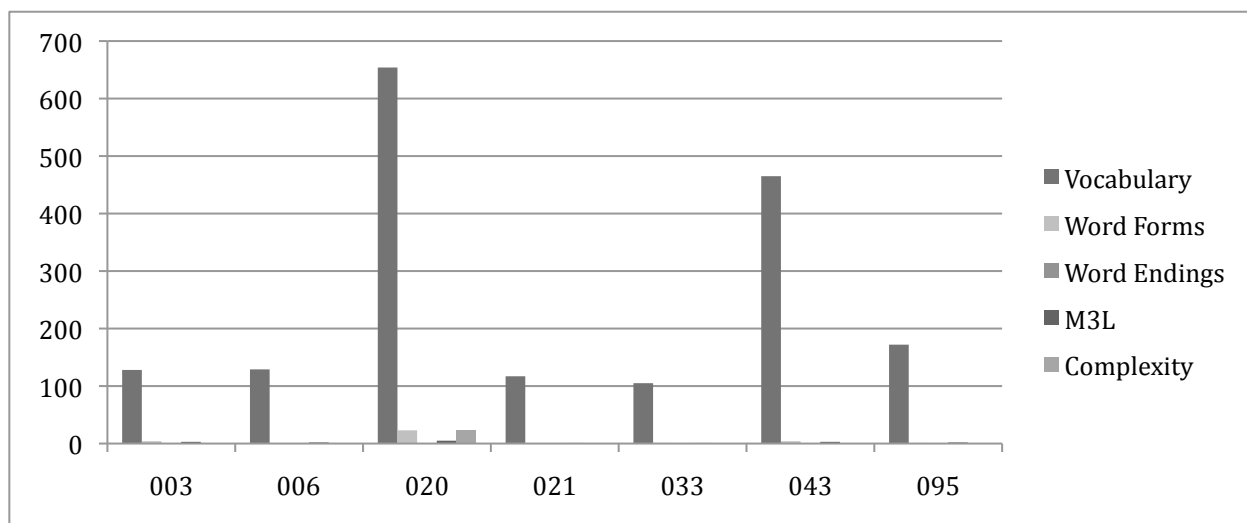
Analysis of the MacArthur Bates CDI: Words and Sentences showed a variation across vocabulary and word forms. However, there are noted similarities between participants in word endings and mean length of three utterances. Table 4 and Figure 3 present the results obtained for this measure.

Table 4
Summary of raw scores from the MacArthur Bates CDI-WS

Participant #	Vocabulary	Word Forms	Word Endings	M3L	Complexity
003	128	4	0	3	0
006	129	0	0	2	0
020	654	23	1	5	23
021	117	0	0	1	0
033	105	1	0	1	0
043	465	4	0	3	0
095	172	1	0	2	0

Total raw score: V = 680; WF = 25; WE = 45; C = 37

*M3L = number of morphemes of child's three longest sentences



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Figure 3. Summary of raw scores from the MacArthur Bates CDI-WS

Variability was noted in all participants for current vocabulary size. The vocabulary checklist consisted of a variety of words ranging from sound effects to connecting words (conjunctions) for a total score of 680. Scores of the participants ranged between 105 and 654. All participants showed difficulties in the sections on quantifiers, articles, helping verbs, and connecting words. Possible reasons for vocabulary differences could be associated with age, use of sign language, family interaction/involvement, and type and amount of therapy.

Findings based on word forms for children with SMS showed inconsistency in the particular words used, however percentiles of number of words used were similar. The score was based out of 25 irregular nouns and verbs that a child above the age of 24 months should begin to acquire. All participants fell within the 0 to 4 raw score range with one outlying score of 23 from participant #020. It is noted that the participant with a word form score of 23 out of 25 received 18 hours of therapy a week with highly involved family members. Scores may suggest that the hours of therapy and family interaction affect the amount of word forms that a child with SMS acquires.

The word ending score was based on use of the past tense -ed ending and plural -s on words in expressive language. The highest score that a participant could receive was 25. Six of the seven participants received a word ending score of 0 (#003, 006, 021, 033, 043, and 095). The one participant (# 020) with a score of 1/25 only over generalized the plural -s on socks. Socks became “sockses.” Data from the sample in this study suggests that children with SMS between the ages of 24-56 months have difficulties in the development and use of morphemes, characterized by a lack of past tense use and pluralization.

Analysis of the M3L showed variability in sentence and utterance length. The M3L was calculated by counting the number of morphemes of the three longest utterances produced by each participant. The total amount of morphemes was divided by three, providing an average of

the typical length of the longest utterances produced by each child. The six participants who received similar scores in the words ending measure (#003, 006, 021, 033, 043, and 095) demonstrated utterance lengths varying between 1 and 3 morphemes. Three participants had a M3L of 1, two had a M3L of 2 and two had a M3L of 3; averaging between 2-3 M3L. Brown (1973) stated that a child with a Mean Length of Utterance (MLU) between 2-3 utterances falls within stage II and III and should be 27 to 34 months of age. This supported the evidence that children with SMS have language development delays. It is to be noted that the participant #020 had a M3L of five, showing typical morphological development.

Areas of Agreement Between Language Use Inventory and MacArthur Bates CDI

Scores from the Language Use (LUI) Inventory and MacArthur Bates CDI: Words & Gestures and Words and Sentences (CDI-WG & CDI-WS) were examined to determine the validity of the LUI as a reliable form for determining language delays in children. Comparisons using the CDI were selected because the MacArthur Bates CDI is an empirically well-validated assessment of language and communication development in young children (Dale, 1991; Thal et al., 1999). The LUI specifically concentrates on identifying communication through gestures and words, and longer sentences produced. In comparison, the CDI-WG seeks to identify understanding of first signs and actions and gestures and the CDI-WS with words used and sentences and grammar. Due to participant ages falling outside of the normative ranges, scores were evaluated by percentage of correct and not by percentiles.

Comparing sections focused on gestures & words and complex sentences, percentages for communication showed variability between the LUI and the CDI. Percentages taken from the LUI raw scores for communication through gestures were closely related, showing typical development of gestures (77%-100%). However, when compared to the CDI section for total gestures, scores differed. CDI total gestures showed percentages closely related and possible

developmental language delays (57%-78%). Scores from the LUI communication through gestures could be interpreted incorrectly and may provide misleading impressions of more advanced gesture use than is actually present.

Analysis and comparison of the raw scores for the LUI and CDI focused on communication through words, revealing percentage variability, but were similarly related. Although half of the percentages from the LUI and CDI differed by almost half (e.g. LUI: 64% and CDI: 36%), analysis of percentiles were in agreement. Percentiles for the CDI-WG were extrapolated, since participants' ages fell outside the norms provided by the CDI. Since participants' ages and raw scores fell below 50% out of the total words assessed, participants' communication through words was extrapolated to be ≥ 1 percentile. It was also noted that two of the participants' raw scores (#020 and 043) were closely related, LUI = 100%, 93% and CDI = 97%, 95% confirming agreement between communication through words in both the LUI and CDI.

Complexity of sentences and sentence length were analyzed through percentages calculated from raw scores obtained through the protocols. Comparison of percentages of the LUI and CDI were closely related. Scores differed by an average of 8%, with the exception of two. The two participants (#020 and 043) who had conflicting percentages were the oldest of the all, 52 and 56 months, respectively, which probably explains the variance. However, for participants (4/7) that fell within the norms for the LUI, percentiles were obtained. Raw scores showed all participants falling just above the first percentile. It was concluded that the percentile in which the participants fell within for the CDI-WS from raw scores were below the 5th percentile regardless of age, indicating significant communication delays in this sample of children with SMS.

Similarities Observed in the Interviews and the LUI

Analysis of the phone interviews and Language Use Inventory questions completed by participants revealed commonalities in language and speech. All participants communicated through the combination of natural gestures (pointing, pulling), sign language and 1-3 utterances, with the exception of one. For the individual who did not use natural gestures (#020), it was confirmed in both the interview and LUI that the child used 4-5 words with occasional signs for communicating. Communication for all participants consisted of naming and independently requesting items, as well as asking and answering simple concrete questions. It was also confirmed that participants imitated gestures, words, and sentences when prompted by others.

Chapter 4: Discussion

The present sample constitutes the first study focused specifically on identifying the speech and language characteristics exhibited by children with Smith-Magenis syndrome between the ages of 25 and 56 months of age. It is to be noted that it is not clear how representative the present sample is of the whole population for individuals with SMS falling within this age group.

All children in the present study had speech and language delays as reported by their parents, which is consistent with previous reports of the communication abilities of individuals with SMS. Analysis of the interviews, LUI, and CDI confirmed significant deficits in participants' speech and language development. Data showed that the children exhibited similarities in their language, which were characterized by an average of 1-3 word utterances with the combination of natural gestures, sign language, and words with a variation in vocabulary size. Prior research and widely used language milestone markers by Brown (1973), states that a typically developing child between 27 to 34 months of age should have a Mean Length of Utterance (MLU) of 2-3 words. In contrast, one participant obtained a MLU of five, showing typically developing morphological development. This difference may well be the result of the extensive therapy which the individual received on a weekly basis and reported higher IQ of 105.

Along with a smaller utterance length, children with SMS have difficulties in the development and acquisition of complex language. Morphological and syntactic errors were characterized by a lack of past tense use and pluralization. Comparison of the language development with Brown's Morphemes (1973), the expected age of mastery of regular plurals and irregular plural words are between 28 and 42 months of age. Mastery of past tense -ed is between age 40 and 46 months. Sentence context contains nouns and directional words with the

lack of articles, verbs, adjectives, and conjunctions. It appears that individuals with SMS easily attain concrete language, however, abstract language appears to be more of a challenge.

As reported in a study by Wolters et al. (2009) and Solomon et al. (2002), individuals with SMS have speech impairments, however not specified. Findings from this study, per parent report, identified that children with SMS exhibit articulation errors of substitutions, distortions, and consonant deletions. These speech errors could be partially secondary due to hypotonia and/or decreased muscle coordination in gross and fine motor skills, which was also identified and confirmed. Such findings can help with the development of intervention strategies targeting articulation and motor speech.

Procedures and intervention strategies currently utilized by speech-language pathologists were variable and inconsistent. The apparent lack of knowledge of this syndrome, as well as possible outcomes of speech and language development in this population, may have an impact on the selection of the most appropriate and most effective treatment approaches to be used when working with clients with this specific syndrome.

Examination of the LUI in relation to the CDI as a reliable predictor for speech and language delays in children was confirmed. The LUI as a parent report of their child's speech and language development is accurate in identifying delays in communication. Percentiles obtained by participants on the LUI and CDI were in agreement with the expectation of Part 1 of the LUI, which focused on the use of gestures. Results showed that raw scores on the LUI, when compared to the CDI, were much higher (LUI range = 77 to 100%, CDI range = 66 to 78 %). Thus the LUI may over-estimate the gestural communication abilities of children with SMS. With the exception of gestural abilities, it appears that the LUI can be used as a quick and accurate protocol for determining the speech and language development of children with SMS.

More detailed assessment would still be needed to verify any speech and language delays once the LUI was administered.

Implications for working with families with SMS suggest immediate early intervention once diagnosis is confirmed. Based on the significant expressive language and speech motor delays noted in this sample of children with SMS, it is recommended that appropriate alternative/augmentative communication systems be implemented at an early age.

Implementation of these systems may decrease communication frustration for the child and improve behavioral regulation. Sessions could also focus on the development of oral motor abilities in order to encourage the development of intelligible speech. Through parent report, it is recommended that sessions are more likely to have success in maintaining the child's interest through highly interactive activities and duration of an activity for no more than five minutes. Therapy should build on any areas of strength exhibited by individuals, as well as use of materials to improve sensory integration.

Chapter 5: Limitations and Recommendations for Further Research and Action

Limitations/Delimitations of the Study

The results of the study should be considered preliminary due to the small sample size of children with SMS. Data collected demonstrates a sampling of speech and language characteristics observed in children with SMS through parent interview and report. Additional limitations to the study include that the participants' speech and language abilities were not assessed by a certified speech and language pathologist due to distance and time constraints, and that the diagnosis of SMS was assumed because of participation in the PRISMS group, the National Smith-Magenis Foundation, and parent report. However, this will be one of the first studies of young children with SMS explicitly examining their language and communication deficits. Despite the lack of normative data for the percentiles for the protocols, measureable scores and information were obtained that indicated markedly consistent descriptions of the speech, language, and communication abilities of the children with SMS in this study. Parent reports of their children's abilities also provided similar descriptions of speech, language, and communication abilities of the children with SMS in this study.

Future Research

Future studies need to continue to examine the speech and language characteristics exhibited by children with SMS of all different age groups, creating more clearly defined descriptions of communication. In addition to parent report, more in-depth assessment of the speech and language abilities of these children is needed. Further, more studies should be conducted on the efficacy of augmentative and alternative communication devices as an efficient and effective form for improving the communication of individuals with SMS. Lastly, studies comparing the speech and language abilities of children with SMS to children with other forms of developmental challenges should be completed. Due the speech and language characteristics

of children with SMS constitute a discrete sub-type of severe language and communication impairments, or are these characteristics representative of a more general impairment in communication in children, regardless of the diagnosis? For instance, are the severe communication impairments in autism spectrum disorders, fetal alcohol syndrome, etc. different from those observed in SMS? If so, then different assessment and intervention strategies need to be implemented.

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Appendix A: Informed Consent

Anna Hesson, B.S.

Bill Cupples, Ph.D., CCC-SLP

I agree to participate in three interviews for a research study about the speech, language, and communication characteristics of children with Smith Magenis Syndrome. The first two interviews, the Language Use Inventory and the MacArthur-Bates Communicative Development Inventory will be mailed for you to complete. The other interview will be conducted by Anna Hesson, a graduate student in the Speech and Language Pathology program, at Eastern Michigan University.

I also understand that my confidentiality will be protected at all times and that fictitious names will be assigned to me in any research presentations and written reports. In addition, I may request copies of my recorded interview and/or transcriptions of that interview. I may also request portions of the recording or transcriptions be deleted if I find that necessary. Although unlikely, I may experience mild emotional concern as a result of talking about my experiences with a child with Smith Magenis Syndrome. By choosing to participate in the study, I will be contributing to the understanding of the speech, language and communication characteristics of children with Smith Magenis Syndrome.

I understand that my participation is completely voluntary and that I may withdraw from the project at any time if I choose without negative consequences.

I understand that the results of the interviews can be used in presentations at research meetings, conferences, and in publications by Anna Hesson and Dr. Cupples.

If I have any questions or concerns, I may contact Anna Hesson or Dr. Cupples (contact information below).

ahesson@emich.edu

(586)453-3375

Researcher Signature: _____ Date: _____

Participant Print Name: _____ Date: _____

Participant Signature: _____ Date: _____

If I have any concerns about this project, I may also contact the faculty supervisor:

Bill Cupples, Ph.D., CCC-SLP

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This research protocol and informed consent document has been reviewed and approved by the Eastern Michigan University Human Subjects Review Committee (UHSRC) for use from 1/01/14 to 1/01/15. If you have questions about the approval process, please contact the UHRSC at human.subjects@emich.edu or call 734.487.0042. Please refer to the College of Education Human Subjects Review Committee, Dr. Beth Kubitskey, administrative co-chair, 734-487-1416, mkubitske1@emich.edu.

Appendix B: Phone Interview Questions

How is your child similar to other children his/her age?
How is your child different from other children her/his age?
What are some areas that you feel your child is successful or good at.
What are some areas that you feel are challenging for you to manage with your child.
How does your child express his/her feelings and emotions?
How does your child recognize feelings and emotions in others?
Does your child describe her/his feelings and emotions? If yes, describe how.
How does your child PREFER to communicate?
 Gestures, words, sentences, pictures, an electronic device, a combination? Any other ways?

Language Use Inventory

In the Language Use Inventory you described three favorite activities of your child: (restate the caregiver's response)

Describe what she/he does with these toys, or in these games, or activities.

On a scale of 1 to 5, how long will your child participate in these activities?

(1.) Briefly, (2.) short period of time, (3.) Appropriate amount of time, (4.) Will do it until I tell him/her to stop, (5.) Difficult to make him/her stop)

How frequently does she/he engage in these activities?

(How many times a day? How many days a week?)

Does he/she request the toys, games or activities independently?

If so, how does she/he request them?

Does she/he prefer to do them alone or with others?

If with others, describe how your child engages with others.

IF GESTURE USE IS INDICATED IN THE LUI:

1. You stated in the Language Use Inventory that your child uses gestures to communicate.

Tell me more about how your child uses gestures in these activities.

How frequently does your child use gestures in these activities? (rarely, sometimes, often)

Does your child use gestures independently in these activities?

Does your child imitate gestures in these activities?

IF WORD USE IS INDICATED IN THE LUI:

2. You stated in the Inventory that your child uses words to communicate.

Tell me more about how your child uses words in these activities.

How frequently does your child use words in these activities? (rarely, sometimes, often)

Does your child use words independently in these activities?

Does your child imitate words in these activities?

IF SENTENCE USE IS INDICATED IN THE LUI:

3. You stated in the inventory that your child uses sentences to communicate.
Tell me more about how your child uses sentences in these activities.
How frequently does your child use sentences in these activities? (rarely, sometimes, often)
Does your child use sentences independently in these activities?
Does your child imitate sentences in these activities?

Role of the Speech-Language Pathologist:

Has your child seen a speech therapist or is your child currently seeing a speech therapist?
How long? How frequently? Duration of sessions?

Is there anything the speech therapist has suggested that has been particularly helpful?
Is there anything the speech therapist has suggested that has not been particularly helpful?
Please describe any therapy approach that you feel has been beneficial for your child.
Has a particular kind of therapy been tried that you feel has not been beneficial for your child?
What would you like to happen next with your child to improve his/her speech, language, and/or communication abilities?