**Title**: Communication, Cognition, and Behavior in Smith-Magenis Syndrome: Qualitative Data from a Scoping Review

**Authors**: Jamie Linert1, Miriam Kornelis 1, & Lizbeth H. Finestack1

**Introduction**: Smith-Magenis syndrome (SMS) is a developmental disorder associated with a genetic deletion at chromosome 17p11.2 and pathogenic RAI1 variants (Elsea et al., 1997; Slager et al., 2003). It is estimated that SMS occurs once in every 15,000-25,000 live births (Elsea & Girirajan, 2008; Smith et al., 2005). The profile of common features such as severe challenging behaviors, sleep disturbances, and a varied cognitive profile are well represented in the literature (Dykens et al., 1997; Greenberg et al., 1996; Finucane et al., 2001; Martin et al., 2006; de Leersnyder, de Blois, et al., 2003). The language profile of people with SMS, however, is not nearly as well understood even though language impairment is present in nearly all cases of SMS (Smith et al., 2019). The purpose of this review is to synthesize the research on the language profile of individuals with SMS and identify specific topics in language and SMS to motivate future research. The specific research questions we will address in this presentation are: 1) What domains of language and communication in people with SMS are being measured qualitatively? 2) What are common themes and findings? This presentation will answer these questions, discuss the implications of the themes that are represented in the literature, and suggest future research.

**Method**: Our initial search yielded 523 articles related to SMS research involving human participants. We used the online screening tool *Rayyan* (Ouzzani et al 2016) to screen these articles. To be included, articles must have featured people with SMS as the primary participants and reference language or communication. Two of the authors independently conducted a title and abstract screening and a full-text screening. After reconciliation, 77 studies were included in the review. The first author read the articles, extracted data, and assessed the quality of the studies using Mixed Methods Appraisal Tool Version 2018 User Guide (Hong et al., 2018). The third author independently did the same for 20% of the articles, which were selected randomly. We then compared and reconciled our independent data extraction. To address this presentation’s specific research questions, we excluded studies that did not use qualitative measures. “Qualitative” is defined as descriptions about an individual or individuals that are based entirely on observation or experience. In other words, descriptions with no associated quantitative data. Descriptions can be made by researchers, doctors, clinicians, or caregivers. The total number of studies included in this presentation is 42. We conducted descriptive analyses on the extracted data to answer our research questions.

**Results**: Forty-two of 77 (55%) of studies included qualitative measures of communication. Thirty-six of those 42 (86%) reported expressive language skills, six (14) reported receptive language, twenty-one (50%) reported observations of speech, and 27 (64%) reported cognitive features. Additionally, 37 (88%) included reports of behavior, and 13 (31%) included reports of social ability. Common themes in the expressive language domain were delayed language skills and the use of manual communication such as signs or gestures, especially in early childhood. Delayed abilities were also commonly reported for receptive language. In the speech domain, description of participants’ voices as hoarse and/or low were the most common theme; developmental differences in articulators and difficulties with articulation were also commonly reported. In the cognitive domain, common themes were the presence of intellectual disability, global developmental delay, and the presence of learning disabilities. Low attention spans were frequently reported, but long-term memory was reported as a relative strength. Deeper analyses of these themes, as well as analyses of the behavioral and social themes, will be completed in the upcoming months.

**Discussion:** These results confirm and reflect trends in quantitively measured SMS communication literature (Linert et al., 2023, 2024). That is, language and communication differences are incredibly common in people with SMS, but there is very little specific information about the nature of the communication differences. With a few notable exceptions (which will be explored further when analyses are complete), much of this data come from case reports which note that delays in communication and cognitive development are present, but do not give further detail. However, the qualitative nature of these data highlight the importance of investigating the communication profile to those in the SMS community. Effective communication is necessary for the quality of life for individuals with SMS and their caregivers. Detailed data on the communication profile of people with SMS will be an invaluable resource to Speech-language pathologists and other clinicians who provide services for clients with SMS.

**References:**

Dykens, E. M., Finucane, B. M., & Gayley, C. (1997). Brief report: cognitive and behavioral profiles in persons with Smith-Magenis syndrome. Journal of Autism & Developmental Disorders, 27(2), 203–211. <http://ovidsp.ovid.com/ovidweb.cgi?T=JS&PAGE=reference&D=med4&NEWS=N&AN=9105971>

Elsea, S. H., & Girirajan, S. (2008). Smith-Magenis syndrome. European Journal of Human Genetics : EJHG, 16(4), 412–421. https://doi.org/https://dx.doi.org/10.1038/sj.ejhg.5202009

Elsea, S. H., Purandare, S. M., Adell, R. A., Juyal, R. C., Davis, J. G., Finucane, B., Magenis, R. E., & Patel, P. I. (1997). Definition of the critical interval for Smith- Magenis syndrome. Cytogenetics and Cell Genetics, 79(3–4), 276–281. http://ovidsp.ovid.com/ovidweb.cgi?T=JS&PAGE=reference&D=med4&NEWS=N&AN=9605871

Greenberg, F., Lewis, R. A., 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. http://ovidsp.ovid.com/ovidweb.cgi?T=JS&PAGE=reference&D=med4&NEWS=N&AN=8882782

Hong, Q., Pluye, P., Fàbregues, S., Bartlett, G., Boardman, F., Cargo, M., Dagenais, P., Gagnon, M.-P., Griffiths, F., Nicolau, B., Rousseau, M.-C., & Vedel, I. (2018). MIXED METHODS APPRAISAL TOOL (MMAT) VERSION 2018 User guide. http://mixedmethodsappraisaltoolpublic.pbworks.com/

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 Developmental and Behavioral Pediatrics : JDBP, 27(3), 188–192. <http://ovidsp.ovid.com/ovidweb.cgi?T=JS&PAGE=reference&D=med6&NEWS=N&AN=16775514>

Linert, Jamie, Miriam Kornelis, and Lizbeth Finestack (2024). How Do We Measure Language Ability of People with Smith-Magenis Syndrome? Data from a Scoping Review. Poster. *56th Gatlinburg Convention.* Kansas City, Missouri.

Linert, Jamie, Miriam Kornelis, and Lizbeth Finestack (2023). Language and Smith-Magenis Syndrome: A Scoping Review. Poster. *55th Gatlinburg Convention.* Kansas City, Missouri.

Martin, S. C., Wolters, P. L., Smith, A. C. M., SC, M., PL, W., & ACM, S. (2006). Adaptive and maladaptive behavior in children with Smith-Magenis syndrome. Journal of Autism and Developmental Disorders, 36(4), 541–552. <http://login.ezproxy.lib.umn.edu/login?url=http://search.ebscohost.com/login.aspx?direct=true&AuthType=ip,uid&db=rzh&AN=106294404&site=eho> st-live

Ouzzani, M., Hammady,H., Fedorowicz, Z., and Elmagarmid, A. Rayyan — a web and mobile app for systematic reviews. Systematic Reviews (2016) 5:210, DOI: 10.1186/s13643-016-0384-4

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. http://ovidsp.ovid.com/ovidweb.cgi?T=JS&PAGE=reference&D=med5&NEWS=N&AN=12652298

Smith, A. C., Boyd, K. E., Brennan, C., Charles, J., Elsea, S. H., Finucane, B. M., Foster, R., Gropman, A., Girirajan, S., & Haas-Givler, B. (2019). Smith-Magenis Syndrome. In Gene Reviews. https://www.ncbi.nlm.nih.gov/books/

Smith, A. C. M., Magenis, R. E., & Elsea, S. H. (2005). Overview of Smith-Magenis syndrome. Journal of the Association of Genetic Technologists, 31(4), 163–167. <http://ovidsp.ovid.com/ovidweb.cgi?T=JS&PAGE=reference&D=prem1&NEWS=N&AN=16354942>

University of Minnesota