PRISMS Clinic
and Research
Consortium

STRATEGIES TO ADDRESS EMOTIONAL AND BEHAVIORAL CHALLENGES IN SMITH-MAGENIS SYNDROME (SMS)

Recognition and Recommendations for Treatment

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PARENTS AND RESEARCHERS INTERESTED IN SMITH-MAGENIS SYNDROME

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The treatment and intervention recommendations for Smith-Magenis syndrome (SMS) are written by professionals, with input from parents/caregivers, and with the understanding that they will be published online by Parents and Researchers Interested in Smith-Magenis syndrome (PRISMS), read by parents, caregivers, and other family members, and shared by parents and caregivers with healthcare professionals. Do not initiate any medications or dietary changes without first consulting a licensed healthcare provider.

The documents are recommendations only and are not formal medical guidelines or scientific papers. They are based on the consensus of clinicians with expertise working in the SMS field. The documents are not referenced in the way articles published in medical journals would be referenced. This was purposefully done to make them more understandable for non-medical individuals. The terminology used here is USA-focused and may be adapted, as appropriate, for use in other countries.

The PRISMS Clinic & Research Consortium was founded in 2020 and exists to improve the delivery of clinical services to families impacted by Smith-Magenis syndrome and to develop a research infrastructure for advancing the development and implementation of new and improved treatments. Please contact PRISMS for more information (www.prisms.org).

Barbara Haas-Givler, BCBA,[1] Kerry Boyd, MD, FRCPC,[2] Christine Brennan, PhD, CCC-SLP,[3] Mary Beall, MEd,[4] Hanna Hildebrand, MSOTR/L,[5] Cora M. Taylor, PhD,[1] and Rebecca H. Foster, PhD,[6 7]

[1] Geisinger-Bucknell Autism & Developmental Medicine Institute, Geisinger Health System, Lewisburg, PA, USA
[2] McMaster University, Department of Psychiatry and Behavioural Neurosciences, Hamilton, Ontario, Canada
[3] Department of Speech, Language, and Hearing Sciences, University of Colorado Boulder, Boulder, Colorado, USA
[4] PRISMS speaker and Regional Representative, Dallas, Texas, USA
[5] Department of Occupational Therapy, National Institute of Health (NIH) Clinical Center, Bethesda, MD, USA
[6] Department of Psychology, St. Louis Children's Hospital, St. Louis, Missouri, USA
[7] Department of Pediatrics, Washington University School of Medicine, St. Louis, Missouri, USA

Correspondence: Barbara Haas-Givler, BCBA, Autism & Developmental Medicine Institute, Geisinger Health System, 120 Hamm Drive, Suite 2A, Lewisburg, PA, USA 17837. Email: bahaasgivler@geisinger.edu

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[5] The section of this article on sensory processing in SMS was prepared by Hanna Hildenbrand MSOTR/L in her personal capacity. The opinions expressed in this article are the author's own and do not reflect the views of the National Institutes of Health (NIH), the Department of Health and Human Services, or the United States government.

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Parents and Researchers Interested in Smith-Magenis Syndrome

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Approaches to understanding emotional and behavioral challenges in Smith-Magenis syndrome (SMS) vary but should begin with an awareness of traits commonly observed among individuals with SMS. Educators, clinicians, and others who work with children and adults with SMS often find them to be energetic, enthusiastic, and endearing. Although strengths exist, individuals with SMS also commonly demonstrate significant challenges with emotional and behavioral dysregulation across the lifespan, which can often pose difficulties for individuals with SMS, their families, professionals, and community members who interact with them.

The following guidelines offer recommendations for parents and those caring for individuals with SMS to approach emotional and behavioral challenges by understanding the person, the syndrome, and effective practices.

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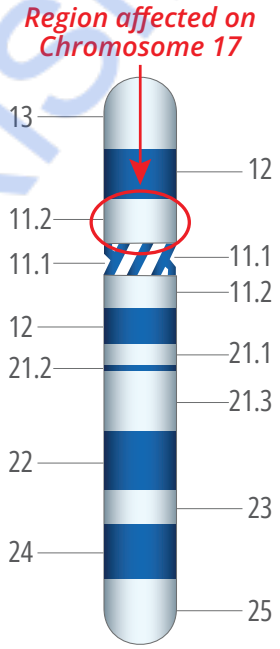
What is Smith-Magenis syndrome (SMS)?

SMS is caused by a missing piece or change in the genetic information within a specific region of chromosome number 17 (Smith et al., 1986; Finucane et al., 2009; Smith et al., 2022). The diagnosis is made through highly accurate DNA laboratory testing. Most cases of SMS (about 90%) are caused by a deletion (a missing piece) of genetic material on the 17th chromosome. The missing portion includes an important gene called RAI1. Some individuals with SMS (about 10%) do not have a deletion; instead, they have a spelling change in the letter code of their genetic material (DNA) called a sequence variant in the RAI1 gene (Slager et al., 2003).

While there may be differences in emotional and behavioral features based on having a deletion or sequence variant, both result in challenges that benefit from interventions. Individuals with SMS exhibit a wide range of medical complexities, developmental delays, sensory processing challenges, and intellectual disability as well as emotional and behavioral concerns. (See Figure 1, page 2.)



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Impact of Health on Behavior

Physical health problems associated with SMS (e.g., neurological, medical, sensory, motor) can have a direct impact on emotional and behavioral functioning (see SMS Medical Management Checklist by Smith et al., 2018; see SMS GeneReviews® by Smith et al., 2022). Sometimes, physical aspects of SMS contribute to behaviors, but some behaviors may continue to occur for non-medical reasons. For example, severe nail picking in SMS is related to abnormal function in nerves going to the hands and feet (also known as peripheral neuropathy); however, individuals with SMS quickly learn that nail picking can generate a lot of attention from adults, such as teachers and caregivers (Haas-Givler & Finucane, 2014).

The physical features of SMS may contribute to challenging behaviors but should not be considered in isolation. Instead, a comprehensive approach should be utilized that examines both contributing medical concerns and the environmental responses that may motivate or reinforce those behaviors. Moreover, addressing SMS-associated medical issues can have a dramatic positive effect on daily functioning (e.g., treating ear infections, managing constipation); (Haas-Givler & Finucane, 2014). Recognizing and treating physical health problems can lead to helpful accommodations or therapies and thereby reduce behavioral and emotional dysregulation.

Figure 1. Developmental and Behavioral Features

COGNITION AND DEVELOPMENT
• Mild to moderate intellectual disability • Speech and language delay and articulation problems and/or long term deficits, hearing loss • Fine and gross motor delay and/or long term deficits • Stereotypies, sensory concerns (may be similar to behaviors observed with autism spectrum disorder)[1] • Atypical social development, socioemotional deficits
SLEEP
• Disrupted sleep patterns including wakefulness at night and daytime sleepiness • Night eating • Elopement at night
BEHAVIOR
• Attention-seeking behaviors • Prolonged outbursts/tantrums • Self-injurious behaviors, skin picking • Aggression • Poor frustration tolerance • Attention problems, hyperactivity, impulsivity • Difficulties with changes in routine
MOOD
• Anxiety symptoms • Mood lability • Exaggerated emotional responses to stressors

Note. Some of the above require a formal diagnosis.

[1] Evidence-based strategies for autism spectrum disorder (ASD) may be appropriate regardless of whether there is a confirmed diagnosis of ASD.

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Cognitive Ability and Emotional Development

While there is a wide range of cognitive abilities among people with SMS, most individuals have mild to moderate intellectual disability (Foster et al., 2013). The emotional and behavioral development of individuals with SMS is usually much younger than their chronological age. Many aspects of the SMS behavioral profile could also describe behaviors that are commonly observed in two- or three-year-old children, including:

- a low tolerance for frustration
- negativity/irritability
- mood dysregulation
- a need to be independent to an extreme
- short attention span
- tantrums
- anxiety about separation from loved ones
- resistance to changes in routine
- repeated questioning.

Like very young children, individuals with SMS often become emotionally attached and possessive of caregivers, need frequent reassurance about upcoming events, and have a poorly developed sense of time. Behavioral outbursts in adults with SMS can look very much like the temper tantrums seen in young children, with the person throwing oneself to the floor, kicking, and crying (Haas-Givler & Finucane, 2014).

Individuals with SMS of all ages may exhibit behaviors that are often seen in earlier development in typically developing children (Finucane et al., 1994; Haas-Givler & Finucane, 2014). For example, many typically developing young children go through a period where they insert objects into their noses or ears; this is a relatively common behavior among older children and adults with SMS. Likewise, there exists an endearing, tic-like behavior in SMS that involves arm or hand-squeezing during periods of happiness or excitement, often referred to as "self-hugging" (Finucane et al., 1994). While this SMS "self-hug" may seem unusual in an older child or an adult, a similar behavior can be observed among typically developing young children when they are excited.

As individuals with SMS age, the gap between intellectual ability that is more advanced than emotional development increases, and this difference contributes to significant behavioral challenges (Haas-Givler & Finucane, 2014). **Developmental asynchrony is at the core of what makes SMS so challenging** (Haas-Giver & Finucane, 2014; Simon et al., 2014).

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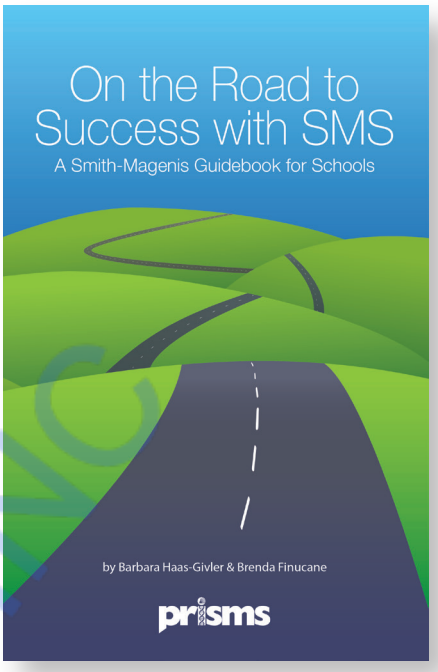
Strategies to Address Common Challenging Behaviors



Please note that much of this section was excerpted from "On the Road to Success with SMS: A Smith-Magenis Guidebook for Schools" (Haas-Givler & Finucane, 2014).

Behaviors need to be approached in the context of the person's developmental level, cognitive abilities, and communication skills, as there are often multiple contributing factors (Haas-Givler & Finucane, 2014; Simon et al., 2014). It can be helpful to break down and systematically consider biopsychosocial factors including frustration triggers, significant life events or traumas, and medication side effects.

In SMS, aggression, attention-seeking behaviors, prolonged outbursts, and self-injury begin in early childhood and can pose significant challenges into adulthood for many individuals with SMS and their caregivers and educators (Haas-Givler & Finucane, 2014; Simon et al., 2014). It is likely that the parent or familiar caregiver will need to provide resources to staff who are working with their child or adult with SMS. Syndrome specific information may assist the practitioner in prioritizing and targeting challenging behaviors and examine these behaviors in the context of the syndrome rather than solely in relation to antecedents (i.e., causes of behaviors) and consequences (i.e., outcomes of behaviors).

Strategies for addressing behavioral issues in children with intellectual and neurodevelopmental disorders include the use of positive reinforcement for appropriate behavior, shaping, reward systems, time-outs, loss of privileges, and ignoring maladaptive behavior (Haas-Givler & Finucane, 2014; Simon et al., 2014). Such strategies are widely used in home, educational, and residential settings. These approaches are "consequence-based" in that they are implemented after a behavior has occurred. **While useful in shaping, reinforcing, increasing, or maintaining positive behaviors, these approaches are less effective for decreasing inappropriate behaviors among individuals with SMS**. Preventative or antecedent strategies facilitate learning new skills, rather than addressing inappropriate behaviors that have already occurred. **A behavior plan that is "front-loaded" with antecedent strategies is recommended so that the person with SMS is set up to engage in appropriate behaviors, which will then be reinforced by positive consequences**.

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A functional behavioral assessment (FBA), conducted by a board-certified behavior analyst (BCBA) or psychologist, should be utilized to develop an individualized behavior support plan (BSP) and/or behavior intervention plan (BIP). Most school districts and agencies have guidelines and documents to utilize to conduct the assessments. Outcomes of the FBA include:

- a clear definition of the challenging behavior(s),
- a hypothesis about the function (or reason) for the behavior,
- identification of setting events/antecedents/triggers, and
- current consequences for the behavior.

It is extremely important to identify and implement changes in the environment that will reduce challenging behaviors (Haas-Givler & Finucane, 2014). Of utmost importance is to select and utilize proactive, strength-based strategies to teach alternative, appropriate behaviors, and consequences that will reinforce these appropriate behaviors.

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Attention Seeking Behaviors, Tantrums and Aggression

Most people with SMS are highly adult-oriented, with an often insatiable need for individual attention (Haas-Givler & Finucane, 2014). As a result, they often compete with peers and siblings for adult attention and may react with aggression and/or self-injury when attention is withdrawn (Taylor & Oliver, 2008; Wilde et al., 2013). Prolonged tantrums lasting hours, with self-injury, property destruction, and physical aggression, are common through adolescence and into adulthood. Behavioral outbursts are generally precipitated by a need for attention, an unexpected change in routine, denials (e.g., food or activity), fine motor activities (which can be especially challenging for individuals with SMS), or a lack of clear expectations or structure, so these potential triggers need to be considered when determining the best course of intervention in any given situation.

Strategies across the Home, School, and Work Settings:

Environment and structure

- Implement structure, routine, and well-defined clear limits
- Provide a variety of durable materials, toys, manipulatives; monitor safety as many items may be destroyed and objects could become dangerous and need to be replaced
- Set realistic expectations for the length of time on tasks; modify task length to align with the attention span for the individual and give short breaks as needed
- Provide multi-sensory, high interest, hands-on activities that are appropriate for age/setting (e.g., writing in shaving cream, music, stampers, stickers, white boards with markers)
- Identify and minimize known triggers and arrange the environment to set individuals up for success; for some individuals, seating next to a preferred parent/caregiver at dinner table or staff in school or at work is most calming
- Class periods may need to be abbreviated for school-aged children (e.g., the student with SMS is scheduled for a regular education classroom for 25 minutes and then returns to special education class)
- Provide photo or written menu of food/snacks/beverages that are scheduled for meals

Staff or caregiver approach

- Provide consistency among caregivers /staff as to expectations, limits, and responses to behaviors; follow positive behavior plan and/or treatment plan
- Offer choices that all result in task completion, but limit to 2 or 3 choices
- Provide frequent reinforcement with a variety of reinforcers (e.g., specific positive praise, thumbs up, sticker charts)
- In response to inappropriate behavior, when possible, remove the audience to avoid inadvertent positive reinforcement through attention

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Supports and strategies

- Use a visual schedule with photos, pictures, and words to help individuals see the order of the day
- Use First/Then language and charts (e.g., "First you get dressed, and then you can pick a game to play.")
- Use timers to signal a preferred activity is available in addition to using timers to communicate transitions to the next activity
- Identify powerful, immediate reinforcers with knowledge that attention from a preferred adult may be the most desired reinforcer
- Develop and implement an individual behavior system across settings (e.g., a home-based system that mirrors a successful school -based system)
- Pair a non-preferred activity with a preferred activity (e.g., allowing music when brushing teeth, putting away toys in the home, or engaging in challenging task at school/work)
- Identify pre-cursor behaviors and interrupt behavioral chains before escalating to aggression or property destruction
  - Distract, engage in appropriate behavior, and reinforce appropriate activity
- Ensure appropriate staff to student/ client ratio in schools, day programs, or living arrangements
  - **There is often a need for a 1:1 staff person for individuals with SMS**
- Behavior plans and supports need to recognize the delayed emotional development in SMS but also match the interest and skills of the older child or adult
- Implement numerous visual supports in the home, classroom, or work area that are beyond what would usually be provided in a high school or adult day program or residence
- If necessary, use locks on the refrigerator and/or pantry to address food seeking behaviors
- Physically or chemically restrain only when necessary for safety and only when utilized under the direction of trained staff and/or medical professionals; attention is attention, and restraint provides attention so these strategies may be necessary from a safety perspective but should also be used only if absolutely needed. When required, safety protocols of schools, work environments, and other institutions must be followed precisely

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Sleep

Disrupted 24-hour sleep patterns are a hallmark of SMS. Most children and some adults with SMS exhibit significant sleep disturbance (Gropman et al., 2006; Smith et al., 2019). Frequent waking at night and early wake-up times are typical. Night wandering is common and can lead to potentially dangerous situations (e.g., eloping, food foraging, unsupervised cooking).

Excessive daytime sleepiness, irritability, and "sleep attacks" have all been described in SMS. During the day, individuals with SMS sometimes fall deeply asleep in the middle of an activity due to a chronic sleep debt. Not surprisingly, sleep difficulties can also exacerbate emotional and behavior problems, and these problems can negatively affect the entire family as well as staff and teachers at school, day programs, or in vocational activities. Families and staff report a correlation between insufficient sleep at night and behavior problems the next day. Parents, caregivers, and staff frequently note that after a nap the individual with SMS is refreshed and often more amenable to completing tasks that were previously refused.

Some research has shown abnormal profiles of the hormone melatonin in people with SMS (Smith et al., 2001). Specifically, in SMS melatonin levels are unusually elevated during the day and significantly low at night; this equates to an inverted circadian melatonin pattern that is consistent with observed sleep disruptions in this disorder (De Leersnyder et al., 2001; Potocki et al., 2000). Melatonin supplements have normalized sleep patterns in some children with SMS, although the effect is often transient. Parents/caregivers should make sure to inform their prescribing physician and seek guidance as to dosage and formula when considering use of melatonin or any medication used to aid in sleep (Smith et al., 2019).

Strategies for School and Work

- Parents/caregivers are encouraged to keep a sleep log and share it with school or day program staff
- Staff should consider providing a schedule with additional preferred activities and planned breaks when the person with SMS is noted to be more tired
- Children/adults with SMS will often need to rest or sleep sometime during the day, often in the early afternoon
  - An area in the classroom or day program should be designated for the person to take a nap when needed
  - A cot, bean bag, or a recliner chair is sufficient for some individuals

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Strategies for Home

- Set and maintain a consistent bedtime routine
- Provide safe, quiet activities that can be played within the bedroom to encourage remaining in the bedroom until morning
- The use of an enclosed or tented bed for younger children has been helpful for some families. Be sure to follow weight and other guidelines provided by the manufacturer
- In-home security cameras can allow parents to monitor behavior without going in the room and inadvertently reinforcing disrupted sleep
- Bed alarms, door/window alarms, refrigerator and pantry locks, locks for microwaves/stoves, and/or other alarms/locks may be warranted
- Provide a positive reinforcement system with a visual support that encourages the person to remain in their room, engaging in safe activities until it is morning
- Visual "clocks" with a red light/green light system can be used to signify when it is okay to get out of bed or leave the bedroom
- A sleep study may be recommended to assess sleep quality/quantity as well as evaluate for sleep apnea (e.g., periods of time when breathing stops during sleep)

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Self-Injury

Self-injurious behaviors (SIB) are nearly universal in SMS and typically include head-banging, face-slapping, and skin picking (Dykens & Smith, 1998). While these behaviors are distressing, parents and caregivers rarely report self-injury as the most difficult aspect of raising a child with SMS. Significant self-injury does not usually take place daily, and these behaviors tend to occur in cycles with varying severity.

Polyembolokoilamania is a term used to describe insertion of foreign objects into bodily orifices, such as the nose and ears (Dykens & Smith, 1998; Finucane et al., 2001). This behavior sometimes requires surgical removal of beads, food, and other items from the ears or nose. Rectal or vaginal object insertion usually occurs in older individuals with SMS (Finucane et al., 2001). Self-insertion of objects in the vagina or anus can be mistaken for sexual abuse.

Strategies to address self-injury

- Do not give more attention to self-injury than is necessary to ensure safety
- If necessary, remove other students or siblings from environments in which self-injury is occurring to limit attention and prevent secondary trauma
- Remove and/or lock up items that can be obvious sources of self-injury (e.g., sharp objects, medications, cleaning products, items that can be tied around the neck, firearms)
- Pad objects often used for headbanging such as bedrails
- Distract, prevent, engage, and reinforce alternative appropriate activities
  - Provide options for counter stimulation (e.g., fidgets, items with preferred textures or temperatures)
- Do not provide screens as a form of distraction when self-injury is occurring, as they can be broken and used to self-injure
- Alert staff in school, work, and community programs to monitor small objects (e.g., beads for arts/crafts or work jobs) or other objects that could be used for insertion if the person has exhibited this behavior
- Seek professionals (e.g., psychologist, psychiatrist, behavior analyst) to advise specific interventions and strategies

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Skin Picking

Picking at finger- and toenails is a common behavioral feature of SMS and may be related to atypical or decreased sensation in the extremities due to peripheral neuropathy (Finucane et al., 2001; Gropman et al., 2016; Haas-Givler & Finucane, 2014). Over half of children and adults with SMS engage in *onychotillomania* (i.e., picking at finger- and/or toenails to the point of bleeding and often with complete removal of the nail). Dry skin can lead to or exacerbate skin picking behaviors.

Strategies to address skin picking

- Redirection to using a manipulative or fidget object works for some individuals who engage in skin picking or nail pulling
  - Use of non-toxic putty can be helpful because this can be used to mimic picking behaviors without making a substantial mess
- Distraction and engagement in an alternate fine motor activity when the individual starts to pick at skin with minimal attention placed picking behaviors
- Keep nails short to reduce temptation to pick at loose cuticles or hangnails
- Gel nail polish manicures may be helpful in reducing the ability to pick; this should not be utilized if the person will attempt to pick off the nail polish and consume it
- Some adults opt to wear gloves to prevent themselves picking skin on their hands
- Moisturizing products should be applied to hands and feet daily to reduce dry, cracked skin, which exacerbates the tendency to pick
  - If tolerated, use of lotion with gloves when sleeping can be helpful to moisturize skin
- In the event the individual pulls off a finger- or toenail, staff and parents should react as neutrally as possible and use plain bandages to avoid inadvertently reinforcing the behavior through an emotional response or use of "character" bandages (e.g., cartoon characters)
- Occupational and physical therapy on a direct or consultative basis is recommended for children and adults with SMS

Due to the complexity of their behavioral profile, it is unrealistic to expect that all inappropriate behaviors can be eliminated (Finucane et al., 1994; Haas-Givler & Finucane, 2014; Simon et al., 2014; Taylor & Oliver, 2008). Minor behavioral issues such as lack of cooperation with adult directives, low frustration tolerance, and negative reactions to change, are daily occurrences for many children and adults with SMS. However, under optimal circumstances, significant aggression, self-injurious and disruptive behaviors can be reduced to a manageable number of episodes a year. For caregivers, it may become important to first address behaviors that pose a safety concern, while other less serious behaviors are given minimal attention to reduce risk of inadvertently providing attention that reinforces behaviors (Haas-Givler & Finucane, 2014).

Please refer to Table 1 (page 30) for additional behavior resources.

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Impact of Speech and Language and Auditory Concerns on Emotional and Behavioral Regulation

Speech and language delays are noted in most, if not all, children with SMS (Smith et al., 1986). Toddlers with SMS were found to have moderate to severe delays in expressive language (i.e., using speech to express and share information/ideas, ask questions, and respond to questions asked by others) with average social skills (Wolters et al., 2009). Compared to individuals without SMS, individuals with SMS demonstrate deficits in verbal working memory, verbal comprehension, and vocabulary (Elsea & Girirajan, 2008; Greenberg et al., 1996; Osório et al., 2012; Udwin et al., 2001).

In contrast to these noted deficits, individuals with SMS were found to have relatively unimpaired long-term memory such that vocabulary concept learning is a relative strength (Osório et al., 2012; Udwin et al., 2001). Individuals with SMS often have difficulties with expressive language and receptive language (i.e., comprehension of language including understanding directions, explanations, stories, and information spoken by others; Brennan, 2021; Brennan et al., 2022; Brennan et al., 2024).

Patient data related to speech, language, and hearing have been collected through the SMS Patient Registry, an initiative of PRISMS. A current study analyzing data from the SMS Patient Registry found that many children with SMS have delayed speech and language development, with first words being spoken around 2-2.5 years of age and ranging from 1-6 years (note: neurotypical children begin speaking their first words at about 12 months of age; Brennan, 2021; Brennan et al., 2022; Brennan et al., 2024).

Preliminary analyses from the SMS Patient Registry have revealed some subtle differences between individuals who have the RAI1 variant version of SMS versus the more common SMS chromosome 17 deletion (Brennan & Baiduc, under review). For example, more individuals with the RAI1 variant used speech to communicate, while individuals with the more common deletion used alternative communication methods such as sign language, picture communication, and/or a speech generating device. Additionally, individuals with the RAI1 variant began speaking earlier and a greater percentage have established or emerging literacy skills (reading and writing) (Brennan & Baiduc, under review).

It is important to be aware that difficulty communicating can result in frustration and lead to challenging behaviors, but even individuals with SMS who have strong communication skills may still struggle with challenging behaviors. When communication needs lead to behavioral dysregulation, speech and language intervention should aim to improve communication. For those with stronger speech, language, and communication abilities, intervention should target ways they can more effectively use their communication to resolve issues and decrease challenging behaviors.

Individuals with SMS also tend to have a voice with a characteristic hoarse quality (Greenberg et al., 1991; Hidalgo-

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De la Guía et al., 2020). A recent study focused on vocal quality in those with SMS suggested that the hoarseness may result from a high level of tension in the larynx (Hidalgo-De la Guía et al., 2020), although earlier research found vocal polyps and nodules in those with SMS (Greenberg et al., 1996).

In many cases, vocal quality may not impact behavior; however, if vocal quality of individuals with SMS interferes with their ability to be understood by others, frustration may lead to challenging behaviors. Unfortunately, there is no research indicating what intervention approaches for individuals with SMS can be used to improve vocal quality, although based on previous findings, strategies that decrease tension and promote relaxation may be worth trying if vocal quality creates a barrier to effective communication.

Individuals with SMS are described as having a high interest in communicating and will attempt to communicate their wants/desires (Martin et al., 2006), but difficulties with expressive language, social skills, and speech intelligibility (i.e., how easy it is for others to understand what is being said) can lead to emotional and behavioral dysregulation. These deficits may create barriers to effective social engagement. Many children start communicating using natural gestures (such as pointing or waving), and speech-language pathologists may recommend teaching children with SMS to use sign language or a picture exchange system to augment spoken words.

It is important for parents and caregivers to know that using sign, pictures, or a speech generating device to communicate will not create further delays in the development of natural speech. Not only do these types of Augmentative and Alternative Communication (AAC) provide a child a means to communicate, but they also promote the development of natural speech (Smith et al., 2016). In fact, teaching a child to communicate using signs, picture exchange, or a speech generating device (any AAC system) will help develop vocabulary, language, and communication skills that would otherwise be at risk of further delay if no communication system was established (Beukelman & Mirenda, 2005).

Behavior is a form of communication. It is important to recognize what the individual with SMS is trying to communicate through behavior. Strategies should aim to replace undesired forms of communication (tantrums, self-injury, harming others) with more desirable forms of communication such as using natural speech, signs, gestures, pictures, or an AAC device.

Hearing loss has been found in 80% of individuals with SMS (Brendal et al., 2017). Parents and other caregivers should be aware that ear infections can cause fluctuating and/or even minimal hearing loss. Fluctuating hearing loss can have a harmful impact on behavior, language development, auditory processing, academic performance, and social–emotional development (Yoshinaga-Itano et al., 2008); therefore, routine audiological evaluations should be a standard care for those with SMS. Additionally, hyperacusis (i.e., an inappropriate or exaggerated intolerance to sound that is not typically uncomfortable or threatening) has been noted in 73.5%

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of individuals with SMS in one study evaluating the auditory phenotype (Brendal et al., 2017). Individuals with SMS who have hyperacusis may exhibit challenging behaviors when sounds are uncomfortable.

Hearing Strategies

- Early evaluation of hearing (newborn infant hearing screening and testing again before 12 months of age) should be conducted by an audiologist
  - This testing should be followed by routine audiological evaluations, which is recommended as part of standard clinical care
  - The frequency of routine audiological evaluation may be yearly or biannually based on the results of initial testing, the frequency of middle ear infections, and any other concerns or issues related to hearing and heating health
- Caregivers and professionals often need to work closely with children/adults with SMS to wear hearing aids
  - Your audiologist, occupational therapist, and behavior therapist are resources to work on increasing tolerance and gradually increasing length of time wearing hearing aids
  - Use of hearing aids reduces the risk of progressive dementia in adults with cognitive impairment (Bucholc et al., 2021)
- Chronic ear infections and hearing loss are more common in those with SMS and parents/caregivers should consult with an otolaryngologist or ENT (ear, nose, and throat physician) to address chronic ear infections regardless of whether intermittent hearing loss is also occurring
  - Intermittent hearing loss can have a negative impact on speech and language development and speech intelligibility
  - Consult the SMS Medical Management Guidelines regarding medical issues addressed by an ENT
- Individuals who have hyperacusis may benefit from changes in the environment or access to more quiet spaces
  - When background noises cannot be avoided, the use of noise-cancelling headphones may make participation easier and may minimize challenging behaviors that occur due to hyperacusis

Speech-Language Strategies

- As early as possible, parents/caregivers should have an evaluation of speech, language, and communication completed. This evaluation should be done by a speech-language pathologist (as young as possible or as soon as possible following the SMS diagnosis) followed by speech-language intervention (again, as early as possible). Intervention should target development of functional communication, improve speech production and intelligibility, improve language formulation and comprehension, and improve social conversational and pragmatic skills.

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- For children/adults who cannot communicate effectively using natural speech (i.e. speaking), an evaluation for Augmentative and Alternative Communication (AAC) is recommended. This evaluation is usually completed by a specialized speech-language pathologist. An AAC evaluation can be conducted by a school-based clinician or one who is a community-based clinic/ hospital. Some schools may have AAC specialists who are special education teachers specializing in assistive technology. It is important that if an evaluation for AAC or assistive technology is not completed by a speech-language pathologist, then it should be accompanied by a speech-language evaluation conducted by a speech-language pathologist.

- AAC systems can include high tech speech-generating devices (iPads or dedicated speech generating devices) and/or lower tech systems, such as picture exchange, picture choice boards, and use of gestures and/ or signing. For individuals with SMS who are not using natural speech to communicate by age 3 years, a dedicated speech-generating device is recommended. A dedicated device is tailored for use by one individual (and only that individual) across all settings (e.g., home, school, therapy). Signing can also be used, but this will limit who the individual with SMS can communicate with as this form of communication will require others to know sign to understand and interact with the individual.

- Some individuals with SMS may require school-based accommodations that allow the child to use AAC to communicate. School-based interventions should also capitalize on the high social interest of those with SMS, both allowing and encouraging social interaction with others while modeling and reinforcing appropriate conversational and social interaction skills.

- Allow choices for what form of communication the individual with SMS wants to use.

- Incorporating such technology in a creative way may also motivate those with SMS to have better levels of cooperation and engagement when communication is needed.

- Consult with an otolaryngologist or ENT to address velo-pharyngeal insufficiency (i.e., a problem with the soft palate) and polyps (i.e. benign growths) or nodules on the vocal folds. It is unclear how prevalent velopharyngeal insufficiency and vocal fold polyps or nodules are, but the presence of these problems could potentially have a negative impact on speech intelligibility. Consult the SMS Medical Management Guidelines regarding medical issues addressed by an ENT.

Please see Table 1 (page 30) for additional speech, language, and hearing resources.

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Sensory Processing in SMS

Sensory processing differences are present and persist at least through the early teens in children with SMS, and some of these differences may become more prominent with increased age (Hildenbrand & Smith, 2012). In the study cited, most children with SMS presented with auditory, vestibular, touch, and multisensory processing as more/much more different than others (compared to national sample for the Sensory Profile (SP; Dunn, 1999, 2006). Based on the SP, these differences reflect parent-reported behaviors that may impact typical daily activities such as:

- being distracted when a lot of noise is around,
- seeking movement,
- expressing distress and discomfort during activities that involve grooming and oral care/hygiene, respectively,
- touching people and things, and
- noticing all actions around them

Some other differences that emerged in most children with SMS reflected challenges meeting performance demands, sustaining performance, and regulating emotional responses (Hildenbrand & Smith, 2012). Specific examples of associated behaviors reported by parents on the SP (Dunn, 1999) include:

- tiring easily and having poor endurance,
- needing more protection (e.g., emotional or physical safeguarding),
- being overly affectionate,
- having emotional outbursts with task failures,
- doing things inefficiently, and
- having difficulty with changes (in plans, expectations, and routines)

Children with SMS had differences across all SP categories compared to the national sample. In addition, they presented with a mixed pattern of thresholds for response to stimuli and a fluctuation in how they managed their needs (Hildenbrand & Smith, 2012). According to Dunn's Model of Sensory Processing (Dunn, 1997, 1999), children with SMS experienced heightened sensitivity sometimes and missed sensory input at other times, fluctuating between active and passive self-regulation. They actively added or reduced sensory input, passively did not notice aspects of their surroundings, or responded only after things happened around them.

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Using a sensory processing perspective to understand behaviors in SMS, it is not surprising that unexpected or extreme behaviors surface during daily activities and those that take an extended period of time (Dunn 1997, 1999, 2006, 2014. Missing sensory cues or poorly modulating them provides context to giving a pet an unintentionally hard squeeze or shampoo unexpectedly gushing out of the bottle. The fluctuating use of active or passive self-regulation also adds insight as to why tantrums or over-reactions can be unpredictable, surprising even the most skilled service provider or beloved caregiver.

As individuals with SMS interact with others in the home, school, and community and engage in a variety of daily activities, they are sure to encounter situations that cannot be predicted or controlled. Assessment of sensory processing difficulties paired with information about participation in daily activities and environments can assist with identifying strengths, needs, and possible barriers. For example, heightened sensitivity or noticing the smallest details that many others miss and responsiveness to routine tasks and established schedules, can serve as strengths in well-matched activities or environments. By understanding sensory processing attributes or differences, environments can be structured or adapted to enrich positive behavior and participation in the daily lives of individuals with SMS.

While sensory processing difficulties or differences are seen in SMS, it is not expected that every child with SMS will have the exact same presentation (Gropman et al., 2006, 2007; Hicks et al., 2008; Hildenbrand & Smith, 2012; Smith & Duncan, 2005; Smith, Hildenbrand, & Smith, 2009; Smith et al., 2010). Assessing the individual's sensory processing as part of a comprehensive OT assessment is essential to identifying their strengths and needs aimed at providing individualized sensory-informed care to each person with SMS.

Occupational Therapy (OT) Strategies

- Early evaluation of sensory processing (infant assessment of sensory processing between 0 and 6 months of age) by an OT
- Clinical consultation to address unexpected sensory processing and concerns related to daily routines for children 0 to 6 months as well as a follow up assessment of sensory processing between age 7 to 12 months (Dunn, 2014) and subsequently as needed
- School assessment of sensory processing paired with information about participation in daily activities and environments is recommended for preschool and school age children
- Developmental and functional assessment factored into individualized care and educational plan

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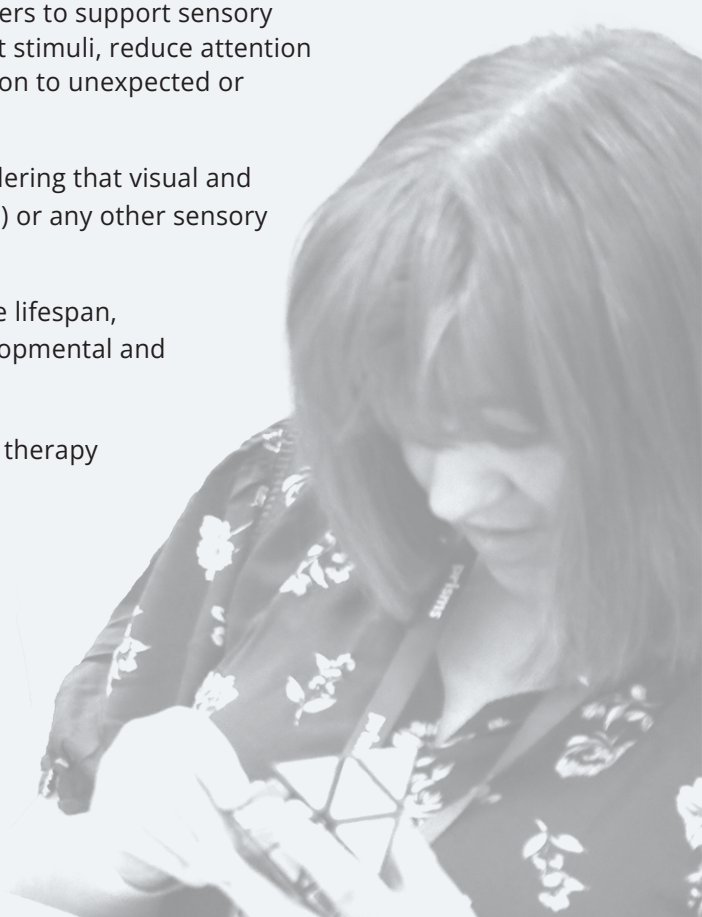
- OT intervention focused on self-regulation, self-advocacy, and participation in daily activities
- Working closely with the speech language pathologist and/or AAC specialist to integrate communication systems/strategies necessary to foster self-regulation skills and autonomy as children age as well as strengthen carry over as activities and environments change
- On an ongoing basis, parents/caregivers and educational/health professionals communicating observations of challenges and successes to work together on expanding effective supports/strategies across activities and environments, with emphasis on being flexible to meet the individual's needs
- School-based and work-related accommodations that support participation in the least restrictive environment and reflect individualized adaptation of activities, modification of the environment, and facilitated engagement, for example:
  - Reduced duration of activities or activity breaks
  - Reduced auditory and visual distractions
  - Movement opportunities and/or postural supports
  - Use of electronic technology (to support academic work and job duties)
  - Visual cues and support from informed bystanders to support sensory modulation (i.e., enhance attention to important stimuli, reduce attention to stimuli unrelated to task, or support adaptation to unexpected or irritating stimuli)
- Capitalizing on sensory processing strengths, considering that visual and oral sensory processing (Hildenbrand & Smith, 2012) or any other sensory modality may be revealed as an individual strength
- Reassessment of sensory processing throughout the lifespan, particularly leading up to or during periods of developmental and role transitions

Please see Table 1 (page 30) for additional occupational therapy and sensory processing resources.



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Emotional and Behavior Concerns: A Comprehensive Approach

Please note that much of this section was excerpted from "HELP with Emotional and Behavioural Concerns in Adults with Intellectual and Developmental Disabilities" presented by the Developmental Disabilities Primary Care Program of Surrey Place (Bradley et al., 2020).

People living with SMS and those who care for them may seek mental health and psychiatric services for various reasons. As discussed throughout this document, often help is sought for emotional and behavioral concerns. While there are no quick fixes, there are some helpful guidelines and models of care. HELP for emotional and behavior problems is one such model that identifies potential influences of Health, Environment, Life Stresses and Psychiatric symptoms or conditions (Bradley et al., 2020).

A systematic assessment of emotional and behavioral concerns is recommended to help understand the multiple contributing factors (Bradley et al., 2020). Clinicians and caregivers should assess for potential health issues, paying particular attention to what is known about the SMS medical vulnerabilities. Consider potential contribution of environmental triggers (e.g., sensory concerns). Evaluation should also ensure that support needs are met, and expectations are well-aligned with abilities (e.g., cognitive abilities, developmental levels) and inclinations. Individuals with SMS can present with psychiatric signs and symptoms that could point to co-occurring conditions.

While inattention, distractibility, impulsivity, anxiety, and mood features are often reported in SMS, caregivers and professionals (e.g., educators, allied health, other specialists) work together, typically with psychologists and/or physicians (e.g., primary care physician, pediatrician, neurologist, psychiatrist) to clarify formal diagnoses and individualized management plans (e.g., attention deficit hyperactivity disorder (ADHD), anxiety and mood disorders).

While there are no psychiatric medications specifically for SMS, pharmacological treatment for these conditions can improve emotional well-being and reduce behaviors of concern. There are cautions about use of psychiatric medications in an individual presenting with a significant change in behavior without a thorough assessment. Medication may be inappropriate for the underlying problems, mask symptoms, quell behaviors that signal distress, or have side effects (especially when more than one psychiatric medication is used). It is recommended that families engage in a multi/interdisciplinary assessment with specialized teams, where available, with a goal of then determining appropriate evidence-based interventions, which may or may not include psychoactive medications.

Help for Assessing and addressing behavioral challenges in the context of SMS

Challenging behaviors often communicate distress (Bradley et al., 2020). The experience of sorting out what is underlying distressed and distressing behaviors can be overwhelming.

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There is helpful guidance from the HELP framework found within the Developmental Disability Primary Care Program (DDPCP; Bradley et al., 2020). Thinking through the "HELP" model provides systematic consideration of important, and sometimes multiple, factors influencing behavior. When there is a change or increase in behaviors it becomes necessary to review timelines, presentation across people/settings, and factors which may be contributing. This may involve multidisciplinary team detective work including observing and listening to the person, thinking together as a circle of care, and embarking on further investigation and involvement of professionals as indicated. The "HELP" framework prompts further assessment of potential Health, Environmental, Life stressors and Psychiatric concerns to aid in behavioral regulation. It is a helpful way of approaching behaviors that challenge our ability to understand and respond.

Health: Consider SMS-related medical or physical concerns (Smith et al., 2018; see SMS Medical Management Checklist). Look for common health problems like infections or pain. Gastrointestinal (e.g., gastro-esophageal reflux/heartburn, constipation) discomfort is often worth considering in SMS. Hormonal/endocrine or pubertal changes may be adding to or exacerbating problems as well. Sleep problems require perpetual assessment and consideration (e.g., disrupted and often disruptive; circadian rhythm disturbance, obstructive sleep apnea). There may be missed or undertreated conditions as well as side effects from medications that require review.

Environment and expectations: Sensory processing issues are common, and there may be distressing environmental triggers for a person with SMS (Hildenbrand & Smith, 2012). The charm of someone with SMS may serve to mask intellectual

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disability, with resultant frustration or harm to self and others when expectations are too high. Similarly, expectations that are too low and result in boredom can be reasons for unanticipated disruptive behaviors as well.

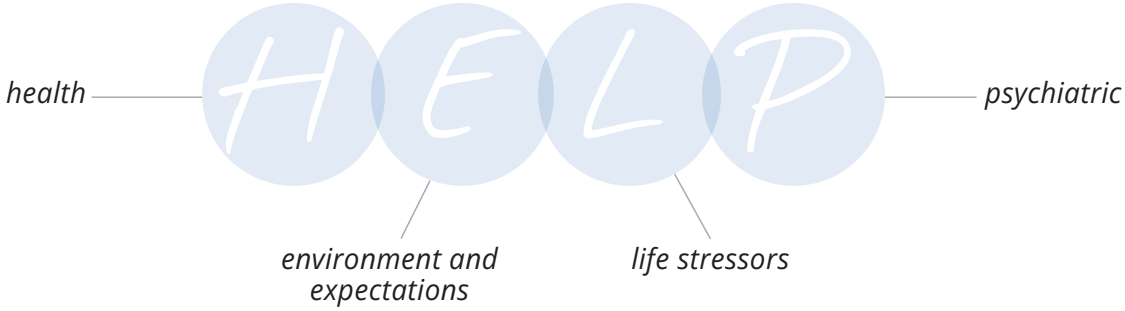
Life Stressors: Individuals with SMS may not be able to communicate when they are in uncomfortable or distressing situations (Finucane et al., 1994; Foster et al., 2013; Haas-Givler & Finucane, 2014; Simon et al., 2014; Taylor & Oliver, 2008). Behaviors can be a signal of maltreatment, bullying, or exclusion. Trauma can also result from losses or major life transitions (e.g., loss of the structure and connections of school transitioning to "the adult world"). Family conflict, strain, and losses are very important to understand and garner necessary support from parents, siblings, extended family, teachers, and one's entire care team.

Psychiatric: Consider mental health holistically (i.e., cognitive, emotional, spiritual, behavioral) and contributors to mental illness (Bradley et al., 2020). People with SMS are more likely to experience adjustment disorders (i.e., situations that cause emotional distress in various forms), anxiety, or mood symptoms. They may also be experiencing symptoms of a psychiatric disorder like major depressive disorder. People with developmental delays and intellectual disability in general tend to be treated with more psychoactive medications, sometimes without a comprehensive evaluation and understanding of the SMS phenotype and other contributing factors.

Mental Health Assessment and Intervention Recommendations

- Think of mental health holistically form physical, psychological, social, and spiritual perspectives
- Seek medical assessment for potential health contributors including primary care provider and specialists as indicated
- Engage in a multi/interdisciplinary assessment with specialized teams where available
- Treat specific problems specifically (e.g., sleep, ADHD, anxiety, depression) with evidence-based strategies, supports, and targeted management including possible psychoactive medications while regularly monitoring for effectiveness and side effects

Please see Table 1 (page 30) for additional information on the HELP model and addressing mental health needs (Bradley et al., 2020).



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Navigating Behavior and Emotional Concerns During the Transition From School Age to Adult Services

Adults with SMS continue to develop and progress as they age, but emotional and behavioral issues usually remain (Finucane & Haas-Givler, 2009; Foster et al., 2013). Impulsive actions and reactions are among the biggest challenges to successful adult living. With evidence-based interventions, emotional and behavioral dysregulation can usually be managed.

The transition to adulthood is complex and multifaceted for individuals with SMS, making it important to consider practical and emotional/behavioral support needs and to get on waiting lists for adult services as early as possible (Ally et al., 2018; Wehrlen et al., 2015). In the U.S., every state has programs to assist adults with an intellectual disability (Berens et al., 2020). There are programs to help with job placement, supported living, and rehabilitative and psychological therapies but these services are not automatically offered (e.g., Roos & Søndenaa, 2020). Families must apply for such resources through the local Department of Developmental Disabilities or other local and national organizations. There are usually long wait lists, and it can take years to obtain resources (Franklin et al., 2019); this can be distressing and overwhelming for individuals with SMS and their caregivers. Overall, transition planning to ensure appropriate, comprehensive adult care, including meeting behavioral support needs, must be completed in a thoughtful, individualized manner that ensures ongoing care needs throughout the lifespan (Ally et al., 2018; Berens et al., 2020, Goslink et al., 2022; Nathenson & Zablostsky, 2017; Varshney et al., 2022).

While some milder behaviors may be evident daily (e.g., being too loud, frequent questioning, perseveration, attention-seeking), it is important to note that the most difficult outbursts and prolonged tantrums for many adults do not happen frequently (Finucane & Haas-Givler, 2009; Foster et al., 2013). Particularly in adulthood, there can be long periods of relative calm, when large crises become rarer. This may be because of maturation but also because of consistent interventions and support. Some adults with SMS go years without a major behavioral disturbance.

While behavioral challenges do improve over time for many adults with SMS, over-reactions are a part of SMS, and therefore not always easily controlled (Finucane 2008; Finucane et al., 1994; Finucane & Haas-Givler, 2009). Adults with SMS may be as confused by their own behavior as the caregiver, making it difficult to explain what happened or choose alternate adaptive behaviors without prompting or other support. Negative consequences and punishments can backfire because the individual with SMS may already feel helpless and defeated. They may not have the skills to control their own behavior, even if that is their desire. Instead, caregivers and families will need to find ways

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to change the environment to make it easier for individuals with SMS to be successful.

People who work with adults with complex cognitive and emotional care needs, such as individuals with SMS, should be well-trained, compassionate, and consistent (Ally et al., 2018; Berens et al., 2020; Finucane & Haas-Givler, 2009; Foster et al., 2013; Goslink et al., 2022; Nathenson & Zablostsky, 2017; Varshney et al., 2022). All caregivers will need to have training in positive behavioral support and the unique challenges of SMS. Developmental asynchrony continues (Finucane 2008; Haas-Givler & Finucane, 2014). Therefore, strategies like distraction and patience for tantrums need to be taught to caregivers (Finucane et al., 1994). Using the specialized services of a behaviorist to prepare, train, and oversee implementation of a behavioral support plan may be needed (Haas-Givler & Finucane, 2014). It is particularly important that everyone who works with an adult with SMS learn strategies to remain "neutral" and not let their own emotions and reactions make a situation worse.

Job training and job coaching should include an understanding of the potential behaviors with a long-term plan to continue intervening as problems arise (Roos & Søndenaa, 2020; Wehrlen et al., 2015). Often, job skills are learned quickly, but the social/behavioral expectations and reactions are a much bigger problem. With that said, workplaces sometimes consider their charming personalities to be an asset.

Throughout their lives, individuals with SMS will likely continue to need assistance with managing their emotions and behaviors (Finucane et al., 1994; Finucane & Haas-Givler, 2009; Foster et al., 2013). The best chance at success is having consistent environments (e.g., job, classes, home, day program) where their SMS challenges are respected, and modifications are made with a goal of being supportive rather than punitive (Wehrlen et al., 2015).

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Strategies for Success for the Adult with SMS

- All environments including home, school, work should be planned with known SMS strategies in mind
- Consider developmental and emotional age when setting expectations
- Staff should be trained specifically in what works to help everyone manage emotions/behaviors
  - Consult with families, long term caregivers, and successful teachers
- Access transition services while enrolled in school
  - Consider taking advantage of extended high school and skill based high school programs whenever available
- Sign up early for waitlists for Adult Services (e.g., years prior to turning 18)
- Apply for funding such as Supplemental Security Income and Home and Community Based Services (HCBS)
- Consult your State Department of Intellectual Disability and Developmental Delay (ID/DD)
- Consider medical alert bracelets, particularly for older individuals with SMS who are more active in the community
- Celebrate and maximize the adaptive traits and strengths that come with SMS
- Behavioral problems in adulthood are often not consistent and constant, so when problems arise, analyze what can change to better support success

Please see Table 1 (page 30) for additional information and resources for adult transition.

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Conclusion

Parents, caregivers, and those working with individuals with SMS are encouraged to review these guidelines and recommendations, coupled with seeking the input of the therapists, educators, and clinicians who work with and treat their children and adults with SMS. While there is no universally standard program that addresses all challenges for all individuals, evidence-based strategies can lead to improved emotional regulation and behavior. There is hope that with increased knowledge and research about SMS, interventions will be identified which will in turn improve the quality of life for children and adults with SMS.

Family support services and resources should be included as essential components of a holistic management plan for people with SMS. (Finucane & Haas-Givler, 2009). Respite services may provide caregivers with opportunities to care for themselves and other family members (Foster et al., 2010). Parents and caregivers are encouraged to seek support through PRISMS and connect with other local/regional SMS support groups.

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TABLE 1
Supplemental Online Resources Highlighted Within the Behavior Guidelines

Overall SMS Resources	Website
SMS Medical Management Guidelines	www.prisms.org/wp-content/uploads/pdf/mmg/PRISMS_Medical_Management_Guidelines2018.pdf
SMS GeneReviews®	www.ncbi.nlm.nih.gov/books/NBK1310/
Parents and Researchers Interested in Smith-Magenis syndrome (PRISMS)	www.prisms.org/education/publications-and-resources/ • Includes access to: — *A Guidebook for Families Newly Diagnosed with Smith-Magenis Syndrome* by Leah Baigell, Barclay Daranyi, Melissa Haley and John Mayer — *On the Road to Success with SMS: A Smith-Magenis Guidebook for Schools* by Barbara Haas-Givler and Brenda Finucane — *A Smith-Magenis Guidebook: Exploring Adult Residential Living* by Deborah Wehrlen, John Mayer, Leah Baigell, Barclay Daranyi, Mary Beall, Brandon Daniel, and Emily Fields — Previous PRISMS Spectrum Journals
Behavior Resources	**Website**
Overview of Applied Behavior Analysis (from the Behavior Analyst Certification Board)	www.bacb.com/about-behavior-analysis/
PRISMS – Behavior	www.prisms.org/about-sms/living-with-sms/behavior/
Association for Science in Autism Treatment	asatonline.org/for-parents/
American Academy of Pediatrics	www.healthychildren.org
Parent-to-Parent (family support network)	www.p2pusa.org/
Educational Resources	**Website**
U.S. Department of Education - Positive, Proactive Approaches to Supporting Children with Disabilities: A Guide for Stakeholders	sites.ed.gov/idea/idea-files/guide-positive-proactive-approaches-to-supporting-children-with-disabilities/
Wright's Law (special education law and advocacy)	https://www.wrightslaw.com/
What Works Clearinghouse (managed the Institute of Education Sciences within the US Department of Education)	ies.ed.gov/ncee/wwc/

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Speech, Language, and Hearing Resources	Website
Augmentative and Alternative Communication (from the American Speech-Language-Hearing Association)	www.asha.org/public/speech/disorders/aac/
Augmentative and Alternative Communication (from the International Society for Augmentative and Alternative Communication)	isaac-online.org/english/what-is-aac/
Key facts for Families New to Speech and Language Services (from the American Speech-Language-Hearing Association)	www.asha.org/news/2022/back-to-school-key-facts-for-families-new-to-speech-and-language-services/
Occupational Therapy Resources (Including Sensory Resources)	**Website**
Occupational Therapy Best Practice Guidelines (from the American Occupational Therapy Association)	www.aota.org/practice/practice-essentials/evidencebased-practiceknowledge-translation/aotas-top-10-choosing-wisely-recommendations
Mental Health/Psychiatric Resources	**Website**
HELP Model (from the Surrey Place Developmental Disabilities Primary Care Program)	ddprimarycare.surreyplace.ca/wp-content/uploads/2020/05/HELP-with-Emotional-Behavioural-Concerns-4May2020.pdf
Behaviors that Challenge (from the Surrey Place Developmental Disabilities Primary Care Program)	https://ddprimarycare.surreyplace.ca/guidelines/mental-health/behaviours-that-challenge/
A Guide to Understanding Behavioural Problems and Emotional Concerns (Psychiatric Symptoms and Behaviours Screen from the Surrey Place Developmental Disabilities Primary Care Program)	http://ddprimarycare.surreyplace.ca/wp-content/uploads/2018/04/Guide_to_Understand_Bx_Probs-2.pdf
American Academy of Child and Adolescent Psychiatry	www.aacap.org/AACAP/
Adult Transition and Family-Based Resources	**Website**
Supplemental Security Income	www.ssa.gov/ssi/
Home and Community Based Services (HCBS)	www.medicaid.gov/medicaid/home-community-based-services/index.html
Medicaid Waivers	medicaidwaiver.org/
U.S. State Vocational Rehabilitation Agencies	askearn.org/page/state-vocational-rehabilitation-agencies
Ticket to Work	choosework.ssa.gov/index
Got Transition	www.gottransition.org/
The Arc	www.thearc.org

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