

Smith-Magenis syndrome at a Glance

Smith-Magenis syndrome (SMS) is considered a developmental disability syndrome. SMS is characterized by distinctive facial features, developmental delay, intellectual impairment and distinct behaviors. Most individuals with SMS have a mild to moderate intellectual disability. The distinctive behavior includes an engaging and affectionate nature but may also include sleep disturbances, repetitive and ritualistic behaviors, and self-injurious behaviors. SMS is caused by a deletion of genetic material on part of chromosome 17. It is estimated that 1 in 15,000-25,000 children will be born with SMS.

Major physical traits and behaviors:

- Characteristic facial appearance (may be subtle)
- Low muscle tone (hypotonia)
- Oral-sensory motor dysfunction
 - Poor suck/swallow
 - Decreased tongue strength and movement
 - Open mouth posture
 - Frequent drooling
 - Possible aversion to certain textures
- Below normal or absent reflexes
- Peripheral neuropathy (weakness, numbness, and pain typically in hands and feet)
- Distinct, broad-based gait (walking pattern)
- Decreased sensitivity to pain
- Short stature
- Small hands and feet
 - Markedly flat or highly arched feet
- Abnormalities of the larynx
 - Polyps
 - Nodules
 - Edema
 - Partial vocal cord paralysis
 - Horse deep voice
 - Hypernasal speech

- Velopharyngeal insufficiency (improper closing in the soft palate) and/or structural vocal fold abnormalities
- Middle ear abnormalities
 - Hearing loss
 - Ear infections can lead to hearing loss
 - Hyperacusis (oversensitivity to certain frequencies and sounds) is common
- Missing teeth
- Major developmental and behavioral features (>75% of affected individuals)

Children with SMS may have some or many of the following:

- Positive behaviors
 - Endearing/appealing personalities
 - Excellent long term memory for names, places, events
 - Great sense of humor
- Developmental delay and variable intellectual disability (usually mild to moderate)
- Tendency to mouth objects or hands. This may persist beyond early childhood.
- Speech delay and articulation problems
- Autism or autistic-like features
- Sensory integration issues
- Tendency to grind teeth
- Delayed toileting skills
- Challenging behaviors
 - Hyperactivity
 - Impulsivity
 - Attention seeking (especially from adults)
 - Easy excitability or distractibility
 - Sudden mood shifts
 - Explosive outbursts
 - Prolonged tantrums
 - Aggressive or destructive behavior
- Chronic sleep disturbances
 - Frequent nighttime awakenings
 - Increased daytime sleepiness/naps
 - Early morning waking times (5:30 a.m. - 6:30 a.m.)
- Other stereotypic or repetitive behaviors include
 - Arm hugging/hand squeezing when excited
 - “Lick and flip” behavior (compulsively lick their fingers and flip through pages)
- Self-injurious behaviors:

- Head banging
- Hand biting
- Picking at skin, sores, and nails
- Pulling off fingernails and toenails
- Inserting foreign object into ears, nose, or other body orifices.

Less common (present in 25-50% of those with SMS) traits:

- Congenital heart defects
- Scoliosis (curvature of the spine)
- Eye problem
 - Strabismus
 - Nearsightedness
 - Small cornea
 - Iris anomalies
- Constipation
- High cholesterol and high triglycerides
- Abnormal EEG without seizures
- Thyroid function abnormalities
- Immune function abnormalities

Occasional traits:

- Kidney differences
- Cleft lip/palate
- Forearm differences
- Retinal detachment

1. Medical and Dietary Needs

What you need to know

- The severity of any one of the possible medical conditions varies widely between individuals. Therefore, it is important to ask parents about their child's specific medical issues.
- School age children with SMS may have multiple doctor and specialist visits to monitor medical conditions.
 - Regular neurodevelopmental and developmental/behavioral evaluations are important.

- Regular hearing evaluations, to monitor for conductive and/or sensorineural hearing loss, will likely be conducted.
- Be aware, or ask parents, if the child has a medical alert bracelet.
- No special diet is required for SMS although a well-balanced diet is important.

What you can do

- A yearly check-up and studies as needed should occur in the child's Medical Home.
- Be aware of any changes in behavior or mood that seem out of line with the situation and notify the parents.
 - It is important to be aware of any academic changes. Contact parents when any differences are noticed.

2. Education Supports

What you need to know

It is important to have HIGH LEARNING EXPECTATIONS for children who have Smith-Magenis syndrome. Encourage use of the core educational curriculum and modify it in order to meet the individual needs of the child.

There is a wide degree of variability in cognitive and adaptive function with the majority of individuals having mild to moderate intellectual disabilities. An individual with SMS often has a cognitive profile that includes a relative weakness in sequential processing and short-term memory. Individuals may also have relative strengths in long-term memory and perceptual closure (where an incomplete visual picture is perceived to be complete).

Therapies should include speech/language, physical, occupational, and sensory processing. Early childhood intervention programs and special education supports are important. With teens and adults, vocational training is important.

Therapeutic goals often include: increasing sensory input, increasing oral motor endurance, and decreasing hypersensitivity. These are needed to develop skills related to swallowing and speech/language production. Use of sign language and total

communication programs (i.e. computer assisted devices and tablets) can help improve communication skills and help with behavior.

- As children with SMS grow older, their unique style will likely become more evident.
- Take the time to learn the child's preferred learning style, and identify and develop its strengths.
- Hoarseness and hypernasal voice quality may be caused by issues that affect the child's ear, nose, and throat. These voice differences may contribute to delays in expressive language.

Students diagnosed with SMS may have some of the following traits and characteristics:

Positive traits and characteristics:

- Engaging and endearing personality
- Good eye contact
- Appreciative of attention, excitable
- Enjoy interaction with adults
- Learn and use names of teachers and students
- Use social expressions "please" and "thank you"
- Often demonstrate self-hugging or hand-squeezing motions
- Responsive to structure and routine
- React positively to consistency
- Follow classroom routine, especially with visual cues
- Motivated by a variety of reinforcers, e.g., food, stickers, attention
- Eager to please
- Communicative
 - Verbal
 - Sign language
 - May use a picture board
- Enjoy a variety of activities
 - Music
 - Water play
 - Puzzles
 - Electronics
 - Toys
 - Computers
- Well developed sense of humor
- Identifiable causes of tantrums and aggression

- Often due to changes in routine
- Predictable triggers
- Child may be able to verbalize what's wrong
- Tantrums and aggressive behaviors can often be redirected

Challenging traits and characteristics:

- Attention seeking
 - May demand a lot of one-on-one attention
 - May be aggressive towards others
 - May have tantrums
- Self-injurious behavior
 - Picking at nails and skin
 - Inserting foreign objects into body orifices
 - Head banging
- Poor impulse control
 - Jumping out of seat
 - Grabbing at things
- Negative reaction to change in routine
 - Provide cues and/or warning when a change is going to occur
- Attention deficit disorder
 - With or without hyperactivity
- Perseveration (repetition)
- Sleep disturbances

What you can do

- Adapt activities
- Modify the environment
- Work on facilitating appropriate and supportive social interactions
- Ensure a total communication program that includes sign language;
 - This approach can improve communication skills and also have a positive impact on behavior.
 - Development of expressive language appears dependent upon the early use of sign language and intervention by speech/language pathologists.
 - Communication skills help children to express their needs and wants, and to interact with adults and peers.
 - Speech often develops by school age but articulation problems may persist.

- Be aware that oral, motor, feeding, and speech language disorders occur in more than 75% of individuals with SMS.

3. Behavioral and Sensory Support

What you need to know

Children with SMS usually have significant sleep disturbances, stereotypies, (repetitive movement, posture, or utterance), maladaptive, and self-injurious behaviors.

It is important to balance your experience of a child's challenging behaviors with the recognition of his or her many positive attributes. Many people with SMS have an engaging and endearing personality.

Individuals with SMS typically have inattention, distractibility, hyperactivity, and impulsivity. Individuals may also have obsessive-compulsive disorder (OCD), oppositional defiant disorder (ODD), and difficulties processing sensory information. Individuals with SMS crave the attention and company of adults.

The frequency and type of behaviors will vary between individuals with SMS.

Possible behaviors

- Frequent outbursts
- Temper tantrums
- Attention seeking
- Disobedience
- Aggression
- Toileting difficulties
- Lick and flick – meaning that they lick their fingers and flick pages over and over again
- Teeth grinding
- Body rocking
- Spinning or twirling objects
- Self-injurious behaviors
 - Self-hitting,
 - Self-biting
 - Skin picking
 - Insertion of foreign objects into body orifices

- Yanking fingernails and/or toenails
- Mouthing of hands or objects persists from early childhood
- Quick upper body squeeze or self-hug highly associated with SMS
- Many individuals will have autism or autistic-like features

Other factors that affect behavior

- Differences in level of intellectual functioning and emotional maturity may contribute to maladaptive behaviors.
- Sleep disturbance is also known to be a significant contributor to negative behaviors in people with SMS.
- Sensory processing challenges may be present and persist throughout childhood

Adolescence and beyond

The SMS behavioral profile often escalates with puberty. The gap between intellectual attainment and emotional development appears to widen for many with SMS and this can pose challenges for older children and adults.

Adolescents may experience:

- Rapid mood shifts
- Increased impulsivity
- Increased anxiety with or without fright/fight reaction
- Aggressive outbursts are common and escalate with age
- Pubertal onset of seizures may occur, especially with females having menses
- Insertion of objects into body orifices may increase

Outbursts often result from frustration over:

- Communication difficulties
- Fine motor challenges
- Changes in routine
- Many people with SMS crave interaction with adults and react negatively if attention is withdrawn or directed toward others
- People with SMS frequently have rigid ideas about their world. A prolonged tantrum may result if things do not go exactly as expected or envisioned.

Information on interventions:

- Published data about the optimal intervention and behavioral strategies in SMS are limited to anecdotal and experiential findings.

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- Use of psychotropic medication may increase attention and/or decrease hyperactivity. No single regimen shows consistent efficacy.
- Therapeutic management of the sleep disorder in SMS remains a challenge for physicians and parents.

What you can do

- Provide a calm, consistent classroom. A small class size is preferred.
 - Use reinforcement and motivation techniques
 - Use visual cues
 - Use pictures and charts for transition, tasks, schedules, etc.
 - Be aware that the child may have difficulty with processing information in a sequential or step-by-step manner
- Be aware that transitions from one activity to another or unexpected changes in the school routine can be difficult and provide advance warning before transitions or special events.
- Learn and utilize appropriate behavioral therapies – they are integral in behavioral management.
 - Special education techniques that emphasize individualized instruction, structure, and routine can help minimize behavioral outbursts in the school setting.
 - Positive behavioral supports and a functional behavioral assessment should be considered.
- Work with the child’s parents to ensure a comprehensive and consistent support plan is in place both at home and in school.
 - A structured school program with one-on-one support is important.
- Be aware that children with SMS may use of medication to treat hyperactivity.
- Utilize behavioral techniques that emphasize individualized instruction, structure, and routine. These can help minimize behavioral outbursts in school.
- Help create the optimal environment for the child. This may include respite care and family psychosocial support.

4. Physical Activity, Trips, Events

What you need to know

- Any change in routine may produce anxiety, fears, and/or worry.

What you can do

- Offer anticipatory guidance and information to prepare for a change in routine such as a field trip.
- Create a picture story about the upcoming event. The child can rehearse it alone or with others.
- If the child has any sensory, hearing, or vision issues, he/she may need preferred seating.
- If you live in New England (USA) and qualify, Northeast Passage offers Therapeutic Recreation and Adaptive Sports programming (www.nepassage.org).

5. School Absences and Fatigue

What you need to know

Absences

- Children with SMS may be absent due to illness and/or medical appointments

Fatigue

- Sleep disturbances are characterized by fragmented and shortened sleep cycles with frequent nocturnal and early morning awakening and excessive daytime sleepiness

What you can do

- Help to make transitions in and out of school as seamless as possible.
- Help provide a supportive educational environment. This may include finding a space where the child can rest or take breaks throughout the day.
- Be aware that a child with SMS may need to take medication to manage their sleep disorder.

6. Emergency Planning

What you need to know

- If necessary, work with the child's parents and medical team to develop an emergency plan that is tailored to the child's specific needs and environment.

7. Resources

Genetics Home Reference

<http://ghr.nlm.nih.gov/condition/smith-magenis-syndrome>

Consumer-friendly information about human genetics from the U.S. National Library of Medicine

Parents and Researchers Interested in SMS (PRISMS)

<http://www.prisms.org/>

<http://www.prisms.org/us/living-with-sms/education>

This organization is dedicated to providing information and support to families with SMS. The site also strives to increase awareness and understanding of SMS. The site offers specific tips and information to aid educators.