

- **Regional Representative Program**—a support and mentoring program connecting new families with experienced families from around the world.
- **Publications and Resources**—Curated guides written by SMS experts including “On the Road to Success with SMS: A Guidebook for Schools” and “A Smith-Magenis Guidebook: Exploring Adult Residential Living.”
- **SMS Clinics**—PRISMS’ Clinic and Research Consortium is a network of clinics across the US dedicated to providing expert, multi-specialty care to persons with SMS.

AWARENESS

- **International SMS Awareness Day** (every November 17th)—PRISMS awareness campaigns and support of community fundraisers.
- **PRISMS Awareness Materials**—bookmarks, wristbands, compassion cards and other tools to create awareness and understanding of SMS.

RESEARCH

- **PRISMS Professional Advisory Board**—leading medical, clinical and research SMS experts who collaborate with PRISMS and each other to drive further knowledge and research.
- **SMS Research Symposium**—connecting SMS researchers and clinicians around the world to share updates, best practices and interventions, and to develop future research plans and goals.
- **Patient Registry**—PRISMS-funded natural history research to further understand this rare disorder and inform treatment and management best practices.
- **Research support and funding**—fostering and supporting SMS researchers to answer key questions about SMS.

PRISMS Mission

PRISMS is dedicated to providing information and support to families of persons with Smith-Magenis syndrome (SMS), sponsoring research and fostering partnerships with professionals to increase awareness and understanding of SMS.

PRISMS Vision

PRISMS is a leader of the Smith-Magenis syndrome community and engages, inspires and supports families, physicians, educators, researchers, and others so they can improve the lives of everyone affected by SMS.

GET CONNECTED WITH PRISMS

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Can You Support PRISMS?

Much of PRISMS’ work is carried out by committed parent volunteers committed to serving families nationally and around the world. PRISMS is a nonprofit 501(c)(3) organization and receives no government funding. The critical work of PRISMS would not be possible without our generous donors.

To donate please visit:



www.prisms.org

Or mail your donation to:

PRISMS
c/o Wood & Huston Bank
PO Box 724
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PARENTS AND RESEARCHERS
INTERESTED IN SMITH-MAGENIS SYNDROME

Understanding Smith-Magenis Syndrome



What is Smith-Magenis Syndrome (SMS)?

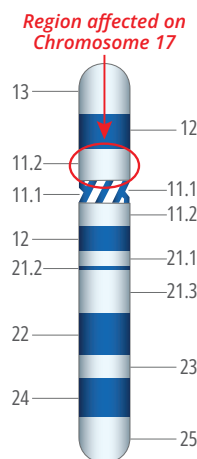
Smith-Magenis syndrome (SMS) is a neurobehavioral disorder caused by genetic changes on chromosome 17, specifically chromosomal region 17p11.2, which contains the gene RAI1. In most cases, SMS is due to a missing piece of the 17p11.2 region. This is called a deletion. Less commonly, SMS is caused by genetic changes, called mutations, in the RAI1 gene itself.

SMS is a rare disorder that occurs in about 1:15,000 to 1:25,000 births. Although the number of individuals diagnosed with SMS has increased in recent years due to better diagnostic tools, the syndrome is likely underdiagnosed.



Diagnosis

The diagnosis of SMS is usually made using a genetic test called a chromosomal microarray analysis (CMA), also known as a microarray. A microarray can detect extra or missing pieces of genetic material. This test is typically ordered as part of the standard genetic work-up for children with developmental delays, with or without behavioral symptoms. When SMS is suspected, more targeted genetic tests, such as FISH (fluorescence in situ hybridization) or RAI1 mutation analysis can be ordered instead of a microarray, to look specifically for the SMS deletion or mutation respectively. Although SMS is genetic, it usually is not inherited, which means it does not run in families.



Common Features of SMS

- Global developmental delay/intellectual disability
- Hypotonia (low muscle tone)
- Poor gross motor and fine motor skills
- Feeding problems in infancy (poor suck/swallow); failure to thrive or difficulty gaining weight in early years
- Speech delay
- Characteristic facial appearance
- Short fingers and toes
- Distinct, broad-based gait (style of walking)
- Scoliosis (abnormal curvature of the spine)
- Vision problems (strabismus; nearsightedness)
- Middle ear abnormalities; chronic ear infections; hearing impairment
- Hoarse, deep, sometimes nasal voice
- Constipation
- Sleep disturbances (frequent nighttime awakenings; daytime sleepiness)
- Self-injurious behaviors
- Prolonged tantrums; explosive outbursts; impulsivity
- Arm hugging/hand squeezing
- Endearing and engaging personality
- Excellent long-term memory for names, places, events
- Great sense of humor
- Eagerness to please and sensitivity towards others

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It is important to remember SMS is a variable disorder, and not every individual with SMS will have all the features listed above. Some may have just a few, whereas others may have many.



30 Years of Education, Awareness, Research

For over 30 years, Parents and Researchers Interested in Smith-Magenis syndrome (PRISMS) has provided support for families as well as medical, educational and other professionals involved in the care of people with SMS. PRISMS is a distinguished global resource for information about SMS, providing a range of educational and support services, including:

EDUCATION

- **PRISMS.org**—a comprehensive resource with information and support for individuals and families living with SMS. The website offers medical management recommendations, research articles, and other resources.
- **Medical Management Guidelines**—PRISMS works with its Professional Advisory Board of experienced SMS clinicians and researchers to maintain current recommendations for medical management of SMS. This guide is an invaluable tool for newly-diagnosed individuals and all families.
- **Telephone and email support**—PRISMS staff and parent volunteers are available to support families and direct them to resources appropriate for their child's age and needs.
- **Educational Webinars**—virtual events on important topics for medical management and family support.
- **International Conference**—a multi-day conference with presentations by SMS researchers and professionals. A place to learn and connect with families from around the country and around the globe.

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