Parents and Researchers Interested in Smith-Magenis Syndrome

EDUCATION | AWARENESS | RESEARCH

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SMS Awareness Fact Sheet

This SMS Awareness Day, join us in spreading awareness of Smith-Magenis Syndrome by posting facts about SMS to your social media pages throughout the month! We've made it easy for you by pulling together 17 facts that you can share every day leading up to SMS Awareness Day. You can share them as-is or make your posts more personal by sharing a picture of your family with each fact or adding a story about how the fact has impacted you and your individual with SMS. However you choose to use them, thank you for spreading the word about Smith-Magenis Syndrome with us this month!

The Facts

Each post has a long version and a short version. Choose whichever version makes sense for your social media! The short version can be posted to Twitter.

Don't forget to add our hashtags #SMSAwareness #SmithMagenisSyndrome #SMSAwarenessDay #MilesofSmiles #PRISMS

Day 1

Long Version

SMS Awareness Day is November 17th. In honor of SMS Awareness Day, I will be sharing facts throughout the month about Smith-Magenis Syndrome. Learn more and join in by visiting https://www.prisms.org.

Smith-Magenis Syndrome (or SMS) is a chromosomal disorder characterized by a recognizable pattern of physical, behavioral, and developmental features. It is caused by a missing piece of genetic material from chromosome 17, referred to as deletion 17p11.2.

Short Version

SMS Awareness Day is November 17th. In honor of SMS Awareness Day, I will be sharing facts throughout the month about Smith-Magenis Syndrome. Learn more and join in by visiting https://www.prisms.org/awareness/sms-awareness-day/

Day 2

Long Version

Common features of Smith-Magenis Syndrome include: characteristic, yet subtle, facial appearance, infant feeding problems, low muscle tone, developmental delay, variable levels of intellectual disability, early speech/language delay, middle ear problems, skeletal anomalies and decreased sensitivity to pain. The syndrome also includes a distinct pattern of neurobehavioral features characterized by chronic sleep disturbances, arm hugging/hand squeezing, hyperactivity and attention problems, prolonged tantrums, sudden mood changes and/or explosive outbursts and self-injurious behaviors

Short Version

Common features of SMS include: characteristic, yet subtle, facial appearance, infant feeding problems, low muscle tone, developmental delay, variable levels of intellectual disability, early speech/language delay, middle ear problems, skeletal anomalies and decreased sensitivity to pain.

Day 3

Long Version

Although the exact incidence is not known, it is estimated that Smith-Magenis Syndrome occurs in between 1 out of every 15,000 to 25,000 births. SMS is likely under-diagnosed. As genetic testing becomes more accessible and awareness of SMS increases, the number of people identified grows every year.

Short Version

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Day 4

Long Version

Most people diagnosed with SMS are born with a small deletion of one member of their 17th pair of chromosomes. It is the lack of this specific section, known as 17p11.2, which causes the features of SMS.

The genes commonly deleted in persons with SMS have been narrowed to a “critical region” encompassing approximately 25 genes. All deletion cases include deletion of the RAI1 gene. Small changes in this gene can also cause SMS.

Approximately 90% of the cases of SMS are due to deletion, with the remaining 10% due to a mutation that occurs within the RAI1 gene.

Short Version

Most people diagnosed with SMS are born with a small deletion of one member of their 17th pair of chromosomes. It is the lack of this specific section, known as 17p11.2, which causes the features of SMS. This area includes the gene RAI1, which has been linked to SMS.

Day 5

Long Version

The diagnosis of Smith-Magenis Syndrome is usually confirmed through a clinical blood test called a chromosome microarray analysis (CMA). Diagnosis can also be made through a cytogenetic test called FISH (fluorescence in situ hybridization). These tests can detect the deletion on the 17th pair of chromosomes that is the most common cause of SMS.

In cases where a deletion is not detected, next generation sequencing tests can be used to detect changes in the RAI1 gene.

Short Version

The diagnosis of SMS is usually confirmed through a clinical blood test called a chromosome microarray analysis (CMA). Diagnosis can also be made through a cytogenetic test called FISH (fluorescence in situ hybridization) or through next generation sequencing tests.

Day 6

Long Version

Although SMS is caused by a deletion or change of genetic material, it usually does not run in families. In most cases, the deletion or change occurs randomly in a child around the time he or she is conceived, without being inherited from either parent. For this reason, we can say that SMS is clearly genetic, but not usually familial. The risk to siblings depends on the results of parental chromosome analysis. If parental chromosomes are normal, the risks to subsequent pregnancies are extremely low. Families are advised to consult a genetics counselor or specialist for further advice regarding their own particular family situation.

Short Version

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Day 7

Long Version

Melatonin is a hormone secreted by the pineal gland that helps control body rhythms and sleep wake cycles in relation to daylight. Individuals with SMS have an inverted circadian rhythm, which means they produce melatonin during the day. This can lead to nighttime disturbances and poor sleep, which in turn contributes to sudden naps during the day and behavioral issues from being sleep deprived. This can also lead to nighttime safety concerns, as some individuals with SMS will venture out of their bedrooms while others in the house are sleeping.

Short Version

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Day 8

Long Version

Many individuals with Smith-Magenis Syndrome share similar facial characteristics, like a broad forehead, short upturned nose, and “tent” shaped upper lip. While subtle during early childhood, these features become more distinctive as they age.

Short Version

Many SMS individuals share similar facial characteristics like a broad forehead, short upturn nose, and “tent” shaped upper lip. While subtle during early childhood, they become more distinctive as they age.

Day 9

Long Version

Characteristics of Smith-Magenis Syndrome include short stature, abnormal curvature of the spine (scoliosis), reduced sensitivity to pain and temperature and a hoarse voice. Some people with this disorder have hearing abnormalities. Affected individuals may have eye abnormalities that cause nearsightedness (myopia) and other problems with vision. Heart and kidney defects also have been reported in people with Smith-Magenis Syndrome, though they are less common.

Short Version

SMS has several other identifying characteristics such as short stature, high pain tolerance, and a hoarse voice among others. SMS individuals are often monitored for other possible health concerns such as hearing and eye abnormalities.

Day 10

Long Version

Oral motor, feeding and speech-language disorders occur in more than 75% of individuals diagnosed with Smith-Magenis Syndrome. The oral motor and swallowing/feeding difficulties typically begin within the first year of life and improve throughout childhood with therapeutic interventions. For speech, although progress may be slow, most persons with SMS are able to eventually speak using a variety of interventions to enhance communication. Individuals with SMS may have speech delay, yet they are always able to tell you what they want in the own ways!

Learn more about speech and language in SMS here: https://www.prisms.org/about-sms/living-with-sms/speech-language/

Short Version

Individuals with SMS may have speech/language delays, yet they are excellent communicators in their own way. Learn more about speech and language in SMS here: https://www.prisms.org/about-sms/living-with-sms/speech-language/

Day 11

Long Version

Challenging behaviors occur at a much higher rate among children and adults with SMS than they do in people with other types of developmental disabilities. Common behavioral differences in individuals with SMS include attention-seeking, self-injury, attention deficits, with or without hyperactivity, explosive outbursts, prolonged tantrums, destructive and aggressive behavior. The reason for these behaviors is unclear, although they appear to be related to underlying physical and developmental differences associated with SMS.

Learn more about challenging behaviors in SMS here: https://www.prisms.org/about-sms/living-with-sms/behavior/

Short Version

Challenging behaviors are often seen in SMS at any age. Some common behaviors can include self-injury, attention seeking, prolonged tantrums, and aggressive behavior. Learn more about challenging behaviors in SMS at https://www.prisms.org/about-sms/living-with-sms/behavior/

Day 12

Long Version

Despite their very difficult behaviors, children and adults with SMS are very affectionate and have engaging personalities and much untapped potential. This combined with them not always knowing their own strength makes for some very sweet and affectionate but sometimes strong hugs. Our community is proud to have adopted the motto “Hug or be hugged!”

Short Version

Despite their very difficult behaviors, individuals with SMS are very affectionate and have engaging personalities. This combined with them not always knowing their own strength makes for some very sweet but strong hugs. Our community has adopted the motto “Hug or be hugged!”

Day 13

Long Version

People with SMS can expect to accomplish many of the things their "typical" peers do - attend school, engage in hobbies, and even be successfully employed. They do, however, need a significant amount of support from their families, school, work and residential service providers to achieve these goals. Early intervention plays a significant role.

See what is possible by watching our SMS Possibilities video from our most recent family conference: https://youtu.be/mZ12cg285Mo

Short Version

With the right amount of support, people with SMS can expect to accomplish many of the things their "typical" peers do - attend school, engage in hobbies, and even be successfully employed.

See what is possible! https://youtu.be/mZ12cg285Mo

Day 14

Long Version

There is no cure for Smith–Magenis Syndrome. Children with SMS often require several forms of support, including physical therapy, occupational therapy and speech therapy. Support is often required throughout an affected person's lifetime.

PRISMS has curated a series of medical management guidelines and treatment recommendations to help support families and the clinicians who care for their individuals with SMS. Learn more by visiting https://www.prisms.org/about-sms/living-with-sms/medical-management-guidelines/

Short Version

Children with SMS often require several forms of support, including physical therapy, occupational therapy and speech therapy. Support is often required throughout an affected person's lifetime.

Day 15

Long Version

Smith–Magenis Syndrome is named after the two pioneering clinicians who described the condition in 1986, namely, Ann C. M. Smith, a genetic counselor at the National Institutes of Health, and R. Ellen Magenis, who was a pediatrician, medical geneticist and cytogeneticist at the Oregon Health Sciences University. Other names people may use to describe Smith-Magenis Syndrome include chromosome 17p deletion syndrome, deletion 17p syndrome, 17p11.2 monosomy, partial monosomy 17p, SMS and 17p- syndrome.

PRISMS Clinic Research Consortium, (PCRC), provides comprehensive care for people with SMS, who have the opportunity to consult with a multi-disciplinary team of experts. Learn more about the PCRC here: https://www.prisms.org/about- sms/living-with-sms/sms-clinics/

Short Version

SMS is named after the two pioneering clinicians who described the condition in 1986: Ann C. M. Smith, a genetic counselor at the National Institutes of Health, and R. Ellen Magenis, who was a pediatrician, medical geneticist and cytogeneticist at the Oregon Health Sciences University.

Day 16

Long Version

While there has been no formal study of the life expectancy of individuals with SMS, they appear to have a normal life expectancy. The oldest known person with SMS lived into her late 80's.

Life expectancy is one of the many topics we hope to learn more about through research. You can help us learn more about SMS by joining our Patient Registry. Learn more here: https://www.prisms.org/research/sms-patient-registry/

Short Version

While there has been no formal study of it, individuals with SMS appear to have a normal life expectancy. The oldest known person with SMS lived into her late 80's. The SMS Patient Registry will help answer this question: https://www.prisms.org/research/sms-patient-registry/

Day 17

Long Version

Happy Smith-Magenis Syndrome Awareness Day! We observe SMS Awareness Day on 11/17 to celebrate people with SMS and raise the awareness and understanding of this complex syndrome.

Thank you for joining me this month to learn more about Smith-Magenis Syndrome and our SMS individuals.

There is an amazing community of parents, researchers, teachers, medical professionals that strive to find therapies and other support solutions for these amazing individuals. If you would like to be a part of this community and help PRISMS in continued support of these individuals, please visit https://www.prisms.org/donate/

Short Version

Happy Smith-Magenis Syndrome Awareness Day! Thank you for joining me this month to learn more about SMS. If you would like to be a part of this amazing community and help support individuals and families like mine affected by SMS, please visit https://www.prisms.org/donate/

Thank you for helping spread SMS Awareness!