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National Disability Forum
November 7th, 2017

- PRISMS (Parents and Researchers Interested in Smith-Magenis Syndrome) is dedicated to providing information and support to families of persons with SMS, sponsoring research and fostering partnerships with professionals to increase awareness and understanding of SMS

- PRISMS is the only holistic support organization focusing on the education, awareness, and research needs of the entire SMS community, internationally

- PRISMS conducts national and international awareness campaigns
- Funds and promotes SMS research
- Hosts informational and supportive international conferences for SMS patients, their families and the professionals who serve them
- Creates and distribute educational resources and materials
- Organizes scientific meetings
- Advocates on behalf of SMS patients and their families

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Parents and Researchers Interested in Smith-Magenis Syndrome

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SMS is a chromosomal disorder characterized by a recognizable pattern of physical, behavioral, and developmental features caused by a missing piece of genetic material from chromosome 17, referred to as deletion 17p11.2

Experts estimate that SMS occurs in somewhere between 1 in 15,000 and 1 in 25,000 births.

With this incidence we can estimate 16,000 individuals in the US and 370,000 individuals globally are affected by Smith-Magenis Syndrome

ICD 9 CM Code: 758.33

- A distinct pattern of physical and behavioral characteristics are present in people with SMS. An individual with SMS may have just a few or many of the features listed below
    - Low muscle town
    - Oral-sensory motor dysfunction
    - Hyporeflexia
    - Peripheral neuropathy
    - Decreased sensitivity to pain
    - Developmental delay
    - Intellectual disability
    - Speech delay
    - Chronic sleep disturbance
    - Inverted circadian rhythm
    - Self-injurious behaviors
    - Constipation
    - Scoliosis
    - Hearing impairment
    - Congenital heart defect
    - Low immune function
    - Seizures
    - Thyroid abnormalities
    - Retinal detachment

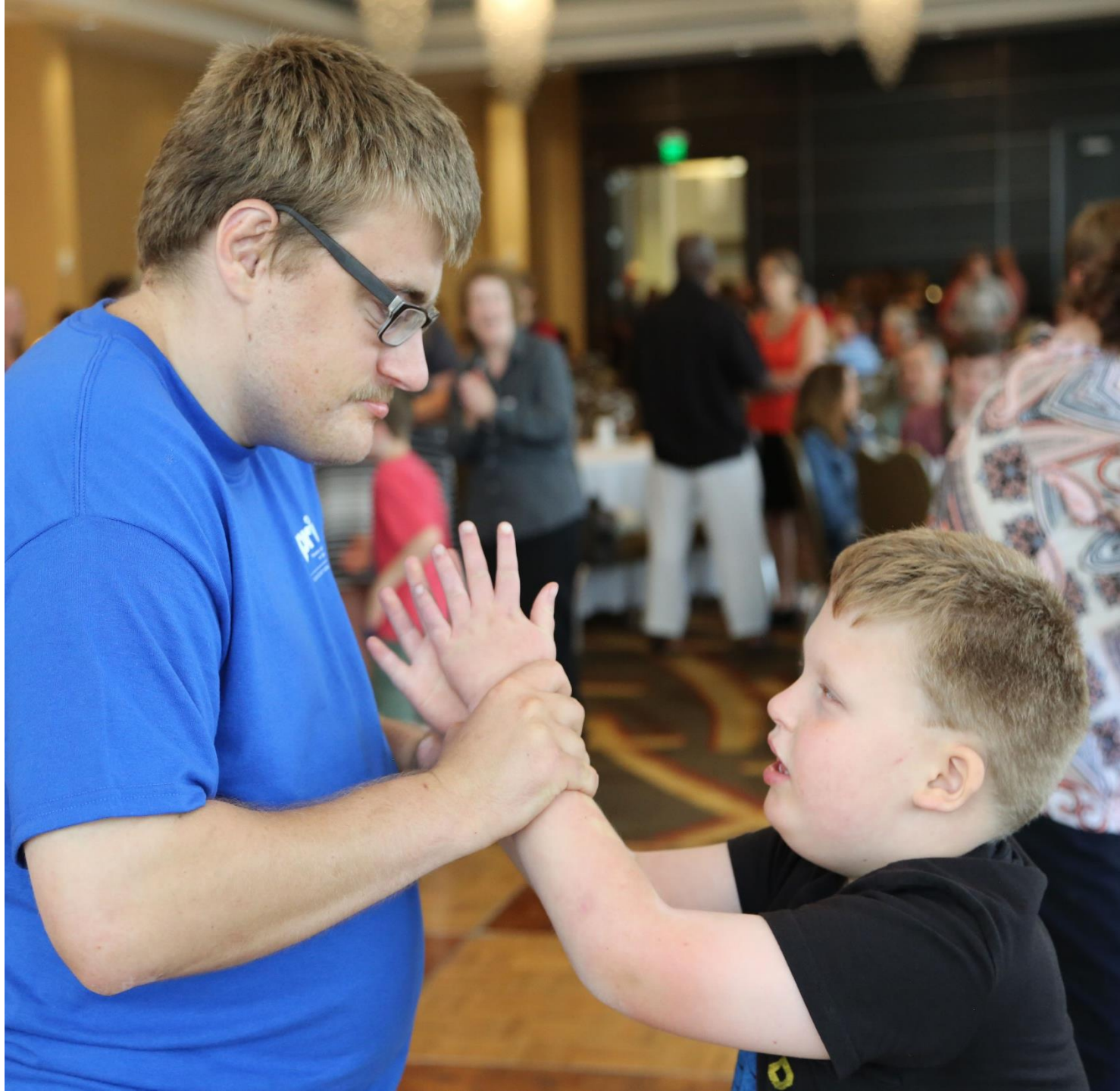
The diagnosis of SMS is confirmed through a clinical blood test – chromosome analysis.

- Cytogenetic test
- FISH
- Chromosome microarray analysis

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

The genes commonly deleted in persons with SMS have been narrowed down to a “critical region” and encompass approximately 25 genes.

All deletion cases include deletion of RAI1 gene



Management of SMS involves treatment to mitigate symptoms.

To establish the extent of the disease in an individual diagnosed with SMS, the following is part of the recommended evaluations:

- Review of systems
- Neurologic examination
- Renal ultrasound
- Echocardiogram
- Spine radiograph
- Blood chemistries
- Audiologic evaluation
- Speech/language pathology evaluation
- Neuroimaging
- EEG
- Sleep history

Treatment of Symptoms

- Ongoing pediatric care with regular immunizations
- Early childhood intervention programs
- Sign language and total communication programs
- Psychotropic medication
- Behavioral therapy
- Therapeutic management of sleep disorder

Thank You!

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