



Sotos syndrome, also known as cerebral gigantism, is a brain-based condition resulting in developmental delays in early childhood. At present the diagnosis is made by observing a host of physical characteristics and developmental delays in

early childhood. Errors in a gene called NSD-1 apparently cause Sotos syndrome in some individuals. Genetic testing is available but may not be definitive since it appears that some people with Sotos do not have errors in the NSD-1 gene. An MRI (magnetic resonance imaging) of the brain may reveal specific marginal anomalies which are characteristic of Sotos syndrome.



Typical characteristics:

Birth

Presence of high arched palate (roof of the mouth is narrow and arched upward), poor suck and low muscle tone often produces feeding problems. Jaundice occurs frequently.

Craniofacial

Facial features include a large head with a tall, narrow skull, wide set down slanting eyes, flat-bridged nose, early eruption of teeth (as early as 3 months of age), thin hair, pointed chin, prominent forehead, and "receding hairline."



Skeleton

An advanced bone age (the bones grow and mature faster than expected for the child's chronological age) is common. The hands and feet may be large in comparison with the rest of the body. Flat or pronated (collapsed inward) feet are common, and scoliosis may develop.

Development

Motor delays are common due to hypotonia. Prolonged drooling and mouth breathing may be present due to poor tone of facial muscles. Delays in gross and fine motor movement are marked in early childhood and improve in the school years. Coordination problems may persist into adulthood. Receptive language ability (understanding others) tends to be more advanced than expressive language (formation of words), setting the stage for frustration. The child may whine and scream to demonstrate desires or emotions. Older children seem to develop competence and normal speech patterns. Borderline to average intelligence is expected, with learning deficits noted in language, math, and socialization. Intellectual, social and emotional maturity may evolve on widely different timetables.

Growth

Rapid growth is common during the first five years of life. Thereafter growth continues to parallel the 97th percentile or above. Head circumference has been documented well about the 98th percentile.

Other Features

Any of these may or may not be present:

- ▶ Behavioural patterns, including phobias, aggression, obsessions, adherence to routine, autistic-like behaviour, attention deficit disorder, above-average memorization skills
- ▶ Enlarged ventricles of the brain, hydrocephalus (rare), abnormal EEG's and seizures (with at least 1/3 of seizures being fever-related)
- ▶ Frequent ear infections and upper respiratory infections, asthma and allergies
- ▶ Constipation, megacolon
- ▶ Delayed toilet training, urogenital anomalies
- ▶ Nystagmus, strabismus
- ▶ Increased sweating
- ▶ Hyperthyroidism, hypothyroidism
- ▶ Possible increased risk for tumours

Other Syndromes/Disorders

These syndromes or disorders should be considered because of shared physical features:

- ▶ Fragile X syndrome
- ▶ Beckwith-Wiedemann syndrome
- ▶ Weaver syndrome
- ▶ Bannayan, Ruvalcaba, Riley-Smith syndrome
- ▶ Marshall-Smith syndrome
- ▶ IODM (infant of a diabetic mother)
- ▶ Gorlin syndrome
- ▶ Pituitary gigantism
- ▶ Pineal tumour
- ▶ Simpson-Rosen-Golabi-Behmel syndrome
- ▶ Lysosomal storage disorders

Early Intervention Programs

Programs such as infant stimulation, occupational therapy, physical therapy, speech therapy, and adaptive physical education play a significant role in the nurturing of a child with Sotos syndrome. In a structured environment, the child is able to practice necessary skills without excessive distractions. Alternative strategies for effective movement and communication can give a child additional mobility and the language to encourage self-help skills. As with any child, self-esteem in a child with Sotos syndrome is bolstered by successful completion of tasks and mastery of developmental skills.

The information in this pamphlet is from the Sotos Syndrome Support Association (SSSA). The SSSA is made up of families, physicians, genetic counsellors, and health care agencies. They organize an annual conference for professionals and families to hear experts in genetics, neurology, psychology and education as well as a chance to meet other parents of children with Sotos.

SSSA: P.O. Box 4626, Wheaton, IL 60189 USA - SSSA@SotosSyndrome.org - www.SotosSyndrome.org - Toll free (888) 246-7772