

Sotos Syndrome For Healthcare Providers

This is a customized health care provider version of our website. Please visit the main website to find more comprehensive information for families and schools (www.gemssforschools.org).

Physical characteristics and/or symptoms

Note: not all people with Sotos syndrome will have all of these features.

Primary features include:

- **Characteristic facial features:**
 - Malar flushing
 - High prominent forehead with receding hairline
 - Down slanting palpebral fissures
 - Long narrow face
 - Prominent jaw with narrow chin and a high, narrow palate
- **Learning difficulties:**
 - Developmental delay
 - Motor and fine motor skill delays
 - Often very “clumsy”
 - Expressive language delay
- **Overgrowth:**
 - Over 90% of children have a height and/or head circumference 2 + SD above the normal
 - Birth weight is usually normal
 - Height may normalize in adulthood, but head size remains large
 - Disproportionately large hands and feet

Other common features:

- Advanced bone age
- Cardiac anomalies
- Cranial MRI/CT abnormalities
- Joint hyperlaxity/*pes planus*

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- Maternal preeclampsia
- Neonatal complications
- Renal anomalies
- Scoliosis
- Seizures
- Dental abnormalities
- Frequent upper respiratory issues
- Behavior problems
 - Anxiety
 - Depression
 - Phobias
 - Sleep disturbances
 - Tantrums
 - Irritability
 - Stereotypies
 - Inappropriate speech
 - Withdrawal
 - Hyperactivity

Less common findings:

- Feeding difficulties/Reflux
- Dislocated hips/club feet
- Autonomic dysfunction
- Constipation
- Eye problems
- Conductive hearing loss
- Thyroid disorders
- Glucose intolerance

Recommended Routine Surveillance

- Surveillance for scoliosis
- Monitor for seizures
- Screening for cardiac and renal abnormalities
- Monitor development

Emergency Protocols

There are no specific emergency protocols for this particular condition as it is not typically associated with episodes of sudden and serious medical decompensation.

- Emergencies should be handled as with any child.
- If seizures are present, the following seizure action plan may be useful:
- https://www.aap.org/en-us/Documents/Seizure_Action_Plan_for%20School.pdf

Specialists Who May Be Involved

Follow up is needed on a case-by-case. A multidisciplinary team approach to best meet the child's individual needs is recommended.

- Cardiologist
 - Screening for cardiac anomalies and blood pressure
- Developmental evaluation
 - Speech therapy
 - Physical therapy
 - Occupational therapy
- ENT
 - Otitis media
 - Hearing loss
- Gastroenterologist
 - Gastroesophageal reflex
 - Constipation
- Geneticist / Genetic Counselor:
 - Diagnosis
 - Coordination of care
 - Genetic risk for family
 - Clinical trials
- Neurology:
 - Monitor seizures
 - Cranial MRI/CT abnormalities
- Orthopedists
 - Scoliosis
 - Joint laxity
- Ophthalmologist
 - Astigmatism/Myopia
 - Cataract
- Nephrologist
 - Monitor renal anomalies

Sample Forms

Sample paragraph to be used for Letters of Medical Necessity or Letters to the school

My patient _____ has been diagnosed with Sotos syndrome. Learning disabilities, characteristic facial features, and overgrowth characterize Sotos syndrome. Medical complications with Sotos syndrome include management of cardiac anomalies, renal anomalies, scoliosis, seizures, gastrointestinal reflux, and constipation. Because of these, _____ needs the following accommodations.

Seven Important Aspects of School Life

“[Sotos Syndrome at a Glance](#)” will help you talk with parents and schools about:

- Medical / Dietary Needs
- Education Supports
- Behavior & Sensory Supports
- Physical Activity, Trips, Events
- School Absences & Fatigue
- Emergency Planning
- Transitions



Resources

Gene Reviews: Sotos syndrome

<http://www.ncbi.nlm.nih.gov/books/NBK1479/>

Genetic Home Reference

<https://ghr.nlm.nih.gov/condition/sotos-syndrome>

Sotos Syndrome Support Group

<http://sotossyndrome.org>