

Sotos Syndrome at a Glance

Sotos is a genetic condition caused by a change on chromosome 5. Children diagnosed with Sotos syndrome have characteristic facial features, overgrowth in height, weight and head size, and delays in their motor, cognitive and social development. Their speech is difficult and they have low muscle tone, which can affect feeding, and movement. The prevalence of Sotos is 1 in 14,000. Sotos is sometimes referred to as cerebral gigantism.

Sotos is usually diagnosed months or years after birth.

Learn more

Primary features include:

- **Characteristic facial features:**
 - Rosy coloring over face and nose
 - High prominent forehead with receding hairline
 - Down slanting eyelid openings (palpebral fissures)
 - Long narrow face
 - Prominent jaw with narrow chin and a high, narrow palate
- **Learning difficulties:**
 - Developmental delay
 - About 15-20% have normal intelligence
 - Ranges from mild to severe
 - IQ is in normal range (>70) in most individuals
 - Motor skill delays due to:
 - Large size
 - Hypotonia
 - Poor coordination
 - Some children will have feeding difficulties as newborns
 - Expressive language delay
- **Overgrowth:**
 - Over 90% of children have a height and/or head circumference 2 + SD above the normal
 - Height may normalize in adulthood, but head size remains large
 - Disproportionately large hands and feet

Other common features

- Advanced bone age
- Poor fine motor control
- Cranial MRI/CT abnormalities
- Joint laxity
- Dental abnormalities
 - Soft enamel
 - Premature tooth eruption
- 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
 - Flushing
 - Excessive sweating
 - Poor temp control
- Congenital cardiac anomalies – about 20%
- Renal anomalies
- Scoliosis – about 40%
- Seizures – about 30%
- Constipation
- Eye problems
 - Astigmatism

- Cataracts
- Myopia
- Nystagmus
- Strabismus
- Conductive hearing loss
- Thyroid disorders
- Glucose intolerance
- Hemihypertrophy (uneven limb length or body mass on one side)

1. Medical / Dietary Needs

What You Need to Know

No special diet is required for Sotos syndrome although a well-balanced diet is important. The severity of any one of the possible medical conditions varies widely between individuals. Therefore it is important to ask the parents about the medical issues in their child

School age children with Sotos may have multiple doctors and specialist visits to monitor medical conditions.

What you can do

- A yearly check-up and studies as needed should occur in the child's Medical Home.
- Be aware of any changes in behavior or mood that seem out of line with the situation and notify the parents.
- It is important to be aware of any academic changes. Contact parents when any differences are noticed.
- No special diet is required for Sotos syndrome although a well-balanced diet is important.
- Be aware, or ask parents, if the child has a medical alert bracelet.

2. Education Supports

What You Need to Know

Individuals with Sotos have a wide degree of variability in cognitive and adaptive function with the majority of individuals in the mild to moderate range of intellectual disability. Individuals often have difficulties with speech and language, particularly in expressive language and articulation. The level of intellectual impairment remains stable throughout life. Individuals with Sotos often look older than their age, therefore it is important to remember their age and treat them as such.

Most individual with Sotos have a delay in developmental milestones. This includes delays in walking and talking. Clumsiness, lack of coordination, low muscle tone (hypotonia) and lax joints are common. Physical and occupational therapy may help gross motor and fine motor skills. Activities that require good hand eye control (i.e. writing, drawing, and painting) may present difficulties.

Mathematics often causes difficulties for children with Sotos, as they are unable to understand abstract concepts and have problems with spatial awareness, sequencing, proportions, and time.

What you can do

Educational supports:

- Help with organization.
- Present information in concrete manner.
- Repetition will help children remember basic facts.
- Verbal explanations are often more effective than visual demonstrations, diagrams and models.
- Provide information in brief, organized, and specific manner.
- Simplify information presented on worksheets.

- Tasks like copying letters and figures takes longer.
- Geometric designs may be difficult.
- Written homework may need to be modified.
- Consider having child work with an occupational therapist.
- A step-by-step approach is most effective paying attention to the student's abilities.
- They may have difficulty remembering locations of objects in space (i.e. number lines may be ineffective).
- Math fractions, geometric shapes and formulas are hard.
- Individuals need help lining up numbers to do calculations.

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- Poor executive function can lead to difficulties in calculations requiring more than one step.
- Word problems may be challenging.

To help develop speech and language:

- Ensure the child has a reliable way to communicate
- Speech and language therapy and supports that is individualized to the child may be helpful

Physical Therapy

- Develop gross motor abilities
- Improve strength
- Assess modifications and adaptive equipment for school and home

Occupational Therapy

- Fine motor
- Strength
- Dexterity

Psychological supports

- As needed, to help with emotional support

3. Behavioral and Sensory Support

What you need to know

Children with Sotos may appear taller, heavier, and have larger heads than their peers. They often have a wide range of behavioral problems that are common at all ages. Peer group relationships and social situations may be challenging due to large size, lack of awareness of social cues, and emotional immaturity.

Behavioral Problems may occur:

- May have a secondary diagnosis of autism spectrum disorder
- Phobias
- Aggression
- Anxiety
 - New situations
 - Separation anxiety

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- Hyperactivity
- Irritability

What you can do

- Consider treatments
 - Behavioral supports
 - Counseling
 - Medication
- Be proactive with behavioral supports. Discuss involvement of behavioral or mental health professionals, or medications with the parents as needed.
 - Firm directions, rules, and clear expectations are helpful.
 - They may benefit from positive behavioral interventions.
 - They may need a functional behavioral assessment to identify causes/triggers of behaviors and develop a plan for supporting changes
- Many children have difficulty regulating emotions and behavior. This is especially true when handling unplanned changes.
 - Talk through expected changes.
 - They usually thrive with consistency and routine. They can be easily upset with disruption.
 - Prepare for any change in schedule.
 - Provide a safe area to share emotions.
 - Teach and model use of words and/or pictures in sharing emotions.
 - Stories may help with transitions
 - Teach, emphasize, and reinforce behaviors you want to see.
 - Make sure they have an effective communication system.
- Support social skills development
 - Misconceptions of abilities can cause insecurity and anxiety in social situations
 - Make sure teaching strategies being used are appropriate for children who are already socially engaged.
- Provide social cues and coaching.
 - Provide information to and discuss differences with the child's peers.
 - Help develop confidence and focus on strengths.
 - Provide positive reinforcement.
 - Teach appropriate social behaviors/skills (e.g., how to ask a friend to play).
 - Teach how to recognize facial expressions, body language, and moods in others.
 - Teach how to regulate own body – sensory strategies may be helpful.

- Foster friendships with neighbors, schoolmates and relatives so they will have friends and companions for their entire life.
- Teach them how to read body language, and moods in others.

4. Physical Activity, Trips, Events

What you need to know

Physical Education

- Individuals may be clumsy or have an awkward gait
- Temperature control and/or excessive sweating maybe an issue for the child
- If you live in New England (USA) and qualify, Northeast Passage offers Therapeutic Recreation and Adaptive Sports programming (www.nepassage.org).

Field trips

- Any change in routine may produce anxiety, fears, and/or worry.
 - Offer anticipatory guidance and preparation to prepare for a change in routine such as a field trip.
 - Create a picture story about the upcoming event. The child can rehearse it alone or with others.
- If a child has any sensory, hearing or vision issues, he/she may need preferred seating.

What You Can Do

Physical Education

- Exercise is important and should be encouraged, especially those exercises that build muscle mass and motor skills.
- Adaptive physical education may be appropriate
- Coordination may be a problem and make individuals a target for teasing.
- Practice can help.
- Support the child emotionally
- Encourage hydration and allow frequent water breaks
 - Provide rest opportunities as appropriate.

Field Trips

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- Offer anticipatory guidance and preparation to prepare for a change in routine such as a field trip.
- Offer supports as needed for vision and hearing issues.
- Use social stories and pictures to help them understand the change.
 - Encourage use of their communication system to help them process concerns.

5. School Absences and Fatigue

What you need to know

- Sleep disturbances may be common
- Children with Sotos may be absent due to illness and/or medical issues.

What you can do

- Help to make transitions in and out of school as seamless as possible
- Children with Sotos may be tired and require rest opportunities or breaks in their day

6. Emergency Planning

What you need to know

- Develop an emergency plan if necessary, depending on the needs of individual children.

7. Resources

Sotos Syndrome Support Group

The parent support group with information for parents and health professionals
<http://sotosyndrome.org/>

Genetics Home Reference

Consumer-friendly information about human genetics from the U.S. National Library of Medicine
<https://ghr.nlm.nih.gov/condition/sotos-syndrome>

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Behavioral and emotion characteristics in Children with Sotos syndrome and learning disabilities

An article that discusses psychological counseling and educational supports for individuals with Sotos syndrome.

<http://onlinelibrary.wiley.com/doi/10.1111/j.1469-8749.2003.tb00926.x/pdf>

Classroom Accommodations for Students with Visual Issues

Classroom Accommodations for Students with Visual Issues is a resource from Boulder Valley Vision Therapy, P.C. (<https://www.bouldervt.com/>), in Boulder, Colorado. It describes a variety of variety of issues and offers strategies.

<https://www.bouldervt.com/wp-content/uploads/sites/478/2015/12/227-Course-Handout-CCIRA-2016.pdf>

GeneReviews

learn more about the genetics of Sotos syndrome

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

National Organization of Rare Disorders

<https://rarediseases.org/rare-diseases/sotos-syndrome/>

Meet a Child with Sotos Syndrome – *Getting the Scoop on Koop*



Growing up in a family of 4, Kooper has many nicknames including, Scoop Mc Lovin, Big Cat and Koop! At age 4, Koop is a cuddly, touchy-feely boy who is growing very tall quite quickly. “He is almost as tall as I am!” says his mother, Rebecca.

Kooper was born about 5 weeks prematurely and had surgery to place a feeding tube at 10 days of age. Due in part to his small jaw, feeding and swallowing were difficult and he began early intervention at 6 months after Sotos syndrome was diagnosed. By two years of age, he was talking and his mother remembers him saying to a cherished pediatrician, “You took my teeth!” (He had his teeth removed because the enamel didn’t grow well and they decayed.) He was also diagnosed at two years of age with autism. His ability to speak has dropped off but he

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does nod and he understands everything according to his mother. When he recently saw that same pediatrician, now retired, he climbed onto his lap, stroked his face and gave everyone a chance to see the depth of his care and feelings.

Koop likes women and is very friendly, but on his own terms. He likes to pull people to get them to do something he wants to have done, like change the channels. He nods his head for yes and gets upset when he means no, and uses facial expressions to express his feelings. Since starting on a new medication, he is starting to babble again and mouth words.

Koop loves school and, although he likes watching and being near the other children, he is tentative about playing with them. In his preschool, he has therapy as part of the program. He has many sensory issues and his low tone makes some fine motor tasks challenging so he is working on developing those skills. He is also working with an iPad and loves to read stories and listen to songs. He also has a jogger/stroller to use when he needs it, especially if he needs it for sensory reasons.



Rebecca says they treat him like everyone else in the family. “We expect him to be part of dinner time and his brothers and sister let him know if he needs to shape up.” She notes that he is so much fun and loves to break into dance when he hears music. “He loves to make people laugh!”

Rebecca is finding it helpful to connect with other parents and has friends via a list serve that “took me under their wing” when Koop was diagnosed. These friends, whom she might never meet in person, live in England. Locally, she has become friendly with another mother who lives about 20 minutes away.

Rebecca’s advice to parents:

- Trust your instincts. She had to work through her fears about letting him go to school and is very glad she did. However, she stayed with him in class for several sessions until she felt they knew him well. She was nervous that the staff wouldn’t know him because he was non-verbal. And she was also fearful that, because of some of his behaviors and his larger size, he might scare the other children.

Rebecca’s advice to school staff:

- Never be afraid to ask a question to a parent. She knows teachers and therapists have so much knowledge, but “we all have something to learn every day.”

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