Sotos syndrome

Child Growth Foundation

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ABOUT US

The Child Growth Foundation (CGF) is a UK charity, focused on the support, understanding and management of rare growth conditions and concerns.

We work to improve the lives of everyone affected by a growth condition. We support children, young people, adults and their families – whether or not they have a diagnosis.

About this booklet

This booklet is for anyone affected by Sotos syndrome and for healthcare professionals, to explain in simple terms what the condition is and how it can affect someone.

WHAT IS SOTOS SYNDROME?

Sotos syndrome is a rare genetic condition that was first described in 1964 by Dr Juan Sotos. It is a variable condition, which means that people can be affected in different ways and to different degrees.

Sotos syndrome is estimated to affect between 1 in 14,000 and 1 in 15,000 births. However, it is possible that, as growing awareness has led to more diagnoses, we may learn that Sotos syndrome is more frequent than this..

Sotos syndrome is primarily caused by mutations in the *NSD1* gene, responsible for regulating growth. Variants within the *NSD1* gene or deletions affecting it are identified in approximately 90% of individuals with Sotos syndrome. Genetic testing can be done to ensure accurate diagnosis.

Alex

Sotos syndrome is often sporadic (de novo). This is where the *NSD1* variant has arisen for the first time and the affected individual is the first person in the family to receive a diagnosis of Sotos syndrome.

If inherited from a parent, Sotos syndrome follows an autosomal dominant pattern of inheritance, which means that an adult affected with Sotos syndrome has a 50% chance of passing on the *NSD1* gene variant and therefore Sotos syndrome, to their children.

Parents of a child with Sotos syndrome, who themselves are not affected, have a low chance of having another child with Sotos syndrome due to a rare condition known as 'germline mosaicism'. This is where the gene variant is present in some of the sperm or egg cells but not present or detectable in the blood.

Due to germline mosaicism, healthcare professionals will advise that there is a 1% chance of a subsequent child inheriting the *NSD1* gene variant and being affected by Sotos syndrome.

PHYSICAL CHARACTERISTICS

Sotos syndrome can be identified from birth; either through recognising a distinct set of clinical features, by performing genetic testing, or both.

The key clinical features of Sotos syndrome, present in more than 90% of individuals, are: increased growth with tall stature and/or macrocephaly (large head); learning difficulties; and distinct facial features.

There is some clinical overlap between Sotos syndrome and other genetic syndromes, such as Weaver syndrome and Beckwith-Wiedemann syndrome, making an accurate diagnosis important.

Increased growth	Children with Sotos syndrome typically experience rapid linear growth in early childhood leading to tall stature along with a larger head circumference (macrocephaly).
Facial features	Characteristic facial traits include a prominent forehead, prominent chin, flat nasal bridge, and downward slanting eyes.
Learning difficulties	A variable learning difficulty, from mild to severe, is common, with moderate learning difficulties most frequently reported. Children with Sotos syndrome often have speech delays and language difficulties.

LESS FREQUENT CHARACTERISTICS

Although the distinctive facial features and overgrowth are the usual indications of Sotos syndrome, there are other, less frequent physical characteristics that may be seen in some individuals. But remember Sotos syndrome is a variable condition, and individuals can be affected in different ways and to different degrees.

Cardiac	There have been reports of individuals with Sotos syndrome having congenital heart conditions (a heart condition from birth).
Dental	Dental issues such as delayed eruption of teeth or malocclusion (misalignment of teeth) may be present.
Feeding difficulties	Infants with Sotos syndrome may experience feeding difficulties due to gastro-oesophageal reflux, muscle-tone issues, high palate, or coordination problems.
Hands & Feet	Individuals may have larger hands and feet. They may also experience joint hypermobility and flat feet.
Hypotonia	Some individuals may experience low muscle tone, leading to muscle weakness, which can affect motor skills and coordination.
Motor coordination challenges	Some individuals may experience difficulties with motor coordination and fine motor skills.
Neurological	In a minority of cases, individuals with Sotos syndrome may experience afebrile seizures.
Renal	In rare instances, individuals may have renal anomalies, such as structural abnormalities or issues with kidney function.
Skeletal	Advanced bone age can be present and scoliosis (curvature of the spine) can sometimes develop, either from birth or over time.

TREATMENT AND MANAGEMENT

The support and treatment for someone diagnosed with Sotos syndrome is dependent on which features of the condition are present – the frequency and severity of each characteristic can vary greatly. Often a number of healthcare professionals may be involved, as shown in Figure 1 below.

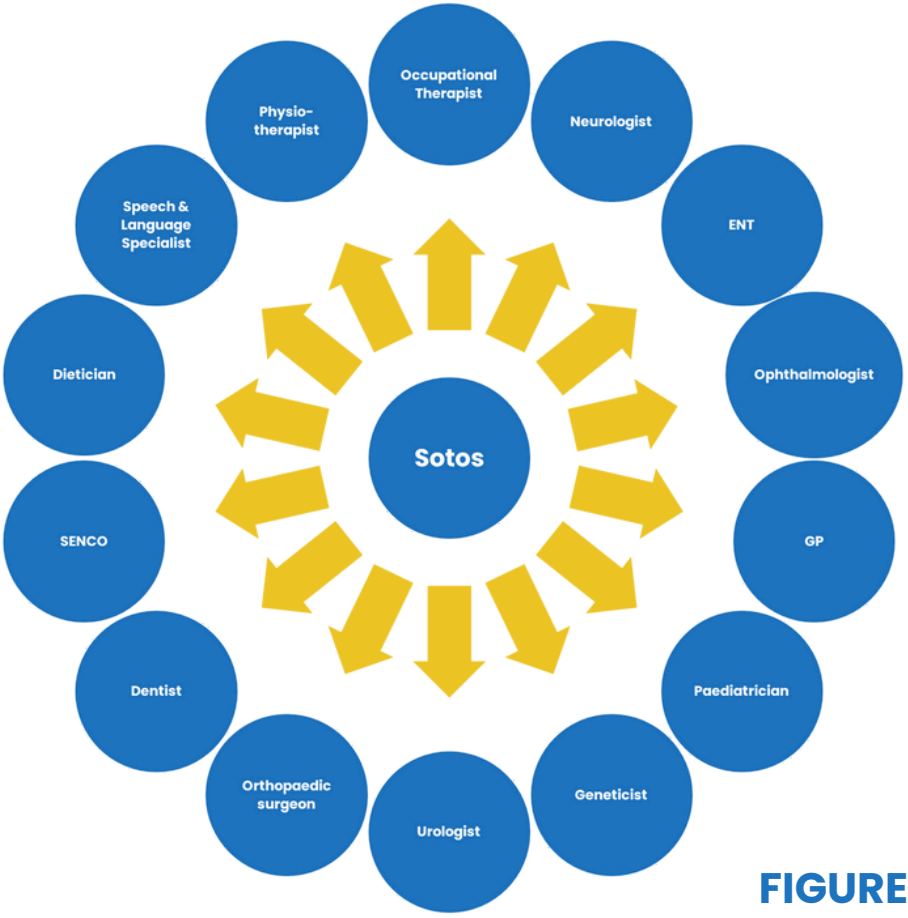


FIGURE 1

Children with Sotos syndrome can have a high pain threshold so when assessing any medical issues, it is important to remember that they might not display the same level of discomfort as other children.

It may be your child's paediatrician or geneticist that, alongside you, implements a plan for the investigation and management of any of the medical problems described. However, evidence suggests that baseline investigations into kidneys (renal ultrasound scan) and heart (echocardiogram) should be undertaken at diagnosis to establish if there are any complications that require further treatment.

Management and treatment into adulthood very much depends on how the individual is affected by Sotos syndrome, but all of the different features of the condition should be considered and managed as needed.

INFANCY

Many babies with Sotos syndrome are long at birth (birth length>2 standard deviations above the mean). The birth weight may not be proportionately increased and so babies with Sotos syndrome are usually long and thin.

Feeding problems could occur due to the challenges with coordination and low muscle tone. This may be seen in infancy, with babies struggling to coordinate and gather the strength to feed from the bottle or breast. When weaning, foods that require chewing could be problematic.

CHILDHOOD

The level of medical input in childhood depends on any identified health issues. Regular medical review and monitoring will be required regardless and care is usually overseen by a General Paediatrician.

Any learning difficulties that arise could require support and input from the special educational needs coordinator (SENCo) in the school setting and an education and health care plan (EHCP) might be needed. Assessment before starting school may be necessary so that the right level of support can be planned as necessary.

PUBERTY

Before puberty, most children with Sotos syndrome have height and head circumference bigger than average (greater than 2 standard deviations above the mean). The progression of puberty is usually in line with peers.

After puberty, the final height of individuals with Sotos syndrome can vary. Some young adults heights will be within the expected range, others may be slightly taller than their target height range, which is based on parental heights.

Macrocephaly (larger head) is typical in both children and adults with Sotos syndrome.

LEARNING DISABILITY

Learning disabilities in Sotos syndrome can range from mild to severe, or non-existent.

A learning disability can be varied but tends to mean that those affected by them could take longer to learn and may need support to develop new skills, understand complicated information or interact with other people.

Research suggests that individuals with Sotos have much better verbal skills than non-verbal reasoning, and they have strengths in visuospatial memory, which means they may be more likely to remember information if it is presented as diagrams or pictures.

Individuals with Sotos syndrome may have relative strengths in verbal skills, with good vocabulary and a good understanding of language, but this may not always be apparent during social interactions, as communicating appropriately may be a struggle. For instance, they might have difficulties in using words in the right context or order.

AUTISM

Like a learning disability, autism is a lifelong condition. Autism is understood as a spectrum because each autistic person has a unique combination of characteristics. Individuals with Sotos syndrome may also have a diagnosis of autism.

There are some common features of autism which might affect the way a person interacts with others in a social situation, is able to communicate with others and experiences the world around them. However, the signs of autism will be different for everyone, and affect different people in different ways, in different environments and to different degrees.

Children with Sotos syndrome tend to be very sociable, but often prefer the company of adults rather than their peers. They can have challenges in understanding social norms and how to interact appropriately within certain social situations and struggle to pick up on what others are thinking or feeling. They may enjoy repetition and discussing the same topics many times.

Attending a special school may be of benefit to some children with Sotos syndrome, while others might attend mainstream schools.

There can be varying degrees of academic ability and while some young people may achieve independent living and employment, others may need support and assistance.

More information on behavioural differences and neurodiversity in Sotos syndrome can be found in resources on our website at: childgrowthfoundation.org/sotos or by scanning the QR code, right.

TRANSITION INTO ADULTHOOD

Transition into adulthood can be a worrying time for everyone and a young person with Sotos syndrome may need health and social care input into adulthood. Therefore, transition planning should start by school year 9, with full involvement of families/carers and young people but led by health and social care practitioners. If your child is 10-11 years old, speak to your lead healthcare professional and ask what transition will look like for your child and how this process works.

NICE (National Institute for Health and Care Excellence) has published a quality standard for transition from children's to adults' services. This document covers the period before, during and after a young person moves from children's to adults' health and/or social care services. You may want to become familiar with this document to help you understand what professionals should be working to. The standard and statements focus on planning transitions and having a coordinated approach across all services involved in providing care to the young person.

You can read more about this here: nice.org.uk/guidance/qs140 or by scanning the QR code, right.

Transition can be a difficult time for young people and their families or carers because it is a lengthy process and involves various practitioners and sometimes, several services with different timescales. A single point of contact – preferably a person that the young person knows and trusts – can coordinate care and signpost for appropriate support.

SUPPORT AVAILABLE

It can be helpful for families to explore local and national resources, including Disability Living Allowance (DLA), that can provide additional support, including respite care and financial assistance.

Social services can often help with guidance on the types of benefits and services that are available and would be beneficial to access locally.

Sotos syndrome often requires a multidisciplinary approach for diagnosis, and ongoing management. This can include developing long-term plans for education, healthcare and social inclusion. Genetic counselling should be made available to all families and individuals affected by Sotos syndrome.

Access to Work (DWP) funding may be available to support young people in the workplace. The application process is relatively straight forward but carers/parents will be expected to source support independently.

Connecting with other families facing similar challenges can provide emotional support and enable practical advice to be shared. Counselling and support groups can also assist families in coping with the emotional challenges associated with Sotos syndrome.

CGF RESOURCES

Facebook groups

The CGF maintains and oversees a number of closed Facebook groups covering the conditions we support. These offer peer support, which can be vital in helping those living with rare conditions.

Sotos group:
facebook.com/groups/sotoscgf or scan the black QR code, right.

Overgrowth group:
facebook.com/groups/overgrowthcgf or scan the yellow QR code, right.

CGF's Sotos webpage

Our website has a dedicated Sotos syndrome page at: childgrowthfoundation.org/sotos or via the blue QR code, right. This page has specialist information and resources, plus personal stories from families affected by Sotos and our 'Spotlight on' Sotos syndrome feature.

MY SOTOS STORY

The CGF has written and published My Sotos Story – a colourful picture book created especially for primary-aged children with Sotos Syndrome, their peers, families, carers and teachers.

With bright, quirky illustrations and fun rhymes, it's the ideal way to help anyone understand Sotos syndrome – what it is, how it happens, and what it's like to live with.

"This story is beautiful. I thought it would be nice to send this book into preschool for my son. Also going to read it to my daughter as she knows her brother is 'a hard-work baby' as she says. I think it will really help her and others gain a little more understanding." - reader feedback

"My 8-year-old son absolutely loves it! Read it all in one go and took it to school to share! It's so amazing that he can relate to it all." - reader feedback

Order now from the CGF shop at: childgrowthfoundation.org/my-sotos-story or by scanning the QR code below. Worldwide shipping is available.

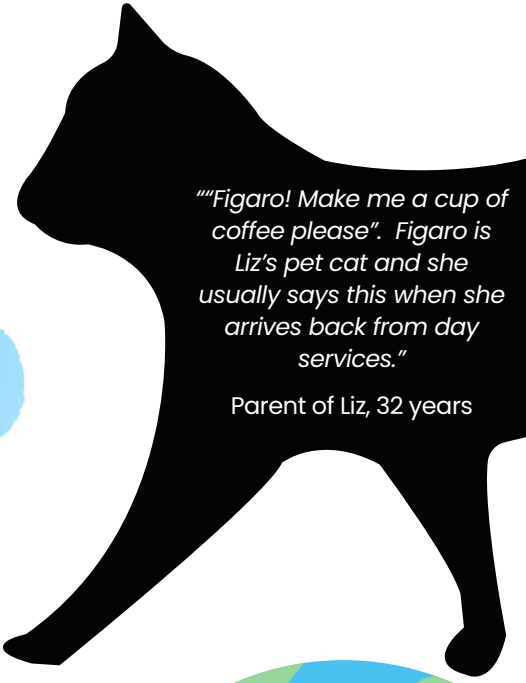
QUOTES FROM THE SOTOS COMMUNITY

"Getting a clear diagnosis has changed everything — I can finally let go of the worry and just enjoy my little boy as he is. Happy, lovely and huge in heart as well as stature!"

Parent of Jack,
4 years

"Living with a rare condition like Sotos can be frustrating, and advocating for a child with Sotos really teaches you how to fight, but there are also so many moments of joy and discovery. I've loved learning to see the world through Ivo's eyes, and I still marvel at the way his memory works every day."

Parent of Ivo, 12 years



""Figaro! Make me a cup of coffee please". Figaro is Liz's pet cat and she usually says this when she arrives back from day services."

Parent of Liz, 32 years

"Alex is pure sunshine with a twist - showing that life with Sotos Syndrome can be full of laughter, wonder, and unstoppable hope. He doesn't just grow taller; he grows hearts bigger everywhere he goes."

Parent of Alex, 17 years

"Sotos syndrome makes me taller than my peers, but that just helps me to see my own world better!"

Molly (via her Mum), 3 years

GLOSSARY OF TERMS

Afebrile seizures - A seizure which happens without an elevated body temperature/fever.

Autosomal dominant pattern of inheritance - Autosomal means that the gene in question is located on one of the numbered, or non-sex, chromosomes. Dominant means that a single copy of the mutated gene (from one parent) is enough to cause the condition.

Bone age - Is determining the age of your bones by taking an x-ray of your hand/wrist which is then compared to your actual age.

Beckwith-Wiedemann syndrome - An overgrowth syndrome, present from birth with variable characteristics, caused by abnormalities at chromosome 11p15.

Cardiac echogram - An ultrasound scan of the heart.

DWP - Abbreviation for the Department for Work & Pensions.

Gastro-oesophageal reflux - When food and drink is swallowed normally, but some of the mixture of food, drink and acid travels back up the food pipe, instead of proceeding to the stomach.

Gene deletion - Loss of genetic material from the genome (a genome is a complete set of genetic material) is known as a deletion.

Gene duplication - Where a section of the genome has been copied.

Gene mutations - An alteration in the genetic code found in DNA (The molecule that encodes genetic information) that changes the specific instructions of the gene.

Germline mosaicism - When some of the sperm cells in the testes or some of the egg cells in the ovaries carry a pathogenic variant that is not found in other cells of the body.

Hypotonia - Low level of muscle tone.

Hypoglycaemia - Low levels of glucose (sugar) in the bloodstream.

GLOSSARY OF TERMS

Jaundice - A medical condition arising from excess of the pigment bilirubin, causing yellowish colouring of the skin and whites of the eyes.

Joint Hypermobility - Where some or all of a person's joints having an unusually large range of movement.

Macrocephaly - A larger head size/circumference than expected.

Multidisciplinary - Combining and involving several professionals, with different specialisms.

Renal - Relating to the renal system in the body, also known as the urinary system and is responsible for the production, storage, and elimination of urine, which contains waste products filtered from the blood.

Scoliosis - Where that the spine is curved abnormally when viewed from the front or the back.

Standard Deviation Score (SDS) - A measure of the amount of variation or spread of a set of values around the average. SDSs on growth charts assess whether a measurement is normal by comparing it with the range of measurements for other children of the same age and sex.

Urinalysis - A test of your urine that can detect a wide range of disorders, such as infections, kidney disease and diabetes.

USS/Ultrasound Scan - A type of scan which uses sound waves to take pictures of inside the body.

Weavers syndrome - An overgrowth condition caused by a mutation in the EZH2 gene.

Vesico-ureteric reflux - The abnormal flow of urine from the bladder back into the ureters.

Visuo-spatial memory - A cognitive ability that allows individuals to recall and manipulate visual information and understand spatial relationships between objects.

FURTHER SUPPORT FROM THE CGF

"The CGF are unique as they support people with a wide range of growth-affecting conditions. Their team, supported by Medical Advisors, encompass extensive expertise and importantly includes those with lived experience. The CGF is a very friendly and approachable charity that provides numerous opportunities for patients and families to connect with each other and Medical Advisors."

-Professor Helen Storr,
Professor and Honorary Consultant in Paediatric Endocrinology

Our charity supports hundreds of children, young people, adults and families each year through our in-person and virtual events, and we help thousands of people in the UK and beyond with our online information, guidance and support.

We provide peer support including our Virtual Cuppa & Chats run by our nurses and involving parents and carers, alongside closed Facebook groups, and also opportunities to meet other families in the child growth community through our in person Meet Ups and convention - and much more! See our full range of support at: childgrowthfoundation.org

"I just wanted to say a big thank you. The information was so helpful. It was a comfort to talk to you and the other parents, I feel less alone with what we are going through."

-Virtual Cuppa & Chat attendee feedback

GET IN TOUCH

If you have any questions regarding the information contained in this booklet, or any other queries, please get in touch with our friendly team.

How to contact our nurse led Support Line:

-Call our dedicated Support Line number: 020 8995 0257
-Complete our Support Line online contact form: childgrowthfoundation.org/supportline
-Email us at: support@childgrowthfoundation.org
-Book a slot at: childgrowthfoundation.org/bookings
-Scan the QR code, below:

By contacting the Child Growth Foundation Support Line, you are providing consent for us to collect, process and store your data to provide you with the information or services you are contacting us about, in line with our Support Line Privacy Statement childgrowthfoundation.org/supportlineprivacy and our charity's Privacy Policy at: childgrowthfoundation.org/privacy

GET INVOLVED

The CGF is an independent charity that relies entirely on the generosity of individuals, groups and organisations to continue our work.

Make a donation

Become a member

Fundraise

Volunteer

Help us continue making a difference wherever growth is a concern. Find out more on how to get involved at: childgrowthfoundation.org/get-involved

Acknowledgements

With thanks to:

- Professor Kate Tatton-Brown, Consultant Clinical Geneticist.
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Disclaimer

We have taken every care to ensure the accuracy of the information contained in this booklet. The information enclosed should not be used as a substitute for the advice from a clinician, GP or other healthcare professional.

Feedback

Your feedback helps us to ensure we are delivering information to the highest standard. If you have any comments or suggestions, please contact us at info@childgrowthfoundation.org or on 020 8798 2139.

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