

# WHAT IS SOTOS SYNDROME?



**Child  
Growth  
Foundation**

Sotos Syndrome is a genetic overgrowth condition that was first described in 1964 by Dr Juan Sotos. In 2002, a team of Japanese scientists discovered that it is caused by a change in the gene known as NSD1.

## Frequency

Approximately

**1 IN 14,000  
PEOPLE**

both male and female.

## What are the signs?

Sotos Syndrome is a variable condition – different people can be affected in different ways, and to different degrees.

### COMMON THINGS TO LOOK OUT FOR:

**Rapid growth** in childhood: tall stature, large head, long limbs and large hands and feet.

**Distinctive facial features**, including a broad, prominent forehead, flat nasal bridge, downward-slanting eyes with a wide distance in between, and a prominent chin.

**Learning difficulties**, ranging from mild to moderate, with associated cognitive challenges affecting memory, verbal and social skills.

CLINICALLY

GENETICALLY

## How is Sotos Syndrome diagnosed?

Sotos Syndrome can be identified from birth:

**Clinically** – by recognising distinctive physical signs  
**Genetically** – with a simple genetic test

Often, getting a diagnosis will involve both.

## Less frequent indications in babies and children

- Neonatal jaundice
- Recurrent ear, respiratory, and/or urinary infections
- Impaired motor skills or poor muscle tone (hypotonia)
- Epilepsy
- Curvature of the spine (scoliosis)
- Heart or kidney problems
- Behavioural difficulties



## How do you get it?

Sotos Syndrome is most often the result of a random (de novo) mutation soon after the egg is fertilised. This means that non-Sotos parents who have a child with Sotos Syndrome are no more likely to have future children with the condition than anyone else.

**50%**

A person with Sotos Syndrome has a 50% chance of passing it on to their children.

NEED MORE INFORMATION, ADVICE OR SUPPORT?

Call 020 8995 0257

Visit [www.childgrowthfoundation.org](http://www.childgrowthfoundation.org)