WHAT IS SOTOS SYNDROME?

Child Growth Foundation

Sotos Syndrome is a genetic overgrowth condition that was first decribed 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?

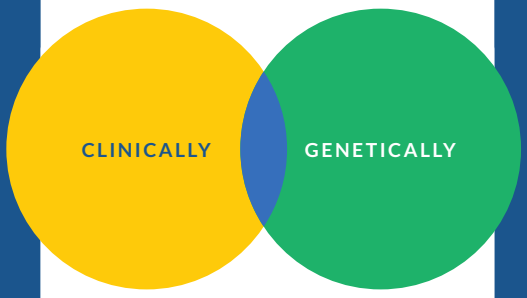
Sotos Syndrome is a variable condition – different people can be affected in different ways, and to different degrees.

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.



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
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