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My46 Trait Profile

Sotos syndrome

Other Names: Cerebral Gigantism

Sotos syndrome is a rare condition characterized by overgrowth, characteristic facial features, and learning disability. It is caused by mutations in the NSD1 gene, which is thought to play a role in regulating growth and development.

Characteristics of Sotos syndrome

Overgrowth in Sotos syndrome is typically noted at birth and persists through early childhood. Individuals with Sotos syndrome are likely to be taller, heavier, and have a larger head size than their peers during this time. While many individuals eventually reach a normal adult height, head circumference may remain above average throughout one's life. Rapid growth along with decreased muscle tone can also lead to coordination problems and slower achievement of motor milestones; however, these issues may improve with time as the child's growth rate slows.

Common facial features seen in individuals with Sotos syndrome include: widely spaced eyes that are slightly down-slanting, prominent forehead, receding hairline, rosy cheeks, and a narrow, pointed chin. Large hands and feet and an advanced bone age (closer to achieving adult size/height than would be expected for their age) are also common.

Many individuals with Sotos syndrome have learning disabilities, which can be related to delayed motor development, intellectual impairment, and/or behavioral problems. Speech problems, especially with expressive language, are common. Intellectual impairments vary widely; some individuals can succeed in mainstream schooling and live independently while others require lifelong support. Behavioral problems also vary between individuals but can include: ADHD, tantrums, phobias, irritability, and impulsivity.

Other less common features of Sotos syndrome include: scoliosis (curvature of the spine), heart defects, kidney abnormalities, hearing loss, seizures, and loose joints. Although less frequent, these issues may be more significant causes of healthcare problems in individuals with Sotos syndrome.

Diagnosis/Testing

Most individuals with Sotos syndrome have a change or mutation within one of the copies of their NSD1 gene, which is located on chromosome 5. However, Japanese individuals with Sotos syndrome are more likely to have a deletion (missing piece) or duplication (extra piece) of a part of chromosome 5 that includes the NSD1 gene. This gene is thought to make a protein that regulates the expression of other genes involved in growth and development. Clinical genetic testing for either the mutation within the gene or deletion/duplication of genetic material yields a positive result in about 90% of individuals with a diagnosis of Sotos syndrome.

Management/Surveillance

Management of Sotos syndrome can include physical therapy, occupational therapy, speech therapy, and educational/behavioral interventions as necessary.

Due to the wide range of symptoms, evaluations for some of the less common features (such as heart and kidney abnormalities or hearing loss) may help guide medical management decision-making.

Mode of inheritance

Sotos syndrome is inherited in an autosomal dominant pattern. This means that inheriting one NSD1 mutation (or having only one copy of the NSD1 gene) is enough for an individual to be affected and show signs of Sotos syndrome. Most cases are due to new (de novo) mutations, although the mutation could also be inherited from an affected parent.

Risk to family members

The risk to family members depends on whether the genetic mutation was de novo or inherited. If an individual is the first affected person in the family, then the risk to other siblings and family members is very low. Individuals with Sotos syndrome have a 50% chance to pass on their non-functioning NSD1 gene and have a child who also has Sotos syndrome.

Special considerations

None

Resources

Sotos Syndrome Support Association
http://sotossyndrome.org/
Genetics Home Reference: Sotos syndrome
http://ghr.nlm.nih.gov/condition/sotos-syndrome
NINDS Sotos Syndrome Information Page
http://www.ninds.nih.gov/disorders/sotos/sotos.htm

References

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Updated: mm/yyyy **Edited by:** Karin Dent, MS, LCGC, Seema Jamal, MSc, LCGC