International Journal of Advanced Science and Research
ISSN: 2455-4227
Impact Factor: RJIF 5.12
www.allsciencejournal.com
Volume 3; Issue 4; July 2018; Page No. 26-27

Sotos syndrome also known as cerebral gigantism or sotos-dodge syndrome

Suveesh US
Assistant Professor, Department of Pediatric Nursing, SCPM College of Nursing and Paramedical Sciences, Gonda, Uttar Pradesh, India

Abstract
Sotos syndrome is a genetic disorder occurs due to NSD1 gene mutation. It is an autosomal dominant pattern of inheritance disorder. The child has an abnormal increased in length, height, weight, and head circumference at birth. As the child grows older, the height and weight normalized, but the head remains large. The bone structure is advance for the age of the patient. There's no cure or one specific treatment for Sotos syndrome. Instead, treatment focuses on treating the symptoms.

Keywords: sotos syndrome, SSSA, NSD1 gene, APC2 gene, cerebral gigantism

Introduction
Sotos syndrome is a childhood growth disorder. Professor Juan Sotos, in 1964 described five unusually tall children in the United States. However the exact description given in 2004 by a group of Japanese scientists they linked Sotos syndrome to mutations in a gene called NSD1 (Nuclear SET domain 1).

Definition
Sotos syndrome also known as cerebral gigantism, is a rare genetic disorder caused by Mutations in the NSD1 gene. Characterized by excessive physical growth during childhood.

Incidence
- Sotos syndrome is reported to occur in 1 in 10,000 to 14,000 newborns world wide.
- In Sweden it is estimated that between five and ten children are born annually with the condition and approximately one hundred people have been diagnosed with the disease.
- According to SSSA (Sotos Syndrome Support Association), in India the first case reported in Chandigarh (2004) and then in Tamilnadu, INDIA (2007).

Etiology
- Mutations in the NSD1 gene are the primary cause of Sotos syndrome (The NSD1 protein controls the activity of genes involved in normal growth and development).
- Mutation in the APC2 (adenomatous polyposis coli 2) gene. The APC2 gene is expressed in the nervous system, and is a downstream gene of NSD1 means mutations in the NSD1 gene affects the APC2 gene and results in the neural abnormalities.
- Consanguineous marriages.
- Sotos syndrome has an autosomal dominant pattern of inheritance (Dominant genetic disorders occur when only a single copy of an abnormal variant of a gene is necessary to cause a particular disease)
- The abnormal variant gene can inherited from parents or due to gene mutation (changing) the risk of transmitting abnormal variant gene from an affected parent to an offspring is 50% for each pregnancy.
- The risk is the same for males and females.

Pathophysiology
Sotos syndrome is a genetic abnormality because the NSD1 gene mutation playing main role in this syndrome. The NSD1 gene carries the instruction for protein creation, which acts as a histone methyltransferase, is a type of enzyme that responsible for changing histones (structural protein). The histone attaches to the DNA and is responsible in determining the shape of the chromosomes. Through the process called methylation, a molecule methyl group is added to the histones to form histone methyltransferases, which regulate the activity of a particular type of genes. NSD1 protein that controls the gene's activity, especially the ones that have to do with growth and development.

Some genetic changes that involve NSD1 gene hinder a portion of the gene to release functional protein. The growth and development of the genes are disrupted if the amount of NSD1 protein is not sufficient, which could lead to Sotos syndrome.

Signs and symptoms
- Newborns with Sotos syndrome often reveals large head circumference (`14.5″ versus average 13.5″), body length (23″ versus average 20″) and birth weight (9 lbs. versus 7.5 lbs.).
- Infants and Toddlers have late head control, and poor muscle tone impairs rolling, sitting, crawling, standing and walking. Fine motor activities – grasping, playing with objects, cooing and babbling, even facial expressions – are also delayed. Head size may grow at an alarming rate (e.g., at 6 months, the head size may be that of an 18-month-old).
- In young children, seizures, low muscle tone, advanced bone age, prominent forehead, learning disabilities, heart defects, scoliosis and dolichocephalism are very common.

- Other manifestations are Nystagmus, strabismus, Behavioral disturbance, Poor fine motor control, Apparent hypertelorism, Prominent, pointed chin, Excessive growth in childhood, Macrocrania, Disproportionately large hands and feet

Diagnostic evaluation

There is no definite laboratory procedure that will solely confirm the diagnosis of Sotos syndrome. It can be diagnosed with various investigations that include:

- **Growth:** The child has an abnormal increased in length, height, weight, and head circumference at birth. As the child grows older, the height and weight normalized, but the head remains large.
- **Bone age:** The bone structure is advance for the age of the patient as seen in the x-ray.
- **Appearance:** The patient's head is large, forehead is high, the shape of the face is elongated, and the chin is long. The space in between the eyes is wide (hypertelorism) and the eyes tend to slant downward.
- **Development:** There is developmental delay, both in motor and mental skills. Behavioural problem is common as well.
- **MRI:** showing normal brain parenchyma and large sphenoid, frontal and ethmoidal sinuses and presence of large ventricles, midline anomalies including agenesis or hypoplasia of corpus callosum)
- 2D-echocardiogram revealed floppy mitral valve with mild prolapse.
- Doppler study showed trivial mitral regurgitation.
- At the time of diagnosis it is important that the family is offered genetic counseling. For carrier prenatal diagnoses, as well as pre-implantation genetic diagnosis (PGD) in association with IVF (in vitro fertilization) are important.

Management

Treatment for Sotos syndrome is based upon the symptoms the person is experiencing; there is no standard course of treatment for it. Treatment options include:

- Physical and occupational therapies play an important role in assisting a child with Sotos syndrome
- speech therapy
- counseling
- medications to manage ADHD, irritability, or aggressiveness
- hearing aids for hearing loss
- glasses to correct vision problems

Through these forms of therapy, the child has the ability to practice balance, movement, speech and hand skills

Conclusion

Many families have stated that the parenting of a child with Sotos syndrome becomes very much easier as the years go by and the child is able to be more independent. Health also seems to improve significantly after the first few years.

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