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CEREBRAL GIGANTISM: SOTOS SYNDROME – A CASE REPORT

KEYWORDS: Sotossyndrome,autism, macrocephaly

Dr .Sunil Kumar Agarwalla	Dr.SushreeSmitaBehura	* Dr. Shubhankar Mishra
Associate professor ,Department of pediatrics ,MKCG Medical college, Berhampur ,Odisha	Junior Resident ,Department of pediatrics ,MKCG Medical college, Berhampur ,Odisha	Junior Resident , Department of pediatrics ,MKCG Medical college, Berhampur ,Odisha * Corresponding Author

ABSTRACT *Sotos syndrome/cerebral gigantism , described by Sotos ,is an overgrowth syndrome characterized by increase birthweight ,excessive growth during the first few years of life and distinctive facial features including macrocephaly ,high forehead pointed chin ,long and narrow face etc . They have mild to moderate mental retardation with developmental delay .It is generally diagnosed by clinical criteria . We present a case of sotos syndrome with all characteristic morphological features .He had no features to suggest other forms of overgrowth such as Marfan syndrome, McCune-Albright syndrome or homocystinuria.*

SUMMARY :
Sotos syndrome is a genetic syndrome causing cerebral gigantism .It is one of the causes of tall stature . The incidence is much rare and rarely reported . We found a case of sotos syndrome with all classicalfeatures .All other causes of tall stature ruled out and he was treated for seizure which was his primary presentation .This report will help clinicians to diagnose the disease in future .

INTRODUCTION:
Sotos syndrome is a rare genetic disorder characterized by a distinct facial profile ,overgrowth in childhood and learning disabilities and delayed development.It is (cerebral gigantism) is a rare form of accelerated growth in utero and in the first three to four years of life, after which it proceeds at a near normal rate. It must be differentiated from the normal variant of overgrowth which exceeds 2 SD from the mean. Other forms of pathological overgrowth due to genetic or endocrine disorders should also be ruled out . We present a case of sotos syndrome with all clinical and morphological features which will help pediatricians in diagnosing this rare entity in future .

CASE REPORT:
A four year old male child presented to us with fever for one day followed by one episode of GTCS lasting for 30 minutes and was admitted in status epilepticus .Parents gave similar history of febrile seizure lasting for five minutes before one and half years .The child had history of delayed development of speech and temper tantrums .He was born out of non consanguineous marriage, full term ,delivered vaginally in hospital with no history of birth asphyxia with a birth weight of 3.8 kg .He has two elder siblings (sisters) who are healthy .On examination he was having typical facial profile with macro dolichocephaliy ,prominent forehead ,lowest ears ,long and narrow face with small, pointed chin and flushed cheeks.

Fig-1
The child was taller with long handsand feetas compared to any other child of same age .

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Fig-2

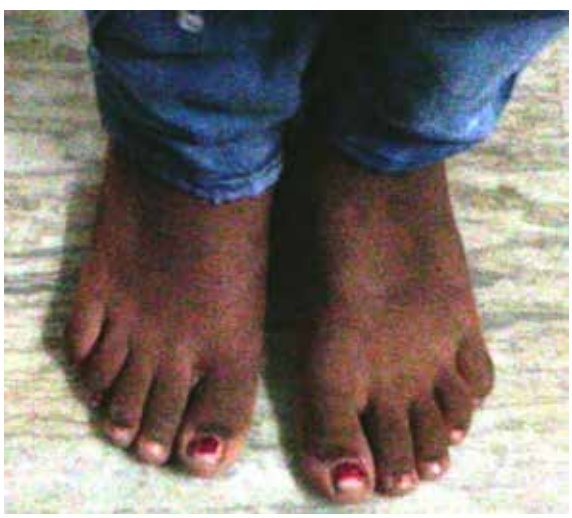

Fig-3

Height of the child was measured to be 116 cm, weight 20 kg, head circumference 55 cms, mid arm circumference 16 cm .Investigations like complete blood counts, serum electrolytes, BUN, creatinine, fasting blood sugar, and liver enzymes were normal.CBC showed features of normocytic normochromic anemia in the peripheral smear. Morning cortisol of 16.8 µg/dL was suppressed to 1.3 µg/dL after giving one mg of dexamethasone at midnight.Oral glucose tolerance test (OGTT) for growth hormone (GH) and insulin response, thyroid function tests were with in normal limits. CSF study was within normal range with two mononuclear cells only and normal protein and sugar contents .CT scan of brain showed no abnormality. Wrist x ray was also normal but showing advanced bone age .

DISCUSSION :

Sotos syndrome is a childhood overgrowth condition ,first described by sotos etal in 1964 [1] .The phenotypic features of the syndrome in more than 200 cases reviewed[1] include an average birth weight of 3.9 kg, length of 55.2 cm, growth especially rapid in the first two to three years, after which growth proceeds at a near normal rate .The four major diagnostic criteria established in 1994 by Cole and hughes[2] .They are

- Overgrowth with advanced bone age
- Macrocephaly
- Characteristic facial appearance
- Learning difficulties

It has been recently identified as a autosomal dominant syndrome with NSD 1 mutations[3] .Apart from typical facial profile these children have large hands and feet though their mid parental height is similar to normal mid parental height [4]. In these children dolichocephaly and marked frontal bossing is accentuated by frontoparental balding with HC greater than 97 centile [5] .Eyes are generally wide set ,mandible is long and narrow inferiorly or pointed .The palate is high arched and premature ,eruption of deciduous teeth observed in >50% of cases .Joint laxity is frequent with pes planus and sometimes rarely they have significant scoliosis .Most patients of sotos syndrome have nonprogressive neurologic dysfunction manifested by unusual clumsiness which improves with age. Delay in expressive language and motor development is seen during infancy followed by attainment of normal and near normal intelligence as they get older. In some cases autism is seen[7] .50% present with seizure mostly febrile convulsion [6]. ADHD may be component in some instances .Other neurological signs as nystagmus, hypotonia may be seen. Sometimes these children can experience facial plethora, kypho-scoliosis, impaired glucose tolerance test .Congenital heart defects have also been discussed along with tumors like wilms tumor, hepatocellular carcinoma and neuroblastoma[8].Sotos syndrome patients have variable mental deficiency with an IQ of 18 to 119. Poor coordination and occasional aggressiveness lead to problems of social adjustment[9]. Metacarpophalangeal pattern profile (MCPP) was determined on 34 Sotos syndrome individuals; the mean hand profile contained a major peak in the proximal phalangeal area and a smaller peak in the metacarpal area, while the distant hand bones were relatively short[10]

DIAGNOSIS AND MANAGEMENT:

The child was diagnosed to be Sotos syndrome basing all the morphological features and anthropometry.He neither had features of genetic disorders with excessive physical growth. Endocrine causes of overgrowth secondary to pituitary or adrenal hyperfunction have been ruled out by biochemical tests and CT .Hand x ray also shows advanced bone age .He was treated for seizure and given continuous prophylaxis of valproic acid .He is now on follow up.

REFERENCE

1. Sotos, J. F., Dodge, P. R., Muirhead, D., Crawford, J. D., & Talbot, N. B. (1964). Cerebral gigantism in childhood: a syndrome of excessively rapid growth with acromegalic features and a nonprogressive neurologic disorder. New England Journal of Medicine, 271(3), 109-116. | 2. Cole, T. R., & Hughes, H. E. (1990). Sotos syndrome. Journal of medical genetics, 27(9), 571-576. | 3. Kurotaki, N., Imaizumi, K., Harada, N., Masuno, M., Kondoh, T., Nagai, T., ...& Matsumoto, N. (2002). Haploinsufficiency of NSD1 causes Sotos syndrome. Nature genetics, 30(4), 365-366. | 4. Char F. Soto syndrome: from childhood to adolescence..7 C/linDysmorphol 1984;2:29-32 | 5. Cohen Jr, M. M. (1989). A comprehensive and critical assessment of overgrowth and overgrowth syndromes. In Advances in human genetics (pp. 181-303). Springer US. | 6. Finegan, J. A. (1998). Study of behavioral phenotypes: goals and methodological considerations. American journal of medical genetics, 81(2), 148-155. | 7. Morrow JD, Whitman BY, Accardo PJ. Autistic disorder in Sotos syndrome: a case report. Eur J Pediatr. 1990;149(8):567–9 | 8. Cohen, M. M. (1999). Tumors and nontumors in Sotos syndrome. American journal of medical genetics, 84(2), 173-175. | 9. Varley CK, Crnic K. Emotional, behavioral, and cognitive status of children with cerebral gigantism. J Dev BehavPediatr 1984;5:132-4. | 10. Butler MG, Dijkstra PF, Meaney FJ, Gale DD. Metacarpophalangeal pattern profile analysis in Sotos syndrome: a follow-up report on 34 subjects. Am J Med Genet 1988;29:143-7. |