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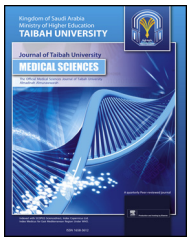

Case Report

Sotos syndrome with developmental co-ordination disorder; report of a case

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الملخص

متلازمة سوتو (العملقة المخية) هو اضطراب وراثي نادر، عادة مايتميز بضخامة الرأس وملامح جيشتالتي مميزة بالوجه. وهناك مجموعة من المشاكل السلوكية والمعرفية المرتبطة به. نقدم حالة طفل يبلغ من العمر ١١ عاما، قدم إلينا مع مجموعة من المشاكل السلوكية وصعوبة في العملية التعليمية مع اضطرابات في التنسيق الوظيفي للجسم. وكان من علامات الفحص السريري طول القامة، وضخامة الرأس مع علامات جيشتالتي مميزة بالوجه. اثبتت عمليات التقييم التعليمية وجود اضطراب في النمو التنسيقي الوظيفي للجسم. وبالقيام بخطوات إضافية للتقييم اتضح أن الطفل يعاني من اضطرابات نقص الانتباه مع فرط الحركة، مما دعانا لعلاجه بعقار الميثيل فينيديت، واستخدمنا نهج متعدد الجوانب يحتوي العلاجات الطبية والسلوكية. لم نتمكن من اثبات علاقة بين هذه المتلازمة واضطرابات النمو التنسيقي الوظيفي للجسم في التقارير المكتوبة على الرغم من أن متلازمة سوتو معروف مصاحبتها للاضطرابات السلوكية.

الكلمات المفتاحية: متلازمة سوتو; اضطرابات سلوكية; اضطرابات نقص الانتباه وفرط الحركة; اضطرابات في النمو التنسيقي الوظيفي للجسم

Abstract

Sotos syndrome (Cerebral Gigantism) a rare genetic disorder is usually characterized by macrocephaly and typical facial gestalt. There is a spectrum of behavioral and cognitive problems associated with it. We present a case of 11-year-old boy who presented with co-ordination issues, academic difficulties along with host of behavioral problems. On examination he was tall and had macrocephaly with typical facial gestalt. His educational assessment revealed the presence of developmental co-ordination disorder (DCD) which was confirmed by Bruininks Otseretsky Motor Proficiency (BOTMP) and developmental co-ordination disorder questionnaire (DCD – 2007). On further assessment he also had an element of attention deficit hyperactivity disorder (ADHD) for which he was started on methylphenidate. In view of presence of both ADHD and DCD we used a multipronged approach while treating him; both medical and behavioral therapies were used. Although Sotos syndrome is well described in literature and known to be associated with several behavioral disorders, we couldn't find any previously reported association with DCD.

Keywords: Attention deficit hyperactivity disorder (ADHD); Behavioral disorders; Developmental co-ordination disorder (DCD); Soto's syndrome

Introduction

Soto's syndrome is a childhood overgrowth condition described in 1964 by Sotos et al. The four major diagnostic criteria were established in 1994 by Cole and Hughes which included somatic overgrowth with advanced bone age, macrocephaly, characteristic facial appearance and learning difficulties.[1] They are associated with NSD1 mutations in most of the cases however 7–35% of them may not have any mutation.[2] There is a spectrum of behavioral disorders

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significantly associated with this syndrome like attention deficit hyperactivity disorder (ADHD), phobias, obsessions and compulsions, tantrums, and impulsive behaviors.[1,2] Neuroimaging findings in these children are distinct enough to allow differentiation of this syndrome from other mental retardation syndromes with macrocephaly. The common abnormalities noted are prominence of cerebral ventricles, prominence of the trigone, prominence of the occipital horns and ventriculomegaly.[3]

Case report

11-year-old boy presented to us with learning difficulty and other behavioral concerns. He had difficulty in coping with academics, was slow in writing and had inattention and hyperactivity. He was born full term by cesarean section in view of large head with normal birth weight for the gestational age. Though there was no history of significant development delay he always had issues with co-ordination and praxis. The parents of the child reported quite a lot of problems faced by the child with the gross motor activities like running, cycling etc. His participation in school sports activities was also restricted. There were host of behavioral problems in him like temper tantrums, inattention, hyperactivity, impulsiveness etc. Physical examination revealed a tall boy with coarse facial features, macrocephaly and prominent forehead. His height was 157 cm which fell between +2 and +3 SD and was obese with BMI of 23.5 as per WHO growth charts (2007). His head circumference was 58.5 cm (>+2 SD) as per Nellhaus charts and thus was macrocephalic.[4]

Secondary sexual characteristics were normally developed and there were no problems noted in other systems. The bone age was derived from X-Ray of the hand which turned out to be two years in advance compared to his chronological age. In view of typical facial gestalt, macrocephaly, advanced bone age a diagnosis of Sotos syndrome was made. The neurological examination revealed problems in fine motor area, co-ordination and balance like presence of illegible handwriting, inappropriate pencil grip, inappropriate pressure while writing, dysdiadochokinesia, difficulties during finger nose test and standing on one foot. The neuroimaging revealed mild dilatation of lateral ventricles, vermian hypoplasia and large arachnoids cyst in posterior fossa. Intelligence quotient (IQ) testing showed verbal quotient of 107, performance quotient of 84 with full scale IQ of 95. The low score thus obtained was partly attributed to upper motor co-ordination problem. On educational assessment he had dysgraphia secondary to upper motor co-ordination affliction. His handwriting was illegible with improper spacing and size of letter cases. As per DSM V criteria and parent and teacher rating NICHQ Vanderbilt assessment scales he was diagnosed to have attention deficit hyperactive disorder (ADHD) and was started on medications.[5] The motor evaluation using Bruininks Otseretsky Test of Motor Proficiency (BOTMP) showed affliction of balance, co-ordination, response speed, visuo-motor control, upper limb speed and dexterity.[6] DCD questionnaire was administrated which confirmed the presence of developmental co-ordination disorder in him.[7,8] The child was treated with methylphenidate as well as various task specific behavioral therapies as he was suffering from DCD along with ADHD. The child initially was started on low dose of methylphenidate (0.5 mg/kg/d) however due to persistence of symptoms the dose was increased to 1 mg/kg/d. In view of behavioral issues like short temper and aggression he was also put on behavioral modification therapy. There was improvement noted in these domains after the initiation of therapy.

Clinical Photographs of the Patient (Figure 1):

Figure 1: Showing front and lateral view of patient with typical facial features of sotos syndrome.

Radiological Images: – MR Images (Figure 2)

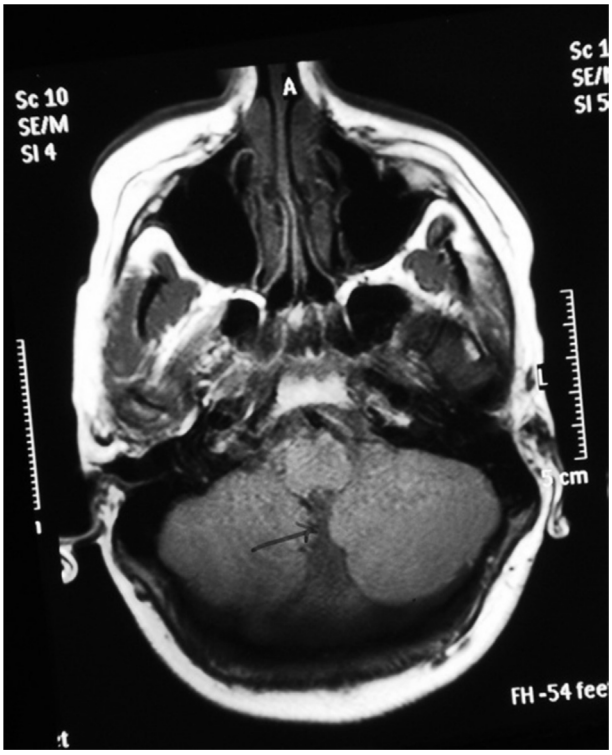

Figure 2: T1 W Axial Images of 11 years old male child showing suspicious vermis hypoplasia.

Discussion

Soto's syndrome is an uncommon condition that affects approximately one in fourteen thousand children. The cardinal features include typical facial gestalt, macrocephaly, behavioral and social problem and some degree of learning disability. Our patient had typical facial gestalt, macrocephaly and advanced bone age without acceleration of puberty; hence we made a diagnosis of Sotos syndrome which is an overgrowth syndrome. There are quite a few overgrowth syndromes like Weavers syndrome, Klippel Feil Trenauney, Beckwith Weidmenn etc. which can mimic sotos. However almost all except for the weavers do not have typical facial gestalt. The genetic studies that is NSD1 mutations is seen in both the syndromes and is not very helpful in distinguishing amongst the two. As there was no affliction of carpal bone, distal long bone or camptodactyly weavers was an unlikely possibility and thus he was diagnosed to have Sotos syndrome.[1,2,9] They have known to have spectrum of behavioral disorders like very low frustration tolerance, oppositional defiant traits, inattention etc.

Our patient though had inattention and behavioral concerns his dismal performance in academics couldn't solely be explained by them. Due to this he was further evaluated with specialized tests performed by a team of experienced personnel, which helped us clinch the diagnosis of an underlying developmental co-ordination disorder.[6–8]

DCD is a unique neurodevelopmental disorder which may coexist with a host of neurobehavioural disorders like ADHD, autism, dyslexia, etc.[9–11] Though the impact and sequelae of DCD is significant and may persist lifetime it still remains a grossly under diagnosed entity.[10] It can be loosely defined as affection of motor performance predominantly of that of co-ordination which cannot be explained by the chronological age or measured intelligence of the child and is not due to any other medical condition which might affect the same.[11] The afflicted children may experience difficulties in simple day to day activities which may have severe emotional impact on them. All these with associated co-morbidities may greatly compromise their academic performance.[10,11] The various treatment modalities for DCD include process-oriented approach, task-oriented approach, cognitive therapies etc. The literature suggests that methylphenidate therapy for ADHD in such children works better as it also improves the motor skills to a certain extent.[11]

Thus we recommend that physicians must think of DCD as a diagnosis when child predominantly comes with co-ordination issues as it currently stands grossly under-reported. Early diagnosis of this disorder can help alleviate parental anxiety by providing them with varied treatment options. Moreover reporting of such cases may help form uniform consensus statement for diagnosis and treatment of such disorders which even though significant are not readily diagnosed by the medical practitioners.

Conflict of interest

The author has no conflict of interest to declare.

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