Multidisciplinary or Monotherapy? Effects of Multidisciplinary Approach in Sotos Syndrome

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Abstract: *Objectives:* Paper presents impact of multidisciplinary approach in treatment of patients with Sotos Syndrome.

Design: 36 months old female patient with Sotos syndrome was presented to us three years ago with an atypical development of communication and social interaction, inattentivity, hyperactivity and overall difficulty to manage her. She met diagnostic criteria for atipic childhood autism and specific training for autism was applied. At the age of five years and nine months Stanford Binett test was applied with a result of learning disability and the patient started receiving specific training.

Results: Development was shown very quickly, she learned reading and writing, inattentivity and hyperactivity disappeared.

Conclusions: Sotos syndrome is syndrome with multiple anomaly and common developmental and behavioral problems. Patients have a high risk of developing ADHD, temper tantrums and present autistic features. Academic difficulties and learning disabilities occur in most of them. Specific cognitive deficit may be present while levels of general intelligence stays unimpaired.

Keywords: Sotos syndrome, psychiatry, multidisciplinary.

INTRODUCTION

Sotos syndrome is a rare syndrome, and was first described by Juan Sotos and colleagues in 1964 [1]. Three cardinal characteristics are present in more than 90% of individuals with Sotos syndrome [1]. These are pre – and postnatal accelerated growth with advanced bone age and macrocephaly; a typical facial appearance due to frontal bossing, sparse frontotemporal hair, malar flushing, downslanting palpebral fissures, pointed chin and development delay. Other clinical features may include neonatal jaundice, feeding difficulties, heart defects, scoliosis, genitourinary problems, hypothyroidism, large hands and feet, and malignant tumors [1,4]. Level of intelligence in children with Sotos syndrome is variable. Learning disabilities, language impairments and behavioral disorders are frequently seen.

The majority cases are sporadic [1,4], but familial cases have also been reported. NSD-1 abnormailities such as truncating mutations, missense mutations in functional domains, partial gene deletions and 5q35 microdeletions encompassing NSD-1 are identifiable in the majority (>90%) of Sotos syndrome cases [4].

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PHYSICAL APPERANCE

In early childhood, children have a head circumference and height above the 97th percentile. Because of advanced bone age, as a little growth occurs after puberty and they do not generally go on the reach gigantic heights in adulthood and do not require any intervention to limit adult height [1]. Facial features are distinctive with high anterior hairline, macrocephaly, frontal bossing, a long thin face, frontotemporal hair scarcity, downslanting palpebral fissures, and a prominent mandible [1]. Although all characteristic are not manifested in every case, macrocephaly and the chin is the most distinctive feature in adulthood [1,4].

PSYCHIATRIC FEATURES

A wide spectrum of emotional, cognitive and behavioral difficulties occur in patients with Sotos Syndrome. They have high risk of developing attention deficit disorder, hyperactivity disorder and temper tantrums [3]. A report done on 11 children with Sotos syndrome revealed that nine of them had a psychiatric disorders such as ADHD, enuresis, emotional liability and impulse control impairment therefore none of the children functioned well socially [5].

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INTELLECTUAL IMPAIRMENT

Level of intelligence in children with Sotos syndrome is variable. In early reports, mental retardation was considered to be extremely common if not invariable present [1]. Rutter and Cole [5] reported on 16 children with Sotos syndrome from whom five children fell into the moderate range of intellectual disability (IQ51 to 70), four were within borderline range (IQ 70 to 79), four were within the low-average range (IQ 80 to 89), and two children were within the average range (IQ 90 to 109). However, it is difficult to determine the true incidence of cognitive and behavioral disorders associated with Sotos syndrome because there is a conflicting result between different studies.

LANGUAGE DISABILITIES

Problems with speech and language are also commonly presented in patients with Sotos syndrome. Language delay in these patients has been reported in many studies [1, 5]. Affected individuals often have problems with sound production, stuttering and a monotone voice.

LEARNING DISABILITIES

Academic difficulties and learning disabilities occured in most of the patients. Tatton-Brown and Rahman [4] estimated that approximately 96% of their cohort had learning difficulties. In another study, nearly all children whether above or below average intelligence, showed greater difficulty with numeracy rather than with literacy. Matthew and collegues report that the most common features were learning disabilities (90%) and learning disabilities were more severe in patients with a microdeletion than in those with a point mutation [4]. Most individuals have mild-moderate intellectual impairment, but the degree of impairment is extremely broad, ranging from occasional individuals with normal development to children with profound learning difficulties requiring long life care [1].

BEHAVIOURAL AND EMOTIONAL CHARACTERISTICS

Finegan and collegues [6] reported a higher rate of irritable, withdrawn and stereotypical behaviour in those patients with Sotos syndrome than in a comparison group matched for IQ. In another study, anxiety and emotional immaturity may contribute significantly to the poor relationships which are reported by many parents of the children with Sotos syndrome. Most of children have no friends in their class and their neighborhood [3]. Authors suggested that behaviours that indicate anxiety and insecurity in children with Sotos syndrome might be the result of social experiences. Contrary to expectation Finegan and collegues [3] found more withdrawn and irritabl behaviours in children with Sotos syndrome than in a control group of children who were large in size, with developmental delayed and dysmorphic features.

Hyperactivity was reported by 38% of parents in the data presented by Finegan and collegues [3]. Some case reports mentioned conduct disorders present [5]. Some individuals have autistic features [3, 5] and temper tantrums were very common and difficult to manage because of the child's size and strength [1].

One of the patients in the study developed psychosis [6]. Other difficulties that were described are feeding and sleeping problems and also phobias and irritability.

The following case history describes an individual with Sotos syndrome who has autistic features, communication disability, hyperactivity and learning disorder.

A CASE REPORT

The patient, A.N.C is second born child from healthy parents and out of non-consanguineous marriage. Patient has a sister and a brother who are healthy. Pregnancy was complicated because of polyhydroamnios and macrocephaly. Maternal serum screening showed signs for increased risk of Down syndrome so amniosynthesis was applied. Amniosynthesis results came back normal fetal karyotype. She was born at 40 weeks of gestation by normal vaginal delivery. At the time of birth her mother was thirty years old. Patient birth weight was 3100 gr (25 percentile), height 52 cm (91 percentile) and head circumference 34,5 cm (50 percentile) During first two week of life patient stayed in the hospital because of complete cheiloschisis and palatoschisis. In the neonatal period feeding difficulties were present. Ear anomaly was noticed. From early infancy patient's psychomotor development was delayed, so she was referred for pediatric assessment. Brain - imaging studies had revealed thinning of the corpus callosum, cavum septum pellucidium et vergae and asymmetric dilatation of the ventricles. Complete blood counts, serum electrolytes, BUN, creatinine, fasting blood sugar, liver enzyme, growth hormone and thyroid profile were normal. Abdomen USG, echocardiography and EEG were normal. Orthopaedics examination was normal. Other systematic examination was

unmarkable. The patient had characteristic facial appearance; frontal bossing, antimongoloid slant of palpebral fissures, and a prominent jaw. Genetic assessment for NSD1 mutation was positive. As a conclusion she was diagnosed as having Sotos Syndrome at the age of 12 months.

The patient was presented to our clinic at the age of 36 months because of an atypical development of communication and social interaction. Verbal language was not developed, her only expressive language consisted of two words. The non-verbal language was also abnormal and patient had severe difficulties in reciprocal social interaction. The patient used her parent's hands to indicate if she wanted something, she did not use pointing for this and did not share any common interest. The eye contact was not regulated in a normal way. Tendency to be with-drawn and preferrance to play by herself were presented. The patient was particularly revolving around herself in a stereotyped way. Altogether our observations showed that she also met ICD-10 diagnostic criteria for childhood atipic autism; so patient was taken to specific training for atipic autism. At this period, parents reported about inattentivity, hyperactivity and overall difficulties to manage her.

The patient was being monitored regularly for speech and learning disability, mental retardation and other behavioral disorders. At the age of five years and nine months psychometric test, Stanford Binett was applied. During the test period, patient had difficulty to separate from her mother and phonological disorder was remarkable. There was further delay in expressive language development, with an intelligence quote of 68. The patient performed weakness at working memory, interpretation of nonverbal information and processing speed and visuospatial tasks upon testing. Learning disabilities that were seen were incompatible with the degree of intelligence. The patient received special education for learning disabilities and language impairments. At the same time we applied Methylphenidat 10mg/day to cure her inactivity and hyperactivity. However, she is not able to enter a class for normal children. She still has major social problems and behavioral disturbances which shows a clinically coherence with the growth velocity. At those intervals we prefer to support the patient with methylphenidat 20mg/day.

DISCUSSION

Our patient exhibited the craniofacial features characteristics of Sotos Syndrome. Furthermore, she had been diagnosed with the disorder as a child and her work-up at that time excluded other causes of over growth.

All the patients with Sotos syndrome in the presented studies were diagnosed postnatally [1]. However, the diagnosis may be considered in the antenatal period in cases with a normal fetal karyotype where there is increased risk for Down syndrome demonstarted in maternal serum screening, especially when supportive ultrasound findings such as macrocephaly, polyhidroamnios and decreased fetal movements are presented [2].

Sotos syndrome is not a life-threatening syndrome but it is a multiple anomaly syndrome and some of the patients have severe clinical problems. This kind of patients clearly displays a wide range of emotional, cognitive and behavioral difficulties and no clearly identifible pattern has emerged. Some adult patients with Sotos syndrome have been noted to be 'socially isolated' due to social difficulties, medical illness, physical differences or autism [3]. Another study reported that most adults with Sotos syndrome were employed at least part–time, 63% (10/16) lived with their parents and 31% (5/16) had significant psychiatric illness [3].

Knowing that a child has Sotos syndrome, can be predicted that the child is at high-risk for developing learning difficulties, behavior problem, ADHD, social problems, fears, temper tantrums, irritability and sleep disturbances [6].

In our case we try to emphasize the importance of a network triangle; special education teacher, psychiatry clinic and the parents.

This presented report highlights the importance and impact of multidisciplinary treatment, psychological counseling and educational support in patients with Sotos Syndrome. Early psychiatric and physical intervention, speech therapy and educational support can make big change in their life and improve their quality of life.

The need for collaboration across research groups / clinics is apparent from the fewness of Sotos Syndrome cases.

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