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Case Report

A Child with Aarskog Scott Syndrome and Autism Spectrum Disorder

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Abstract: Autism Spectrum Disorder (ASD) is a neurodevelopmental disorder that is increasingly common in society, characterized by limitations in social communication and some repetitive behaviors. Many genetic factors such as genetic syndromes and single gene mutations and many environmental factors play a role in the etiology of ASD. Many genetic syndromes accompanying ASD have been reported. Aarskog-Scott Syndrome is a rare genetic disorder characterized by facial, digital, and genital anomalies, including a broad upper lip, anteverted nostrils, ptosis, hypertelorism, shawl scrotum, short stature, flat feet, and genu recurvatum. Cases with learning difficulties, hyperactivity, or cognitive retardation have been reported to date with Aarskog Scott syndrome. However, to our knowledge, one case report of ASD and Aarskog Scott syndrome has been reported. In this report, we will present a case with Aarskog Scott syndrome and Autism Spectrum Disorder.

Keywords: Autism, Aarskog Scott syndrome, Genetic, Syndrome

1. INTRODUCTION

Autism Spectrum Disorder (ASD) is a neurodevelopmental disorder that is increasingly common in society, characterized by limitations in social communication and some repetitive behaviors, and is seen in 1 in 36 children 1,2. Many genetic factors such as genetic syndromes and single gene mutations and many environmental factors play a role in the etiology of ASD 3. Many genetic syndromes accompanying ASD have been reported 4,5.

Aarskog-Scott Syndrome is a rare genetic disorder characterized by facial, digital, and genital anomalies, including a broad upper lip, anteverted nostrils, ptosis, hypertelorism, shawl scrotum, short stature, flat feet, and genu recurvatum 6,7. It was first described by Aarskog in 1970 6. Symptoms of ligament laxity such as flat feet and genu recurvatum were also added by Scott in 1971 7. Although its inheritance pattern is still unknown, it is thought to be X-linked because it is more common in male patients 6. There are also cases where it shows autosomal recessive and autosomal dominant inheritance patterns 8,9. Phenotypic symptoms show a wide spectrum 10. The diagnosis of Aarskog Scott syndrome is generally made clinically 11. Cases with learning difficulties, hyperactivity, or cognitive retardation have been reported to date with Aarskog Scott syndrome 12. However, to our knowledge, a case report of Autism Spectrum Disorder and Aarskog Scott syndrome have been reported 13. In this report, we will present a case with Aarskog Scott syndrome and Autism Spectrum Disorder.

2. CASE

M.Y., a 3-year-old boy, was brought by his father with the complaint of not making eye contact and not playing with children. Written and verbal consent was obtained from the patient's family. According to the psychiatric examination and the interview with the father, he did not make eye contact, was not interested in his peers, had limited affective participation, had about 20 words and could not form sentences, and did not know how to play games like his peers. The child was spinning around, shaking, and was sensitive to the sound of a vacuum cleaner. The hearing test result requested for the child was normal. The child received a score of 34 on the Childhood Autism Rating Scale (CARS). The Turkish validity and reliability study of the CARS scale was conducted by İncekaş et al14. In this study, the

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Cronbach alpha coefficient was determined as 0.95 and the cut-off score was determined as 30. Children who scored 30-36.5 on the scale were considered to have mild-moderate autism, and those who scored 37-60 were considered to have severe autism. The patient was diagnosed with ASD according to DSM-5. The child's developmental stages were also behind. He started walking at 1.5 years old and his first word was at 2 years old. He was not yet toilet trained. In the Denver test applied to the child, general development, gross motor and fine motor development were found to be around 2 years old, and language development was around 1.5 years old. Psychoeducation was given to the family. The child was directed to education. Since the child had short stature, long philtrum, hypertelorism and brachydactyly, he was referred to the genetic department due to a dysmorphic facial appearance and cognitive retardation. The patient was diagnosed with Aarskog Scott syndrome by the genetic department. Additionally, it was learned that the child had a scarf scrotum. No pathology, including Fragile X, was detected in the patient's requested chromosome analysis. The complete blood count, biochemistry, thyroid functions, metabolic tests, EEG and brain MRI requested from the child were normal. The child is followed at three-month intervals. The patient started special education during the 3-month follow-up period. There was a partial increase in eye contact and interest in peers after starting special education. The word count reached approximately 40 words in the 6th month. The child's turning has partially decreased. The delay in motor skills continues.

3. DISCUSSION

In this case report, we presented a boy with ASD and Aarskog Scott syndrome. Cases with learning difficulties, hyperactivity, or cognitive retardation have been reported to date with Aarskog Scott syndrome 12. However, to our knowledge, one case report of Autism Spectrum Disorder and Aarskog Scott syndrome has been reported 13. Three cases were presented in a single study. The first case is a 10-year-old male, the 2nd case is a 5-year-old male, and the 3rd case is a 9-year-old male. There was hyperactivity in all of these cases and severe cognitive impairment in 2 of the cases. A previous case report discussed the challenge of diagnosing ASD in individuals with severe cognitive impairment 13. In none of these cases, a scale was used to diagnose ASD, and the diagnoses were made clinically according to the DSM. In our case, CARS was applied to support the diagnosis of ASD. In our case, there was also mild cognitive developmental delay, consistent with the literature. The strengths of our case are that it was not accompanied by a confounding psychiatric symptom such as hyperactivity, moderate-severe cognitive retardation, and the scale was used in making the diagnosis.

Aarskog Scott syndrome is a rare genetic syndrome. A small number of psychiatric disorders accompanying this syndrome have been reported 12,13. There is a need for further investigation of psychiatric diseases accompanying Aarskog Scott syndrome. This syndrome mostly shows X-linked inheritance6. ASD is also more common in boy3. Another genetic disorder that resembles Aarskog-Scott Syndrome in terms of dysmorphic features is Noonan syndrome. Noonan syndrome is a genetic disorder characterized by folded, folded, thick, nape skin, short stature, a longer than normal distance between the eyes, a downward sloping eye line, a deep area between the two thin vertical lines connecting the nose and the upper lip, and dysmorphic facial findings such as the lower jaw being positioned backwards relative to the upper jaw, heart defect, vision/eye problems, chest deformity, cryptorchidism, lymphatic dysplasia, delayed puberty, and many systematic findings such as autosomal dominant inheritance, supported by 80% genetic mutations, and mutations in genes that produce codes in the RAS/mitogen-activated protein kinase signaling pathway 15. The genetic inheritance pattern of Noonan syndrome, the fact that its genetic mutations have been determined, and that it is a disease with many systematic findings other than dysmorphic findings are distinctive. Aarskog-Scott Syndrome is a clinically diagnosed genetic disorder. Possible genetic mechanisms between ASD and Aarskog Scott Syndrome remain unclear. For this reason, there is a need to further

investigate the relationship between ASD and Aarskog Scott syndrome in terms of etiological mechanisms. Additionally, we would like to emphasize the importance of genetic evaluation in children with dysmorphic facial appearance accompanying ASD. From the clinician's perspective; we would like to state that in order to establish early diagnosis, follow-up and individualized treatment plans for children diagnosed with ASD, with cognitive delay or dysmorphic facial appearance, it is a multidisciplinary team work that should be evaluated by other departments such as genetics, in addition to child psychiatry.

Article Information Form

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The Declaration of Conflict of Interest/ Common Interest

No conflict of interest or common interest has been declared by author.

Artificial Intelligence Statement

No artificial intelligence tools were used while writing this article.

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