Nablus Mask-Like Facial Syndrome with Moderate Developmental Delay

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ABSTRACT

Nablus mask-like facial syndrome (NMLFS) is defined by distinctive craniofacial appearance including tight-appearing glistening facial skin, blepharophimosis, telecanthus, severe arched eyebrows, flat and broad nose, long philtrum, distinctive ears, unusual hair patterns, mild developmental delay and "happy" disposition. We aim to report a 7-year-old boy diagnosed with NMLFS and moderate developmental delay. Literature emphasis that Intellectual Disability is common in this syndrome though it has been diagnosed to only a few people worldwide.

Keywords: Children with special needs, developmental delay, microdeletion syndromes

Introduction

Nablus mask-like facial syndrome (NMLFS) is characterized by a distinctive craniofacial appearance featuring tight-appearing glistening facial skin, blepharophimosis, telecanthus, severe arched eyebrows, flat and broad nose, long philtrum, distinctive ears, unusual hair patterns on the scalp, mild developmental delay, and "happy" disposition [1]. The initial clinical report of a case revealed by Teebi in 2000 caused the recognition of a new syndrome [1]. It is very rare, and there are currently approximately 12 cases of NMLFS published in the literature [2-4]. NMLFS is a rare microdeletion syndrome, in which deletion of chromosome 8q22.1 is significant, but not sufficient, for development, and this contributes to additional clarification of the critical region [1-3].

Case Report

The patient was a 7-year-old child born at a normal gestational age. At birth, his height was 49 cm, and weight was 3500 grams. When his age was 7 months, he was suspected to have NMLFS owing to a phenotypic appearance specific to the syndrome and was diagnosed with NMLFS; 22q11 deletion was revealed in the genetic test conducted at another center. He was referred to our centre with hyperactivity, irritability, and self-spinning. According to his developmental history and mental examination, it was found that he never breastfed, started to walk at 2.5 years, spoke his first words when he was 4 years old, was able to make short sentences recently, and was toilet trained for 1 year. In addition, non-verbal communication was appropriate based on his age there were no echolalia and stereotype; there were no comorbid depression and anxiety symptoms. Autism Diagnostic Observation Schedule (ADOS- 2) was administered to the patient, and autism spectrum disorder was not detected. Results of the psychometric tests and mental examination revealed that language development and fine and gross motor skills were −2.5 SD behind his peers. Based on the results of the clinical examination and IQ test Wechsler Intelligence Scale for Children-Revised (WISC-R) conducted for the patient, he was diagnosed with moderate intellectual disability. Written informed consent was obtained from the patient and his family.

Discussion

This syndrome, which has been diagnosed in only a few people worldwide, has drawn attention owing to its typical phenotype and microdeletion syndrome as well as good social communication skills of the patient. Although literature emphasizes that mild intellectual disability is common in patients with this syndrome, the patient included in our case report was diagnosed with

moderate intellectual disability, and to date, only one case with autism has been described [5, 6]. Consequently, it should be considered that the level of intellectual disability in these children may vary.

Informed Consent: Written informed consent was obtained from the patient and patient's parents who participated in this study.

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