Letter to the Editor

DOI: 10.5152/eurasianjmed.2019.18448

Nablus Mask-Like Facial Syndrome with Developmental Delay

Bahadır TURAN

Department of Child and Adolescent Psychiatry,

Agrı State Hospital,

Agrı, Turkey

0000-0003-1190-9589

Mehmet Akif AKINCI

Department of Child and Adolescent Psychiatry,

Ataturk University Faculty of Medicine,

Erzurum, Turkey

0000-0002-6436-4394

İbrahim Selçuk Esin

Department of Child and Adolescent Psychiatry,

Ataturk University Faculty of Medicine,

Erzurum, Turkey

0000-0001-5598-2097

Onur Burak Dursun

Department of Child and Adolescent Psychiatry,

Health Science Univercity, Faculty of Medicine,

Trabzon, Turkey

0000-0003-2990-9851

Correspondence to: İbrahim Selçuk Esin, Department of Child and Adolescent Psychiatry, Ataturk University Faculty of Medicine, Erzurum Turkey. e-mail: esinibrahimselcuk@gmail.com

Conflicts of Interest and Source of Funding: This manuscript was not funded. The authors declare that they have no conflict of interest.

This case report was presented at 27.Turkish Child and Adolescent Psychiatry Congress, 10-13 May 2017 Ilıca Çeşme, Turkey

You may cite this article as: Turan B, Akinci MA, Esin IS, Dursun OB. Nablus Mask-Like Facial Syndrome with Developmental Delay. Eurasian J Med 2019; 10.5152/eurasianjmed.2019.18448.

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, upwest frontal hairline, mild developmental delay and "happy" disposition. We aim to report a 7-year-old boy diagnosed with NMLFS and moderate mental retardation. Literature emphasis that Intellectual Disability is common in this syndrome though it has been diagnosed to only a few person worldwide.

Keywords: Pediatric Psychiatry, Developmental/Behavioral, Pediatrics

INTRODUCTION

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

CASE REPORT

The patient is a 7-year-old child was born at a normal gestational age. At birth, his height was 49 cm, weight was 3500 grams. While he was 7 months old he was suspected of having NMLFS due to phenotypic appearance specific to the syndrome and diagnosed with NMLFS. 22q11 deletion was revealed in the genetic test in another center. He was consulted to us with hyperactivity, irritability and self-spinning. According to his developmental history and mental examination he had never breastfed, started to walk at 2,5 years, used his first words at 4 years old, could make short sentences recently, toilet trained for 1 year. Also, non-verbal communication was at the age level, there were no echolalia and sterotype, and there were no comorbid depression and anxiety symptoms. The patient was administered ADOS-2 and Autism Spectrum Disorder was not detected. As a result of the psychometric tests and mental examination, it was seen that language development, fine and gross motor skills were psychosocial development were -2,5SD behind his peers. In terms of the clinical examination and IQ test (Wisc-r) of the patient, he was diagnosed with moderate intellectual disability. Written informed consent was obtained from the patient and her family.

DISCUSSION

This syndrome, which has been diagnosed to only a few people worldwide, draws attention with its typical phenotype and microdeletion syndrome and good social communication skills. While Literature emphasis that Mild Intellectual Disability is common in this syndrome, our case was diagnosed with Moderate Intellectual Disability, and up until now one case with autism is described (5, 6). As a consequence, it should be considered that the level of intellectual disability in these children may show variation.

Acknowledgement: The paper had been published as a poster, on 27. Congress on Child and Adolescent Mental Health 2017, Izmır, Turkey.

REFERENCES

1. Teebi AS. Nablus mask-like facial syndrome. American journal of medical genetics. 2000;95(4):407-8.
2. Jamuar SS, Duzkale H, Duzkale N, Zhang C, High FA, Kaban L, et al. Deletion of chromosome 8q22. 1, a critical region for Nablus mask-like facial syndrome: Four additional cases support a role of genetic modifiers in the manifestation of the phenotype. American Journal of Medical Genetics Part A. 2015;167(6):1400-5.

3. Debost-Legrand A, Eymard-Pierre E, Pebrel-Richard C, Gouas L, Goumy C, Giollant M, et al. A new case of 8q22. 1 microdeletion restricts the critical region for Nablus mask-like facial syndrome. American Journal of Medical Genetics Part A. 2013;161(1):162-5.
4. Overhoff J, Rabideau MM, Bird LM, Schweitzer DN, Haynes K, Schultz RA, et al. Refinement of the 8q22. 1 microdeletion critical region associated with Nablus mask-like facial syndrome. American Journal of Medical Genetics Part A. 2014;164(1):259-63.
5. Jain S, Yang P, Farrell S. A case of 8q22. 1 microdeletion without the Nablus mask-like facial syndrome phenotype. Eur J Med Genet. 2010;53(2):108-10.
6. Raas-Rothschild A, Dijkhuizen T, Sikkema-Raddatz B, Werner M, Dagan J, Abeliovich D, et al. The 8q22. 1 microdeletion syndrome or Nablus mask-like facial syndrome: report on two patients and review of the literature. Eur J Med Genet. 2009;52(2-3):140-4.

This article has been accepted for publication and undergone full peer review but has not been through the copyediting, typesetting, pagination and proofreading process, which may lead to differences between this version and the Version of Record. Please cite this article as: Turan B, Akinci MA, Esin IS, Dursun OB. Nablus Mask-Like Facial Syndrome with Developmental Delay. Eurasian J Med 2019; 10.5152/eurasianjmed.2019.18448.