Knowledge of Nursing Journal Spring 2024, 2(1):57-61

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Original Article Open Access

Moebius Syndrome in a 14-Year-Old Boy: A Case Report

Seyed Maryam Mousavi[1], Sima Vadaei[2], Farimah Rezaie[3], Sepideh Nasrollah[4*]

1. Department of Nursing, Golpayegan Branch, Islamic Azad University, Golpayegan, Iran.
2. Department of Nursing, Golpayegan Branch, Islamic Azad University, Golpayegan, Iran.
3. Master's degree student in nursing، Karaj Branch, Islamic Azad University, Karaj Iran.
4.Assistant professor, Department of Nursing, TeMS.C., Islamic Azad University, Tehran, Iran ***(Corresponding Author)****.*

Abstract

Moebius syndrome is a rare neurological disorder that causes paralysis of facial muscles, especially those responsible for facial expression and eye movement. The cause of this syndrome is unknown, but genetic and environmental factors are thought to play a role in this regard. The aim of present study is to report a case of Moebius syndrome in a 14-yers-old boy in order to better understand the syndrome and familiarize nurses, especially pediatric nurses, with this rare syndrome. This would help pediatric nurses to provide better care, education and support to affected patients. The patient studied in this report was a 14-year-old boy, who was the result of an unrelated marriage and a natural term delivery without complications. At birth, he had a masked face and was unable to suckle at his mother's breast. He also had sialorrhea and a limited abduction and adduction of the eyes, which led to strabismus. The definitive diagnosis was made at 4 months of age. Since there is no definitive treatment for Moebius syndrome, recognition of this rare syndrome, timely diagnosis and provision of supportive and symptomatic treatment can prevent the development of severe complications in such cases.

Keywords: Moebius syndrome, facial paralysis, congenital disorders, cranial nerve disorders, neurological disorders, case report.

Corresponding author: Sepideh Nasrollah, ORCID ID: 0000-0001-7084-926X. Email: S.nasrollah@iautmu.ac.ir, **Received**: February 2024, **Accepted**: May 2024, **ePublish**: Spring 2024. **Citation**: Mousavi SM, Vadaei S, Rezaie F, Nasrollah S, Moebius Syndrome in a 14-Year-Old Boy: A Case Report, Knowledge of Nursing Journal. 2024;2(1):57-61.

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[DOI: 10.66224/knjournal.2.1.57]

Introduction

Moebius syndrome, which is among rare congenital neuromuscular disorders of unknown etiology, has gained attention in recent years following the discovery of genetic mutations [1]. It was introduced by Paul Julius Moebius, a German neurologist in 1888 [2].

This syndrome is an asymmetric, non-progressive, and often bilateral congenital facial palsy accompanied by limited abduction of the eyes, which can lead to strabismus, limb deformities, motor deficits, impaired mental development, speech and swallowing problems, mental retardation, and even autism [3-4]. Hereditary disorders, vascular disorders, infections, neuronal degeneration, and muscular dystrophy have been implicated in the pathogenesis of this syndrome [2]. Its prevalence has been estimated to be between 0.002% and 0.0002% of live births, with equal incidence in boys and girls [5].

Diagnosis is based on clinical symptoms, patient history, and paraclinical tests such as brain scans or MRI [3]. Treatment is based on the patient's symptoms and needs. Physiotherapy, occupational therapy, corrective surgery, speech therapy, and nutritional education can help to improve complications and quality of life in affected people [6].

Case presentation

The patient in this report is a 14-year-old boy, the third child of the family, and the result of an unrelated marriage and a natural term delivery without complications. He was born in a hospital in the city of Arak. His weight at birth was 2850 grams. He also has two older sisters who are genetically healthy.

The mother had a normal pregnancy with no history of drug and medication use, rubella, or attempted abortion. Her only complaint during pregnancy was severe headaches. During pregnancy, the mother experienced severe shock after hearing the bad news.

After birth, the patient was admitted to Sarem Hospital in Tehran, the capital of Iran, due to his inability to suck breast milk. After tests were performed at this hospital, a definitive diagnosis of Moebius syndrome was given at 4 months of age.

The patient has now been referred to a hospital in Arak for periodic tests, and members of the research team are getting to know him at the hospital.

Currently, on physical examination, the patient has normal facial muscle tone and volume without microsomia, but his face is mask-like and has bilateral nerve palsy. Sialorrhea is present due to the mouth being open. There is severe limitation of abduction and adduction movements in the eyes, but vertical movements are normal, which has led to strabismus. There is corneal dryness due to the eyes being open during sleep. Both eyes have decreased vision. The patient has no problems with blinking, tongue, nose, teeth, swallowing, and gag reflex movements, but he chews food with his mouth open. He has difficulty pronouncing some letters. The limbs, head, chest, and abdomen are normal on examination. Sensory and cognitive disorders and decreased IQ are not evident.

According to the documentation in the patient's file, in clinical tests, the levels of

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[DOI: 10.66224/knjournal.2.1.57]

creatine phosphokinase and lactate were increased, and in genetic testing, 46+XY was reported. No abnormal findings were found in other clinical and paraclinical tests.

Discussion

The causes of Moebius syndrome are unknown, but it is thought to be related to environmental and genetic factors [6]. Dotti et al. (1989) [7], in a report talked about families in which more than one person had this syndrome, highlighting that the method of inheritance in these families was autosomal dominant. In the present report, as in the study of Ghosh et al. (2017) [8], we observed no family history of this syndrome.

Studies by Vauzelle et al. (2013) [9], Miller et al. (2009) [10], and Singham et al. (2004) [11], indicated that drug and alcohol use, some medication (such as misoprostol, thalidomide), and attempted abortion could be risk factors for Moebius syndrome. Lima et al. (2009) [12], in a study referred to rubella and birth injuries as the risk factors for this syndrome. In the present report, as in the Souni et al. (2023) study, there was none of the above factors in the maternal birth record [13].

Respiratory effort, apnea, nasal insufficiency, snoring, eating and swallowing problems that cause coughing, choking and aspiration are common in affected people [14]. In the study of Gheorghe et al. (2022) [15], the patient had many respiratory and eating problems. In the study of Renault et al. (2020) [16], out of 32 patients, suckling ability was absent or weak in 30 patients, swallowing was impaired in 25 patients, eating disorders were severe/moderate in 25 patients, and respiratory complications occurred in 17 patients.

In the present report, as in the Picciolini et al. (2016) study, the affected child had eating problems at birth and was unable to suckle at mother's breast. However, later the patient's body mechanics were activated and he was able to eat, chew and swallow food completely with his mouth open [17].

Growth delay, speech and language delay (11), sleep disturbances, and some degree of mental retardation (14) may be present. The child in our study had normal development, normal IQ and no sleep disturbances. However, he had difficulty pronouncing some letters such as "b" that requires the closure of lips, which was improved by speech therapy sessions.

In the study of Picciolini et al. (2016) [17], in a sample of 50 patients, 15% had cognitive impairment and developmental delay, 31% had language delay, 42% had speech impairment, and 28% had sleep disturbances. In the study of Ghosh et al. (2017) one patient had normal growth and IQ, while the other one had delayed growth and average IQ [8].

The patient has limited eye’s abduction and adduction movements, dry cornea, and decreased vision. He also had strabismus, which improved with physiotherapy. In all the studies reviewed, there was some degree of ocular abnormalities in affected people. For example, in the study of Ramraika et al. (2022) [18], a 13-year-old patient had inability to close the right eye, dry cornea, decreased vision, complete abduction of the right eye, moderate abduction of the left eye, adduction of both eyes, nystagmus, and strabismus.

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Conclusions

Mobius syndrome is a neurological disorder with multiple and severe complications. Given the complex needs of affected children and their families, providing collaborative and family-centered treatment, care and education by pediatric physicians and nurses, and referring patients to multidisciplinary teams (pediatrics, otolaryngologists, radiologists, neurologists, neurosurgeons, psychologists, physiotherapists, occupational therapists, speech therapists) are essential for early and ongoing diagnosis and management at an early age [6, 14].

Pediatric nurses play a central role in providing care, education and support to patients with this syndrome. The values emphasized in the pediatric nursing curriculum include continuous and dynamic learning, and provision of quality care, counseling and support to children and families. These nurses need to be familiar with rare syndromes and diseases in order to perform their professional duties optimally.

The purpose of this report is to familiarize pediatric nurses with this rare syndrome. Through early recognition and diagnosis, pediatric nurses can provide preventive guidance and recommend resources to parents of children with this neurological disease. The cost of treating these children is also very high. In Iran, there is no independent association to help these children, while early rehabilitation improves their performance and quality of life. Therefore, we recommend health managers to pay special attention to this issue.

Acknowledgement

The research team would like to thank the patient and his family for their sincere cooperation. The patient's family expressed their consent for their child's syndrome to be published in this article.

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