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SELÇUK TIP DERGİSİ

A Rare Cause of Facial Paralysis: Moebius Syndrome

Nadir Bir Fasiyal Paralizi Nedeni: Moebius Sendromu

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Özet

Moebius sendromu ilerleyici olmayan tam ya da parsiyel konjenital fasiyal paralizi ile karakterize bir sendromdur. Genellikle orofasiyal malformasyonlar, kas-iskelet sistemi defektleri, beyin sapı displazisi ve diğer kraniyal sinir felçleri ile ilişkilidir. Moebius sendromunun ortalama insidansı 2-20/milyondur. En sık görülme şekli bilateral lateral rektus kası felci ve fasiyal güçsüzlüktür. Sıklıkla 5., 10., 11. ve 12. kraniyal sinirler de tutulur ve öksürük, yutma ve çiğneme güçlüğü ve solunum yetersizliğine neden olabilir. Tam veya parsiyel fasiyal paralizi Moebius sendromu tanısı için şarttır. Dört aylık kız hasta doğumdan itibaren sağ gözünü tam kapatamama ve içe bakış şikayetleri ile kliniğimize getirildi. Hasta sağ gözünü tam kapatamıyordu, dilde atrofi ve mikrognatisi vardı. İlk bakışta her iki gözde içe bakıyordu. Dışa bakış kısıtlılığı, ayaklarda pes ekinovarus deformitesi ve katlantılı kulağı vardı. Dismorfik özellikleri, mikrognati ve dilde atrofi nedeni ile kraniyal manyetik rezonans görüntüleme yapıldı. 3D FIESTA taramada bilateral fasiyal sinirler görüntülenemedi. Bu vaka konjenital fasiyal güçsüzlükle başvuran hastaların ayırıcı tanısında Moebius sendromunun mutlaka akılda tutulmasını vurgulamak amacıyla sunulmuştur.

Anahtar kelimeler: Fasiyal paralizi, çocuklar, Moebius sendromu

Abstract

Moebius syndrome is a rare, non-progressive congenital syndrome presenting with complete or partial facial paralysis. It is usually associated with orofacial malformations, musculoskeletal defects, brainstem dysplasia, and other cranial nerve palsies. Mean incidence is 2-20/million, although there is considerable regional variation. The most common presentation is with bilateral lateral rectus palsies and facial diplegia. Frequently, the 5th, 10th, 11th and 12th cranial nerves are involved and may cause cough, difficulty in chewing and swallowing, and respiratory insufficiency. Complete or partial facial nerve palsy is necessary for a diagnosis of Moebius syndrome. A 4-month-old girl was brought to our clinic with complaints of inability to close her right eye completely and internal deviation in that eye, beginning from birth. She had micrognathia and an inability to completely close the right eye. Both eyes were turned inwards during primary gaze. She had limited lateral gaze, a pes equinovarus deformity in her feet, and flap ears. Cranial magnetic resonance imaging was performed because of the dysmorphic features and revealed micrognathia and volume loss at the tongue. Bilateral facial nerves could not be visualised by a 3D FIESTA scan, suggesting bilateral facial nerve agenesis. This case is presented to highlight Moebius syndrome in the differential diagnosis of cases presenting with congenital facial weakness.

Key words: Facial paralysis, children, Moebius syndrome

INTRODUCTION

Moebius syndrome was first described as bilateral combined 6th and 7th cranial nerve palsies by the German neurologist Paul Julius Möbius (1). It is a very rare genetic disorder, characterised by congenital facial and abducens palsies (2,3). Most patients are diagnosed during infancy or early childhood. The incidence of the disease varies among regions, between 2 and 20 cases per million live births. Most reported cases are sporadic (4,5). Although facial paralysis is a prerequisite for diagnosis, other criteria for diagnosis include extremity abnormalities (syndactyly, brachydactyly, absence of fingers and lumped feet), findings related to involvement of other cranial nerves and presence of orofacial malformations (bifid uvula, micrognathia, and ear abnormalities) (3). Limited abduction in eyes and facial palsy are observed in all cases. Exotropia (54%), epicanthus (51.5%), entropion (22%) and a history of abortive drug use in the first 3 months of pregnancy (28%) were reported in cases (6). Here, we describe a girl who presented with facial weakness and was diagnosed with Moebius syndrome.

CASE REPORT

A 4-month-old girl was brought to our clinic with complaints of inability to close her right eye completely and internal deviation at that eye, beginning from birth. She was born at term as the second live birth from her 36-year-old mother's second pregnancy. Her birth weight was 3200 g and she had no postnatal adaptation problem. Her mother did not use any drugs during her pregnancy. Her mother and father were third-degree relatives. Their family had no history of any similar disease. She had a healthy 4-year-old brother. Her weight was 6500 g (50-75th percentile), height was 65 cm (50-75th percentile), the anterior fontanelle was closed, and head circumference was <3rd percentile. She had micrognathia, and an inability to completely close the right eye. Both eyes turned inwards during primary gaze. She had limited lateral gaze, pes equinovarus deformity in her feet, and flap ears. A

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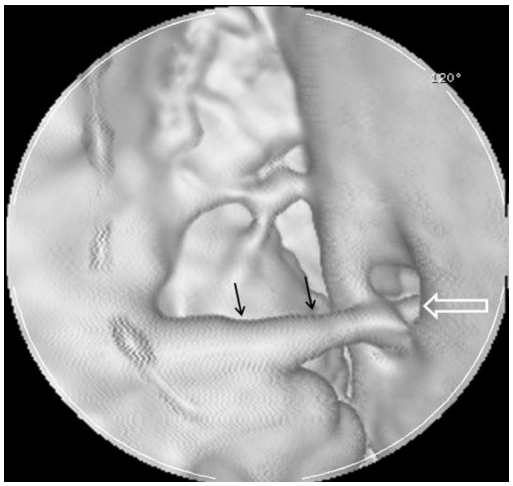
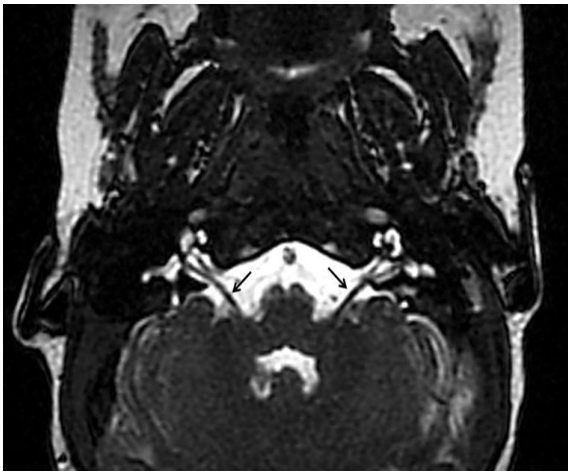
Figure 1. Sagittal MR endoscopic image. Left 8th nerve (black arrow). 7th nerve cannot be visualised. White arrow: internal acoustic canal.



cardiology examination revealed 1/6 cardiac murmur. Other neurological and systemic examinations were unrevealing. In a laboratory work-up, routine blood and urine tests were within normal limits. Her blood was screened for toxoplasma, rubella, cytomegalovirus and herpes virus antibodies; all were within normal limits.

Echocardiography was performed because of the cardiac murmur, and patent foramen ovale and a narrow ductus opening were detected. Cranial magnetic resonance imaging was performed because of the dysmorphic features and revealed micrognathia and volume loss at the tongue. Bilateral facial nerves could not be visualised by a 3D FIESTA scan, suggesting bilateral facial nerve agenesis (Figures 1, 2). The diagnosis of Moebius syndrome was made due to the left facial and bilateral abducens palsies and accompanying dysmorphic features.

Figure 2. Axial plan 3D FIESTA sequence. Eighth nerves were visualised, which run down the brain stem to the internal acoustic canal. Seventh nerves could not be visualised, which should be antero superior to the 8th nerves.



DISCUSSION

Bilateral facia was first described by Von Graefe et al. in 1880. In 1888, after classifying congenital cranial nerve palsies, Moebius differentiated 6th and 7th nerve weakness; since then, the condition has been named Moebius syndrome (1). The severity of Moebius syndrome varies; it can involve multiple cranial nerves and facial and abducens nerves are affected uni- or bilaterally (7). Although sporadic cases are predominant, autosomal dominant, autosomal recessive or X-linked recessive inheritance have also been reported. In genetic studies, abnormalities involving chromosomes 1 and 13 have been found (8). Abnormal development of cranial nerves VI and VII is the main cause of Moebius syndrome. Indeed, Moebius syndrome is usually first detected at birth because of facial paralysis and the absence of lateral movement of eyes. In addition to the facial nerves, cranial nerves V, VI, VIII, and XII are sometimes affected. The incidence of Moebius syndrome is estimated to be 2-20 per million live births (4, 9). Although pathogenesis is still controversial, the most widely accepted hypothesis is brain stem ischemia. Moebius syndrome should be differentiated from other causes of facial paralysis. The most common cause of congenital facial palsies is known to be perinatal trauma (10). The aetiology of Moebius syndrome can be divided to four major categories according to neuropathological findings: (i) hypoplasia or atrophy of cranial nerve nuclei, (ii) necrosis or destruction of cranial nerve nuclei, (iii) primary involvement of the peripheral nerve, and (iv) primary myopathic defects (8,10). After the facial and abducens nerves, the glossopharyngeal and hypoglossal nerves are most commonly involved (8). The MR finding of tongue atrophy suggested hypoglossal nerve involvement in our patient. Neurological, psychiatric, and endocrinological abnormalities, such as motor and mental retardation, epilepsy, autism, isolated ACTH deficiency, and diabetes insipidus have been shown to accompany Moebius syndrome. Furthermore, extremity abnormalities, absence of the pectoralis muscle, peripheral neuropathy, and myopathies may also be associated (10). In addition to these conditions, Kallman syndrome (hypogonadotrophic hypogonadism), Poland syndrome (hypoplasia or absence of the pectoralis major muscles), congenital bilateral paralysis of the vocal cords, brachial malformations, orofacial abnormalities and hearing defects are frequent. There are no specific findings for Moebius syndrome in laboratory tests. Hypoplasia or calcification of the cranial nerve nuclei and sometimes cerebral malformations may be demonstrated with computed tomography (CT) or MRI (4).

In conclusion, this case is presented to highlight Moebius syndrome in the differential diagnosis of cases presenting with congenital facial weakness.

Conflict of Interests

There is no conflict of interests.

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