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ORIGINAL ARTICLE

Eye and systemic manifestations of Mobius syndrome☆,☆☆

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KEYWORDS
Mobius syndrome;
Facial palsy;
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Epicanthus;
Entropion

Abstract
Introduction: Mobius syndrome is characterised by damage in the nucleus of the sixth and seventh cranial nerves, with subsequent facial palsy and abduction limitation of the eyes. The aim of this article is to describe the ophthalmological findings of the Mobius syndrome in Mexican children.
Patients and methods: A cross-sectional, retrospective, observational and descriptive study was conducted. A review was made of the clinical charts of patients with Mobius syndrome who were seen in the National Institute of Pediatrics in Mexico, between the years 2000 and 2010.
Results: A total of 64 charts were reviewed. The most important findings were eye abduction limitation (100%), facial palsy (100%), esotropia (54%), epicanthus (51.5%), entropion (22%), and history of use of abortion inducers in the mother in the first trimester of pregnancy (28%). We also found exotropia and hypertropia in some cases.
Conclusions: Mobius syndrome has a wide spectrum of ophthalmological manifestations that are important to detect early in order to improve function and aesthetics.

PALABRAS CLAVE
Síndrome de Möebius;
Endotropia;
Parálisis facial;
Agentes abortivos;

Manifestaciones oculares y sistémicas del síndrome de Möebius

Resumen
Introducción: El síndrome de Möebius es una enfermedad caracterizada por lesión en los núcleos del sexto y séptimo nervios craneales, produciendo parálisis facial y limitación a la abducción

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Epicanto;
Entropión

principalmente. El objetivo es describir los hallazgos oftalmológicos del síndrome de Möebius en niños mexicanos.
Pacientes y métodos: Estudio retrospectivo, transversal, observacional y descriptivo. Se revisaron expedientes clínicos de los pacientes con síndrome de Möebius del Instituto Nacional de Pediatría de México atendidos entre los años 2000 y 2010.
Resultados: Se revisaron 64 expedientes clínicos. Los hallazgos más importantes fueron limitación a la abducción (100%), parálisis facial (100%) endotropia (54%), epicanto (51,5%), entropión (22%) y antecedente de uso de abortivos en la madre durante el primer trimestre de embarazo (28%). Sin embargo, también se presentaron hallazgos atípicos como exotropia e hipertropia.
Conclusiones: El síndrome de Möebius tiene una amplia gama de manifestaciones oftalmológicas que se deben detectar temprano para mejorar su función y estética.

Introduction

Limitation of eye movements associated with facial palsy was first described by von Graefe in 1880.[1] However, it was Möbius who noticed the association of congenital facial palsy with other malformations between 1888 and 1892.[2–7] Möbius syndrome is a disease characterised by lesions in the facial (VII) and abducen (VI) nerves, with possible involvement of other cranial nerves and traversing long tracts in the brainstem. Its main manifestations are facial palsy, impairment of ocular abduction, and horizontal vergence abnormalities. Since it is a rare congenital disorder, there are few statistical data regarding its incidence.

Its cause remains unknown, and most cases are sporadic. Towfighi et al.[8] proposed a classification system based on neuropathologic findings, but it has not been fully accepted. Two possible aetiologies are hypothesised: a genetic defect leading to abnormal development of the brainstem, and an ischaemic origin possibly caused by intrauterine or environmental toxicity that restricts blood flow to the brainstem during early embryonic development.[9–13]

Cytogenetic studies have found 2 possible loci for the syndrome, 1p22 and 13q12.2-q13.[14–21] Healthy parents of a child with Möbius syndrome have a low probability of having another child with the disease. However, Möbius syndrome may be familial in some patients.[19]

Maldevelopment has been associated with the use of misoprostol in the early stages of pregnancy.[7,9,14,21–29] It has also been associated with exposure to infection, alcohol, cocaine, and thalidomide.[30]

Verzijl[9,11] and Roig Quilis[31] propose that the disease be redefined as a syndrome of rhomboencephalic maldevelopment or brainstem dysgenesis with involvement of motor nuclei and axons, as well as traversing long tracts.

In addition to facial and eye involvement, the syndrome can affect oropharyngeal, craniofacial, musculoskeletal, and neurologic structures.[5,11,13,21–40]

Five patterns of strabismus have been described:[38] bilateral restriction of abduction, bilateral restriction of abduction and adduction, bilateral restriction of abduction and adduction with globe retraction in adduction (as seen in Duane syndrome), asymmetrical or unilateral alterations in ocular motility, and restricted horizontal and vertical movement.

Carta et al.[39] propose 3 patterns of strabismus. Pattern A consists of orthotopia due to damage to cells of the VI cranial nerve nucleus, the axons of which constitute the medial longitudinal fasciculus and synapse on the medial rectus subnucleus, leading to palsy in the lateral rectus muscle and balancing opposing forces so that the gaze is in primary position. Pattern B consists of esotropia due to bilateral lesion of only the ventral portion of the VI cranial nerve nucleus, leading to overreaction of the remaining neurons in the nucleus connecting to the subnucleus of the oculomotor (III) nerve, which innervates the contralateral medial rectus. Pattern C involves exotropia associated to vertical misalignment caused by rostral nerve damage extending into the midbrain, in proximity to the vertical gaze centre.

The first Möbius Syndrome Scientific Conference was held in Bethesda, Maryland, in 2007, during which the minimum diagnostic criteria for the syndrome were established: congenital, nonprogressive unilateral or bilateral facial palsy, and limitation of abduction. Additional findings may include deficits in other cranial nerves, and motor, orofacial, musculoskeletal, social, and neurodevelopmental impairment. Some authors use the term Möbius-*like* syndrome to refer to cases that do not meet the minimum criteria, although the difference between the two syndromes is not well defined.[39]

Since Möbius syndrome is a rare entity, few studies in the world make a detailed description of its ophthalmologic manifestations. We only know of one similar study in Latin America, performed in Brazil,[40] while others are isolated case reports. The aim of our study is to offer a broader understanding of the ophthalmologic manifestations of this entity.

Patients and methods

We conducted a descriptive, observational, cross-sectional retrospective study.

We reviewed the medical histories of patients with Möbius syndrome receiving care at the Instituto Nacional de Pediatría (National Paediatric Institute), Mexico, between 2000 and 2010, who met the minimum diagnostic criteria established at the First Möbius Syndrome Scientific Conference: congenital, nonprogressive unilateral or bilateral facial palsy, and limitation of abduction.