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Charcot-Marie-Tooth disease and bilateral abducens nerve palsy

Charcot-Marie-Tooth hastalığı ve bilateral abdusens sinir felci

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Abstract

Charcot-Marie-Tooth (CMT) disease is the most common hereditary degenerative disorder of the peripheral nervous system. It is generally inherited in an autosomal dominant pattern. Clinical diagnosis is based on family history and characteristic findings on physical examination. Cranial nerve involvement is rare. In this paper, it is aimed to report bilateral abducens nerve palsy involvement in CMT disease.

Keywords: Charcot-Marie-Tooth disease, hereditary neuropathy.

Öz

Charcot-Marie-Tooth (CMT) hastalığı, periferik nervöz sistemin en sık görülen dejeneratif bir hastalığıdır. Genellikle otozomal dominant olarak kalıtılır. Tanı, aile öyküsü ve fizik muayenedeki karakteristik bulgulara dayanır. Kraniyal sinir tutulumu nadir görülür. Bu yazıda, bilateral altıncı sinir tutulumu olan CMT hastalığının sunulması amaçlandı.

Anahtar Sözcükler: *Charcot-Marie-Tooth hastalığı, herediter nöropati.*

Introduction

Charcot-Marie-Tooth disease (CMT), known as hereditary motor and sensory neuropathy (HMSN) or peroneal muscular atrophy, comprises a group of disorders that affect peripheral nerves. The peripheral nerves lie outside the brain and spinal cord and supply the muscles and sensory organs in the limbs. Disorders that affect the peripheral nerves are called peripheral neuropathies, first described in 1886 (1).The main signs and symptoms of Charcot-Marie-Tooth disease are muscle weakness and decreased muscle size. You may also notice decreased sensation in affected areas. Foot deformities such as hammer toes and high arches are common in Charcot-Marie-Tooth disease. Cranial nerve involvement has been included infrequently in descriptions of CMT disease. In this study we describe CMT disease with clinical sixth cranial nerve involvement.

Case Report

Twenty five year-old man was referred because of a five months history of diplopia. Diplopia present in the primer position and to the left gaze. Patient had a year ago the same complaint.

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The patient's had a CMT disease of diagnosed with neurological.

The ocular examination revealed a best corrected visual acuity was 10/10 in both eyes. Intraocular pressure, biomicroscopy, fundoscopy were normal. The ocular motility examination evaluated in nine diagnostic gaze position; abduction, adduction, elevation, depression normal in the right eye and abduction (-3) restricted were in the left eye. 30 PD (prism diopter) esotropia was found in prism cover test near and distance. He had abnormal head posture of turn to face on left. Bilateral abducens palsy (worse in his left eye) found in Hess screen test (Figure-1A).

In this case report for the bilateral abducens nerve palsy, patient was received botulinum type A toxin (2.5 U) injection for his paralytic esotropia. The injection was performed under local anesthesia in the patient. Botulinum type A toxin was injection by grasping the medial rectus muscle both eyes belly through the conjunctiva and then injecting the muscle. After the first injection the symptoms of the patient was decreased but full recovery was not achieved. Then second and third injections were performed. After the last injection the symptoms was better and there was a diplopia in only left gaze on minimal (not in up-left and down-left gaze). After we showed the positive results from botox injections (Figure-1B). Surgery was recommended to the patient (resection of left lateral rectus) for permanent

result but the patient has not accepted surgical treatment yet.

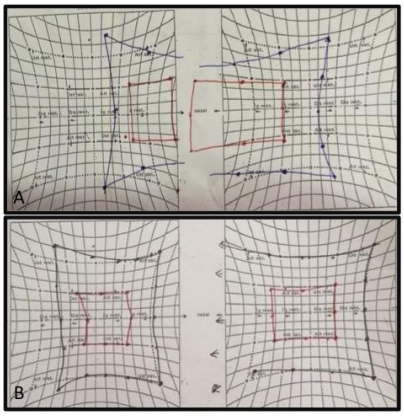

Figure-1. A. Before botulinum toxin A injection.
B. After botulinum toxin A injection.

Written informed consent was obtained from the patient for publishing the individual medical records.

Discussion

CMT diseases are the most common degenerative disorders of the peripheral nervous system. In these disease in generally inherited autosomal dominant pattern but also is inherited in recessive or an X-linked pattern. The disease onset usually occurs in the first two decades of life and subsequently shows a slow progression over decades. Interestingly, central nervous system (CNS) involvement has been also observed in X-linked form of the disease (2).

Symptoms and signs indicative of CMT include: pes cavus (or pes planus, often later progressing to cavus deformity); hammer toes; difficulty in running; twisting of the ankle and tripping; difficulty in walking; foot drop; steppage gait; wasting, weakness, and sensory loss of distal segments of lower and then upper limbs; difficulties in hand manipulation; and reduced or absent deep-tendon reflexes (1,3). Other common symptoms and signs are hand tremors, muscle cramps (particularly of the foot and leg), cold feet, foot callosities, and acrocyanosis. Positive sensory symptoms such as paraesthesias are rare, but pain is common, particularly in the feet, lower limbs, and lumbar spine. Onset can sometimes occur so early that it causes hypotonia (floppy baby syndrome), delayed motor development, and toe walking, whereas in other cases, the onset can occur late in life.

On the basis of nerve-conduction studies and nerve pathology, CMT is subdivided into two main groups: 1) a demyelinating form (CMT1 if autosomal dominant, if autosomal recessive), characterized by slowed nerve-conduction velocities (38 m/s) and prominent myelin abnormalities at nerve biopsy; and 2) an axonal form (CMT2), with preserved or only mildly slowed nerve-conduction velocities (>38 m/s) and pathological evidence of chronic axonal degeneration and regeneration (3,4). CMT3 (HMSN III) is the term sometimes used to indicate Déjèrine-Sottas neuropathy, which was once used to describe severe early-onset hereditary neuropathy with motor delay, very low nerve-conduction velocities, increased concentrations of proteins in the cerebrospinal fluid, nerve hypertrophy, and severe dysmyelination at nerve biopsy; today, Déjèrine-Sottas neuropathy is considered the most severe form of demyelinating CMT (1,3).In the literature, concerning the involvement of cranial nerves in this disorder are rare, although clinical experience has demonstrated an association between CMT and trigeminal neuralgia of uncertain etiology and most often associated with normal nerve size (5). Aho et al. (4) found extensive cranial nerve involvement on neuroimaging in a 64-year-old patient with, including cranial nerves III, V and VII. and Glocker et al. (6) reported a patient with facial nerve involvement. Our case report suggest bilateral abducens nerve palsy involvement in CMT disease.

Koros and coauthors (2) represented the patient a history of CMT1A who developed CNS involvement mimicking multiple sclerosis. The patient was initially referred to their because of a 3-day history of diplopia, present in horizontal gaze positions. Examination in this patient of the cranial nerves showed a right intranuclear ophthalmoplegia with dissociative nystagmus along with right abducens nerve paresis. Our patient was admitted with diplopia history of five months and there were other signs of CMT disease. At present, there is no drug therapy for Charcot–Marie–Tooth disease, and rehabilitation therapy and surgical procedures for skeletal deformities are the only available treatments, although best practice has not been defined. Our approach in our case recurrent botox injection was performed for abducens nerve palsy.

A case report by Shizuka et al. (7) demonstrated thickening of the cisternal trigeminal nerves bilaterally on MR images in a patient with CMT. Interestingly, this patient had no clinical symptoms referable to these nerves. Our patient had bilateral abducens palsy and we were reported Hess Screen test. We found no other reports concerning sixth cranial nerves in this disorder in the literature. Duarte et al (8) in 1999 demonstrated significant enlargement of the orbital segments of the oculomotor nerves in a case report, as well as enlargement of all three divisions of the trigeminal nerves bilaterally on MR images in a patient with CIDP. Aho et al. (4) presented a case, weakness had progressed over time and was associated with significant muscle wasting of both lower extremities and hands. The patient also had bilateral deafness and severe right facial pain diagnosed as trigeminal neuralgia. The trigeminal neuralgia had been treated in the past with radio-frequency ablation and alcohol injection, but the patient had recurrent and severe

symptoms (4). And more rarely, familial trigeminal neuralgia has been described in the setting of hereditary peripheral neuropathy, especially Charcot-Marie-Tooth disease. Coffey et al., describe patients from two different families with Charcot-Marie-Tooth disease and medically intractable trigeminal neuralgia suggests.

In summary, CMT is a predominantly distal sensory and motor neuropathy and the clinical presentation of cranial nerve involvement and asymmetry in a small number of CMT patients. This finding is particularly important for ophthalmologist.

Conflict of Interests

There is no conflict of interests regarding the publication of this paper.

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