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Electrodiagnostic Features of a Molecular Genetic Proven Case of HNPP

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Abstract

Hereditary Neuropathy with Pressure Palsies (HNPP) is an inherited autosomal dominated demyelinating disease that is associated with point mutation at 17p11.2-12 of the gene Peripheral Myelin Protein-22 (PMP22). It usually presents with painless focal sensory and motor dysfunction secondary to mild compression, stretching or focal trauma to nerves. Electro diagnostic evidence is important in diagnosing HNPP.

Here we report a case of a 23-old-man with a 3-month history of numbness in the volar side of the left 3-5 digits. The patient had a prior history of progressive weakness and numbness in the right arm with spontaneous resolution as well as a positive family history. Similar symptomatology was noted in his mother. Electromyography (EMG) and needle electromyotrography demonstrated multi-mononeuropathies at bilateral wrists and elbows. Genetic test was performed following the positive EMG findings and confirmed heterozygous deletion of PMP22 gene on chromosome 17p11.2. Thus, he was diagnosed with HNPP. Our report indicates the importance of electro diagnostic studies in diagnosis HNPP.

Keywords: HNPP; PMP 22 deletion; Molecular genetics and HNPP; Electro diagnostic features of HNPP

Introduction

Hereditary neuropathy with liability to pressure palsies (HNPP) is a hereditary disease that is associated with point mutation at 17p11.2-12 of PMP22 gene. Clinically, it presents as recurrent transient focal sensory or motor mononeuropathy associated with mild compression, stretching or focal trauma to nerves. The patients may or may not have a family history consistent with autosomal dominant inheritance. The first attack usually occurs in the second or third decade [1]. In decreasing order of frequency, the most common sites of involvement are peroneal nerve at the fibular head, ulnar nerve at the elbow, median nerve at the wrist, and brachial plexus and radial nerve.

Case Report

A 23 year-old man without significant medical history visited our hospital for evaluation of a 3-month history of numbness in his left hand over the volar aspect of digits 3 to 5. He reported a prior episode of numbness and weakness in the right arm which occurred after he extended both his arms upward during stretching. The weakness was progressive and eventually leads to decreased bulk of the tricep muscle on the right side, with spontaneous resolution. The patient reported similar symptoms in his mother.

Electromyography (EMG) of bilateral upper extremities revealed bilateral sensor motor median mononeuropathies at the wrist (consistent with Carpal tunnel syndrome); bilateral ulnar mononeuropathies at the elbows with conduction block at the left elbow; left radial mononeuropathy distal to the left elbow (conduction block).

EMG of the lower extremities demonstrated low compound muscle action potentials amplitudes in bilateral lower extremities, prolonged distal motor latencies in peroneal nerves, and slowed conduction velocities in peroneal nerves, as well as the right tibial nerve. Proximal conductions, as measured by F response latencies, were also prolonged. The needle electromyography of lower extremities was normal.

Subsequently, we performed a PMP22 DNA sequencing test which revealed heterozygous deletion of PMP22 gene on chromosome 17p11.2. The patient was diagnosed with HNPP, based on the findings of multiple mononeuropathies at the entrapment sites, positive family history and positive genetic test.

Discussion

The diagnosis of HNPP requires detailed history, electro diagnostic evidence and genetic confirmation. The characteristic electro diagnostic features in HNPP are bilateral slowing of sensory and motor nerve conduction at the carpal tunnel with at least one additional abnormal finding for motor conduction in one peroneal nerve [2]. It is necessary to evaluate sensory conduction in the sural nerve and motor conduction in at least two nerves across the usual entrapment sites, especially the ulnar nerve at the elbow [3]. Prolonged distal motor latencies may be found in the median and peroneal nerves but not in ulnar or tibial nerves [4]. Nerve Conduction Velocity (NCV) can be delayed at the site of compression. General motor NCVs are usually normal (>40 m/s); a few individuals show electrical evidence of a mild diffuse polyneuropathy.

HNPP is an inherited autosomal dominant demyelinating disease.

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It causes painless focal sensory and motor dysfunction.

Compression, stretching or focal trauma to nerves are the underlying mechanisms responsible for the symptoms. The disease is associated with mutations in the peripheral myelin protein 22 gene.

In our patient, the nerve conduction studies of the upper extremities showed bilateral sensorimotor median mononeuropathy at the wrists, bilateral ulnar mononeuropathy at the elbows and left radial mononeuropathy with conduction block. Lower extremity testing showed sensorimotor demyelinating polyneuropathy. The patient's genetic testing confirmed he was positive for HNPP.

Recommendation was to avoid compressing his elbows and to avoid repetitive movement, flexion at the elbows. It was also recommended that he use braces for his wrist.

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