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Case Report

Electrodiagnostic Features of a Molecular Genetic Proven Case of HNPP: A Case Report

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Abstract

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. 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.

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

Case Presentation

Here we 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 [1]. The weakness was progressive and eventually lead to decreased bulk of the tricep muscle on the right side, with spontaneous resolution. The patient reported similar symptoms in his mother.

Electromyography (EMG) bilateral upper extremities revealed bilateral sensorimotor median mono neuropathies at the wrist (consistent with Carpal tunnel syndrome); bilateral ulnar mono neuropathies at the elbows with conduction block at the left elbow; left radial mononeuropathy distal to the left elbow (conduction block). EMG of lower extremities demonstrated low compound muscle action potentials 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 were normal.

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

Discussion

The diagnosis of HNPP requires detailed history, electrodiagnostic evidence and genetic confirmation. The characteristic electrodiagnostic 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 have electrical evidence of a mild diffuse polyneuropathy.

HNPP is an inherited autosomal dominated demyelinating disease. It causes painless focal sensory and motor dysfunction. The mechanism causing the symptoms are contributed to compression, stretching or focal trauma to nerves. The disease is associated with mutations in the PMP 22 gene. In our report, the patient was found to have bilateral sensorimotor median mononeuropathy at the wrists, bilateral ulnar mononeuropathy at the elbows and left radial mononeuropathy with conduction block. He also had sensorimotor demyelinating polyneuropathy in bilateral lower extremities. These findings along with his clinical presentation prompted a genetic test for HNPP which confirmed the diagnosis. The patient was recommended to avoid compressing his elbows, repetitive movement and flexion at the elbows. He was also advised to use braces at night for his wrists.

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