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Case Report

An Uncommon Disease with a Common Presentation: Severe Back pain and Diffuse Neuropathy in Two Cases of Hereditary Neuropathy with Liability to Pressure Palsy

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Abstract

Introduction: Hereditary Neuropathy with liability to Pressure Palsy (HNPP) is an autosomal dominant genetic disorder characterized by multiple episodes of focal weakness and sensory loss caused by compression or trauma. HNPP is rare disorder and often exhibits cranial nerve involvement, sensorineural deafness and scoliosis. Individuals diagnosed with this condition typically exhibit a primary deficit of a single nerve palsy that can result in numbness, paresthesias, or weakness.

Case Presentation: Two unrelated individuals with HNPP were evaluated with clinical, laboratory, electrophysiological and genetic testing. Case 1 was a 32-year-old man who presented with an episode of transient right arm weakness and severe back pain. Case 2 was a 46-year-old woman who presented with severe back pain and diffuse paresthesias, which was diagnosed as HNPP.

Discussion: These cases present common symptoms that are uncommon for the typical HNPP patient. This report serves to contribute new information to the body of evidence describing HNPP.

Conclusion: Although HNPP typically presents with transient compressive mononeuropathies, this study suggests that HNPP can present with various broad clinical presentations, including severe back pain. Neurologists need to broaden their spectrum of clinical presentations to help diagnose these rare conditions.

Keywords: Pressure Palsy; Back Pain; Electromyograph; Neuropathy

Introduction

Hereditary Neuropathy with liability to Pressure Palsy (HNPP) is an autosomal dominant episodic demyelinating neuropathy which typically presents with mononeuropathies after compression. A common presentation is an acute focal sensorimotor mononeuropathy, often the peroneal or ulnar nerves [1]. HNPP can present with a variety of neuropathies, sensorimotor neuropathies and even progressive muscular atrophy [2-4]. The prevalence of HNPP is unknown. It is estimated to be between 7 to 16 in 100,000 individuals. It is likely that the current measures of prevalence are an underestimate due to underdiagnosis. This is likely to increase as testing becomes more widespread. Electrodiagnostic studies typically demonstrate prolonged distal motor latencies, normal to mildly reduced sensory nerve conduction velocities and segmental slowing at typical sites of compression [3,5-7]. HNPP commonly involves a deletion of chromosome 17p11.2 of the Peripheral Myelin Protein 22 (PMP22) gene. Given its variable presentation, it is possible that HNPP can be under-recognized. The following case study demonstrates how two unrelated subjects presented with a common condition such as severe back pain and were found to have a rare condition such as HNPP after further investigation.

Methods

All subjects received care at the AdventHealth Orlando Neuroscience Institute. Both unrelated probands had neurological records evaluated. Electrodiagnostic, radiographic and genetic testing were performed. Genetic testing was completed through the hereditary neuropathy panel offered by Invitae co (San Francisco, CA). Two pathogenic deletions of the PMP22 gene were identified (https://www.invitae.com/en/).

Clinical Presentation

Case 1

This individual is a 32-year-old gentleman who presented with numbness in his hands, back pain and an episode of transient biceps weakness. The episodes of biceps weakness occurred after rotating his arm while stretching. The back pain is described as unbearable pain which radiates and is accompanied by stiffness. These symptoms typically worsen with certain positions and have remained consistent since he was a child. As a child, he had jolting pain and stiffness in his lower back. His symptoms were worse with sitting for prolonged periods of time including driving. Stretching helped his symptoms. The patient denied numbness, saddle anesthesia or bowel/bladder dysfunction. The severity of the back pain was reported to significantly limit mobility of his back on multiple occurrences. He reported no other episodes of compressive neuropathies.

The patient was evaluated at an outpatient neurology clinic. An Electromyograph (EMG) was ordered to assess neuromuscular function. This study showed a left musculocutaneous neuropathy, bilateral median neuropathy and a symmetric sensorimotor polyneuropathy.

Additionally, an MRI lumbar spine was ordered. This study revealed L4-L5 moderate to severe bilateral neural foraminal narrowing; L5-S1, disc bulge with severe left and moderate right neural foraminal narrowing. Genetic testing was ordered. A hereditary neuropathy panel was performed and results were significant for a PMP22 deletion associated with hereditary neuropathy with liability to pressure palsies. The patient was treated with epidural steroid injections, which improved his symptoms.

Case 2

This individual is a 46-year-old woman with a past medical history congestive heart failure and Takayasu's arteritis who presented with many years of burning sensations in her head, feet, hands, knees and groin area. She reported tingling in her fingers, which was positional and sporadic when laying down. She had severe back pain and numbness in her legs that worsen with certain positions especially sitting in a chair.

The patient was initially evaluated at an outpatient neurology clinic. There were no clinical features of pes cavus, sensory loss, or weakness on exam. There was no significant family history. The patient was initially treated with routine medications including pregabalin, neurontin and lidocaine gel. These medications provided minimal relief. Electrodiagnostic testing was ordered. This testing showed evidence of a sensorimotor polyneuropathy with slowing at common compression sites. MRI lumbar spine was also ordered. This revealed minimal degenerative changes. Genetic testing was ordered. A hereditary neuropathy panel was performed and results were significant for a PMP22 deletion associated with hereditary neuropathy with liability to pressure palsies.

Discussion

HNPP is a hereditary neuropathy the involves the formulation of tomaculae, focal, sausage-like enlargement of the nerve. It has been known to be a relatively milder neuropathy with multiple compression at common sites. is an autosomal dominant peripheral neuropathy causing a recurrent mononeuropathy induced by any compression that results from deletion of the Peripheral Myelin Protein-22 (PMP22) gene encoded by chromosome 17p11.2- 12.1. PMP22 is a myelin protein that regulates proliferation and apoptosis of Schwann cells that form compact myelin. HNPP is thought to be under-recognized given the phenotypic variability of the disease and the mild nature of symptoms. Presentations included multiple different forms of neuropathy with motor symptoms presenting in a longer lasting fashion than sensory symptoms. Usually, episodes of weakness are painless and back pain hasn't been reported in previous case studies [5]. Another study, however, published in 2017 suggested that pain is a common feature of HNPP reported by 74% with 72% having pain in the lower back as seen in our

subjects. This pain led to poorer health-related quality of life, higher levels of disrupted employment and more frequent physician visits [8]. In our two subjects, it seems that the mechanical position of the back (prolonged sitting/certain positions) triggered the pain and thus this could be triggering a compressive response involving the sensory nerves. Despite the well-known evidence of neuropathies, very little is discussed in terms of further aspects of the back pain. Case 1 had severe back pain that resolved with steroid injections. Of note, back surgery is not indicated in these patients, as it can cause or worsen focal neuropathies. HNPP has been reported to have associations with ALS. Although three cases of HNPP with Amyotrophic Lateral Sclerosis (ALS) have been reported, little is known about the role of PMP22 in the Central Nervous System (CNS). Additionally, there has been a reported case of HNPP with bulbar palsy and PMA in all extremities, associated with a deletion in the PMP22 gene [2].

Conclusion

Although HNPP typically presents with focal neuropathies, it can also present with a variety of neurological complaints. In these individuals, it presented with a common complaints of severe unrelenting back pain. Research in the future should focus on developing a greater understanding of the underlying pathophysiology of HNPP and how those results in varying presentations. These case reports contribute to the larger body of evidence regarding how the disease can present. This is suggestive of a more complex clinical picture than what has been previously suggested.

Conflict of Interest

The authors have no conflict of interest to declare.

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