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Foot drop as presentation of HNPP in a teenage boy

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Hereditary neuropathy with liability to pressure palsies (HNPP) is a recurrent, episodic demyelinating neuropathy, most commonly caused by a 17p11.2 chromosomal deletion encompassing the PMP22 gene. The peroneal and ulnar nerves are most frequently affected nerves.

This presentation describes case of 15 years old boy presenting with foot numbness and difficulty to lift the front part of the foot, with no trauma history and with history of prolonged sitting with his legs crossed while playing video games. The family history is negative in respect to the disorder of the patient. Lumbosacral spine MRI was done and showed discus herniation but at this time, the protrusion wasn't significant enough to put pressure on the nearby nerves.

Subsequently, family history investigation revealed that the patient's mother, grandfather and aunt reported similar symptoms earlier in life. Further clinical and electrophysiological investigations revealed neuropathic dysfunction and demyelinating injuries in proband boy. Hereditary neuropathy disorder was highly suggestive. He was treated with pulse corticosteroid therapy and physical therapy, and his condition was improved.

The definitive diagnosis is achieved by genetic analysis.

We should consider HNPP in order to make an accurate diagnose and to provide a good quality of life.

Biography

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