Hereditary brachial plexus neuropathy

Hereditary brachial plexus neuropathy (HBPN) is a rare genetic disorder that affects the brachial plexus, a network of nerves that control movement and sensation in the arm and hand. This condition is typically inherited in an autosomal dominant manner, meaning that a person only needs to inherit one copy of the mutated gene from either parent to develop the disorder.

The brachial plexus is a complex network of nerves that originates from the spinal cord in the neck and controls the muscles and sensation in the shoulder, arm, and hand. In individuals with hereditary brachial plexus neuropathy, there is a genetic mutation that leads to abnormalities in the development or functioning of the brachial plexus.

Common features of hereditary brachial plexus neuropathy may include weakness or paralysis in the muscles of the shoulder, arm, or hand, as well as sensory abnormalities. Symptoms often appear in childhood or adolescence and may progress over time. The severity of the condition can vary among affected individuals.

Diagnosis of hereditary brachial plexus neuropathy involves a thorough clinical evaluation, genetic testing, and imaging studies such as magnetic resonance imaging (MRI) to assess the structure of the brachial plexus.

Management of hereditary brachial plexus neuropathy typically involves supportive care to address symptoms and maximize functioning. Physical therapy may be beneficial to help maintain mobility and strengthen muscles. In some cases, surgical interventions may be considered to address specific issues related to nerve compression or other abnormalities.

It's important for individuals with suspected hereditary brachial plexus neuropathy to consult with healthcare professionals, including genetic counselors and neurologists, to receive an accurate diagnosis and appropriate management. Additionally, ongoing research is essential to better understand the underlying genetic mechanisms and to develop potential targeted therapies for this rare condition.

Parsonage-Turner syndrome and hereditary brachial plexus neuropathy (HBPN) present with indistinguishable attacks of rapid-onset severe shoulder and arm pain, disabling weakness, and early muscle atrophy. Their combined incidence ranges from 3 to 100 in 100,000 persons per year. Dominant mutations of SEPT9 are the only known mutations responsible for HBPN. Parsonage and Turner termed the disorder “brachial neuralgic amyotrophy,” highlighting neuropathic pain and muscle atrophy. Modern electrodiagnostic and imaging testing assists the diagnosis in distinction from mimicking disorders. Shoulder and upper limb nerves outside the brachial plexus are commonly affected including the phrenic nerve where diaphragm ultrasound improves diagnosis. Magnetic resonance imaging can show multifocal T2 nerve and muscle hyperintensities with nerve hourglass swellings and constrictions identifiable also by ultrasound. An inflammatory immune component is suggested by nerve biopsies and associated infectious, immunization, trauma, surgery, and childbirth triggers. High-dose pulsed steroids assist initial pain control; however, weakness and subsequent pain are not clearly responsive to steroids and instead benefit from time, physical therapy, and non-narcotic pain medications. Recurrent attacks in HBPN are common and prophylactic steroids or intravenous immunoglobulin may reduce surgical- or childbirth-induced attacks. Rehabilitation focusing on restoring functional scapular mechanics, energy conservation, contracture prevention,

and pain management are critical. Lifetime residual pain and weakness are rare with most making dramatic functional recovery. Tendon transfers can be used when recovery does not occur after 18 months. Early neurolysis and nerve grafts are controversial [1)]

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Meiling JB, Boon AJ, Niu Z, Howe BM, Hoskote SS, Spinner RJ, Klein CJ. Parsonage-Turner Syndrome and Hereditary Brachial Plexus Neuropathy. Mayo Clin Proc. 2024 Jan;99(1):124-140. doi: 10.1016/j.mayocp.2023.06.011. PMID: 38176820.

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