SEPT9

SEPT9 (Septin 9) is a gene that encodes a protein involved in the formation of septin filaments. Septins are a family of GTP-binding proteins that play important roles in various cellular processes, including cytokinesis, cell division, and the organization of cell shape.

One notable aspect of the SEPT9 gene is its association with certain cancers, particularly colorectal cancer. Aberrant DNA methylation of the SEPT9 gene has been observed in colorectal cancer cells, leading to the development of a diagnostic test known as the SEPT9 DNA methylation test. This test detects specific methylation changes in the SEPT9 gene, and it has been used as a non-invasive method for colorectal cancer screening.

The SEPT9 DNA methylation test is designed to detect abnormal methylation patterns in circulating cell-free DNA in the blood. It has been used as a blood-based screening tool for colorectal cancer, providing an alternative to traditional methods like colonoscopy. However, it's important to note that this test is not a definitive diagnostic tool, and positive results may still require further investigation with additional diagnostic methods.

Research on the role of SEPT9 in cancer and other cellular processes is ongoing, and the gene's functions in normal and pathological conditions continue to be explored.

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.