Intraneural perineurioma

Intraneural perineuriomas are benign peripheral nerve sheath tumors that cause progressive debilitating focal extremity weakness, that does not recur or metastasize.

Etiology

The etiology of perineuriomas is largely unknown.

Klein et al., utilized whole exome sequencing, copy number algorithm evaluation and high-resolution whole genome microarray to investigate for a genetic causal link of intraneural perineuriomas. Ten of 16 (60%) tumor cases had mutations in the WD40 domain of TRAF7, the same location for causal mutations of meningiomas. Two additional perineurioma cases had large chromosomal abnormalities in multiple chromosomes, including chromosome 22q. This study identifies a common cause for intraneural perineuriomas and an unexpected shared pathogenesis with intracranial meningiomas [1].

The proliferation of perineurial cells with unique immunohistochemistry and ultrastructural features, is distinct from other onion bulb Schwann cell-derived entities. Despite harboring molecular abnormalities of the long arm of chromosome 22, intraneural perineurioma has not been associated with neurofibromatosis.

Intraneural perineurioma may be confused with other “onion bulb” Schwann cell entities (localized hypertrophic neuropathy, reactive/demyelinating processes, or inherited polyneuropathies of Charcot-Marie-Tooth/Dejerine Sottas) due to similar clinical, radiologic, and histologic features. Perineurial and Schwann cells can only be differentiated by ultrastructure and immunohistochemsitry.

A systematic review was performed on definitive intraneural perineuriomas identified through Medline. Baylor College of Medicine-affiliated hospitals' anatomic pathology databases yielded 2 illustrative intraneural perineurioma cases.

Intraneural perineurioma inclusion criteria consisted of characteristic histology and confirmation of perineurial cell lineage by either immunohistochemistry (epithelial membrane antigen positive, S100 protein negative) and/or ultrastructural analysis (thin cytoplasmic processes with an incomplete basal lamina, poorly formed tight junctions, and pinocytotic vesicles).

Clinicopathologic data were extracted from all identified articles, with subsequent statistical analysis of the following parameters: age, sex, race, tumor location, tumor size, duration of symptoms prior to diagnosis, treatment modalities and outcomes measures, follow-up assessment for tumor recurrence and metastasis, clinical features (history of trauma, motor/sensory abnormalities, clinical/family history), and diagnostic workup (routine histology, immunohistochemistry, ultrastructural analysis, and molecular/cytogenetic characteristics) [2].

1)
Klein CJ, Wu Y, Jentoft ME, Mer G, Spinner RJ, Dyck PJ, Dyck PJ, Mauermann ML. Genomic analysis reveals frequent TRAF7 mutations in intraneural perineuriomas. Ann Neurol. 2016 Dec 26. doi: 10.1002/ana.24854. [Epub ahead of print] PubMed PMID: 28019650.
2)
Boyanton BL Jr, Jones JK, Shenaq SM, Hicks MJ, Bhattacharjee MB. Intraneural perineurioma: a

systematic review with illustrative cases. Arch Pathol Lab Med. 2007 Sep;131(9):1382-92. Review. PubMed PMID: 17824794.