Spinal neurofibroma

Spinal tumors are observed in about 40% of neurofibromatosis type 1 (NF1) patients and occur within two subgroups:

(1) NF1 patients carrying classical diagnostic criteria and only one or few spinal tumours

(2) patients with few NF1 stigmata but multiple bilateral spinal tumours, an entity called spinal neurofibromatosis [1)].

Spinal neurofibromas are benign peripheral nerve sheath tumors, usually of the localized subtype.

see also Bilateral spinal neurofibroma.

Except in cases of neurofibromatosis, it is very rare for tumors of different pathological types to exist concurrently at the same spinal level, with only 9 cases reported to date, in which spinal meningioma was found with spinal schwannoma in 7 cases and with spinal neurofibroma in 2 cases [2)].

Clinical

Spinal neurofibromas are often asymptomatic.

If symptoms are present, they usually include pain and/or radicular sensory changes due to the typical location along the dorsal sensory roots. Weakness is less common. As multiple lesions are common, patients may present with multiple radiculopathies. Myelopathy may occur if the lesion is large.

When located along spinal nerve roots, they are most frequently encountered along the cervical cord.

Diagnosis

Differential diagnosis

The main differential for spinal neurofibromas includes:

spinal schwannoma.

spinal meningioma.

Case series

Upadhyaya et al. studied 22 spinal neurofibromas derived from 14 unrelated Neurofibromatosis type 1 patients. Seven of these patients satisfied the diagnostic criteria of NF1 while the remaining seven had only few features of NF1. The latter group defined as FSNF harbored significantly higher number of missense or missense and splice-site germline mutations compared to the group with classical NF1. This is the first study to describe NF1 somatic mutations in spinal neurofibromas. Loss-of-heterozygosity (LOH) was identified in 8/22 of the spinal tumors, 75% of LOH observed was found to result from mitotic recombination, suggesting that this may represent a frequent mutational mechanisms in these benign tumors. No evidence for LOH of the TP53 gene was found in these tumors [3].

1)
Carman Kb, Yakut A, Anlar B, Ayter S. Spinal neurofibromatosis associated with classical neurofibromatosis type 1: genetic characterisation of an atypical case. BMJ Case Rep. 2013 Feb 14;2013. pii: bcr2012008468. doi: 10.1136/bcr-2012-008468. PubMed PMID: 23417386; PubMed Central PMCID: PMC3604550.
2)
Zhan Z, Yan X, Nie W, Ding Y, Xu W, Huang H. Neurofibroma and Meningioma within a Single Dumbbell-Shaped Tumor at the Same Cervical Level without Neurofibromatosis: a Case Report and Literature Review. World Neurosurg. 2019 Jun 26. pii: S1878-8750(19)31788-7. doi: 10.1016/j.wneu.2019.06.142. [Epub ahead of print] PubMed PMID: 31254713.
3)
Upadhyaya M, Spurlock G, Kluwe L, Chuzhanova N, Bennett E, Thomas N, Guha A, Mautner V. The spectrum of somatic and germline NF1 mutations in NF1 patients with spinal neurofibromas. Neurogenetics. 2009 Jul;10(3):251-63. doi: 10.1007/s10048-009-0178-0. Epub 2009 Feb 17. PubMed PMID: 19221814.