Neuromuscular choristoma

Desmoid-type fibromatosis (DTF) frequently arises in patients with neuromuscular choristoma (NMC). Maldonado et al. hypothesized that Neuromuscular choristoma-associated DTF occurs in soft tissues innervated by the NMC-affected nerve, and arises from CTNNB1-mutated (myo) fibroblasts within or directly adjacent to the NMC.

A retrospective review of patients treated at Mayo Clinic Rochester, was performed for patients with a biopsy-confirmed diagnosis of NMC-DTF. Clinical presentation, physical examination, electrodiagnostic findings and radiological features (MR and FDG PET/CT images for each NMC-DTF) and pathologic re-review of available materials were analyzed. A literature review was also performed.

Eight patients from this institution met the inclusion criteria. All patients presented with neuropathic symptoms and soft tissue or bone changes in the nerve territory innervated by the NMC. All MR images (N=8 cases) showed the characteristic features of NMC, and also showed direct contact between unifocal (N=5) or multifocal (N=3) DTF(s) and the NMC-affected nerve NMC. FDG PET/CT (N=2 cases) showed diffuse, increased FDG uptake along the entire affected nerve segment, contiguous with the FDG-avid DTF. In all cases, the DTFs arose in the soft tissues of the NMC-affected nerve's territory. No patient developed DTF at any other anatomic site.

These data demonstrate that Neuromuscular choristoma-Desmoid-type fibromatosis arises solely within the NMC-affected nerve territory, and has direct contact with the NMC itself. Based on all these findings and the multifocality of NMC in several cases, they recommend imaging and surveillance of the entire NMC-affected nerve (from the spine to distal extremity) to identify clinically-occult DTF in patients with NMC [1].

1)
Maldonado AA, Spinner RJ, Broski SM, Stone JJ, Howe BM, Carter JM. Neuromuscular choristoma-associated desmoid-type fibromatosis: Establishing a nerve territory concept. Acta Neurochir (Wien). 2020 Jan 2. doi: 10.1007/s00701-019-04178-8. [Epub ahead of print] PubMed PMID: 31897730.