Myxoid chondrosarcoma

A myxoid chondrosarcoma is a rare, intermediate grade sub type of chondrosarcoma (see chondrosarcoma grading). It is found in both bone and soft tissues (see extra skeletal chondrosarcoma).

Epidemiology

Typically affects patients in their 30s to 60s with a male predilection

They may account for as many as 12% of chondrosarcomas of bone.

Clinical presentation

Extra skeletal lesions tend to present with a palpable mass.

Pathology

They are the most common extra skeletal chondrosarcomas, typically occurring in the deep soft tissues of the extremities, with the thigh being most common.

Conventional intramedullary chondrosarcomas can often demonstrate some degree of myxoid degeneration, leading to difficulty in accurate diagnosis of this entity.

Extensive myxoid stroma accounts not only for its name, but also its high water content. Histologically as well as on imaging these tumours are difficult to distinguish from chordomas, and accounting for its alternative name of chordoid chondrosarcoma.

Radiographic features

Going along with its more aggressive clinical course, the lesion is predominantly lytic, with the high water content giving it high signal on T2 weighted images, thus mimicking chordomas on imaging also. Enhancement is usually present but mild.

Case series

Primary intracranial neoplasms with features of extraskeletal myxoid chondrosarcomas (EMC) are extremely rare and poorly characterized tumors with only ~12 cases described, the majority lacking molecular confirmation. There is an urgent need for the integration of molecular studies for correct subclassification of these tumors in order to predict clinical behavior, guide therapeutic decision-making, and provide novel targets for therapy. Clinical and pathologic data of 3 intracranial EMC-like

myxoid neoplasms were retrospectively reviewed. In 2/3 cases, immunohistochemistry showed loss of nuclear SWI/SNF-related matrix-associated actin-dependent regulator of chromatin subfamily B member 1 (SMARCB1; integrase interactor 1 [INI1]) staining accompanied by monosomy of chromosome 22q (fluorescence in situ hybridization [FISH]). These 2 cases had no evidence of any fusion products by next generation sequencing (NGS). The third case had intact SMARCB1 expression and showed instead a rearrangement of the EWSR1 gene detected by FISH, with an EWSR1-CREB1 gene fusion on NGS. None of the cases showed rearrangement of the NR4A3 gene, neither by FISH nor by NGS. This small case series highlights the molecular heterogeneity of intracranial neoplasms in the morphologic spectrum of EMC. Distinct molecular alterations found in tumors with morphologic features of EMC encompass SMARCB1(INI1) loss and EWSR1-CREB gene fusions. None of the cases showed rearrangements of NR4A3 genes, suggesting they are distinct from conventional EMC [1)].

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Velz J, Agaimy A, Frontzek K, Neidert MC, Bozinov O, Wagner U, Fritz C, Coras R, Hofer S, Bode-Lesniewska B, Rushing E. Molecular and Clinicopathologic Heterogeneity of Intracranial Tumors Mimicking Extraskeletal Myxoid Chondrosarcoma. J Neuropathol Exp Neurol. 2018 Jun 18. doi: 10.1093/jnen/nly050. [Epub ahead of print] PubMed PMID: 29924341.