Intramuscular Myxoma

136

Definition

Intramuscular myxoma is a benign neoplasm currently classified among soft tissue tumors of uncertain differentiation.

Epidemiology and Presentation

It occurs mainly in adult or elderly patients (more frequently females) as a painless intramuscular mass. The combination of intramuscular myxoma with bone fibrous dysplasia defines the Mazabraud's syndrome. Magnetic resonance imaging shows a poorly vascularized tumor (which can measure up to 20 cm in diameter) hyperintense on T2-weighted images.

Pathology

Macroscopically, the mass – which may show undefined margins – displays a gelatinous lobulated cut surface and may present fluid-filled cystic spaces.

Miscroscopically, intramuscular myxoma is composed of uniform spindled and stellated cells separated by abundant extracellular myxoid stroma composed of glycosaminoglycans (similar to low-grade myxofibrosarcoma, → see dedicated section). Mitoses, pleomorphism, and necrosis are not present even in the most cellular areas. Intramuscular myxoma with more cellularity is also called cellular myxoma. **Differential diagnosis** may be needed with the following: chondrosarcoma (bone or soft tissue tumor mimicking chordoma with rows of cuboidal cells separated by myxoid background; stains positive for S100 and vimentin, negative for cytokeratins); myxoid liposarcoma (mitotic figures, lipoblasts, positive for FUS-DDIT3 fusion gene); and myxoid leiomyosarcoma (invasive, highly myxomatous; see

S. Mocellin, *Soft Tissue Tumors*, https://doi.org/10.1007/978-3-030-58710-9_136

typical smooth muscle cells alternating with mesenchymal cells); intramuscular cellular myxoma may need to be differentiated from low-grade myxofibrosarcoma (no GNAS mutations).

Biomarkers

At immunohistochemistry, the cells stain variably positive for CD34 and desmin and negative for S100. From the genetics viewpoint, point mutations of the GNAS gene[1] are commonly observed (about 60% of all cases).

Prognosis

Intramuscular myxoma is a benign tumor, although cellular myxomas may recur.

Therapy

Surgical excision is the treatment of choice.

Suggested Readings

Allen (2000) Myxoma is not a single entity: a review of the concept of myxoma. Ann Diagn Pathol 4(2):99–123

Delaney (2009) GNAS1 mutations occur more commonly than previously thought in intramuscular myxoma. Mod Pathol 22(5):718–724

Fletcher (2020) WHO classification of tumours of soft tissue and bone (5th edition)

Vescini (2019) Mazabraud's syndrome: a case report and up-to-date literature review. Endocr Metab Immune Disord Drug Targets 19(6):885–893

[1] GNAS: alternative splicing of downstream exons of this gene (also named GNAS complex locus and guanine nucleotide-binding protein alpha-stimulating activity polypeptide 1) results in different forms of the stimulatory G-protein alpha subunit, a key element of the classical signal transduction pathway linking G protein-coupled receptors (GPCR) with the activation of adenylyl cyclase and ultimately a variety of cell activities.