EUROPEAN OPHTHALMIC PATHOLOGY SOCIETY/
63th Annual Meeting

Date of Meeting: June 11th- June 14th, 2025 Basel, Switzerland

Member's Name: Steffen Heegaard
Eye Pathology Section, Department of Pathology, Rigshospitalet
Frederik d. V's Vej 11
DK-2100, Copenhagen, Denmark
e-mail: sthe@sund.ku.dk
Case number: 24rig060533

Giant Cell Tumour of Soft Tissue of the Lacrimal Sac

Clinical history:
A 72-yer-old male complained of epiphora of the left eye for 18 months. He has experienced bleedings for three months from the lacrimal apparatus (haemolacria) and the bleeding was constant. The patient wakes up in the morning with lots of bleeding in the left eye. The local ophthalmologist has performed flushing of the tear ducts with no obstruction. Local antibiotics and tetracycline were prescribed with no effect. The patient was admitted to our hospital and the visual acuity was 1.0/1.0. The lacrimal punctum was open. In the medial canthus above the medial canthal ligament, it was possible to palpate a little hard tumour. We performed flushing of the tear ducts and there was passage of fresh blood and coagulated blood. MR scanning showed a tumour of the left tear sac. PET-CT showed only localised enhancement. A biopsy was performed and showed a low differentiated carcinoma of the left lacrimal sac. As the tumour was also involving the nasolacrimal duct and with a component in the nasal vestibule, a partial maxillectomi and resection of the inferior concha along with the lacrimal sac and surrounding bone, was performed.

Pathology:
Microscopical examination:
The tumour tissue consists of a mixture of mononuclear and osteoclast-like multinucleated giant cells in a richly vascularized stroma. There are scattered mitoses. There is no necrosis.

Immunohistochemistry: The mononuclear tumour cells are positive for actin. The osteoclast-like giant cells demonstrate positivity for CD68, but are negative for actin. They are also scattered positive for cytokeratin. Immunostainings for CD45, S-100, Melan-A, desmin, CD1alpha, langerin, ERG, D2-40, CK5 and p40 are negative. The tumour is HPV negative

Genetic analysis:
Ion AmpliSeq showed mutations of *PIK3CA* and homozygot deletion of cyclin-dependent kinase inhibitor 2A/B (*CDKN2A/B).*
Archer Expanded Sarcoma panel showed no mutations.

Final diagnosis: Giant Cell tumour of soft tissue of the lacrimal sac

Discussion

Giant cell tumor of soft tissue (GCTST) is extremely rare, locally aggressive and was first described in 1972[1]. The tumour is a so-called fibrohistiocytic tumor. The aetiology is unknown. GCTST is histologically similar to giant cell tumour of bone, but genetically different[2]. Clinically there is a tendency to recur locally. Both primary and secondary malignant GCTST have been reported. Surgical resection is the mainstay of treatment for localized GCTST. Long term follow-up is recommended to monitor for metastases and recurrence.

Clinical characteristics:

There is a peak incidence in the fifth decade and no gender predilection [3]. It typically presents as a painless, superficial soft tissue mass of the upper and lower extremities. Local recurrence occur in 6-21%.

Imaging features:

CT shows a solid, heterogeneous hypodense mass. The underlying bone is typically normal

Pathogenesis:

The vast majority of cases harbour H3.3 histone A gene mutations [4].

Histopathology:

GCTST is a well-circumscribed solid mass and consists of a mixture of mononuclear cells and osteoclast-like multinucleated giant cells [3]. Mitotic figures are found only in the mononuclear cells with an average count of 2-3 mitoses per 10 high –power fields.

Differential diagnosis:

GCTST is often confused with tenosynovial giant cell tumours. Undifferentiated pleomorphic sarcoma with giant cells, formerly known as giant cell malignant fibrous histiocytoma, is the most important differential diagnosis [5]. Undifferentiated pleomorphic sarcoma occurs in older adults and arises in the deep soft tissue of the extremities.

Management:

Surgical resection is the standard treatment for localized GCTST. Radiation therapy can be used to improve local disease control. Pazopanib, lenvatinib and desosumab and also immunotherapy (PD-L1) show promising new treatment modalities for the GCTST.

Conclusion

This is the first described case of a Giant Cell tumour of soft tissue of the lacrimal sac.

References

1 Salm, R.; Sissons, H.A. Giant-cell tumors of soft tissues. J. Pathol. 1972, 107, 27–39.
2 Lee, J.C.; Liang, C.W.; Fletcher, C.D.M. Giant cell tumor of soft tissue is genetically distinct from its bone counterpart. Mod. Pathol. 2017, 30, 728–733.
3 Oliveira, A.M.; Lee, J.C. Giant cell tumour of soft tissue. In World Health Organization (WHO) Classification of Soft Tissue and Bone. Tumours, 5th ed.; International Agency for Research on Cancer (IARC): Lyon, France, 2020; pp. 141–142.
4 Presneau, N.; Baumhoer, D.; Behjati, S.; Pillay, N.; Tarpey, P.; Campbell, P.J.; Jundt, G.; Hamoudi, R.; Wedge, D.C.; Loo, P.V.; et al. Diagnostic value of H3F3A mutations in giant cell tumor of bone compared to osteoclast-rich mimics. J. Pathol. Clin. Res. 2015,1, 113–123.
5 Giant Cell Tumor of Soft Tissue: An Updated Review. Nishio J, Nakayama S, Koga K, Aoki M. J Clin Med. 2024 13;13(10):2870.