Hemangioblastoma

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Definition

Hemangioblastoma is a benign tumor of vascular origin (tumor of grade I according to the World Health Organization).

Epidemiology and Presentation

This is a rare neoplasm of the central nervous system (most often arising in the cerebellum or spinal cord) where it represents about 2% of all tumors. It presents in adults with dizziness, headache, bladder or bowel dysfunction, numbness, weakness, and pain in the upper or lower extremities.

Polycythemia due to the ectopic production of erythropoietin by tumor cells can be present as a paraneoplastic syndrome.

Exceptional cases located outside of the central nervous system (e.g., kidney, retroperitoneum) have been reported.

Etiology and Predisposition

Hemangioblastoma can be sporadic (about 70% of cases) or associated with the **von Hippel-Lindau** (VHL) **disease**, a cancer predisposition syndrome inherited as an autosomal dominant trait and due to the loss of function of the VHL gene that leads to increased activity of hypoxia-inducible factor (HIF), which in turn increases the levels of angiogenic factors such as VEGF. Besides hemangioblastoma of the cerebellum and retina, the VHL syndrome also includes the development cysts of the liver and pancreas, pheochromocytoma, and kidney cancer.

S. Mocellin, *Soft Tissue Tumors*, https://doi.org/10.1007/978-3-030-58710-9_115

Pathology

Macroscopically, it presents as a well-circumscribed mural nodule (which contains the tumor cells) associated with a large fluid-filled cyst (hence, it is important to obtain a frozen section of the mural nodule, and not of the cyst wall).

Differential diagnosis may be needed with renal cell carcinoma (nuclear atypia, mitotic figures, cytokeratin positive, EMA positive, NSE negative).

Biomarkers

Hemangioblastoma stains positive for NSE,[1] reticulin, and CD34. It stains negative for EMA,[2] cytokeratins, and CD10.

Prognosis

The prognosis is good as long as the tumor is amenable to complete surgical excision, which is the case in most patients.

Therapy

Surgical excision is the treatment of choice (microsurgical resection or stereotactic radiosurgery).

Suggested Readings

Aronow (2019) Von Hippel-Lindau disease: update on pathogenesis and systemic aspects. Retina [Epub ahead of print]

Bisceglia (2018) Extraneuraxial hemangioblastoma: clinicopathologic features and review of the literature. Adv Anat Pathol 25(3):197–215

Pan (2018) Stereotactic radiosurgery for central nervous system hemangioblastoma: systematic review and meta-analysis. J Neurooncol 137(1):11–22

[1] NSE: neuron-specific enolase.

[2] EMA: epithelial membrane antigen.