

History

- A 30-year-old male presents with a phthisical left eye and undergoes enucleation. He has some lesions in his right eye that are under ophthalmic surveillance.
- Figure 1 is a low power of the enucleation.
- Figures 2, 3 and 4 are higher power shots of the arrowed lesion in Figure 1.

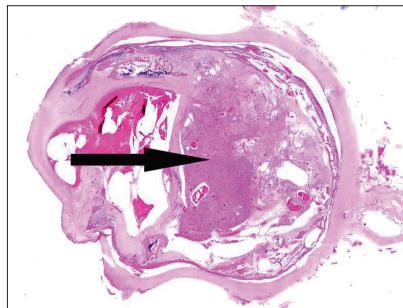


Figure 1

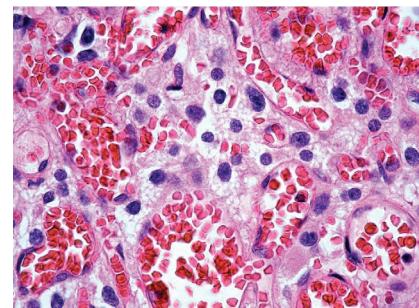


Figure 3

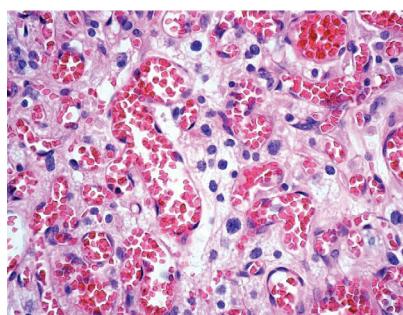


Figure 2

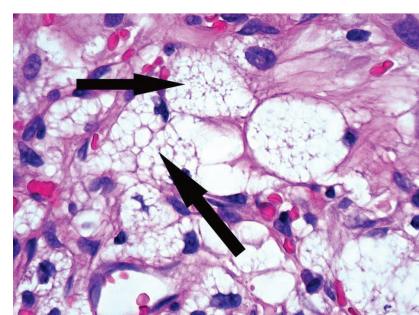


Figure 4

Questions

- What do Figures 2 and 3 show?
- What does Figure 4 show (arrow)?
- What is the likeliest diagnosis?
- Which immuno-stain will highlight the cells in Figure 4?
- What other history may you ask for from the clinicians?

- Other organs: cysts.
- Adenexae: cystadenoma
- Epidiidymis: epididymal cystadenoma
- Endolymphatic sac / duct: endolymphatic sac tumour
- Adrenal gland / paraganglia: phaeochromocytoma / paraganglioma
- Pancreas: cysts, microcystic adenoma and neuroendocrine tumours
- Kidney: cysts and renal cell carcinoma
- Peripherial nerves: haemangioblastoma
- Retina: haemangioblastoma
- The systemic manifestations are:
- 5. This patient has Von Hippel-Lindau disease (hence the lesions in the other eye).
- 4. Anti-VEGF.
- 3. The combination of endothelial and foamy stromal cells are the neoplastic elements.
- 2. Foamy stromal cells between the vascular element.
- 1. A vascular neoplasm.

ANSWERS

Further reading

- <http://emedicine.medscape.com/article/1219430-overview>
- WHO classification of tumours of the central nervous system. Third edition. IARC Press; 2007:215-7.
- Mettu P, Agron E, Samtani S, et al. Genotype-phenotype correlation in ocular von Hippel-Lindau (VHL) disease: The effect of missense mutation position on ocular VHL phenotype. *Invest Ophthalmol Vis Sci* 2010;51(9):4464-70.

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