

History

- A 35-year-old woman had a long-standing left blind eye following extensive exudative retinal detachment in the past.
- She later developed increasing pain in her blind eye.
- Her medical history includes pheochromocytoma resected five years ago, and she is currently on surveillance for renal cysts.
- Her father had renal tumour and died with widespread metastatic disease.
- Fundus examination was impaired by media opacity.
- USB demonstrated a mass in the posterior segment.
- Enucleation was performed.
- Macroscopic examination revealed a partly solid partly cystic yellow-brown mass filling the posterior segment.

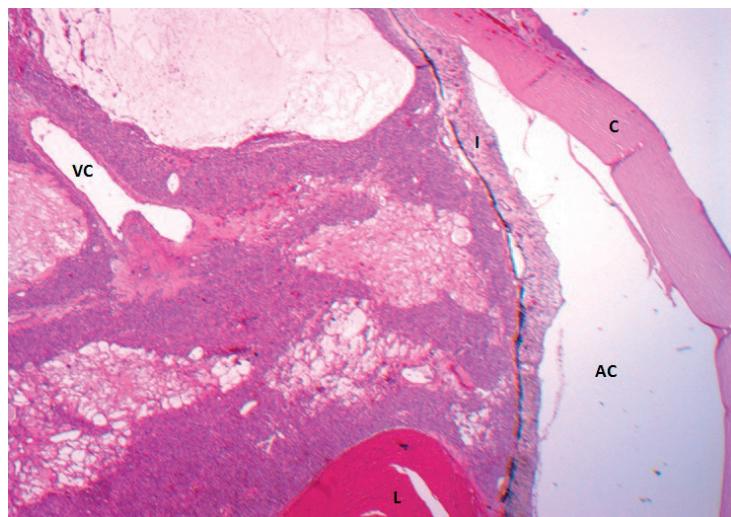


Figure 1

Questions

1. Figures 1, 2 and 3 show representative histological and immunohistochemical sections. How can those be described?
2. What is the most likely diagnosis?
3. Considering the diagnosis on this patient's enucleation and the clinical history provided, is there any other relevant clinical condition to be considered?
4. Are there any further diagnostic steps?

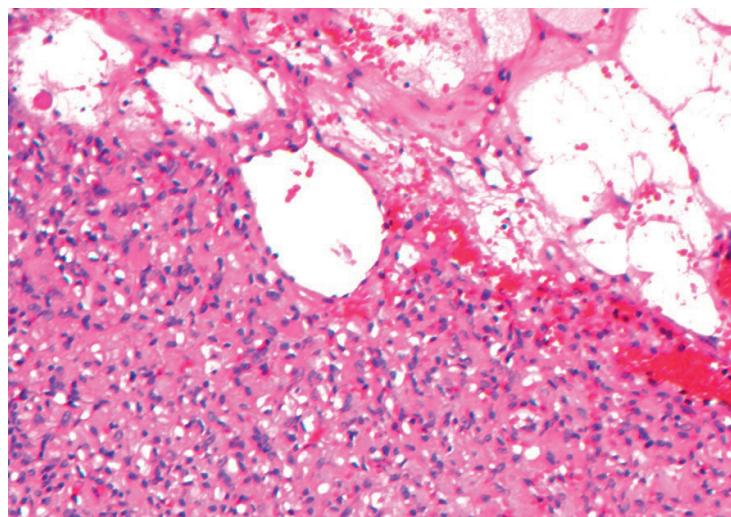


Figure 2

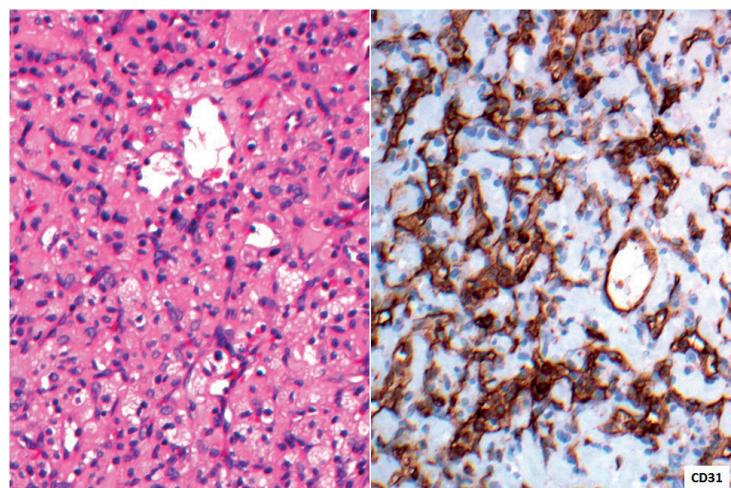


Figure 3

1. Figure 1 shows a solid-cystic mass filling up the vitreous cavity, encircling the lens and being adherent to the posterior surface of the iris. Figure 2: Interface between solid and cystic components which actually represents vascular channels. Note intervening red blood vessels and cystic, solid components with a well developed interface.

2. Figure 3: Left image shows a well vascularised solid area including cells with abundant foamy and eosinophilic cytoplasm. On the right, framework in this rich vascular tumour.

3. The occurrence of retinal haemangioblastoma, phaeochromocytoma and investigation for renal cysts indicates von Hippel-Lindau syndrome.

4. She most likely has a mutation on the VHL gene. The inheritance pattern of renal tumour in her family suggests any sibling and children that the patient may have is essential. The patient also needs to be under close follow-up due to the risk of further neoplasia developing in other organs.



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ANSWERS