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A Rare Tumour in Sight: Surgical Management of an Optic Nerve Hemangioblastoma

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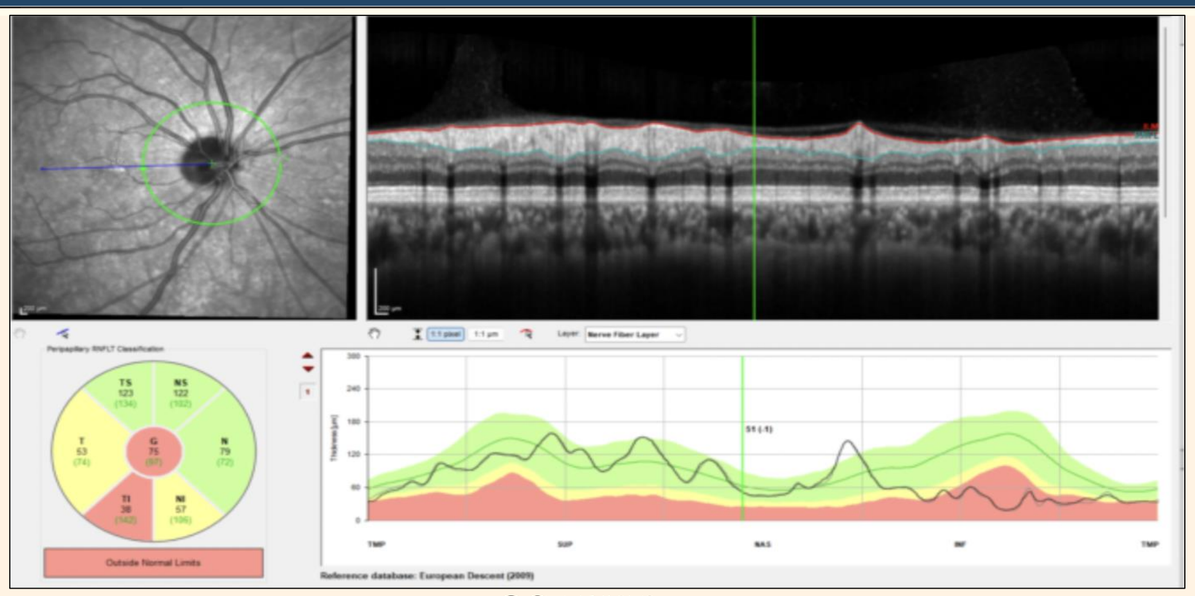
BACKGROUND

- Optic nerve tumours are abnormal growths within, or on, the optic nerve which cause visual dysfunction through infiltration and/or compression of the optic nerve[1].
- Optic nerve hemangioblastomas are extremely rare, with only 40 reported cases in literature as of 2024[2].
- Hemangioblastomas, typically occurring in the cerebellum, brainstem or spinal cord, are benign vascular tumours which are characteristically associated with Von Hippel-Lindau (VHL) disease[3].
- With a non-specific presentation, including loss of vision, proptosis and orbital pain, which mimics other optic nerve tumours, this case highlights the role of imaging, early resection, histopathological assessment and genetic analysis, within a multidisciplinary approach, to manage a rare differential for an optic nerve lesion.

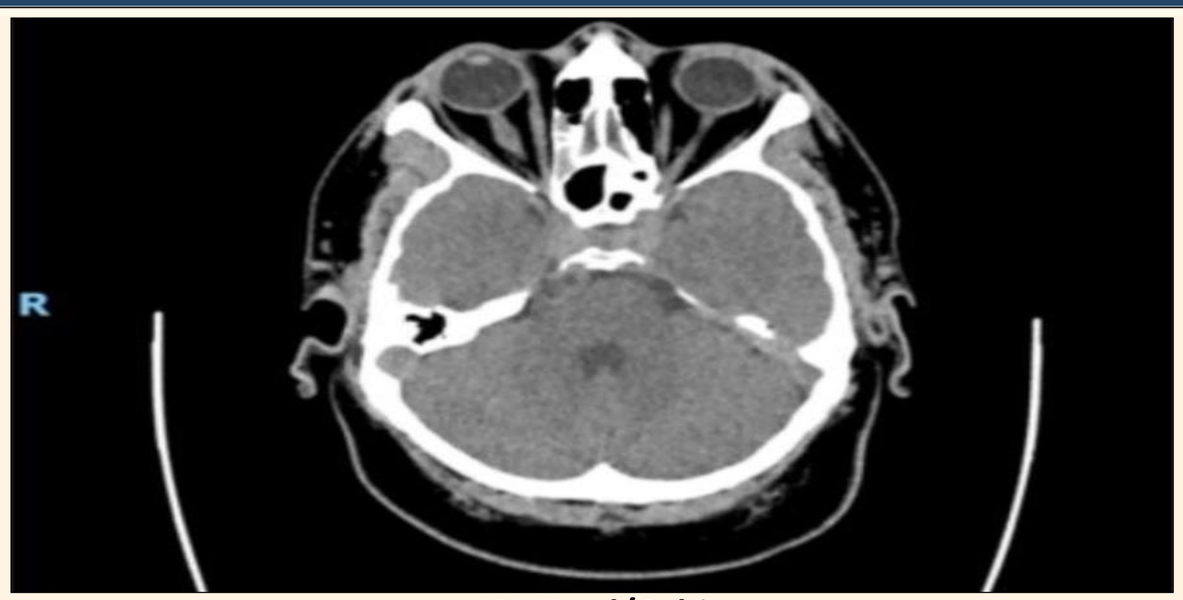
CASE SUMMARY

- **Demographics:** 44-year-old, male
- **Presentation:** Referred after a right superonasal quadrantanopia was found on a routine optician assessment. Otherwise, asymptomatic.
- **Imaging:**
  - OCT-RNFL = inferotemporal thinning of the right optic nerve
  - MRI head/orbit = fusiform thickening of the right optic nerve, with T2 hyperintensity
- **Histology:** WHO Grade I Hemangioblastoma
- **Genetic Testing for VHL Disease:** Negative
- **Management:** Combined approach endoscopic, endonasal surgical excision of the optic nerve lesion with follow-up imaging
- **Follow-up:** Serial MRIs demonstrated no recurrence or residuum of the optic nerve lesion as of 5 years follow-up.

IMAGING



OCT-RNFL



MRI Head/Orbit

SURGICAL PROCEDURE

- History and imaging findings were reviewed in a skull-base multi-disciplinary meeting. Ophthalmology and ENT surgeons opted to collaborate for combined approach endoscopic, endonasal surgical excision of the optic nerve lesion. This involved:

1. Initially performing a conjunctival peritomy.
2. The medial rectus was then identified and hooked, before being released and reflected.
3. Dissection down to the optic nerve, and then a sling was tied around the optic nerve and attached to a navigation probe.
4. An endonasal approach involved ethmoidectomy, sphenoidotomy and resection of the lamina papyracea until the orbital apex.
5. Delivery of the optic nerve into the nasal cavity using the navigation probe. The optic nerve was then coagulated and approximately 3cm resected.
6. Closure involved haemostasis followed by nasal packing and conjunctival sutures.

DISCUSSION

- Symptoms such as loss of vision, proptosis and orbital pain are non-specific. Definitive diagnosis requires neuroimaging and histological assessment[5].
- T2 hyperintensity of MRI is common to optic nerve hemangioblastomas, gliomas and meningiomas. MRI features more specific to optic nerve hemangioblastomas include the presence of flow and the absence of dural attachment[4,5].
- Differentiating between optic nerve hemangioblastomas and more prevalent optic nerve tumours using neuroimaging guides management approach. In optic nerve gliomas, first-line treatment is typically chemotherapy, whereas in optic nerve hemangioblastomas, complete surgical excision to reduce risk of recurrence is typically recommended[6,7].
- Pre-operative suspicion of hemangioblastoma allows for appropriate multidisciplinary planning to minimise intra-operative complications, such as haemorrhage, when on operating on these highly vascular tumours[3].
- Histological diagnosis of hemangioblastoma is characterised by stromal cells and dense capillary networks in a biphasic tissue composition[3].
- Approximately 70% of reported cases of optic nerve hemangioblastomas are associated with VHL-disease[3]. Therefore, all patients with optic nerve hemangioblastomas should undergo genetic testing. This case however highlights optic nerve hemangioblastomas can occur without associated VHL-disease and should remain a differential diagnosis to be excluded in patients with or without VHL-disease.
- Combined approach with ENT was a feasible surgical approach for excision of this optic nerve hemangioblastoma, with no complications or recurrence as of 5 years follow-up.

CONCLUSION

- History alone cannot distinguish between subtypes of optic nerve tumours. A multidisciplinary approach involving neuroimaging, skull-base surgery, histology and genetics is required for definitive diagnosis and appropriate management of optic nerve hemangioblastomas.
- Complete excision reduces risk of recurrence, and a combined approach is a viable surgical option with an appropriate outcome having been demonstrated in this case.

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