

Lower Lid Mass in a Neonate

A 4-week-old boy was referred to our dermatology unit for evaluation of a left lower lid mass which was present since birth. Physical examination revealed a large, non-pulsatile mass in the left infra-orbital region, with bluish discoloration of the overlying skin. In the first 2 months after birth, the mass was noted to show reduction in size, prompting the diagnosis of a rapidly involuting congenital hemangioma (RICH). However, at 5 months of age, the mass showed interval increase in size. In addition, there was increased bluish discoloration of the overlying skin. In view of the unexpected clinical course, further imaging was performed. Ultrasound of the left orbit revealed a well defined, heterogenous soft tissue mass in the medial aspect of the lower eyelid. Minimal internal vascularity was noted on Doppler interrogation (Figs. 1A-1B). Magnetic resonance

imaging (MRI) confirmed the presence of a mildly enhancing heterogenous soft tissue mass, confined within the inferior extra-conal compartment of the left orbit. A small cystic area was seen within its medial aspect but no definite fluid-fluid level or fat was seen. There were also no apparent signal voids on T1-weighted and T2-weighted images to suggest the presence of calcifications. No intracranial extension was demonstrated (Figs. 1C-1E).

What is the most likely diagnosis?

- A. Congenital hemangioma
- B. Venous lymphatic malformation
- C. Dermoid cyst
- D. Epidermoid cyst
- E. Heterotopic neuroglial tissue

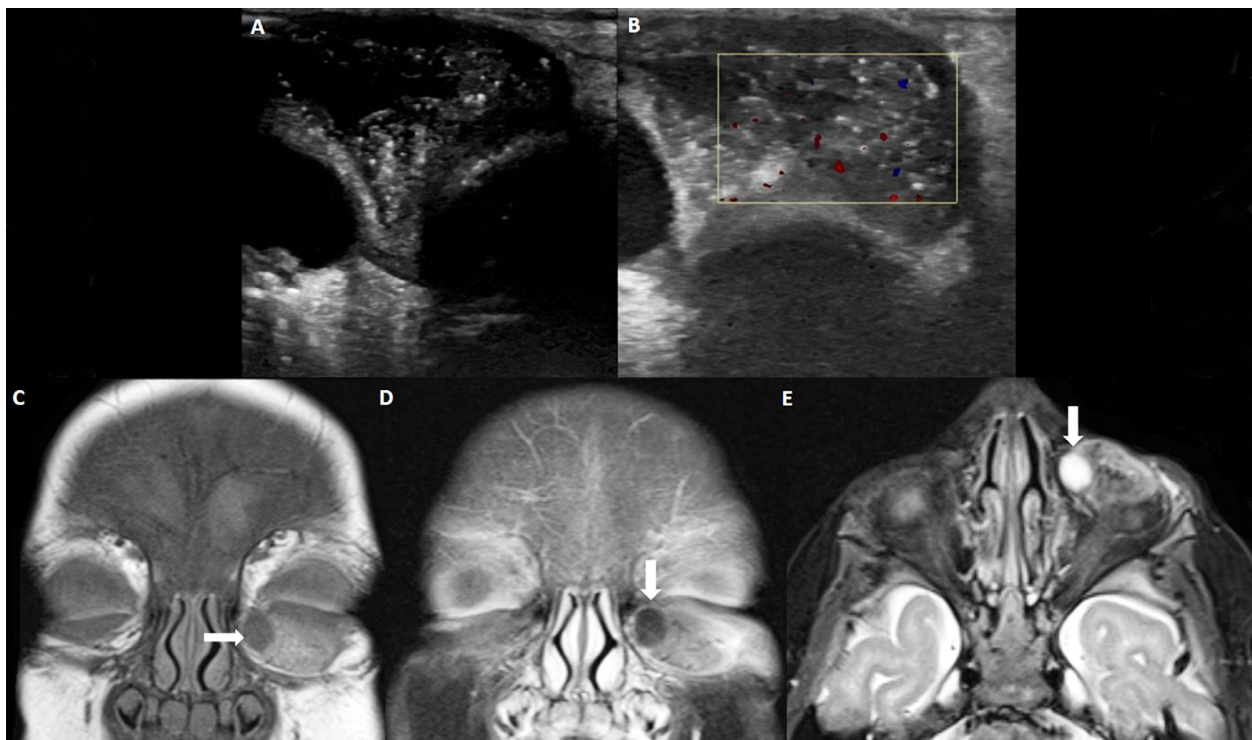


Fig 1. A: Ultrasound (US) of the left orbit showed a well defined, heterogenous soft tissue mass in the medial aspect of the infra-orbital region. B: Minimal internal vascularity was noted on Doppler interrogation. C: Coronal T1-weighted magnetic resonance imaging (MRI). D: Coronal T1-weighted postcontrast MRI. E: Axial T2-weighted MRI confirmed the presence of a mildly enhancing heterogenous soft tissue mass, confined within the antero-inferior extra-conal compartment of the left orbit. A small cystic area (white arrow) was seen within its medial aspect but no definite fluid-fluid level, fat or calcification was seen.

Answer: E

Discussion

Heterotopic neuroglial tissue is an uncommon entity, typically sited along the nasal midline structure. Its occurrence within the orbits is rare, with only few published cases. Presentation within the first year of life is common, and may be mistaken for other entities such as congenital hemangiomas, venolymphatic malformations, dermoid and epidermoid cysts, all of which are common orbital masses in the infantile period. A superficial lesion such as in our case, often presents as a mass while a deeper lesion may present with visual disturbances, proptosis or papilloedema. Diagnosing this lesion on clinical grounds is extremely challenging. Even with high resolution MRI, a definite diagnosis is often elusive due to the lack of characteristic imaging features. The presence of calcifications is not uncommon.¹ It may also appear cystic due to cerebrospinal fluid (CSF) production.² Although imaging may not be diagnostic, it is crucial for preoperative planning, primarily to exclude the presence of bony defects and intracranial communication, of which, if present, may necessitate a different surgical approach.³ Definitive diagnosis requires histological demonstration of neuroglial cells as well as a positive glial fibrillary acidic protein (GFAP) immunohistochemical stain.^{1,3,4} Neurons are usually sparse (as in our case) or absent. If abundant neurons are seen, one must exclude an encephalocele. Heterotopic neuroglial tissue is believed to grow at a similar rate comparable to the normal surrounding tissues. Hence, complete surgical excision is the mainstay of treatment, failing which, a recurrence rate of approximately 10% has been reported. Although there is no reported case of malignant transformation, they may possess low-grade neoplastic potential.

Congenital hemangiomas develop in utero and there is no postnatal growth, unlike infantile hemangiomas and heterotopic neuroglial tissue.⁵ Doppler ultrasound classically reveals high vascularity.⁶ Large flow voids on

the surface of the lesion, arterial aneurysms and arteriovenous shunting may also be present. On MRI, it is seen as a heterogenous mass with inhomogenous enhancement. Cystic spaces and intralesional calcifications may also be present.^{7,8} The presence of postnatal growth as well as absence of high vascularity on Doppler interrogation in our patient make this diagnosis unlikely.

Venous lymphatic malformation is another common orbital lesion within the paediatric population. Children with orbital venous lymphatic malformation typically present in childhood with slowly progressive proptosis, periorbital swelling and displacement of globe. It appears on MRI as an enhancing mass that crosses anatomic boundaries, such as the conal fascia and orbital septum. It usually has a predominant cystic component with fluid-fluid levels,^{7,8} not seen in our patient. Phleboliths may also be present.

Dermoid cysts are benign heterotopic neoplasms termed 'choristomas' and account for up to 9% of paediatric orbital tumours. They occur in 3 primary locations in the head and neck—in the frontotemporal, periorbital and naso-glabella regions. Within the periorbital region, the lateral orbit (adjacent to the lateral canthus) is the most common location. MRI appearance is variable and depends on the specific contents of the cyst. If there is lipid material within the cyst, it will appear hyperintense on T1-weighted imaging. Cysts containing higher levels of protein can appear hyperintense on both T1 and T2 imaging. Occasionally, calcifications may be present in dermoid cyst. Ruptured dermoids may also show adjacent inflammatory changes.⁸ Epidermoid cysts, also a choristoma, are seen as cystic lesions, typically with restricted diffusion on diffusion-weighted imaging.

As the lesion was present at birth, demonstrated postnatal growth and appeared as a solid mass with minimal internal vascularity on imaging, ectopic neuroglial tissue was deemed the most likely diagnosis. Excisional biopsy was performed and microscopic examination revealed disorganised collections of mature neurons and glial cells (Fig. 2).

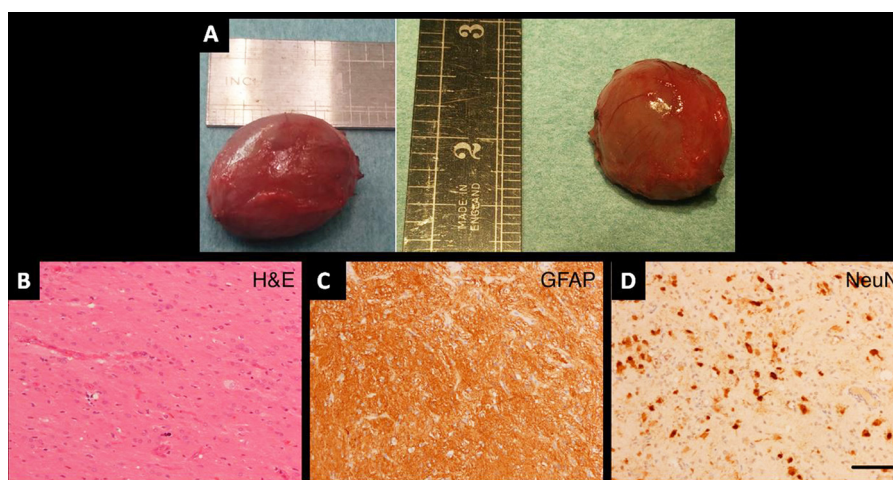


Fig. 2. A: Gross examination of the specimen showed a firm nodule measuring approximately 2.0 cm x 2.5 cm x 1.5 cm. B: Histopathological section revealed disorganised collections of mature neurons and glial cells. C: Diffusely positive glial fibrillary acidic protein (GFAP) on immunohistochemical staining was noted. D: Mild neuronal nuclei (NeuN) immunoreactivity was observed due to the presence of neurons. Findings were in keeping with neuroglial heterotopia.

Several interposed areas of haemorrhage and hemosiderin deposition were seen. Diffusely positive glial fibrillary acidic protein (GFAP) on immunohistochemical staining was noted. Mild neuronal nuclei (NeuN) immunoreactivity was also observed, due to the presence of neurons. Findings were in keeping with heterotopic neuroglial tissue.

Conclusion

Although rare, awareness of heterotopic neuroglial tissue as a cause of periorbital mass is important in expediting the diagnosis so as to enable an effective management strategy.

REFERENCES

1. Scheiner AJ, Frayer WC, Rorke LB, Heher K. Ectopic brain tissue in the orbit. *Eye (Lond)* 1999;13:251-4.
2. Nasr AM, Huaman A, Johnson T, Weatherhead R. Disjunct cephalocele (bilateral orbital cystic glial tissue): a clinico-pathologic entity. *Orbit* 1992;11:99-107.
3. Sitaula R, Shrestha GB, Paudel N, Shrestha S, Shah DN. Glial heterotopia of the orbit: a rare presentation. *BMC Ophthalmol* 2011;11:34.
4. Bajaj MS, Kashyap S, Wagh VB, Pathak H, Shrey D. Glial heterotopia of the orbit and extranasal region: an unusual entity. *Clin Experiment Ophthalmol* 2005;33:513-5.
5. Krol A, MacArthur CJ. Congenital hemangiomas: rapidly involuting and noninvoluting congenital hemangiomas. *Arch Facial Plast Surg* 2005;7:307-11.
6. Rogers M, Lam A, Fischer G. Sonographic findings in a series of rapidly involuting congenital hemangiomas (RICH). *Pediatr Dermatol* 2002;19:5-11.
7. Bhat V, Salins PC, Bhat V. Imaging spectrum of hemangioma and vascular malformations of the head and neck in children and adolescents. *J Clin Imaging Sci* 2014;4:31.
8. Chung EM, Murphey MD, Specht CS, Cube R, Smirniotopoulos JG. From the Archives of the AFIP. Pediatric orbit tumors and tumorlike lesions: osseous lesions of the orbit. *Radiographics* 2008;28:1193-214.

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