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Clinical Image

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Ocular coloboma with microphtalmia

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Clinical image

A 3-year-old child was referred for small eyes and always keeping them closed. MRI revealed dysmorphic ocular globes with loss of their spherical shape and a cystic polilobulated lesion communicating with the globe; hypointense on T1-weighted images and hyperintense on T2-weighted images without contrast enhancement associated with a microphthalmia. Findings are suggestive of a retrobulbar colobomatous congenital cyst (Figure 1).

Discussion

A coloboma is a developmental abnormality resulting from the failure of closure of the embryonic choroidal fissure, that occurs between gestational days 35 and 41. Presenting as a cleft that may involve the retina, choroid, sclera, iris, ciliary body, lens, or optic nerve head. A coloboma can be an isolated finding in an otherwise healthy individual or part of a complex malformation syndrome of known or unknown etiology [1]. Colobomas may be associated with reduced ocular globe size or with normal size and are bilateral in more than 60% of cases [2]. Both sexes are equally affected [3].

On MRI, these cysts vary in size and appear as hypointense on T1-weighted images and hyperintense on T2-weighted images. The exact communication site between the cyst and the globe may not always be clear on imaging and the differential diagnosis to consider in imaging is staphyloma [4].

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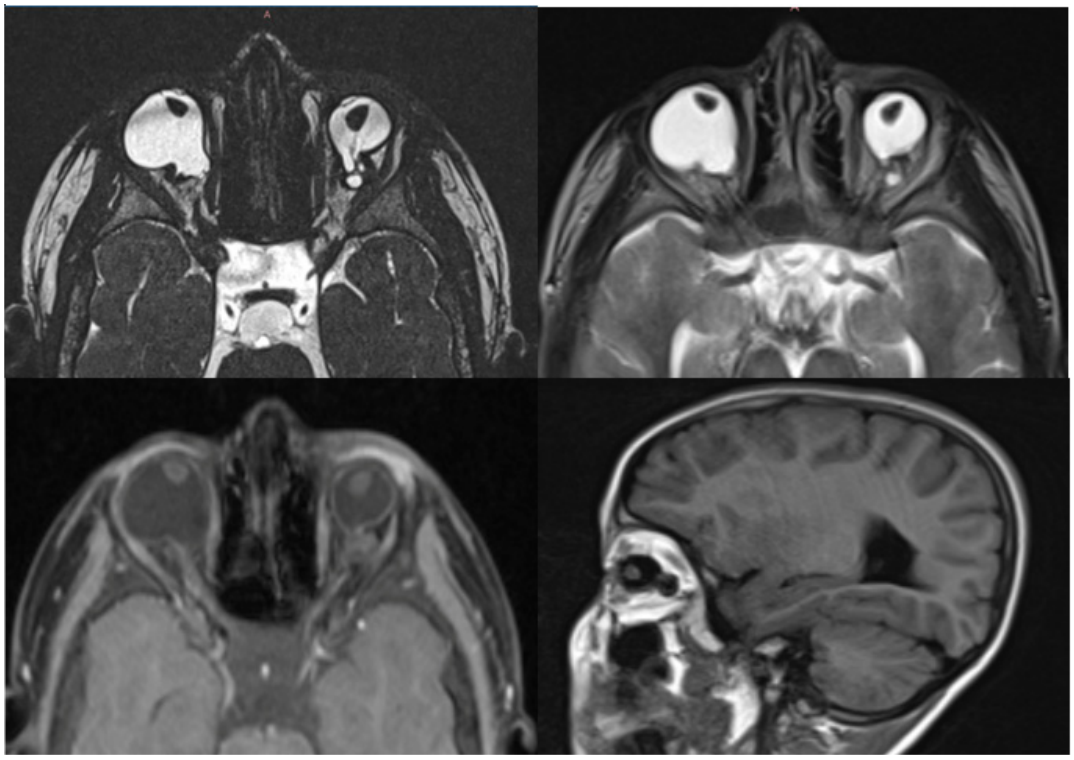

Figure 1: Axial CISS, T2, contrast-enhanced T1, and sagittal T1 MRI sequences reveal dysmorphic ocular globes with cysts communicating with the ocular globes associated with left microphthalmia.