Tuberomammillary Fusion and Moya-Moya Vasculopathy Associated with PHACE Syndrome

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Neuropediatrics

A 1-year and 5 month-old female child with a large right facial hemangioma (►**Fig. 1**) presents recurrent seizures. Brain magnetic resonance imaging (►**Fig. 2**) demonstrated posterior fossa malformations, with a right colobomatous cyst and an extensive sub-occlusive vasculopathy with a Moya-Moya pattern consistent with PHACE syndrome.

The PHACE syndrome is a phakomatosis also known as cutaneous hemangioma–vascular complex syndrome or Pascual-Castroviejo type II syndrome. There are some extracerebral and intracranial vascular abnormalities, including the Moya-Moya arteriopathy[1] which affects less than 7% of the patients.[2] Also, there are no previous papers reporting the association with tuberomammillary fusion in this clinical scenario.

This uncommon association of PHACE syndrome is extremely relevant for neurologists, neuropediatricians, and neuroradiologists, expanding the imaging phenotype features.

Fig. 1 Photos of the patient's face demonstrating the right segmental forehead and facial hemangioma at birth (**A**), proliferative phase (**B**), and the involutional phase (**C**). Note made for a right ocular prosthesis in the last image.

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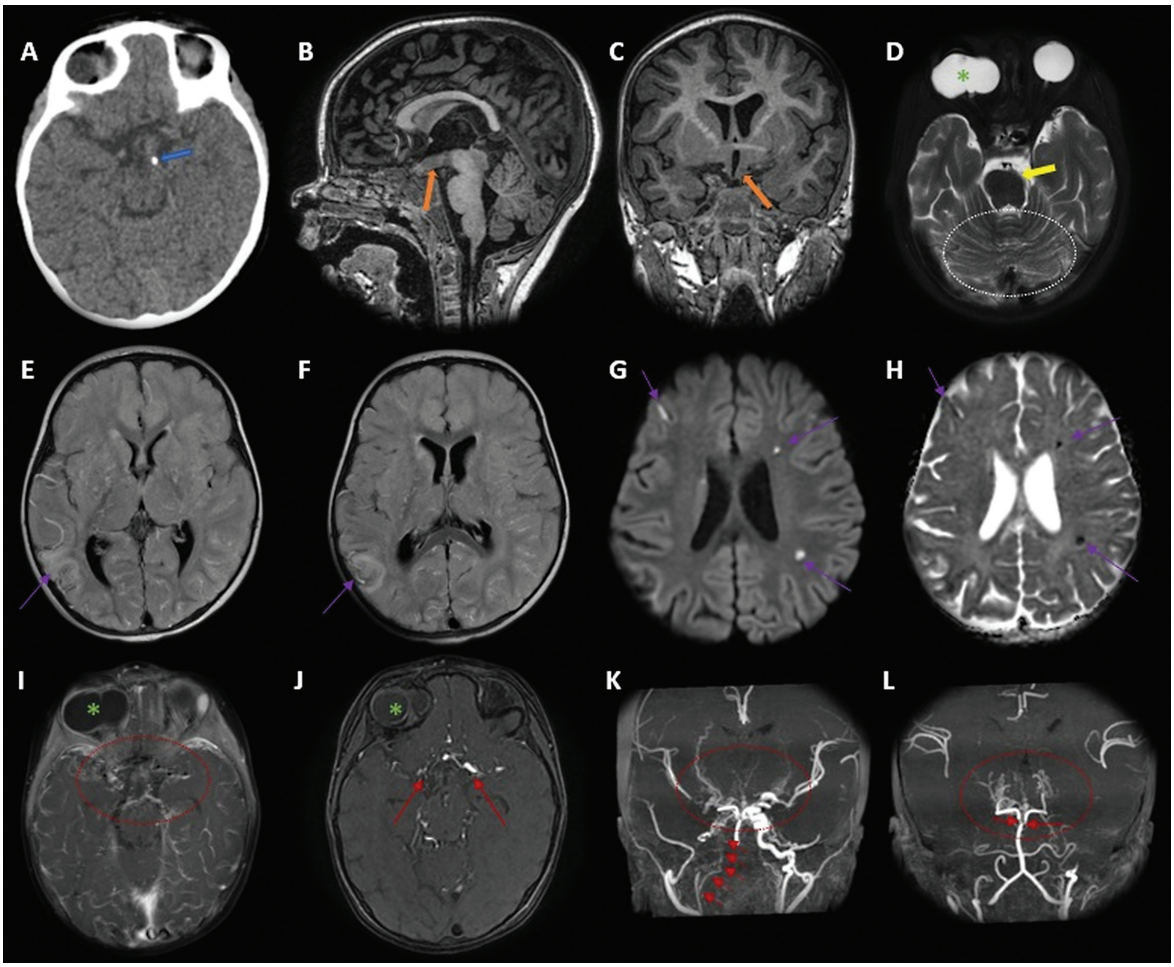

Fig. 2 CT head without contrast (**A**) and 3.0 Tesla MRI and MRA of the brain (**B–L**). T1 MPRAGE-weighted images (B and C), T2 (D), FLAIR (E and F), DWI (G), ADC map (H), T1 post-gadolinium (I), and 3D-TOF (J–L). Multiple features of PHACE syndrome. Focal calcification in the proximal left PCA (blue arrow), left-sided tuberomammillary fusion (orange arrows), large colobomatous cyst in the right eye globe (green asterisk), left anterior pons hypoplasia (yellow arrow), cerebellar dysplasia (white dashed circle), diffuse "ivy"-sign (E and F), external watershed zone infarcts (purple arrows), and hyperleptomeningeal enhancement (I). Moya-Moya pattern of steno-occlusive vasculopathy, with involvement of the carotid and vertebrobasilar systems (red arrows), pseudo-occlusion of the right ICA (red dashed arrows), and multiple pial collaterals (red dashed circles). 3D-TOF, three-dimensional time of flight; CT, computed tomography; DWI, diffusion-weighted imaging; FLAIR, fluid attenuated inversion recovery; ICA, internal carotid artery; MRA, magnetic resonance angiography; MRI, magnetic resonance imaging; PCA, posterior cerebral artery.

Author Contribution

1. Case report project: A: conception; B: organization; C: execution.
2. Manuscript: A: writing of the first draft; B: review and critique.

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Ethical Statement

Full consent was obtained from the patient for the case report publication.

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Conflict of Interest

The authors have no conflict of interest.

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