Tuberomammillary Fusion and Moya-Moya Vasculopathy Associated with PHACE Syndrome

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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 arteriopathy¹ 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.
**Author Contribution**

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

Freitas L.F.: 1A, 1B, 1C, 2A, 2B.
Miranda E.C.: 1A, 1B, 1C.
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**Ethical Statement**

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

**Financial Disclosure**

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Dr. Miranda reports no disclosure.
Dr. Amaro reports no disclosure.

Dr. Narvaez reports no disclosure.
Dr. Duarte reports no disclosure.

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**Conflict of Interest**

The authors have no conflict of interest.

**References**