



Cerebrofacial vascular metamerism syndrome associated with Moyamoya syndrome: a rare case report

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Abstract

Introduction Neural crest and mesoderm cell dysfunction of certain metamerism level result in vascular malformations, i.e., cerebrofacial arteriovenous metamerism syndrome (CAMS) and cerebrofacial venous metamerism syndrome (CVMS). Moyamoya disease is a progressive steno-occlusive disease in the terminal portions of the bilateral internal carotid artery. The patient in this case report was a child with cerebrofacial vascular metamerism syndrome, associated with moyamoya syndrome.

Case report Child, 7 months old, female, admitted to the emergency department with seizures, hemangioma on the right half of the face (forehead, upper eyelid, and upper lip), and left hemiparesis. The magnetic resonance imaging of the skull indicated increased myelination in the right hemisphere (T2) and atrophy compatible with Sturge-Weber syndrome. Cerebral angiography indicated vasculopathy with bilateral moyamoya pattern, associated with other arteriovenous malformations compatible with cerebrofacial vascular metamerism syndrome. Moyamoya syndrome was treated with indirect revascularization (pial synangiosis) achieving good outcomes.

Discussion Vascular malformations can involve the orbits, face, and brain simultaneously. CAMS with forebrain or hind-brain involvement can be classified into subgroups: I, II, and III. On the other hand, venous malformations in Sturge-Weber syndrome or encephalotrigeminal angiomatosis can be considered CVMS. Moyamoya disease is called syndrome when related to another clinical condition, such as the present case, i.e., neurocutaneous Sturge-Weber syndrome. The association of chronic moyamoya vasculopathy with cerebrofacial vascular metamerism syndrome is rare. Further studies are required to establish the best treatment approach.

Keywords Vascular malformations · Hemangioma · Moyamoya · Arteriovenous

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Introduction

Studies have pointed out that the cells of the neural crest and mesoderm of certain transversal or metamerism level will occupy the same territory in the face and in the brain and will also help in myogenesis and vasogenesis. The early dysfunction of these cells during migration and differentiation results in vascular malformations, which can be a genetic defect or somatic mutation [1, 2]. This way, the names “cerebrofacial arteriovenous metamerism syndrome” (CAMS) and “cerebrofacial venous metamerism syndrome” (CVMS) have been suggested, corresponding to arterial and venous malformations, respectively, involving the face and the brain at the same time [3].

In turn, Moyamoya disease is considered a progressive steno-occlusive disease occurring in the terminal portions of the bilateral internal carotid artery, with the development of

collaterals, called “moyamoya vessels,” to ensure the circulation channels. When associated with other pathological conditions, it is called syndrome [4]. The present report describes the case of a child with cerebrofacial vascular metameric syndrome, rarely associated with moyamoya syndrome.

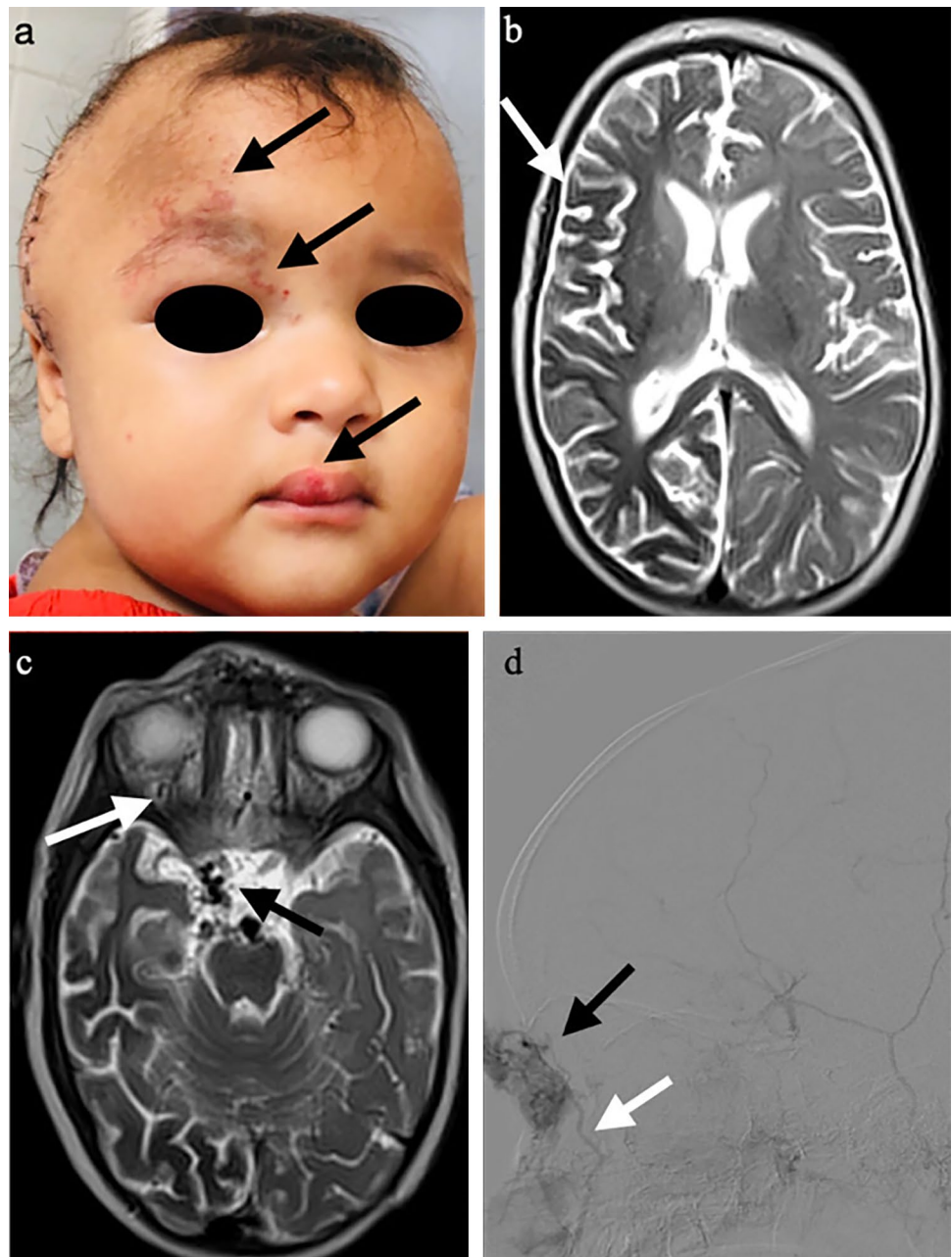
Case report

This is the case of a 7-month-old girl, daughter of non-consanguineous parents, admitted to the emergency department with convulsions and weakness on the left side of the body. Upon examination, she exhibited hemangioma on the

right half of the face (forehead, upper eyelid, and upper lip) (Fig. 1a), irritability, and left hemiparesis. The other systems (cardiovascular and respiratory) had no alterations. The child's birth had been at term, with normal labor and no complications. The progression was normal, and she was immunized for her age. There was no family history of the condition.

Magnetic resonance imaging (T2) indicated subcortical white matter hypointensity (Fig. 1b), with accentuation of the myelination pattern of the entire right cerebral hemisphere, and atrophy compatible with Sturge-Weber syndrome (Fig. 1a). In addition, there were arteriovenous malformations in the cisterns at the base of the brain (Willis polygon),

Fig. 1 **a** Hemangioma on the right half of the face: forehead, upper eyelid, and upper lip (black arrows); **b** brain magnetic resonance imaging, axial T2 showing hypointensity with atrophy in the right cerebral hemisphere (white arrow); **c** arteriovenous malformations involving cisterns at the base of the brain and hypothalamic region (black arrow) with extension to the right orbit (white arrow); and **d** angiogram showing arteriovenous malformations in the right orbit (black arrow) supplied by branches of the facial artery (white arrow)



with extension to the right orbit through the optic nerve, associated with multiple ectatic and tortuous vessels, thus configuring a CAMS II-type lateral prosencephalic arteriovenous malformation (Fig. 1c, d).

Cerebral angiography indicated vasculopathy with bilateral moyamoya pattern (Fig. 2), associated with bilateral orbital arteriovenous malformation and aneurysmatic formation with dysplastic vessels of the basilar artery and right posterior communicating artery.

In view of the diagnosis of bilateral moyamoya syndrome, with episodes of cerebral ischemia, it was decided to perform surgical treatment. Indirect revascularization was performed through bilateral pial synangiosis (Fig. 3a, c). The procedure was performed without problems. Cerebral angiography was performed 8 months after the surgery. It evidenced signs of anastomosis between branches of the superficial temporal artery, and cortical branches of the middle cerebral artery on the left side (Fig. 3b). However, the procedure on the right side did not present a favorable (Fig. 3d) result because we

could not say whether the ischemic suffering would be Moyamoya or Sturge-Weber syndrome. The child had no seizures (using phenobarbital), with improvement in left hemiparesis after rehabilitation by means of physiotherapy.

Discussion

In 2001, Bhattacharya et al. observed evidence of metameric craniofacial vascular malformations, with typical characteristics and revised embryology [2,5]. This way, a classification of three subgroups determined as CAMS was proposed based on the involvement of the forebrain or rhombencephalon, namely, CAMS I—medial prosencephalic group involving the hypothalamus, pituitary, and nasal regions; CAMS II—lateral prosencephalic group involving the occipital lobe, thalamus, optic tract, retina, and maxilla; and CAMS III—lateral rhombencephalon group involving the cerebellum, nasal bridge, and mandible [5, 6]. According to the

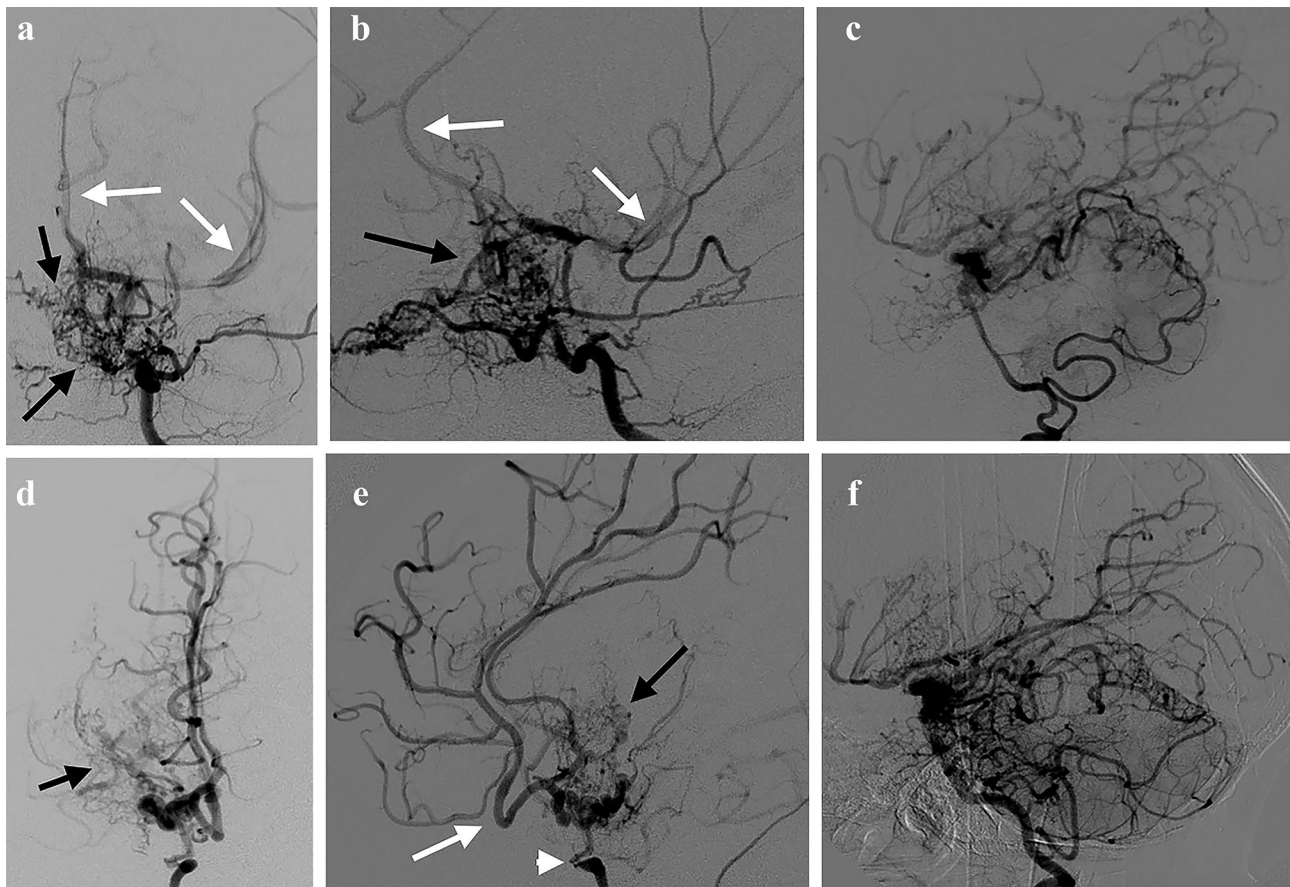
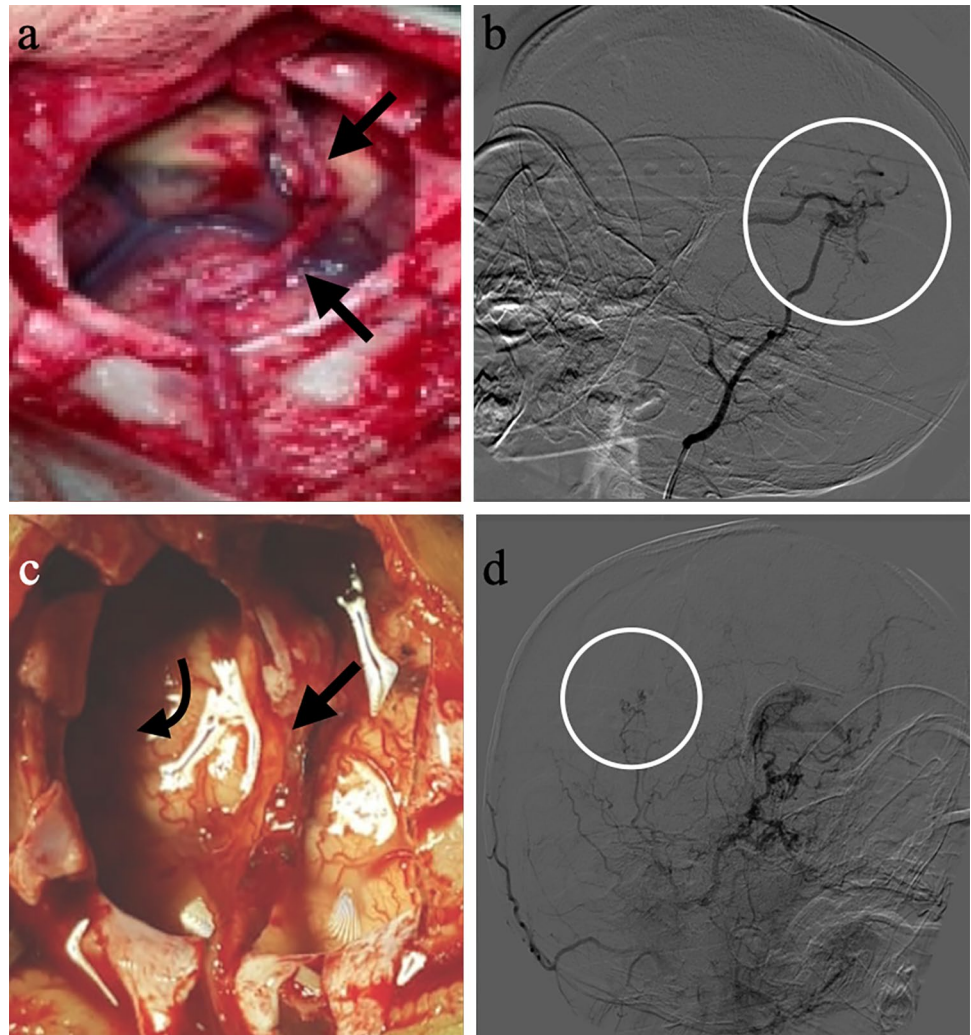


Fig. 2 a AP and b lateral view of cerebral angiography showing a network of collateral vessels or Moyamoya vessels (black arrows) and poor visualization of the terminal branches of the left internal carotid artery (ICA): anterior cerebral and middle cerebral arteries (white arrows); c lateral view of cerebral angiography showing preserved left posterior circulation; d AP and e lateral view of cerebral angi-

ography showing stenosis of the supraclinoid segment of the right internal carotid artery (white arrowhead), with steno-occlusive lesion of the middle cerebral artery, presence of a network of collateral vessels (black arrows) with aneurysmatic formations and normal anterior cerebral artery (white arrow); f lateral view of cerebral angiography showing preserved right posterior circulation

Fig. 3 **a** Left pial synangiosis—branch of the left superficial temporal artery sutured directly to the pia mater (black arrows); **b** postoperative with formation of neovascularization (white ring) between branches of the superficial temporal artery and cortical branches of the middle cerebral artery; **c** right pial synangiosis (black arrow) with cerebral atrophy (curved black arrow); **d** postoperative with formation of mild neovascularization (white ring)



alterations in images of the present case, the CAMS II subgroup would be the most similar.

Sturge-Weber syndrome is a neurocutaneous syndrome that, due to veno-lymphatic malformations on the face and brain, is characterized by maxillofacial port-wine stains, cerebral atrophy, convulsions, cortical vessel and parenchymal calcifications, maxillofacial and cranial bone hypertrophy, ocular abnormalities (glaucoma and choroidal hemorrhage), and leptomeningeal angiomatosis [7, 8]. Venous malformations observed in Sturge-Weber syndrome can be considered CVMS. In the case presented, CVMS with port-wine stains on the face and image alterations characteristic of Sturge-Weber syndrome were evident.

Moyamoya disease is characterized by a progressive steno-occlusive disease in the terminal portions of the internal carotid arteries and their main branches [9]. This disease is related to neural crest pathology or neurochristopathy [10]. It is called syndrome when related to another clinical condition, such as the present case, i.e., neurocutaneous Sturge-Weber syndrome [11].

The metameric arrangement of intracranial arteries suggests that the region of the brain (more specifically derived from the prosencephalon and diencephalon), supplied by the primitive internal carotid arteries, is more prone to intimal thickening. Therefore, it is expected that the cerebral arteries in Moyamoya disease have preexisting structural defects that are not known and remain latent during vasculogenesis [12].

The gold standard image exam is cerebral angiography, which will more accurately demonstrate the stenotic vessels, the network of collateral vessels, in addition to small aneurysms that may be associated [11].

Due to the fact that this disease is characterized by progressive worsening in its evolution, the most appropriate approach would be surgery. There are two surgical techniques, namely, (1) direct revascularization with intracranial-extracranial anastomoses. The great disadvantage of this technique is children with small-sized vessels and (2) indirect revascularization performed with the contact of the vascularized tissue of the external carotid artery (dura mater, temporal muscle, or even branches of the superficial

temporal artery) and the surface of the brain. Unfortunately, this technique should not be used in an emergency, as a time interval is required for neovascularization [11]. The case described was treated with the technique of indirect revascularization by pial synangiosis. Neovascularization was only observed after 8 months.

A rare association between chronic moyamoya vasculopathy and cerebrofacial vascular metamerism syndrome was observed in the present case report. They are severe vascular malformations with few reports in the literature. Therefore, further studies should be conducted with the goal of establishing the best treatment approach.

Author contribution Conception and design: da Silva. Acquisition of data: all authors. Analysis and interpretation of data: all authors. Drafting the article: all authors. Critically revising the article: da Silva. Reviewed submitted version of manuscript: all authors. Approved the final version of the manuscript on behalf of all authors: da Silva

Declarations

Ethics approval This article does not contain any studies with animals performed by any of the authors.

Consent to participate Informed consent was obtained from parents.

Conflict of interest On behalf of all authors, the corresponding author states that there is no conflict of interest.

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