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Case Report

Cerebrofacial venous metameric syndrome type 2+3: face is the index of brain ☆

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ABSTRACT

We describe a rare case of a 20-year-old man who presented with an extensive facial and orbital venous malformation associated with multiple intracranial venous malformations. The co-existence of cerebrofacial venous malformations points towards a common final pathway in development of these malformations. Our findings are consistent with few previous similar case descriptions. In addition, we describe some novel observations which, to the best of our knowledge, have not been described in the literature. This case reinforces the concept of metameric and segmental distribution of cerebrofacial vasculature, and the aberrations thereof leading to the metameric venous malformations, as proposed by Lasjaunias et al.

Introduction

Cerebrofacial venous metameric syndromes (CVMS) include venous vascular malformations involving the head and neck in a metameric distribution [1,2]. These are rare vascular lesions and may be associated with a wide spectrum of intracranial venous abnormalities [3]. Sturge-Weber syndrome is the most common cerebrofacial venous metameric syndrome [4]. However, there are other phenotypical manifestations of CVMS, including venous malformations of the head and neck associated with developmental venous anomaly (DVA), cavernous hemangioma, cavernoma, and dural sinus malformations [3,5,6]. Herein, we present a case of CVMS with an extensive venous malformation involving the scalp and orbit with ipsilateral intracranial venous anomalies. Our findings further strengthen the hypothesis that the cerebrofacial venous malformations and intracranial venous anomalies may develop through a final common pathway, probably triggered by a genetic mutation during the period of embryogenesis and before the migration of the neural crest cells.

Case report

A 20-year-old man presented to us with multiple nodular swellings over the eye lids and over the lateral orbital margins on the right side since birth, which were blue in color, noncompressible, nontender, and non-pulsatile (Fig. 1). There

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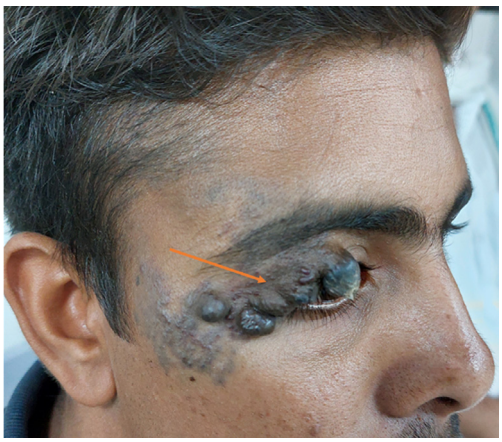

Fig. 1 – Photograph of the patient's face shows multiple nodular lesions involving the right eye lids and periorbital skin (arrow). The lesions have a bluish hue. Bluish discoloration is also noted over the forehead and skin over the right temporal region. These findings are suggestive of a vascular malformation.

was no significant change in their size on change in posture or Valsalva manoeuvre. Also, there was an asymmetry in the scalp with mild bulge over the right temporoparietal region with no palpable thrill over the bulge. The swellings showed proportional increase in size with overall physical growth of the face. The clinical findings raised the possibility of craniofacial vascular malformation and CT angiography of the head and neck was requested. Arterial phase of CT angiography did not reveal any enlarged arterial feeders or early draining veins within a large ill-defined swelling involving the subcutaneous tissues over the right frontal, temporal, and parietal region, extending over the eye lids on the right side (Fig. 2a). Multiple nodular soft-tissue attenuation lesions were also noted within the intraconal compartment in the right orbit (Fig. 2b). There was progressive enhancement of these lesions on subsequent venous phase, suggesting venous vascular malformations (Fig. 2c). Within the right cerebral hemisphere, prominence of the medullary veins was noted, which were seen merging into a common collector venous channel, suggesting a DVA (Fig. 2d). DVA were also noted in right periatrial white matter (Fig. 2e). Bilateral cerebellar DVA were also noted, draining into the vein of Galen (Fig. 2f). Abnormal

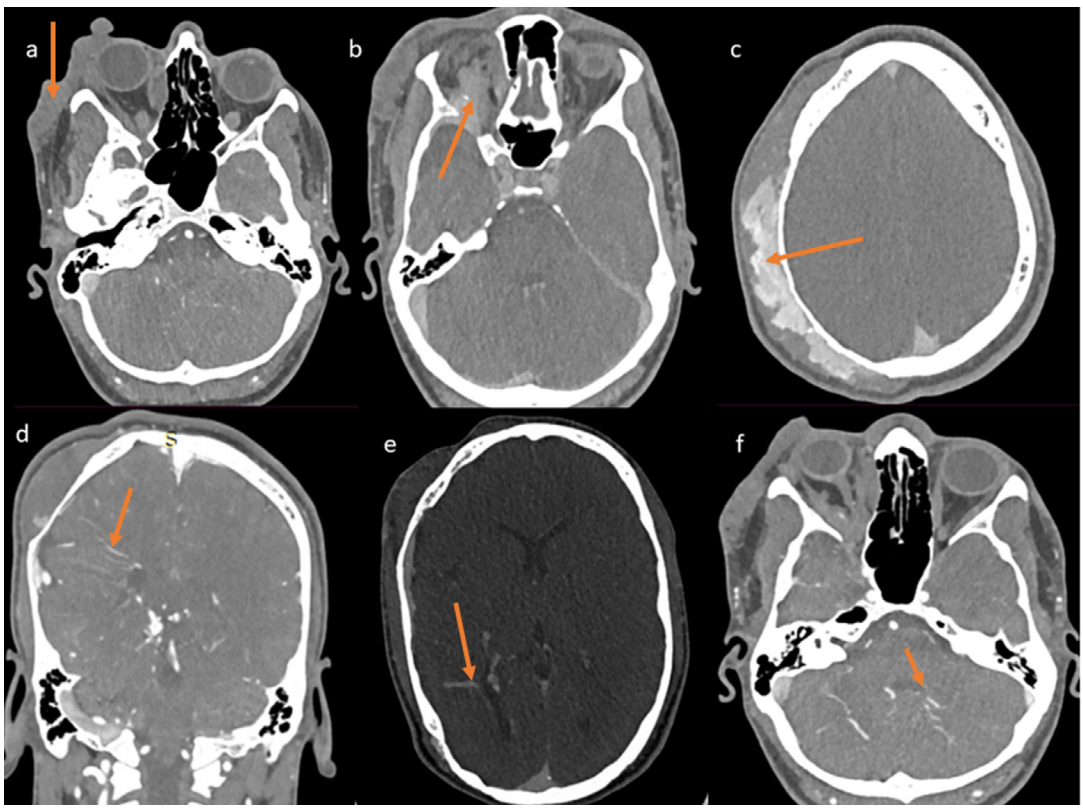

Fig. 2 – (a) Axial CT image shows nodular soft tissue thickening over the right periorbital and right temporal region (arrow). (b) Soft tissue nodular lesion also noted in the intraconal compartment of the orbit on right side. (c) Postcontrast axial CT in venous phase shows progressive enhancement of the soft tissue lesion in right frontoparieto-temporal region which is suggestive of a venous malformation (arrow). (d) Coronal CT venogram shows multiple prominent medullary veins (arrow) converging towards a common collector vein in right periatrial region, suggestive of a developmental venous anomaly. (e) Another right periatrial developmental venous anomaly is noted (arrow) on axial CT venogram. (f) Axial CT venogram shows developmental venous anomaly involving the left cerebellar hemisphere (arrow) which was seen draining into the Vein of Galen.

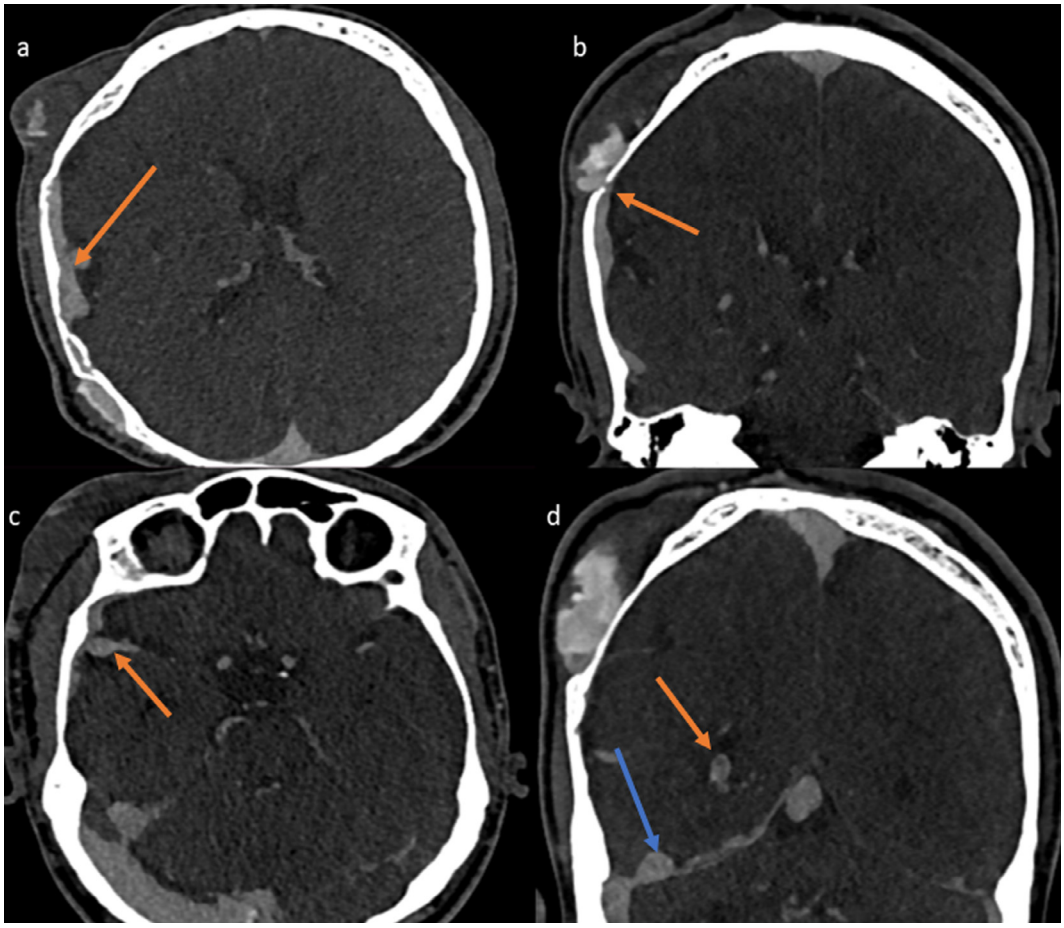

Fig. 3 – (a) Axial CT venogram shows an abnormal venous structure along the right frontoparietal convexity (arrow) which could represent a meningeal venous angioma. (b) Coronal CT venogram shows the abnormal venous structure along the right frontoparietal region communicating with the extracranial venous malformation through transdiploic venous channels (arrow). (c) Axial CT venogram shows venous ectasia (arrow) along the course of the right superficial middle cerebral vein within the right Sylvian fissure. (d) Coronal CT venogram shows prominence of right choroid plexus (orange arrow) and abnormal venous channels along the right tentorial leaflet (blue arrow).

venous structures were noted along the right frontotemporo-parietal convexity (Fig. 3a) which were seen communicating with the soft tissue thickening over the scalp through transdiploic venous channels (Fig. 3b). We considered these to represent meningeal venous angiomas. Remodeling of the calvarium was noted adjacent to these lesions. The right basal vein of Rosenthal and right superficial middle cerebral vein (SMCV) were prominent on the right side with venous ectasia along the right SMCV (Fig. 3c). The choroid plexus on the right side appeared prominent and abnormal venous channels were noted along the right tentorial leaflet (Fig. 3d). There was no hemiatrophy of the right cerebral hemisphere and no parenchymal calcifications were noted. MRI demonstrated multiple fluid-fluid levels within the orbital and scalp venous malformation on T2W images (Fig. 4a). Multiple DVAs were appreciated on SWI sequence, correlating with the CT findings (Figs. 4b–d). Phase contrast MR venogram showed the abnormal venous structures along the right frontotemporo-parietal convexity, communicating with the scalp venous malformation through transdiploic venous connections (Fig. 4e). The posterior part of superior sagittal sinus and right transverse sinus were abnormally enlarged (Fig. 4f). There was no suggestion of cavernoma on MRI. The patient did not give a history of seizures. There was no family history of similar complaints. Considering the distribution of the craniofacial venous malformations, involvement of the calvarium and intracranial venous abnormalities involving the supratentorial and infratentorial compartments, a diagnosis of CVMS type 2+3 was made.

Discussion

The cerebrofacial vascular metameric syndromes have been classified into 3 groups by Lasjaunias et al., depending on the topographical distribution of the vascular malformations with the median prosencephalic group involving the nose and hypothalamus, the lateral prosencephalic group involving the optic nerves, optic chiasm, thalamus, maxilla and retina, and the rhombencephalic group involving the cerebellum, Pons, and mandible [7]. This classification is based on the concept of segmental organization of the neural tube into 34 somites

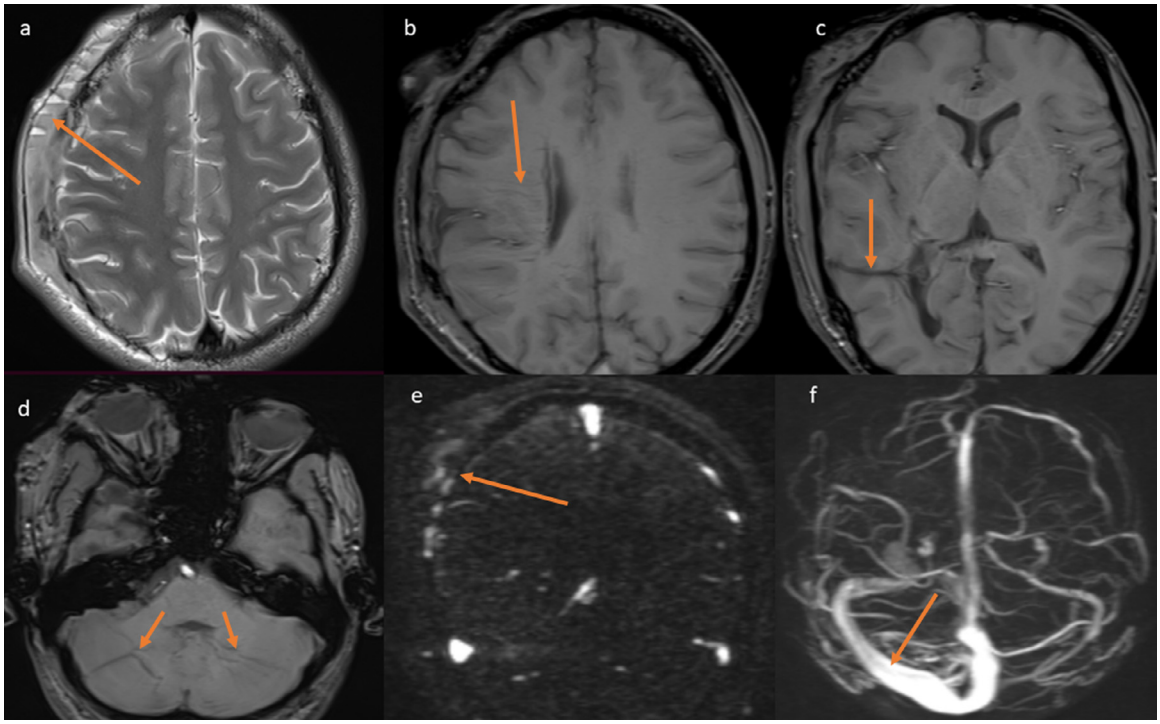

Fig. 4 – (a) T2W axial images show evidence of fluid-fluid levels within the venous malformation along the scalp in right frontoparieto-temporal region (arrow). (b) Axial susceptibility weighted image (SWI) shows prominence of medullary veins in right periventricular white matter (arrow) converging towards a common collector vein and forming a developmental venous anomaly. (c) Axial SWI image shows a developmental venous anomaly in right periatrial region (arrow). (d) Axial SWI image shows developmental venous anomalies in bilateral cerebellar hemispheres (arrows) draining into the Vein of Galen. (e) MR venogram shows transdiploic communication (arrow) between the venous angioma along the right parietal convexity and extracranial scalp venous malformation. (f) Maximum intensity projection (MIP) of the MR venogram shows abnormal dilatation of the right transverse and sigmoid sinuses (arrow) suggestive of dural venous sinus malformation.

(3 for the brain and 31 for the spinal cord) with subsequent migration of the neural crest cells in a metameric distribution [7]. Couly et al. described the regional origin of the endothelial cells in the cephalic region. The endothelium of the blood vessels is derived from the mesoderm, while the media is derived from the neural crest [8]. Due to the segmental distribution of the neural crest and mesoderm, the neural crest cells and the mesoderm at any segmental level occupy the same facial vascular territory, thus explaining the metameric nature of vascular malformations [8]. With the advances in research in the field of genetics, many genetic pathways involved in the development of vascular malformation have been unearthed, with multiple genes such as GNAQ, mTOR, PI3K, etc., which ultimately regulate the transcription of the growth factors, and mutations in one or more of these genes may result in specific venous metameric syndromes [3,9].

The association between the vascular malformations of the head and neck and intracranial vascular malformations has been demonstrated in previous studies. Brinjkji et al. [10] reported a significant association between DVA and facial venous malformations, with a prevalence of 28.6% of their study population (42 patients with facial venous malformations). It is noteworthy that in most of these patients, the DVA was ipsilateral to the facial venous malformation and along the same metamere. Similar results were reported by Boukobza et al. [11], who found a prevalence of 20% for patients with superficial venous malformations of the head and neck. In the index case which we have presented here, multiple DVAs were noted associated with an extensive venous malformation involving the right orbit and the scalp. However, we found an involvement of more than one metamere, that is, lateral prosencephalic as well as rhombencephalic, since DVA were found in the bilateral cerebellar hemispheres as well. This finding may be explained by the observations of Couly et al. that the metameric regionalization may allow some flexibility and is not strict, thus co-existence of a combination of CVMS may be seen in a given case, such as the one reported by Agid et al. [8,12]. Brinjkji et al. [3] also have described a spectrum of CVMS, ranging from various combinations of CVMS with associated DVA, "forme fruste" Sturge Weber Syndrome to classical Sturge Weber Syndrome. Our case has certain novel radiological findings which, to the best of our knowledge, have not been described previously. One, the DVAs seen in our case appears to be more extensive, both in number as well as the cerebral and cerebellar venous territory involved. An abnormal venous structure noted along the right cerebral convexity, with transdiploic communication with the external venous malformation along the scalp noted in the index case may represent meningeal venous angiomatous malformation. Leptomeningeal venous angiomas have been de-

scribed in SWS. However, in our case clinical and radiological findings are not consistent with SWS. The transdiploic connections between the intracranial and extracranial venous malformations may have a relevance as far as the injection sclerotherapy for facial and orbital venous malformations is concerned, since there may be a risk of intracranial migration of the embosylates through these connections, the clinical repercussions of which is yet to be reported in the literature. Thus, the findings in the index case reinforce the concept of metameric regionalization of cerebrofacial structures during the embryonic development, since the malformation involving the scalp, meninges, and calvarium in contiguity points toward their development from a common precursor. Ectasia along the course of the right superficial middle cerebral vein in association with CVMS is also a novel finding, hitherto not described in patients with CVMS.

In the index case, we noted abnormal morphology of the right transverse and sigmoid sinuses with prominent venous channels along the right tentorial leaflet. Such association between cervicofacial venous malformations and dural venous sinus abnormalities have been described previously by Brinjkji et al. [6]. Co-existence of DVA, facial venous malformations and dural venous sinus malformations may be explained by thrombosis or occlusion of veins during fetal development as a common pathway leading to these lesions [3,5,6]. In this regard, presence of coagulopathy and elevated D-dimer levels has been suggested as one of the factors leading to the development of venous malformations by Dompmartin et al. [13,14].

Conclusion

Our case is an example of the concept of continuum regarding cerebrofacial venous metameric syndrome. We have described few novel radiological findings in this case such as meningeal venous angiomatous malformation with transdiploic communication with extracranial scalp venous malformation and prominence of ipsilateral choroid plexus *sans* Stugre Weber Syndrome. We emphasize the need for screening the brain in any given case of vascular malformation involving head and neck.

Patient consent

The authors declare that an informed and written consent was obtained from the patient for the publication of this case report

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