Cerebrofacial Venous Metameric Syndrome 2 plus 3: Facial and Cerebral Manifestations

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Summary

A unique case of Cerebrofacial Venous Metameric Syndrome (CVMS) in a 25-year-old woman is described with venous malformations involving simultaneously the brain and the face. This case represents CVMS 2 plus 3 according to the classification described by Lasjaunias et Al. The metameric distribution of the cerebrofacial venous syndromes is well illustrated in this case. The extracranial and intracranial involvement is described and the appearance on MRI imaging is detailed. In our opinion this case demonstrates that cerebrofacial venous metameric syndromes include a wide spectrum of possible phenotypes including Sturge Weber syndrome and cases such as we describe here.

Case Report

A 25-year-old woman presented at the age of six years with facial asymmetry. This started at infancy as a small mark on the left face and developed into a very large orbito-maxillo-facial venous malformation (VM). She underwent partial surgical debulking of this VM at six and 15 years of age. Due to the orbital involvement, her left eye was shut, leading to progressive poor vision and finally blindness of this eye. Several months ago she had a prosthesis implanted to replace her left globe. At the age of two years she developed right-sided focal motor seizures that are treated with phenobarbital

with complete control and no seizures since six years of age. Due to mild retardation she needed special education and currently works as a volunteer in a kindergarten. There is no family history of venous malformations or seizures.

Clinical examination revealed a physically well-developed 25-year-old right-handed woman. She had an asymmetry of the face due to swollen left cheek and partially shut left eye (figure 1). Apart from left eye blindness she is neurologically intact. EEG examinations at ages 18 and 24 years demonstrated consistent amplitude asymmetry of the electrical cerebral activity with lower amplitude over the left hemisphere compared to the right. A single potentially epileptogenic paroxysmal discharge was recorded over the left temporo-parietal area.

Imaging MRI examinations at ages 16, 20 and 25 years (figure 2) demonstrated a left sided facial venous malformation involving the soft tissues of the forehead, eyelid, cheek and mandible and extending into the deep structures of the orbit, infra-temporal fossa and hard palate on the left. MRI of the brain reveals loss of volume of the left cerebral hemisphere. Multiple large developmental venous anomalies (DVA) were noted throughout the left cerebral hemisphere but also involving the cerebellum bilaterally. Abnormalities of the dural sinuses were also noted with an abnormally large posterior aspect of the superior sagittal sinus. The normal straight sinus is missing and instead a complex of several large

Figure 1 Our patient at age 25 years with facial asymmetry due to left sided facial venous malformation. Her left eye is still partially shut although her left eye globe was replaced by a prosthesis several months earlier.

DVAs are noted to drain the internal cerebral veins, the basal veins of Rosenthal and the cerebellum. Cavernous malformations are noted in the left aspect of the pons and in the left temporal lobe. These cavernomas became more evident on the later examination. The cavernoma located in the left aspect of the pons had acutely bled at age 16 and shows normal evolution post bleed at the follow-up studies.

Discussion

In 1995 Couly demonstrated that endothelial cells of the cephalic region have a regionalized origin providing blood vessels to specific regions of the face and brain¹. The neural crest and mesoderm cells arising from a certain transverse level occupy the same facial territories². Since the endothelium and the media of blood vessels are derived from mesoderm and neural crest respectively, Lasjaunias' group used this approach to regroup the neural tube and neural crest derivatives to associate between certain maxillo-facial and central nervous system malformations^{2,3,5}. They have suggested the name Cerebrofacial Arterial/Venous Metameric Syndrome (CAMS or CVMS) for

conditions with arterial or venous malformations simultaneously involving the brain and the face. This association, although recognized in the literature for many years was previously thought to be related to the course of the fifth cranial nerve as first suggested by Cushing in 1906⁶. According to the new classification three subgroups are identified within CAMS or CVMS. CAMS 1 or the "median prosencephalic group" - involves the hypothalamus and the hypophysis as the intracranial component and the nose as the facial component. CAMS 2 or the "lateral prosencephalic group" involves the occipital lobe, thalamus, optic tract, and retina intracranially and the maxilla as its facial component. CAMS 3 - or the "lateral rhombencephalic group" involves the cerebellum and pons intracranially and the mandible as the facial component⁷. Using this classification our case represents CVMS 2 plus 3. The metameric distribution of the cerebrofacial venous syndromes is well illustrated in this case.

There are relatively many reports of Sturge Weber syndrome in the literature^{3,8} which qualify for the CVMS criteria. Our case does not represent Sturge-Weber syndrome since the facial involvement is not a port wine stain and the intracranial involvement is not a leptomeningeal venous thrombosis, cortical enhancement and enlarged choroid plexus^{3,4,7}. Cortical calcifications is also absent in our case. The only similarity our patient has with Sturge-Weber syndrome is the presence of retardation and seizures in the face of slight hemiatrophy of the left cerebral hemisphere. Our patient most likely has an underdeveloped superficial venous system as indicated by the presence of a prominent deep venous system. This could explain the decreased brain volume on the left.

We did not find many reported cases of CVMS describing facial venous malformations accompanied by intracranial DVAs, cavernous malformations, ectatic dural sinuses and deep draining veins such as reported here. The only such report is by Boukobza et al⁹. This group studied cerebral DVAs in 40 patients with extensive venous malformations of the head and neck. They found DVAs in eight of these patients (20%), four of them were found to harbor multiple intracranial DVAs. One patient (2.5%) had an intracerebral cavernoma but no DVA. None of their patients had both DVAs and cavernomas. Four patients had unilateral facial venous malformations. Although the au-

Figure 2 A-K) MRI and MR Venography (MRV) of the head at 25 years of age.

Facial involvement (A-C): Coronal T2-weighted images (A-B) and axial T1-weighted image (C) show extensive left sided venous malformation, hyperintense on T2 and isointense on T1 weighted images. The malformation overlies the left forehead, maxillary and mandibular regions and invades the orbit and the infra-temporal fossa (arrows).

Intracranial involvement (D-K): Contrast enhanced MRV (D-E) in 3D reconstruction (D) and source coronal image (E) demonstrates an abnormally dilated posterior aspect of the superior sagittal sinus (star), as well as absence of the straight sinus with a complex of several large DVAs draining the internal cerebral veins, the basal veins of Rosenthal and the cerebellum (arrow heads). Axial contrast enhanced T1 weighted images (F-I) shows the multiple DVAs in the left cerebral hemisphere and bilaterally in the cerebellum (arrowheads). Coronal T2 weighted images (J-K) reveal three cavernous hemangiomas (bold arrows) - two in the left temporal lobe and one in the left middle cerebellar peduncle. Images H and K also demonstrate the enlarged left lateral ventricle and the slight volume loss of the left cerebral hemisphere.

thors of this paper mention that one of the “hemifacial patients” had bilateral intracranial DVAs, they did not correlate the laterality of the facial findings with that of the intracranial findings. The findings in our patient are strikingly similar to those found in Boukobzas’ report regarding atypical parenchymatous cerebral venous drainage⁹. This includes multiple draining vessels in the white matter converging in a fanlike pattern toward enlarged venous channels as well as enlargement of the basal vein and anomalous tributaries of the vein of Galen confluence. As opposed to the patients in Boukobzas’ report, our patient presented with clinical neurological findings. These are attributed to bleeding from one of her cavernomas and to the volume loss of her left cerebral hemisphere presenting with seizures.

The intracranial findings in our case, although predominantly ipsilateral to the facial involvement, do involve to a lesser extent the contralateral cerebellum. This could be explained by Couly’s comment that the regionalization of the cephalic neural crest and mesoderm is not strict¹. In his opinion this may allow some fluidity in the final phenotypic expression. This is supported by similar bilaterality reported by Boukobza et Al in one of their unilateral facial malformation patients⁹.

This case as well as the cases described by Boukobza et Al and Sturge Weber syndrome all match the criteria of CVMS and most likely represent part of a wide spectrum of possible phenotypes included in the cerebrofacial venous metameric syndromes.

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