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Heterogeneous Continuum of Cerebral and Cervicofacial Venous Malformations

In this issue, Brinjiki et al¹ demonstrate a remarkable association between cervicofacial venous malformations (VMs) and cerebral developmental venous anomalies (DVAs), as well as between cervicofacial VMs and dural venous sinus anomalies in a retrospective study involving healthy age- and sex-matched controls. In this study, the venous malformations involving the face and neck drain into the external jugular system and fit into the following criteria: septate lobulated mass hyperintense on T2 and hypointense on T1 MR images without mass effect, the presence of phleboliths and fluid-fluid levels, the absence of vascular flow voids on spin-echo sequences, infiltration of the lesion into tissue planes, the absence of arterial or early venous enhancement, and the presence of diffuse enhancement on delayed MR images. Intracranial venous drainage was evaluated by the presence of a DVA, superficial-versus-deep drainage of a DVA, side and location of a DVA, and the presence of cavernous malformations and dural venous sinus anomalies such as dural ectasia and persistent falcine sinuses (PFS). The authors have also examined the combined VM-DVA cases for their occurrence in the same metamere, either medial prosencephalic, lateral prosencephalic and/or otic (rhombencephalic/mesencephalic) as the case may be. Their findings suggest that VMs result from a segmental in utero insult to cells involved in cerebral/cervicofacial venous development.

They found a significant association with facial venous malformations and ipsilateral DVAs, concordant with the existing literature, and a novel observation of dural venous sinus anomalies associated with the PFS and torcular ectasia in 6% and 10% of VMs, respectively. Of note, the authors should be commended for examining over 550 venograms of patients 0–20 years of age who did not have facial VMs and who did not have any dural sinus anomalies. I would like to present these anomalies as a part of the heterogeneous continuum, which comprises intracranial venous and dural sinus malformations, associated with cavernous angiomas, telangiectasias, and venous ectasias or varices. These are well-elucidated by the seminal article by Lasjaunias (with multiple posterior fossa DVAs, cavernous malformations, and torcular ectasia from dural sinus malformation), as well as many articles published by the prominent cerebrovascular group led by Robert Spetzler, already referenced in the article.

Several interesting observations are presented by the authors:

1. The cervicofacial VMs and intracranial DVAs appear to have occurred along the same side in same metameres. Metameric lesions as in cerebrofacial arteriovenous metamerism syndrome, cerebrofacial venous metamerism syndrome, and spinal arteriovenous metamerism syndrome are already described in literature; however, the underlying genetics and metamerism association of intracranial DVAs need to be studied in a larger prospective study. I would like to compare the authors' work with that of Oza et al,² who demonstrated focal cerebellar atrophy, lip/nasal cleft, and gray matter heterotopias occurring on the same side as a facial hemangioma in posterior fossa malformations, hemangiomas, arterial anomalies, cardiac defects, eye abnormalities, and sternal or ventral defects (PHACES) syndrome.
2. The in utero insult does not explain how the presence of cervicofacial VMs favors development of other listed intracranial malformations in the continuum in infants and adults; moreover, there has been evidence of de novo development of pediatric and adult cavernous malformations and intracranial DVAs in patients on close radiologic surveillance. Cranial irradiation, coexistent vascular malformations, genetic/hormonal factors, previous intracranial surgery, or even apparently unrelated intracranial lesions have been considered risk factors for de novo cavernomas in the brain.^{3–9} There is evidence of multiple bilateral cavernomas with and without a family history, the interval growth being generally attributed to recurrent bleeds (rather than tumor tissue proliferation); however, there are cases of lesion growth reported without hemosiderin in or around these lesions. The cavernomas seen in association with DVAs are most likely of acquired etiology rather than true congenital vascular malformations; this is also supported by the fact that the coexistence of cavernous malformations and DVAs is more common in the adults than children.
3. The authors have demonstrated a subset of venous malformations with dural venous sinuses including PFS and persistent ballooning of the torcula, the latter without obvious laterality, in a limited subset of patients. The associated dystrophy of adjacent brain around venous and dural sinus anomalies is

not adequately studied, given the limited sample size. The juxtaposed developmental brain anomalies as well as the hypometabolic activities of the brain in the vicinity of these venous malformations should be evaluated vividly in symptomatic cases. For example, Manjila et al¹⁰ classified the PFS, with and without the association of supratentorial brain ectasia associated with these venous/dural sinus anomalies. Similarly, Larvie et al¹¹ published metabolic abnormalities in the adjacent brain parenchyma, as assessed by FDG-PET, in more than three-fourths of DVAs in a series of 25 cases.

Future studies on brain ectasia/atrophy and focal metabolic brain activities around DVAs as well as the genetic role in metamer venous malformations (as in *PIK3CA* and *TEK* genes) can provide more useful clinical and radiologic insights into the developmental genetics of these uncommon phenotypes in the heterogeneous continuum of cervicofacial and cerebral venous malformations.

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