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CEREBROFACIAL VENOUS METAMERIC SYNDROME: A RARE VASCULAR MALFORMATION DISORDER

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RESUMO

Introduction Cerebrofacial venous metameric syndrome is a complex vascular malformation disorder, where patients may present multiple malformations involving soft tissue, bone structures, and nerve structures. This disorder is quite rare, accounting for about 0.5% of all cerebrovascular malformations. The classic clinical presentation includes cutaneous, soft tissue, and intracranial lesions within the spectrum of low-flow venous malformations. **Objective** The study aims do show a case of cerebrofacial venous metameric syndrome. **Methods** The case reported is of an 11-month-old female child, followed since birth due to an expansive lesion in the right hemiface. Cranial magnetic resonance showed a heterogeneous lesion in the right hemiface with frontal, zygomatic, and orbital maxillary components, associated with transverse and superior sagittal sinus ectasia with apparent persistence of the falcine sinus, as well as supratentorial dilation. In post-contrast, an anomaly of venous development was confirmed, associated with VAD in the region of the bilateral capsular nucleus. **Results** Cerebrofacial venous metameric syndrome is a complex vascular malformation disorder, where patients may present multiple malformations involving different tissues. It represents less than 1% of cerebral vascular malformations. The pathogenesis involves an early dysfunction of the neural crest and groups of mesodermal cells in a given metamer, which can lead to vascular malformations during differentiation and migration. These changes can be focal or related to multiple segments and tissues. These characteristics favor a classification according to the craniofacial metamers involved, where group 1 is related to the medial forebrain segment, with involvement of the nose, hypothalamus, and orbit; group 2 maintains a relationship with the lateral forebrain segment, with involvement of the parietal, temporal, occipital, optic nerve, retina and maxilla; group 3 is related to the rhombencephalon segment, with involvement of the mandible, cerebellum, and pons. The case described presents an overlapping characteristic of groups 2 and 3. The clinical presentation of these patients occurs through cutaneous, soft tissue, or intracranial lesions of the spectrum of slow-flow vascular lesions, including cavernomas and venous developmental anomalies. The radiographic

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alterations are diverse, mainly showing findings of anomalies and venous malformations that can affect the different cerebrofacial segments, depending on their pathological embryonic origin. In the case reported here, there is a heterogeneous lesion on the right hemiface with frontal, zygomaticomaxillary, and orbital components, associated with transverse and superior sagittal sinus ectasia with apparent persistence of the falcine sinus, as well as supratentorial dilatation. In post-contrast, an anomaly of venous development was confirmed, associated with VAD in the region of the bilateral capsular nucleus. Among the therapeutic proposals, there is embolization or microsurgery, as independent or synergistic treatments, being curative in a minority of patients. However, most malformations of this complexity have extensive collateral involvement and are therefore incurable by any means. **Conclusion** CVMS is a complex disease that has a wide spectrum of manifestations ranging from extensive VADs and associated facial venous malformations, which may not present neurological complications or even have debilitating neurological involvement with cerebral atrophy associated with seizures and developmental delay.

PALAVRAS-CHAVE: CVMS, vascular malformation, neuroradiology, cerebrofacial venous metameric syndrome

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