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CEREBROFACIAL ARTERIOVENOUS METAMERIC SYNDROME: IMAGING FEATURES OF A RARE ENTITY

Antônio Gomes Lima Junior
Clinical Research Department, Icahn Medical School of Medicine at Mount Sinai, New York, NY, United States

Carolina Marques Ribeiro Naccarato
Department of Radiology, Antonio Prudente Hospital, Fortaleza, CE, Brazil

João Paulo Carmo Rodrigues
Department of Radiology, Antonio Prudente Hospital, Fortaleza, CE, Brazil

Carlos Alberto Santillán Vilchez
Department of Radiology, Antonio Prudente Hospital, Fortaleza, CE, Brazil

Marcus Duarte de Carvalho
Department of Radiology, Antonio Prudente Hospital, Fortaleza, CE, Brazil

Nina Maia Pinheiro Abreu
Department of Radiology, Antonio Prudente Hospital, Fortaleza, CE, Brazil

Julliana Melo Gomes
Department of Radiology, Antonio Prudente Hospital, Fortaleza, CE, Brazil

Maria Fernanda Lucena Karbage
Medical School, Unichristus University, Fortaleza, CE, Brazil

Pablo P.A. Coimbra
Department of Radiology, Antonio Prudente Hospital, Fortaleza, CE, Brazil

Abstract: Cerebrofacial venous/arteriovenous metameric syndrome (CVMS or CAMS) is a rare complex vascular malformation disorder, which patients can present with multiple possible malformations involving soft tissue, bone structures, and nerve structures (Brinjikji et al., 2020). The clinical presentation depends on the location and extension of the lesion, and the classification is based on the embryological origin of the affected structure (Bhattacharya et al., 2001). This article presents a pediatric case of CVMS with overlapping features of two classifications (CMVS 2 and 3).
Keywords: Cerebrofacial arteriovenous metameric syndrome; vascular abnormalities; developmental venous anomalies.

INTRODUCTION

CVMS is rare, accounting for approximately 0.5% of all cerebrovascular malformations (Willinsky et al., 1990). The classic clinical presentation includes cutaneous, soft tissue, and intracranial lesions within the scope of low-flow venous malformations, such as cavernomas or developmental venous anomalies (DVAs) (Brinjikji et al., 2020) (Brinjikji et al., 2018). To assess intracranial vascular abnormalities, contrast-enhanced magnetic resonance imagining (MRI) of the brain should be strongly considered primarily due to its high negative predictive value and specificity, making it a valuable non-invasive tool (Jiarakongmun et al., 2002) to rule in the disease and proceed with further investigation or treatment plan.

CASE PRESENTATION

The case reported is of an 11-month-old female child, followed by a neuro pediatrician and plastic surgeon since birth due to an expansive lesion in the right hemiface. In addition, diffuse microangiomas were present throughout her body. Cranial magnetic resonance was requested to investigate and

delimit the facial lesion, which showed a heterogeneous lesion in the right hemiface with frontal, zygomatic, and orbital maxillary components (Figures 1-4), associated with transverse and superior sagittal sinus ectasia with apparent persistence of the falcine sinus (Figures 3-5), as well as supratentorial dilation (Figures 1,2 and 4). In post-contrast, a DVA in the region of the bilateral capsular nucleus was confirmed.

DISCUSSION

CVMS is a complex vascular malformation disorder, where patients may present multiple malformations involving different tissues (Brinjikji et al., 2018) and different symptoms, depending on the involved metamer and structure. It is a rare disorder, representing less than 1% of cerebral vascular malformations (Willinsky et al., 1990). The pathogenesis of this syndrome involves an early dysfunction of the neural crest and groups of mesodermal cells in a specific metamer caused by somatic mutation (Fernández-Gajardo et al., 2018) (O'Loughlin et al., 2017), which can lead to vascular malformations during differentiation and migration (Bhattacharya et al., 2001) (Agid & Terbrugge, 2007). These changes can be focal or related to multiple segments and tissues (Agid & Terbrugge, 2007). These characteristics favor a classification according to the craniofacial metamers involved, where group 1 (CVMS 1) is related to the medial prosencephalon, with possible involvement of the nose, hypothalamus, and orbit; group 2 (CVMS 2) maintains a relationship with the lateral prosencephalon segment, with involvement of the parietal, temporal, occipital, optic nerve, retina and maxilla; group 3 (CVMS 3) is related to the rhombencephalon segment, with involvement of the mandible, cerebellum, petrous bone, and pons (Bhattacharya et al., 2001) (Brinjikji et al., 2020). The clinical presentation of these patients is related to which areas and metamers are affected (Ramli et al., 2003), occurring through cutaneous, soft tissue, or intracranial lesions of the spectrum of slow-flow vascular lesions, including cavernomas and DVAs (Brinjikji et al., 2020). The radiographic alterations are diverse, mainly showing findings of anomalies and venous malformations that can affect the different cerebrofacial segments, depending on their pathological embryonic origin (Brinjikji et al., 2020). In the presented case, 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, a DVA was confirmed, associated with another DVA in the region of the bilateral capsular nucleus. It is seen that the above patient has mixed characteristics of all three CVMS groups, making it difficult to classify it in just one group (Agid & Terbrugge, 2007) due to distinct lesions' locations. Among the therapeutic proposals, there is sclerotherapy (Brinjikji et al., 2020), embolization (Fernández-Gajardo et al., 2018) or surgery (Boukobza et al., 1996), as independent or synergistic treatments, depending on the patients' presentation. However, most malformations of this complexity have extensive collateral involvement, risk of hemorrhage and complex anatomy areas, generating a challenging condition to treat (Krings et al., 2007).

CONCLUSION

CVMS is a complex disease with a wide spectrum of manifestations such as extensive DVAs with associated facial venous malformations (Brinjikji et al., 2018), which may not present neurological complications or even have debilitating neurological

involvement with cerebral atrophy associated with seizures and developmental delay (Krings et al., 2007). Knowledge of the various manifestations is important for treatment and screening purposes, with contrast-enhanced magnetic resonance imaging of the brain being a valuable tool in the assessment of intracranial vascular abnormalities.

IMAGES

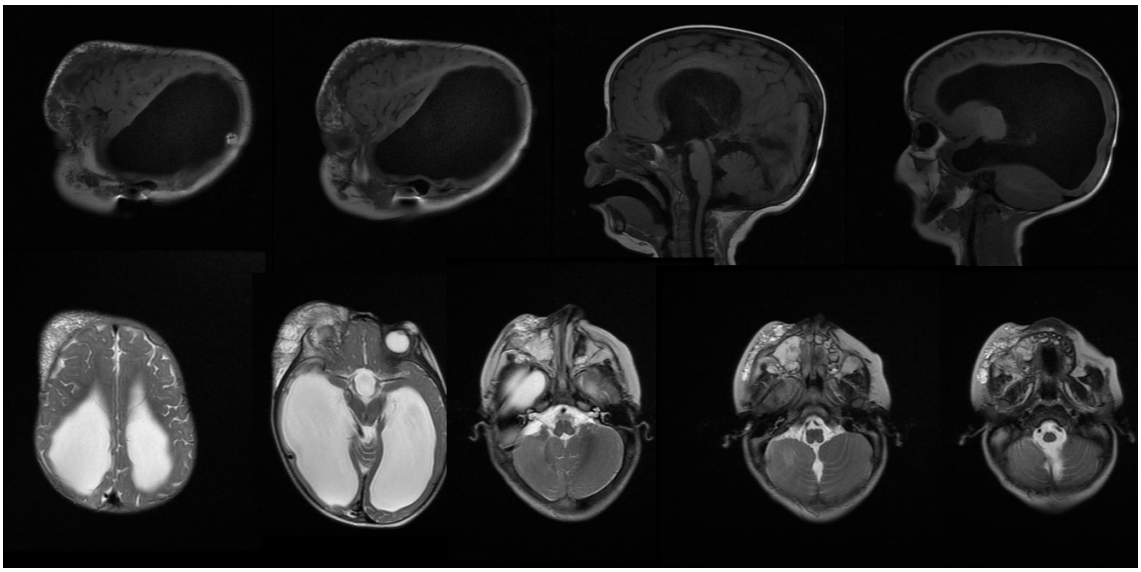

Figure 1

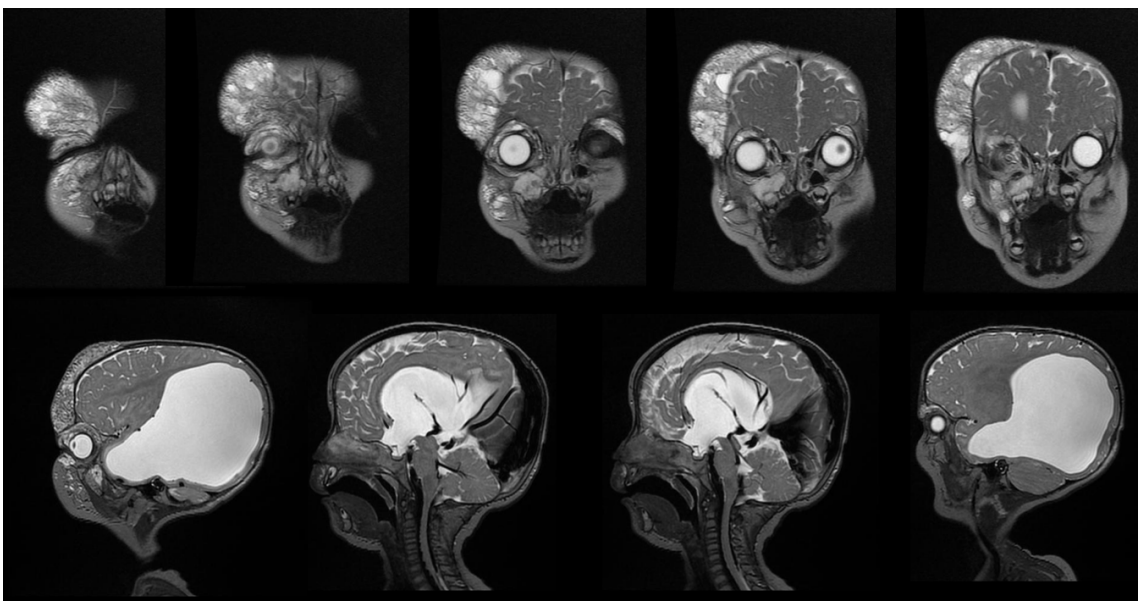

Figure 2

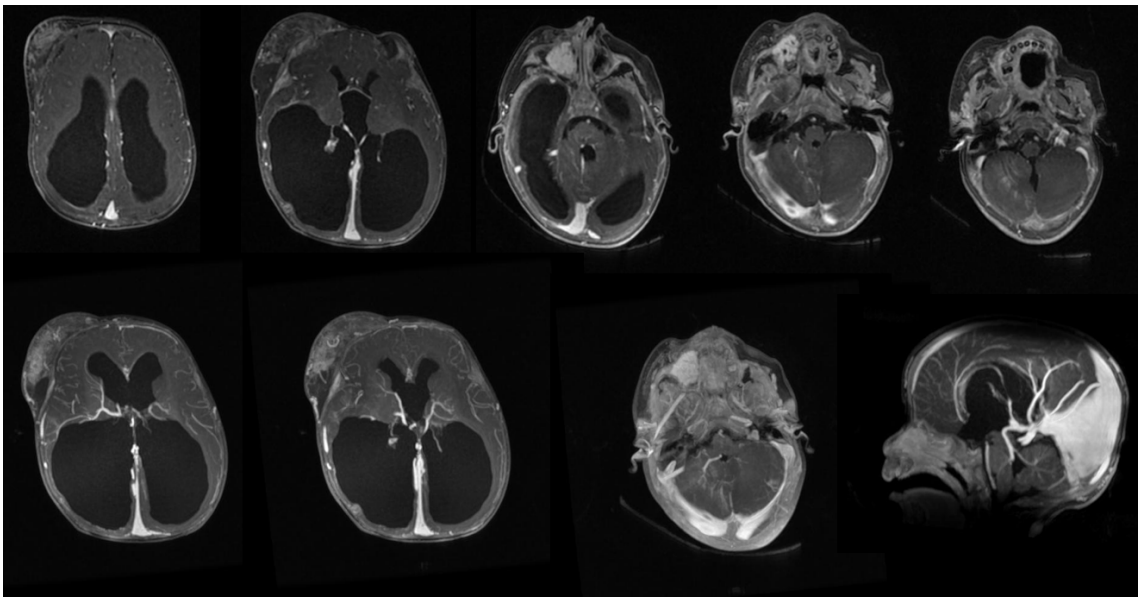

Figure 3

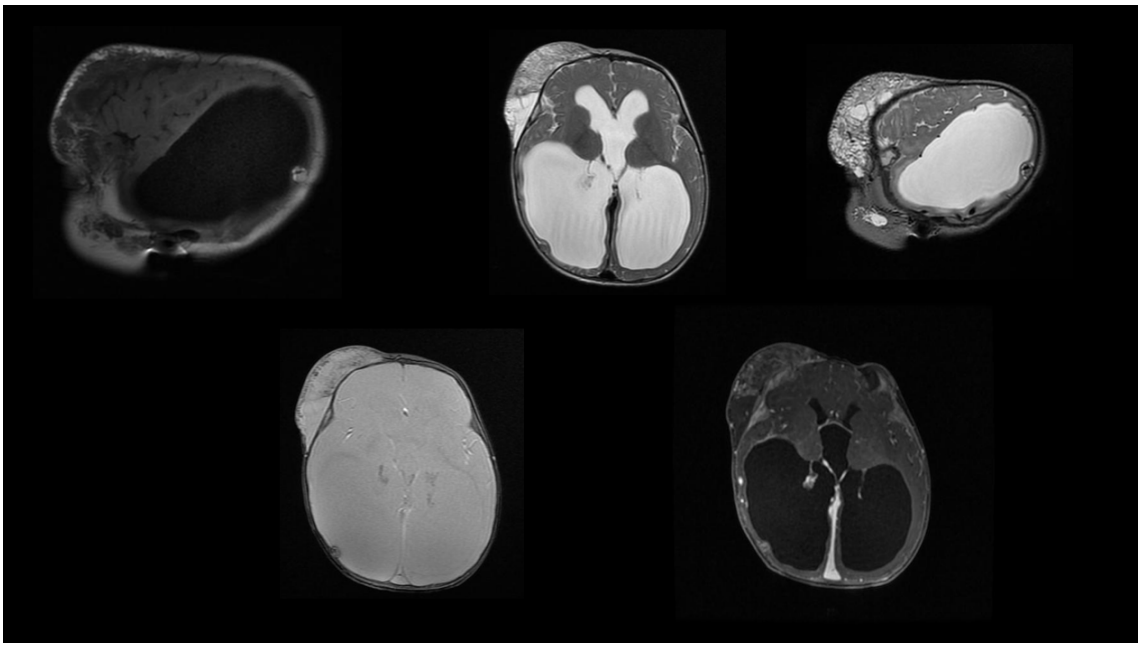

Figure 4

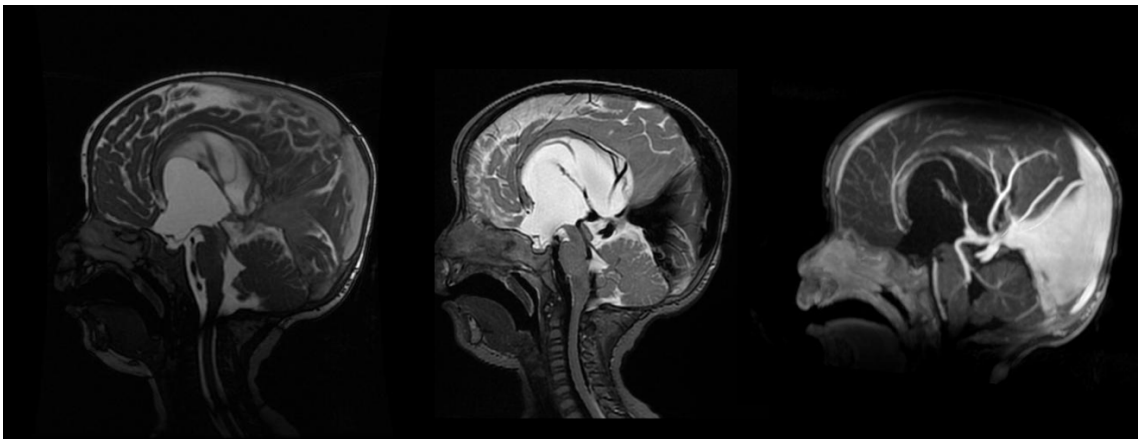

Figure 5

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