

# DYKE-DAVIDOFF-MASSON SYNDROME AND CHIARI TYPE II MALFORMATION

## SÍNDROME DE DYKE-DAVIDOFF-MASSON E MALFORMAÇÃO DE CHIARI TIPO II

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### RESUMO

A Síndrome de Dyke-Davidoff-Masson é uma síndrome associada à epilepsia refratária. A malformação de Chiari II é uma malformação congênita complexa do cérebro. Os autores relatam um caso de uma adolescente de 15 anos apresentando a síndrome de Dyke-Davidoff-Masson associada à malformação de Chiari tipo II. Este caso demonstra uma associação incomum nos exames de neuroimagem que indica a necessidade de avaliar doenças associadas, como mielomeningocele, disgenesia do corpo caloso e a siringohidromielia.

**Palavras-chave:** imagem por ressonância magnética; malformação de Arnold-Chiari; adolescente; encefalopatias

### ABSTRACT

Dyke-Davidoff-Masson Syndrome is a syndrome associated with refractory epilepsy. The Chiari II malformation is a complex congenital malformation of the brain. The authors report a case of a 15 years-old adolescent presenting Dyke-Davidoff-Masson syndrome and Chiari type II malformation association. This case demonstrates an unusual association in neuroimaging tests that indicates the need to evaluate associated diseases, such as myelomeningocele, corpus callosum dysgenesis and syringohydromyelia.

**Keywords:** magnetic resonance imaging; Arnold-Chiari malformation; adolescent; encephalopathies

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## INTRODUCTION

Dyke-Davidoff-Masson syndrome (DDMS) is a syndrome associated with refractory epilepsy and was first described by Dyke, Davidoff and Masson in 1933.<sup>1,2</sup> DDMS was originally characterized by its radiologic features, which include cerebral hemiatrophy and ipsilateral skull hypertrophy with hyperpneumatization of the paranasal sinuses and mastoid cells.<sup>3</sup> Since then, there were few case reports in the literature.<sup>1</sup> Both sexes and any of the hemispheres may be affected but male gender and left hemisphere involvement are more frequent. Age of presentation depends on the time of neurologic insult, and characteristic changes may be seen only in adolescence.<sup>1</sup>

Thus etiology of DDMS may be classified as congenital or acquired. In the congenital type, there is usually no apparent etiologic factor and the symptoms are present at birth or shortly thereafter. In this category, the cerebral damage most likely occurs during intrauterine life which might be due to intrauterine vascular occlusion. In the acquired type, the symptoms are related to central nervous system damage that occurs in the perinatal period or later.<sup>4</sup> Among the etiologic factors are trauma, infection, vascular abnormality, and ischemic and hemorrhagic conditions.<sup>5</sup>

The Chiari II malformation is a complex congenital malformation of the brain, nearly always associated with myelomeningocele, and is the most common serious malformation of the posterior fossa. This condition has skull, dural, brain, spinal, and spinal cord manifestations, including downward displacement of the medulla, fourth ventricle, and cerebellum into the cervical spinal canal, as well as elongation of the pons and fourth ventricle, probably due to a relatively small posterior fossa.<sup>6</sup>

Exact etiology so far is unknown. Chiari II malformation is best explained with the theory of McLone and Knepper, which allows the hindbrain disorder to be conceptualized as resulting from a normal-sized cerebellum developing in an abnormally small posterior fossa with a low tentorial attachment.<sup>7</sup>

## CASE REPORT

A 15 years-old adolescent complaining of imbalance without other comorbidities, performed routine magnetic resonance imaging (MRI) for subsequent control of ventricular shunt. MRI detected two ventricular shunt catheters at right, Chiari II malformation, with shallow posterior fossa, upward transtentorial herniation of the cerebellum, cerebellar tonsils herniation through the foramen

magnum, interdigitation of the sulci and dysgenesis of the corpus callosum, as well as diffuse volumetric reduction of the right cerebral hemisphere, with periventricular white matter gliosis (periventricular leukomalacia) and porencephalic cysts in the thalamus and the frontal lobe, and also thickened diploe of the frontal bone, with enlargement of the ipsilateral frontal sinus, featuring Dyke-Davidoff-Masson syndrome (Figures 1 and 2).

## DISCUSSION

In DDMS, the hemiatrophic cerebral parenchyma will have prominent sulci if the vascular insult occurs after birth or after sulcation is complete.<sup>4</sup> On the other hand, if the vascular ischemia occurs during embryogenesis, when the formation of gyri and sulci is incomplete, no prominent sulci will be present.<sup>4</sup> Encephalomalacia, gliosis, porencephaly, loss of white and grey matter, hypoplastic cerebral peduncle, thalamus and internal capsule, ventricular enlargement and midline shift toward the atrophic side may also be present in the hemiatrophic brain.<sup>4</sup>

The compensatory skull changes reflect adaptations to the unilateral decrease of brain substance and consist of ipsilateral calvarial thickening (diploe space and inner table) with loss of convolutional markings of the inner table of the skull, over-development of the paranasal sinuses (mainly frontal) and mastoid air cells, elevation of the petrous ridge, sphenoid wing and orbital roof, diminished size of the middle/anterior cranial fossae and displacement of falx attachment.<sup>1,4</sup>

Clinically, patients may present with seizures, facial asymmetry, contralateral hemiparesis and mental retardation.<sup>2,3,5</sup> The main differential diagnoses are hemimegalencephaly, Sturge-Weber syndrome (calcification with laminar cortical necrosis and atrophy), and Rasmussen encephalitis (no changes in the skull).<sup>2</sup>

Computed tomography (CT scan) and MRI shows enlargement of the sinuses of the ipsilateral face, thickening of the calvaria, ventricular enlargement and prominence of the grooves between the gyri. Although CT scan is the first examination to be asked, it is considered a MRI exam to evaluate the etiology and extent of involvement of brain parenchyma in patients with a combination of symptoms.<sup>8</sup>

Shen et al proposed three image patterns at RM:<sup>9</sup>  
 Pattern I: diffuse cortical and subcortical atrophy.  
 Pattern II: diffuse cortical atrophy associated with expanded porencephalic cysts.

Pattern III: old infarction with necrosis in the territory of the middle cerebral artery.

The anomalies associated with Chiari II malformations include the following:<sup>6</sup>

- Myelomeningocele (88-100%)
- Dysgenesis of corpus callosum (80-90%)
- Obstructive hydrocephalus following closure of myelomeningocele (50-98%)
- Syringohydromyelia (50-90%)
- Aqueductal stenosis (70%)
- Absence of septum pellucidum (40%)
- Contracted, narrow gyri (stenogyria; 50%)
- Heterotopias
- Diastematomyelia
- Segmentation anomalies (< 10%), incomplete C1 arch
- Malrotation of the posterior arches of C1 and C2
- Low-lying, often-tethered conus medullaris below lumbar nerve L2
- Rare anomalies
- Holoprosencephaly
- Cervical myelocystocele
- Frontometaphyseal dysplasia
- Juvenile distal spinal muscular dystrophy
- Williams syndrome

Chiari II malformations are encountered relatively commonly with an incidence of ~1:1000 live births.<sup>6</sup> The incidence is gradually rising because of increased detection with MRI.<sup>10</sup>

Given the wide range of anatomical severity as well as a large number of associated abnormalities which are sometimes encountered, it should be no surprise that the clinical presentation and the treatment of patients with Chiari II malformations is also varied both in character and severity.<sup>6</sup>

## CONCLUSION

This case demonstrates an unusual association in neuroimaging tests that indicates the need to evaluate associated diseases, such as myelomeningocele, corpus callosum dysgenesis and syringohydromyelia.

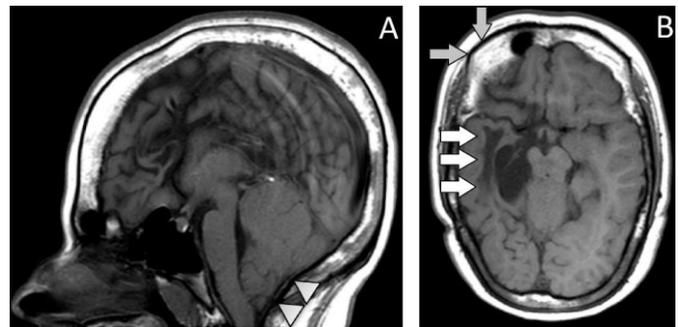


Figure 1: Magnetic resonance imaging in the T1-weighted sequence in the sagittal section in A and in the axial section in B demonstrating the small posterior fossa associated with cerebellar tonsil herniation through the foramen magnum (arrowheads), diffuse volumetric reduction of the right cerebral hemisphere (white arrows), and thickening of the frontal bone diploe and hyperaeration of the ipsilateral frontal sinus (gray arrows).

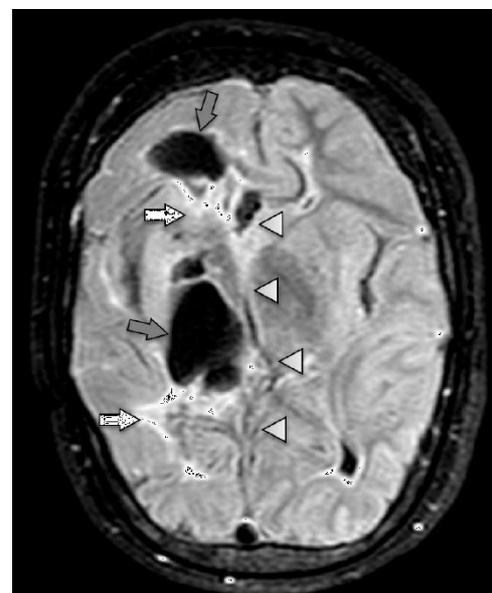


Figure 2: Magnetic resonance imaging in FLAIR-weighted sequence in the axial section demonstrating interdigitation of the grooves and dysgenesis of the corpus callosum (arrow heads), periventricular white matter gliosis (white arrows) and porencephalic cysts in the thalamus and frontal lobe (gray arrows).

## CONFLICT OF INTEREST

The authors declares that there is no conflict of interest.

## REFERENCES

1. Prerna Malik, Rajinder Garg, Anil kumar D Gulia, Joginder Kario, Dyke-Davidoff-Masson Syndrome- a rare cause of refractory epilepsy. *Iran J Psychiatry* 2014; 9(1): 42-44.
2. Roy U, Panwar A, Mukherjee A, Biswas D. Adult Presentation of Dyke-Davidoff-Masson Syndrome: A Case Report. *Case Reports in Neurology*. 2016;8(1):20-26.
3. Dix JE, Cail WS. Cerebral hemiatrophy: classification on the basis of MR imaging findings of mesial temporal sclerosis and childhood febrile seizures. *Radiology*. 1997;203 (1): 269-74.
4. Aguiar PH, Liu CW, Leitão H *et al.* MR and CT imaging in the Dyke-Davidoff-Masson syndrome. Report of three cases and contribution to pathogenesis and differential diagnosis. *Arq Neuropsiquiatr* 1998;56(4):803-7.
5. Singh P, Saggarr K, Ahluwalia A. Dyke-Davidoff-Masson syndrome: Classi-

cal imaging findings. J Pediatr Neurosci 2010;5 (2): 124-5.

6. Ozek MM, Cinalli G, Maixner W. Spina Bifida. Milan: Springer; 2008.

7. McLone DG, Knepper PA. The cause of Chiari II malformation: a unified theory. Pediatr Neurosci 1989; 15(1):1-12.

8. Kirks DR, Griscom NT. Practical pediatric. Boston: Lippincott Williams & Wilkins; 1998.

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9. Shen WC, Chen CC, Lee SK, Ho YJ, Lee KR. Magnetic resonance imaging of cerebral hemiatrophy. J Formos Med Assoc 1993;92:995-1000.

Ganesh D, Sagayaraj BM, Barua RK, Sharma N, Ranga U. "Arnold Chiari Malformation with Spina Bifida: A Lost Opportunity of Folic Acid Supplementation". J Clin Diagn Res 2014;8(12):OD01-3.