

Dyke-Davidoff-Masson syndrome, a rare entity in pediatrics. About a case

Síndrome Dyke-Davidoff-Masson, una entidad poco frecuente en pediatría. A propósito de un caso

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Summary

Dyke-Davidoff-Masson syndrome is a clinical-radiological entity characterized by loss of the volume of a cerebral hemisphere with ipsilateral hypertrophy of the skull, elevation of the sphenoid wing and the petrosal edge of the temporal bone, as well as hyperpneumatization of the frontal sinus and mastoid cells associated with contralateral hemiplegia, facial asymmetry, seizures and intellectual disability. Initially described in 1933 by Dyke, Davidoff, and Masson. It mainly affects the pediatric age, and is attributed to an intrauterine or perinatal insult that affects the perfusion of a cerebral hemisphere. It may be of congenital or acquired origin (infectious, ischemic, hemorrhagic or tumor); its frequency is unknown since the diagnosis is usually established at ages outside the pediatric age range. The correlation between the clinical and neuroimaging characteristics are fundamental for its correct identification. Management is mainly symptomatic, including anticonvulsant medication for epilepsy and infant rehabilitation. We describe an unusual case with clinical and imaging features consistent with an early diagnosis of Dyke-Davidoff-Masson syndrome with maternal history of *in utero* damage risk.

Resumen

El síndrome Dyke-Davidoff-Masson es una entidad caracterizada por pérdida del volumen de un hemisferio cerebral con hipertrofia homolateral del cráneo y senos paranasales, elevación del ala del esfenoides y del borde petroso del temporal, así como hiperneumatización del seno frontal y celdillas mastoideas, asociada a hemiplejia contralateral, asimetría facial, convulsiones y discapacidad intelectual. Fue descrita inicialmente en 1933 por Dyke, Davidoff y Masson. Afecta principalmente en la edad pediátrica, y se atribuye a un daño intrauterino o perinatal que afecta la perfusión de un hemisferio cerebral. Puede ser de origen congénito o adquirido (infeccioso, isquémico, hemorrágico o tumoral); su frecuencia es desconocida, ya que el diagnóstico suele establecerse en edades fuera del rango de edad pediátrica. La correlación entre las características clínicas y de neuroimagen es fundamental para su correcta identificación. El manejo es principalmente sintomático, entre otras, medicación anticonvulsiva para la epilepsia y rehabilitación infantil. Se describe un caso inusual con características clínicas e imagenológicas compatibles con síndrome de Dyke-Davidoff-Masson con antecedente materno de riesgo de daño *in utero*.

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Introduction

Dyke-Davidoff-Masson syndrome (DDMS) is a clinical-radiological entity characterized by volume loss of one cerebral hemisphere with homolateral hypertrophy of the skull and paranasal sinuses, elevation of the sphenoid wing and petrosal rim of the temporalis, associated with contralateral hemiplegia, facial asymmetry, seizures and intellectual disability (1, 2). These manifestations may present in a variety of combinations or degrees of severity depending on the extent of brain damage (2-4); intellectual disability is not always present and seizures may take months or years to appear. It is attributed to brain injury in fetal life, perinatal or early childhood. Mainly, it has been documented in late childhood (4), adolescents and adults (5); few, in neonatal stage. We describe here the case of a 27-week preterm infant, who at 30 weeks of corrected gestational age was detected by ultrasound as having a unilateral parenchymal hypoechoic parenchymal area. Brain magnetic resonance imaging (MRI) performed at 37 weeks corrected gestational age allowed early diagnosis of congenital DDMS.

Clinical case

Patient known to child neurology at 24 days after birth, with 30 weeks of corrected gestational age. Parents not consanguineous. Mother was 35 years old, first time pregnant, with no abnormalities during gestation, negative maternal infectious screening, hospitalized 11 days before delivery for pneumonia secondary to SARS-CoV-2/COVID-19 and preeclampsia. Admission fetal ultrasound showed a vital fetus, weighing 700 g, growing at the 25th percentile. Fetal biometry suggested pregnancy of 24+6 weeks, without brain abnormalities. The mother required 4 days of management in the intensive care unit due to signs of moderate respiratory distress, 2 of them with high requirements of supplemental oxygen with non-rebreathing mask; after her recovery, she was transferred to hospitalization. On the 11th day of hospitalization, she underwent cesarean section for severe preeclampsia with severity criteria. A male baby was born, neonatal adaptation conducted, Apgar 7/9/10, Ballard 27 weeks, birth weight 790 g (10th percentile for gestational age), height 35 cm (50th percentile), head circumference 23 cm, with progressive respiratory distress syndrome, requiring mechanical ventilation, pulmonary surfactant and hospitalization in the neonatal intensive care unit. PCR for COVID-19 was performed in nasopharyngeal sample with negative results. The first days he was in critical condition, with invasive ventilatory support, negative acute phase reactants, negative blood cultures, negative serology for STORCH, brain ultrasound on the second day of birth that showed partial collapse of one of the frontal horns of the lateral ventricles (Figure 1).

In the following days he was able to switch to non-invasive ventilation and then to nasal cannula, but on day 20 he suffered clinical deterioration of pulmonary origin that required high frequency ventilation (HFOV). Bacterial involvement was ruled out, viral panel for respiratory infections was negative. At 23 days after birth, a brain ultrasound was performed, showing an area of porencephaly (Figure 1).

The neurological examination was limited in the first evaluation, due to prone ventilation with HFOV and weight of 1000g. A 3-hour electroencephalogram video was performed with normal results. In the follow-up she evolved slowly towards improvement, with moderate bronchopulmonary dysplasia, brain ultrasounds after day 23 had no major changes, the last one with a slight decrease in the size of the lesion. In the control neurological examination there was evidence of decreased spontaneous movements, axial hypotonia, with sucking disorder that improved progressively, the head circumference always growing in the 3rd percentile. Normal automated auditory evoked potentials, negative total antibodies for COVID-19. At 37 weeks of corrected gestational age a simple brain MRI was performed in 1.5 Tesla equipment, which showed areas of left frontal parasagittal porencephaly and a smaller one in the ipsilateral frontoparietal convexity, with atrophy of the left cerebral hemisphere and ventricular asymmetry due to ipsilateral ex vacuo change (Figure 2).

At 20 months of life, motor developmental delay was noted, with crawling at 15 months and independent walking at 19 months. Physical examination showed mild right hemiparesis and extensor plantar response, but no facial asymmetry; in the social and language sphere no deficits were reported, but microcephaly was observed due to head circumference of 44 cm (below the 3rd percentile), height of 77 cm (below the 3rd percentile) consistent with low height for age and weight of 10 kg, which places him in the 50th percentile for the weight/height for age ratio according to WHO curve criteria. Control images show interhemispheric asymmetry due to loss of left brain volume, ex vacuo changes and parietooccipital white matter lesion due to homolateral sequel gliosis (Figure 3), without evidence of porencephaly areas observed in initial images and without cerebellar alterations (Figure 3).

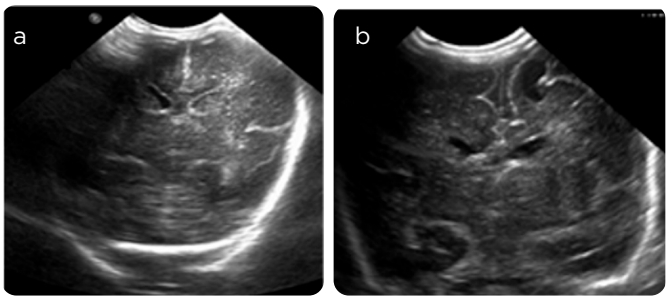


Figure 1. a) Transfontanelar ultrasound on the second day of birth: subtle collapse of the left frontal horn. b) Control at 23 days of life: cortical area of left frontal parasagittal porencephaly of 31 mm, slight dilatation of the left frontal horn and loss of volume of the left hemisphere.

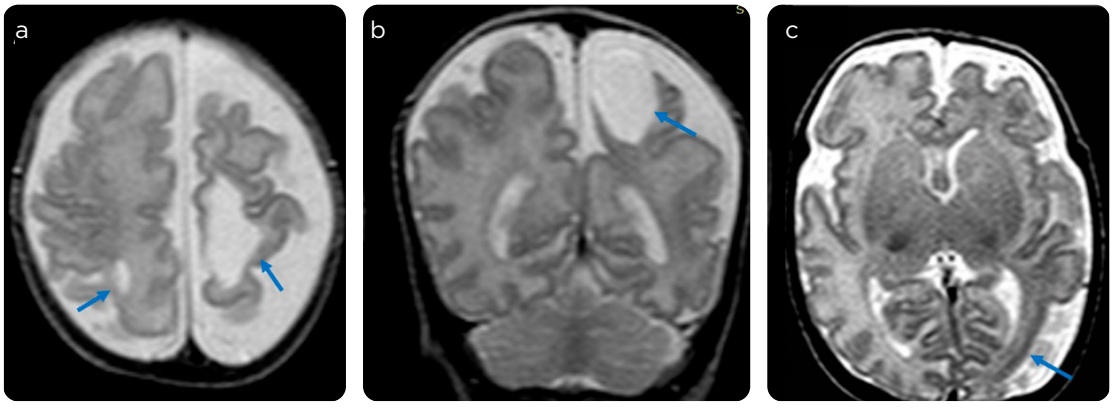


Figure 2. a) MRI with axial T2 information: bilateral parietal porencephalic cysts, larger on the left side. b) Coronal: left parietal porencephalic cyst, in addition, decreased left brain volume. c) Axial in lower slice: loss of left hemispheric volume. Focal hypermyelinization indicated with the arrow.

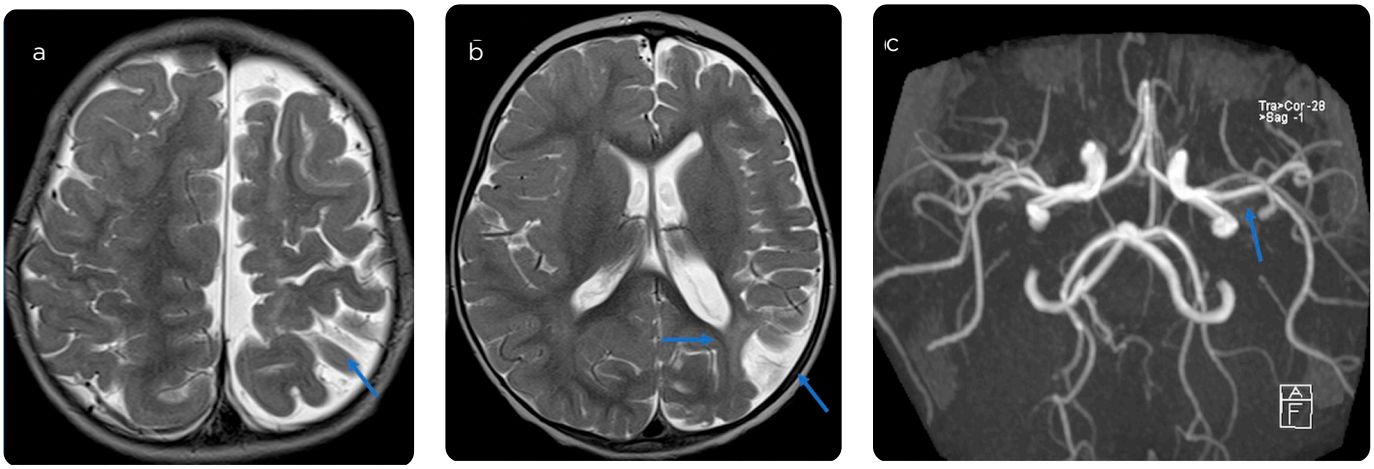


Figure 3. Control MRI of the same patient at 20 months of life. a) Axial T2 sequence: disappearance of the porencephalic cysts is evidenced, with marked decrease of volume of the left cerebral hemisphere. b) Loss of cortical volume persists in the left cerebral hemisphere. There is increased signal in the homolateral parietooccipital periatrial region and increased size of the atrioventricular atrium, ex vacuo changes compatible with sequel gliosis. The thickening of the ipsilateral cranial vault is noteworthy. c) Angioresonance sequence with subtle asymmetry in the middle cerebral arteries with smaller size of the left M1 and its terminal branches, secondary to hemispheric atrophic changes.

Discussion

DDMS was first described in 1933 (6), it is characterized by loss of volume of a cerebral hemisphere due to fetal or early life damage. It can affect both sexes and any cerebral hemisphere, but it is usually more frequent in the male sex and in the left cerebral hemisphere (2), as is the case of the patient described here. DDMS can be classified as congenital or acquired. The former can be caused by conditions experienced in utero or in early childhood, such as a lesion of vascular origin, infections or congenital malformations. The acquired form is usually associated with hemorrhage, trauma or infections occurring after the first month of life (1). The congenital form is characterized on neuroimaging by unilateral cerebral volume loss causing midline deviation towards the atrophic side, ex vacuo ventricular dilatation and enlargement of the convexity sulci on the side with volume loss, as well as atrophy of the ipsilateral cerebral peduncle secondary to Wallerian degeneration; ipsilateral bony compensatory changes of the atrophic hemisphere secondary to the intracranial relative vacuum effect are also found; for example, thickening of the cranial vault on the same side or elevation of the temporal crest (3, 4). In the acquired form there is only atrophy of one hemisphere without compensatory bone changes (3, 4). The patient in this case corresponds to congenital or primary DDMS, with images suggestive of in utero lesion; the cause of the lesion is not clear, but there are risk factors that increase the possibility of the same, since his mother had preeclampsia and COVID-19 pneumonia, which raises the doubt as to whether or not there is direct versus indirect involvement of the virus, despite having negative PCR and total antibodies for SARS-CoV-2. Although they did not describe hypoxia or ischemia events in the mother, there were high requirements for supplemental oxygen with a non-rebreathing mask for some days, which in the context of preeclampsia could worsen the decrease in placental flow and lead to ischemia.

This entity has been documented mainly in late ages. Two case reports are found in neonatal age, the one described by Aggarwal et al. (7) at 29 weeks of gestational age: a unilateral ventriculomegaly was observed in the antenatal ultrasound, in the postnatal cerebral MRI it was confirmed an extensive atrophy of the left cerebral hemisphere, with absence of the left anterior cerebral artery and a very thinned left middle cerebral artery. Piro et al. (8) reported the earliest case of DDMS described at the time, at 32+2 weeks of gestation, the antenatal ultrasound likewise with unilateral ventriculomegaly (8), the cerebral MRI showed an anomaly in the caliber of the middle cerebral artery with concomitant unilateral cerebral hypoplasia. Unlike these patients, the one in the case presented here had an area of unilateral porencephaly in the postnatal cerebral ultrasound performed at 30 weeks of corrected gestational age, that is, not only a unilateral ventriculomegaly can later develop DDMS, a hypoechoic area in cerebral parenchyma found in the ultrasound as well, so these patients will require follow-up with imaging and DDMS should be one of the differential diagnoses that should be excluded.

Congenital cerebral hemiatrophy has been classified into three imaging patterns according to Shen et al (9). Pattern I is a diffuse cortical and subcortical atrophy; pattern II is a diffuse cortical atrophy associated with porencephalic cysts, and pattern III is an old infarct with necrosis in the middle cerebral artery territory. The case presented here corresponds to pattern II, and this finding has not been described before.

The patient›s MRI does not show cerebellar involvement, but it could be found later. There is literature that mentions a loss of volume with signal alteration in the contralateral cerebellar hemisphere that may be present in DDMS, this as a consequence of a contralateral cerebellar hypometabolism originated in a supratentorial damage (3, 10), which would produce destruction of the cortico-ponto-cerebellar and cerebello-rubrothalamic pathways, multifocal neuronal loss, loss of myelination and loss of Purkinje cells, this in relation to a phenomenon called crossed cerebellar diaschisis. The crossed (or contralateral) cerebellar atrophy in the case of DDMS is ipsilateral when the damage occurs before one month of life and contralateral when it occurs after this age, due to anatomical differences during development (10). It should not be forgotten that neuronal migration disorders, such as polymicrogyria or pachygyria, have been described in this type of patients (8).

In the case of comorbidities, epilepsy is the most frequent, between 60% and 70% of cases, with no gender predominance (11). It is important to clarify that the damage is produced in a brain in formation, so that the focal response, both humoral and cellular, will favor the creation of proepileptogenic zones (astrogliosis) with an imbalance in the formation of overexcitatory inhibitory networks in the affected region (12).

There is no definitive treatment for DDMS, it is mainly symptomatic. The main objective is to control comorbidities over time, including epilepsy, and anticonvulsant drugs are useful for this; however, refractoriness may arise over time, and in some cases surgical management, such as hemispherectomy, has been performed with some success in some patients with intractable seizures and hemiplegia. Motor rehabilitation is also important for the restoration of motor functions. As for the prognosis of DDMS, it is better when epilepsy is absent or if it is of late onset (more than 2 years of age) (13).

Conclusion

DDMS is an entity poorly diagnosed at early ages, but should be suggested when areas of porencephaly are found in the antenatal or perinatal ultrasound, or important risk factors that lead to major brain damage; therefore, continuous follow-up is the tool that will allow making an appropriate clinical-radiological diagnosis, in order to offer the patient and his caregiver counseling on the prognosis of his disease. Unfortunately, the clear origin could not be established, and no histological report of placental tissue was obtained, which also invites to consider routinely searching for this type of pathology. This clinical case was presented with the aim of familiarizing the physician with the entity for its timely recognition. Although this condition has no cure, the management of comorbidities will allow the patient to have a multidisciplinary care that will improve their quality of life and the opportunity to participate socially.

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