CASE REPORT

Dyke-Davidoff-Masson syndrome: a clinicoradiological amalgam

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SUMMARY

Dyke-Davidoff-Masson syndrome is a relatively rare syndrome with its typical clinical and radiological features including facial asymmetry, hemiplegia, cerebral hemiatrophy, mental retardation with calvarial thickening, hypertrophy of sinuses and elevated petrous ridge on imaging. We present here a case of congenital type Dyke-Davidoff-Masson syndrome with some additional features in the form of microcephaly, hypospadias and pachygyria.

BACKGROUND

The first description of Dyke-Davidoff-Masson syndrome (DDMS) dates back to 1933, wherein authors described skull radiographs and pneumatoencephalographic changes in a series of nine patients.¹ The radiological features of the skull included asymmetry, ipsilateral osseous hypertrophy of the calvarium and hyperpneumatisation of sinuses.² Initially it was thought to be an isolated radiological syndrome, but various clinical manifestations have been described. These include seizures, facial asymmetry, contralateral hemiparesis, mental retardation and sensorineural hearing loss.³ Clinically, such patients can be classified under hemiconvulsion-hemiplegia and epilepsy (HHE) syndrome. This clinical syndrome was described by Gastaut *et al*,⁴ wherein an underlying vascular insult was considered the commonest cause. We report a case wherein radiological features helped in establishing the diagnosis of DDMS in previously considered HHE syndrome. Additional features such as microcephaly and pachygyria were also encountered in our case; association of which to our best of knowledge is exceedingly rare.

CASE PRESENTATION

A 6-year-old boy presented to us with poorly controlled, recurrent episodes of right-sided focal motor seizures with secondary generalisation for last 4 years. His parents noticed that since childhood his right half of body was weak and underdeveloped. He was born out of a non-consanguineous marriage and was born at full term by normal vaginal delivery in an institutional set-up. All his milestones were delayed, especially the language milestones as he could only utter bisyllables at the age of 6 years. There was no history of perinatal insult, fever with altered sensorium, hypoxic spells or any visual problems. He was treated initially with antiepileptics but was non-compliant on medications, hence seizures were never controlled. There was no history of similar

illness in any of the family members. On examination the patient had short stature, microcephaly and hypospadias. He was playful but could utter only bisyllable words. Bilateral fundi were normal and there was decreased prominence of nasolabial fold on the right side. The rest all cranial nerves were intact. There was hemiatrophy of right upper (figure 1) and lower limb. Tone was increased in the right side with bilateral brisk reflexes and right extensor plantar response. Sensory and cerebellar system examination was however normal. No meningeal signs were seen either.

INVESTIGATIONS

All his routine laboratory parameters, thyroid functions, HIV and vasculitis profiles were normal. His electrocardiogram, ultrasound of the abdomen and two-dimensional echocardiography were unremarkable as well. However EEG showed left-sided predominant, 2 Hz sharp and wave complexes (figure 3D). CT scan of the head showed hemiatrophy of the left hemisphere with thickening of squamous edge of temporal bone with widened mastoid cells, elevation of petrous ridge on the left side (figure 2A) and enlarged diploic spaces in the left side of calvarium (figure 2B). MRI of the brain also showed hemiatrophy with flattening of gyri and sulci on the left side which is suggestive of pachygyria (figure 3A–C). The clinicoradiological presentation was suggestive of Dyke-Davidoff-Masson syndrome.

DIFFERENTIAL DIAGNOSIS

- ▶ Sturge-Weber syndrome
- ▶ Rasmussen encephalitis
- ▶ Hemimegalencephaly
- ▶ Silver-Russell syndrome.

Figure 1 Clinical photograph showing hemiatrophy of the right upper limb.

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Figure 2 (A) CT of the head showing thickening of squamous edge of temporal bone (black arrow) with widened mastoid cells and elevation of petrous ridge on the left side. (B) CT of the head with bony window showing enlarged diploic spaces on the left side of calvarium (black arrows).

TREATMENT

Carbamazepine and clobazam were administered in increasing doses as per the patient's body weight.

OUTCOME AND FOLLOW-UP

The patient showed favourable response to treatment in the form of reduction in frequency of seizures.

DISCUSSION

DDMS is characterised by cerebral hemiatrophy with homolateral hypertrophy of skull and sinuses, elevation of sphenoid

wing and petrous ridge and association with contralateral hemiplegia, seizures and mental retardation. These features may be present in varying combinations or degree of severity. There can be affection of both sexes and any of the cerebral hemispheres, but involvement of male gender and left cerebral hemisphere is more frequent. DDMS can be classified as congenital or acquired. In the congenital type, there is usually no apparent aetiological factor and the symptoms are generally present at birth. The cerebral insult in this type occurs most likely during intrauterine life that might be due to intrauterine vascular occlusion. Whereas in the acquired type, the symptoms are related to

Figure 3 (A–C) MRI T2 weighted axial, coronal and fluid attenuated inversion recovery sequences, respectively, showing left cerebral hemiatrophy, falx displacement and smoothing and flattening of gyri and sulci on the left side suggestive of pachygyria. (D) EEG showing left-sided predominant sharp and wave complexes occurring at a frequency of 2 Hz.

central nervous system damage that occurs during the perinatal period or later. Among the aetiological factors are trauma, infection, vascular abnormality, ischaemic and haemorrhagic conditions, coarctation of the mid-aortic arch and intraventricular haemorrhage of premature infants.^{5–7} This is still one such syndrome wherein a CT scan provides a better insight into the disease as compared with MRI. The amalgamation of clinical and radiological features such as thickening of skull vault, enlargement of sinuses, elevation of petrous ridge, ipsilateral falx displacement and capillary malformations, characterises DDMS. Our case had the clinico-radiological profile of DDMS; and we considered it to be of a congenital variety, as an apparent underlying aetiology was not clear. Generally, three patterns of cerebral hemiatrophy have been described by Shen *et al*⁸ on MRI. MRI pattern I includes diffuse cerebral cortical and subcortical atrophy, pattern II corresponds to diffuse cortical and porencephalic cysts and finally pattern III corresponds to previous infarction and gliosis in the middle cerebral artery territory. Our patient could be classified as having pattern I of unilateral cerebral atrophy. The peculiar features in our case were the presence of microcephaly, hypospadias and pachygyria that has never been mentioned as a part of spectrum of DDMS in earlier reports. Pachygyria is a neuronal migration defect of the developing brain that results in flattening of gyri thereby providing a smooth appearance to the cortex. Treatment of DDMS is generally symptomatic including management of seizures. Functional hemispherectomy is done in cases of refractory epilepsy, which has a good long-term prognosis.⁹ Our patient however showed a favourable response to antiepileptic drugs. DDMS is relatively a rare entity, but a meticulous history taking with radiological features is paramount in establishing a diagnosis.

Learning points

- Dyke-Davidoff-Masson syndrome is a rare epilepsy syndrome with typical clinical and radiological characteristics.
- Predominant features are seizures, hemiatrophy, facial asymmetry, calvarial thickening and hypertrophy of sinuses.
- Aetiology of cerebral hemiatrophy includes perinatal cerebral insult, intrauterine infections, trauma, ischaemia or intracranial haemorrhage.

Contributors BS is the guarantor and all the co-authors were equally involved in developing the content of manuscript.

Competing interests None.

Patient consent Obtained.

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