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Case Report

# Dyke-Davidoff-Masson Syndrome: A Case Report

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## Abstract

**Background:** Dyke-Davidoff-Masson syndrome (DDMS) is a neurological syndrome characterized by the presence of convulsions, facial asymmetry due to palsy of the facial nerve (CN VII), contralateral hemiplegia, and reduced intellectual capacity. **The Case:** We report a case of DDMS in a 20-year-old male who is a previously known case of generalized epilepsy on medication presenting with status epilepticus and initially managed by anticonvulsants. On admission, the seizures manifested again which required the patient to be sedated with injectable anesthetics and intubated. Clinical examination showed no focal neurological deficits or neurocutaneous markers. Imaging studies showed characteristic features of DDMS which were hemiatrophy of the right cerebrum with calvarial thickening, and sinuses showing hyperpneumatization on the same side as hemiatrophy. Previous history of such episodes had been recorded and the patient was kept on strict pharmacotherapy. Failure of adherence to these led to the current presentation. The diagnosis of DDMS was kept and the patient was treated conservatively with anticonvulsants and referred to a higher center for further management. **Conclusion:** DDMS, being a rare but important cause of refractory epilepsy, is easily missed on initial assessment and failure of adequate management leads to higher rates of morbidity and mortality associated with this syndrome. In cases with an atypical presentation, such as this one, a good background in radio-imaging and knowledge of the physical manifestations are required for final diagnosis.

**Key Words:** Seizures; Neuroimaging; Anticonvulsants; Cerebral atrophy; Hemiplegia; Dyke-Davidoff-Masson syndrome (Source: MeSH-NLM).

## Introduction

In the year 1993 three researchers Dyke, Davidoff, and Masson came across peculiar radiographic images of cerebral hemiatrophy and compensatory hypertrophy of calvarium and frontal sinuses in nine patients who clinically presented with seizures, facial hemiparesis, and learning/developmental disabilities - thus forming the typical presentation of this syndrome and named it as Dyke-Davidoff-Masson Syndrome (DDMS).[1] This condition usually results from a perinatal insult, which further leads to the loss of neurons compromising the development of the brain either focally, or as a whole, leading to the spectrum of clinical features.[2] The major concern is the occurrence of such convulsive episodes for which pharmacotherapy alone is insufficient in most of the cases, and where surgical management is eventually advised.[3] We are hereby describing the clinical and radiological features of this syndrome in a young adult presenting to us with refractory seizures.

**Highlights:**

- Refractory seizures are not only problematic to manage in terms of medications but also hamper the quality of life of such individuals not restricted to the pathology of the causative factor but also the adverse effects of ASDs.
- The absence of characteristic features of this syndrome such as hemiparesis, mental retardation, facial palsy makes it easier to miss out on the diagnosis of DDMS with seizures being the presenting feature and its rarity of occurrence in our case.
- Early recognition of this syndrome would lead to better management in terms of both therapeutic as well as rehabilitative, thus improving the quality of life of such individuals by preventing intellectual decline.

## The Case

A 20-year-old male patient presented to our emergency department with sudden onset of involuntary movements of both limbs, upward gazing of eyes, frothing of the mouth, involuntary micturition, and tongue bite. The patient's attendants gave a history of 10-12 seizures since the previous night before arrival to the hospital with episodes of loss of consciousness for more than 30 minutes and post-ictal confusion for a period of 45 minutes. His seizure was managed with a dose of Lorazepam (2mg) followed by Levetiracetam (1g) intravenously. Blood samples were collected and sent for blood sugar levels, complete metabolic panel, and complete hemogram, in order to rule out the common causes of seizures. After stabilization with lorazepam and levetiracetam, the patient was in a state of post-ictal confusion, and admission to the medical intensive care unit was

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taken up for monitoring and further investigations. Further tests for liver and renal functions were conducted.

The patient was a known case of seizure disorders since the age of 17, with left focal onset seizures in his upper limb generalizing to both upper and lower limbs and was on pharmacotherapy (Sodium valproate 300mg BD, Phenobarbital 60mg BD, Clobazam 10mg BD). The seizure episodes started at the age of 3 and were managed under the above-mentioned antiepileptics. Seizures were usually preceded by neck pain, nausea, and involuntary movements of the right hand, diagnosed as idiopathic generalized epilepsy by the local physician, and kept as the diagnosis without further investigations or referral to a higher center. They also gave a history of episodic seizures which were managed by increasing the dosage of Clobazam to 20mg BD instead of regular dosing of 10mg BD. Consanguinity was not seen in the family tree. Uneventful perinatal history was given by the patient's attenders. There were no similar complaints in the immediate family. The parents noted learning difficulties and took him off from schooling in his first grade. He can speak in his mother tongue fluently. Motor developmental milestones were developed at appropriate ages.

On admission to the medical intensive care unit, the patient remained stable shortly for an hour and then presented with the second episode of seizures, initially with focal seizures of the left hand with secondary generalization. Patient was treated with Lorazepam 2mg, Levetiracetam 1g, Sodium Valproate 1g, Phenobarbitone 1g, following sedation with Midazolam infusion at 0.2mg/kg/hr, and mechanical ventilation, due to the seizure not being controlled by the above medications. Mechanical ventilation was continued for the next 4 days, then weaned off and extubated. On extubation, the patient remained stable and vital signs were near normal with no new onset of seizure episodes. Initially sent blood tests showed no significant findings.

The clinical examination of the central nervous system was normal and did not reveal any neurocutaneous lesions. He scored poorly on the Mini-Mental Status Examination (14/30), with brisk tendon reflexes and flexor plantar response. A magnetic resonance imaging (MRI) of the brain was subsequently done, which revealed right cerebral atrophy, with gliotic and encephalomalacic changes together with compensatory thickening of the cranial vault (***Figure 1***), and enlargement of frontal and hemisphenoid sinuses on the right side, with an elevation of the right petrous edge (***Figure 2***). An electroencephalogram (EEG) report, which was performed 3 years ago, showed abnormal EEG changes with generalized seizure discharges and diffuse background slowing.

We accordingly kept a diagnosis of DDMS, managed him conservatively with the above mentioned antiepileptics, and referred him to a comprehensive center for further management upon the patient's attendees' request. The patient's attendees refused surgical intervention due to financial constraints, and are continuing the anticonvulsants. Informed consent for publication was obtained from the patient's representative.

***Figure 1.*** MRI Showing Right Cerebral Atrophy with Gliotic and Encephalomalacic Changes Along with Compensatory Thickening of Cranial Vault.

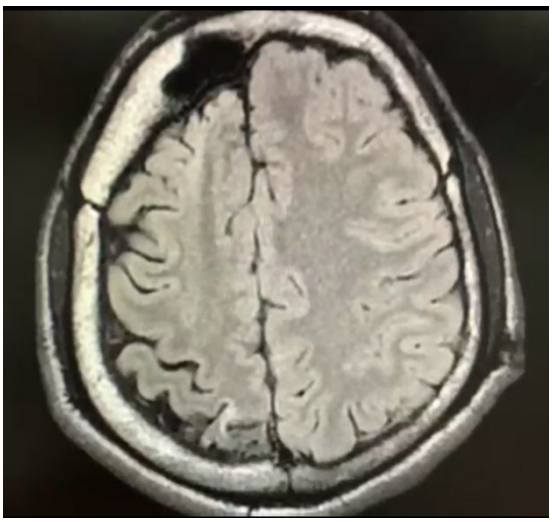
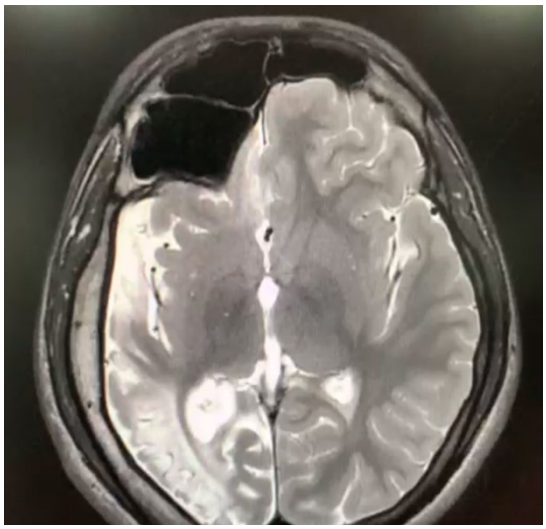


***Figure 2.*** MRI Showing Enlargement of Frontal and Hemisphenoid Sinuses on the Right Side with Elevation of the Right Petrous Edge.



## Discussion

DDMS, which is a rare but important condition commonly associated with refractory seizures, was first documented by Dyke, Davidoff, and Masson in 1933 when they noted radiographic images in a series of 9 patients with similar presentations.[1] Total and subtotal cortical hemiatrophy is the pathognomonic radiological finding in this syndrome, while sometimes unilateral cerebral atrophy is also noted in the cerebral peduncles, thalamus, pons, cerebellar crossings, and surrounding areas. Neuroimaging shows prominent sulcus over

the cerebrum, lateral ventricles dilated in certain parts, increase in the CSF spaces, calvarial thickening, osseous hypertrophy on the same side as the hemiatrophy with hyperpneumatization of the frontal and mastoid sinuses, and an elevated calvarium on the temporal side. Both sexes are equally affected in this case and any part of the brain can be equally involved as well, although left-sided involvement and male preponderance have been more frequently observed in one particular case study.[4] The clinical features of this syndrome are hemiparesis on the same side as hemiatrophy, with an upper motor neuron type palsy of the facial nerve (CN VIII), focal or generalized convulsions, and poor intellect with a delay in the achievement of milestones either occurring alone or in combination based on the side of hemiatrophy.[5]

Refractory epilepsy has many etiologies. These are commonly associated with failure of adherence to antiepileptic drugs, and include seizures that are non-epileptic, misdiagnosed, or inappropriate use of medications such as inadequate dosing, drug-to-drug interactions, and lifestyle choices such as alcohol & drug abuse, stress, and sleep deprivation.[6] Identification of the causative etiology is essential in planning its management, since refractory seizures are associated with high rates of morbidity and mortality. Out of the variety of tests available to investigate epilepsy, neuroimaging is the main tool used in its investigation. We came across this rare case of Dyke-Davidoff-Masson syndrome presenting as refractory seizures alone without the other typical features mentioned above.

Of the two types of cerebral hemiatrophy, the infantile subtype results from perinatal vascular insult usually involving the middle or anterior cerebral artery, coarctation of aortic arch; or common early neonatal sepsis thus presenting with the symptoms subsequently in the age group when the insult had occurred. Other, acquired, subtype of DDMS usually results from hypoxic-ischemic encephalopathy, pyrexic seizures of prolonged duration, traumatic insult, or from neoplastic or infectious etiology, along with hemorrhagic and ischemic causes.[7-8] The classical MRI changes of this disease, which are hemiatrophy and hyperpneumatization of sinuses, are observed radiographically only if the causative factor has acted upon the developing brain before the age of three.[9]

The differential diagnosis of this presentation seen in our case includes Sturge-Weber syndrome and Rasmussen encephalitis. Also, certain syndromes like Fishman syndrome, Silver-Russell syndrome, and linear nevus syndrome have to be kept in the picture as rare but possible causes. These syndromes are recognized through neuroimaging and clinical correlation.[10-11] Sturge-Weber syndrome is presented clinically by port-wine nevus on the face, epilepsy, ophthalmic manifestations primarily being increased intraocular pressure, learning difficulties, and stroke-like features occurring frequently. The underlying pathology is due to intracranial vascular anomaly and leptomeningeal angiomatosis and stasis causing the pathognomonic intracranial tram track calcification with laminar cortical necrosis leading to atrophy.[12] Rasmussen encephalitis, an immune-mediated progressive chronic condition occurring commonly in the younger age group of six to eight years, with the child presenting with intractable focal onset epilepsy and cognitive defects with imaging findings similar to that of hemi cerebral atrophy but no significant calvarial changes.[13] Silver-Russell syndrome is characterized by its unique facial phenotype, poor attainment of physical parameters such as height and bone length, clinodactyly, cerebral hemihypertrophy without affecting the head circumference, and no deranged mental capacity.[14] Fishman syndrome is a neurocutaneous syndrome occurring rarely which presents with unilateral cranial lipomatosis, ophthalmic lipodermoid, along with seizures characterized by radiological features of cortical calcification and hemiatrophy.[15] The hallmarks of linear nevus syndrome are typically facial nevus, recurrent refractory seizures, growth retardation with mental retardation, and unilateral ventricular dilatation resembling cerebral hemiatrophy.[16]

With the clinical features of cerebral hemiatrophy along with supportive radiological evidence of cerebral hemiatrophy, osseous hypertrophy of the skull, and compensatory hyper-pneumatization of the sinuses, DDMS has to be considered as the cause.[17-18] Even though our patient had just refractory seizures and learning difficulties as the clinical features, radiographic assistance is the one that aided in the prompt diagnosis of this syndrome. Commonly affecting the pediatric population, this case is of importance since our patient is in his early adulthood.[19] On further examination, patient was seen to have missed the dosing of the antiepileptics leading to the onset of the above scenario, thus being the causative etiology.

Conservative management of DDMS includes rational use of antiepileptic drugs, usually in combination since they do not easily adhere to monotherapy. If seizures are refractory, cerebral hemispherectomy is the available neurosurgical option which ensures the patient is seizure-free in about 85% of the operated cases.[3] Long-term management also includes adjunctive usage of physiotherapy, occupational and speech therapy. At present, management of epilepsy is still limited to monotherapy or adjunct usage of antiseizure drugs as the first-line management. Prompt diagnosis and early adherence to antiepileptics as the medical management of the seizures along with rehabilitation of both neurological and physical activities are also essential.[20]

## Conclusion

DDMS usually presents in early childhood or adolescents as refractory seizures requiring lifelong pharmacotherapy with anticonvulsants. Due to its rarity of occurrence, it is commonly missed on initial assessment. The relatively high cost of anticonvulsants, upon the background of low socioeconomic status, personal expenses for treatment, facilitates poor adherence to the drugs and thus broadens the treatment gap.[21] Further studies are necessary to identify the natural course of

DDMS, especially in the adult population leading to appropriate and economical management.

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