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SEIZURES IN THE SHADOWS: AN UNEXPECTED DIAGNOSIS

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ABSTRACT

Dyke–Davidoff–Masson Syndrome (DDMS) is a rare neurological disorder characterized by cerebral hemiatrophy associated with seizures, contralateral hemiparesis, and cognitive impairment. The condition results from an insult to the developing brain during the intrauterine period or early childhood. This case report describes a 14-year-old female who presented with recurrent abnormal limb movements for one year and chest pain for one week. The seizure episodes were characterized by sudden onset involuntary movements of bilateral limbs associated with up-rolling of eyeballs, frothing at the mouth, tongue bite, loss of consciousness, and post-ictal confusion. The patient also had a history of bleeding gums, rectal bleeding, developmental delay, and difficulty walking since childhood. Magnetic Resonance Imaging (MRI) of the brain revealed right cerebral hemisphere hemiatrophy with associated calvarial thickening suggestive of Dyke–Davidoff–Masson Syndrome. Electroencephalography (EEG) showed abnormal findings consistent with seizure activity. Laboratory investigations revealed severe anemia and Factor V deficiency. Additional findings included polycystic ovarian morphology on ultrasonography. The patient was managed with blood transfusions, antiepileptic medications, and supportive therapy during hospitalization. This case highlights the importance of recognizing characteristic radiological findings and clinical features for early diagnosis of DDMS. Early identification allows appropriate management of seizures, rehabilitation therapy, and monitoring for associated conditions.

INTRODUCTION

Dyke–Davidoff–Masson Syndrome (DDMS) is a rare neurological condition primarily identified by the presence of unilateral cerebral hemiatrophy, which is often accompanied by compensatory changes in the overlying skull. This syndrome typically results from a significant brain injury or insult that occurs either during the intrauterine period or early stages of childhood development, leading to compromised growth of one cerebral hemisphere. The classic clinical triad includes recurrent seizures, contralateral hemiparesis (weakness or paralysis on the side of the body opposite to the affected brain hemisphere), and intellectual disability or developmental delay. In some cases, patients may also present additional neurological deficits, including speech disorders, learning difficulties, and behavioral issues, further impacting their quality of life.

Radiological imaging, particularly Magnetic Resonance Imaging (MRI) and Computed Tomography (CT) scans, is essential for establishing the diagnosis of DDMS. Characteristic imaging findings often reveal unilateral cerebral atrophy, prominent dilated sulci, enlargement of the lateral ventricle on the affected side, and thickening of the calvarium as a compensatory response. These radiological features, combined with the patient's clinical history and presentation, help differentiate DDMS from other causes of cerebral atrophy. Early identification and accurate diagnosis are crucial for guiding management, which focuses on controlling seizures, providing rehabilitative therapy, and addressing any associated medical or developmental complications. Regular monitoring and a multidisciplinary approach can improve patient outcomes and support optimal development.

Patient Profile

Age/Sex	14 years / Female
Occupation	Unemployed
Residence	Mankhurd
Date of Admission	16/12/2025

Chief Complaints

1. Abnormal movements of bilateral upper and lower limbs for 1 year.
2. Chest pain for 1 week.

History of Present Illness

The patient developed episodic abnormal movements involving upper limbs progressing to lower limbs. Episodes were associated with up-rolling of eyeballs, frothing at the mouth, tongue bite, loss of consciousness, and post-ictal confusion. The patient also reported burning chest pain radiating to the back and epigastric region for one week without associated dyspnea or palpitations. On further inquiry, Patient also companies of bleeding from the gums in the past 2 years for which she went to a local doctor and was transfused PRBC i/v/o low hemoglobin (No documents available). Patient also gives history of bleeding per rectum on and off in the past one month. The patient was born at home with a birth weight of 2.5 kilograms and did not require NICU admission. However, she experienced delayed developmental milestones, notably beginning to walk at age five and continuing to have difficulty walking thereafter. She attained menarche at 12 years old, initially having heavy menstrual periods, but has not had periods in the past year. An outside ultrasound indicated that both ovaries are bulky with echogenic stroma, suggesting polycystic changes, though she has not received any treatment for this. Family history reveals that the mother reports similar complaints on the father's side, with the father's younger brothers' children also exhibiting developmental delays and intellectual disabilities. In terms of personal history, the patient's diet and appetite are normal and mixed; her sleep is disturbed. She reports painful bowel movements with hard stools, while bladder function remains unaltered, and she does not have any addictions. On general examination, the patient was afebrile, with a pulse

rate of 110 beats per minute and blood pressure measured at 110/60 mmHg. Oxygen saturation was 98% on room air. Pallor was noted, and the ECG showed a normal sinus rhythm. Facial asymmetry was observed, along with deformities in both hands and feet. Systemic examination revealed normal heart sounds (S1 and S2) with no murmurs detected. Respiratory assessment found bilateral breath sounds present, clear and equal, with no adventitious sounds. The abdomen was soft and non-tender, with no evidence of organomegaly.

Central Nervous System Examination

Component	Findings
Higher Mental Functions (HMF)	Intact
Cranial Nerves	
Pupils	Bilaterally equal and reactive to light
Extraocular Movements	Strabismus present
Other Cranial Nerves (I–XII)	Normal in function
Motor System	
Muscle Tone	Normal bilaterally
Muscle Power – Right Upper Limb	5/5
Muscle Power – Right Lower Limb	5/5
Muscle Power – Left Upper Limb	4/5
Muscle Power – Left Lower Limb	4/5
Deep Tendon Reflexes	Exaggerated on the left side
Plantar Response	Left: Extensor; Right: Withdrawal
Sensory System	No sensory deficits detected
Cerebellar Signs	Absent
Meningeal Signs	Absent
Autonomic Nervous System	No autonomic dysfunction
Gait	Normal
Involuntary Movements	Absent

Investigations

- MRI Brain: Hemiatrophy of the right cerebral hemisphere with calvarial thickening.
- EEG: Abnormal findings suggestive of seizure activity.
- Ultrasound Abdomen and Pelvis: Bulky ovaries with multiple follicles suggestive of PCOS.
- PR Examination: Internal hemorrhoids present at 3 and 5 O clock position

Laboratory Investigations During Hospital Stay

- Peripheral smear -normal
- PT/INR-4.38
- Appt-215
- Bleeding time -normal
- Thrombin time -18.5 -normal
- H/h/h -negative
- Factor 5 = <1% (normal range: 50-150)
- Fibrinogen: 146 (normal range: 200-400)
- D-dimer: negative.

Investigation	16/12	17/12	18/12	Normal Range
Hb (gm/dl)	5.3	6.5	9.0	13-18
WBC	13,720	9,780	11,510	4-11k
Platelets	5.54 L	4.95 L	4.85 L	1.5-4.5 L
PT / INR	43.8 / 4.38	-		
Na / K	136 / 3.6	-	137 / 4.0	Normal

Management During Hospital Stay

- 4 units Packed Red Blood Cells and 6 units Fresh Frozen Plasma transfused.
- Antiepileptic medications including Lacosamide and Levetiracetam were administered.
- Supportive therapy for gastritis and constipation provided.

Final Diagnosis

Acute Gastritis in a case of Dyke–Davidoff–Masson Syndrome with Factor V Deficiency and Polycystic Ovarian Syndrome.

Dyke–Davidoff–Masson Syndrome

DDMS is characterized by unilateral cerebral hemiatrophy with ipsilateral skull thickening and hyperpneumatization of paranasal sinuses. The condition usually results from vascular occlusion, infection, trauma, or developmental abnormalities affecting the brain during early development.

Prognosis

Prognosis depends on seizure control and the severity of neurological deficits. Early diagnosis and rehabilitation improve functional outcomes. Definition: A rare bleeding disorder caused by a lack of factor V, a protein needed for blood clotting.

Relation to DDMS

The search results do not list factor V deficiency as an etiology for DDMS. DDMS is generally linked to ischemic (clotting/stroke) events rather than bleeding (f actor V deficiency) events, although vascular issues in general are involved.

Possible Indirect Connection

The search results do not mention factor V deficiency but do mention that DDMS can be caused by vascular anomalies and prenatal vascular occlusion.

Potential Risk

Any underlying thrombophilic state (e.g., Factor V Leiden, which causes increased clotting) is sometimes linked to strokes in children, which could potentially lead to brain insults that cause DDMS. However, a deficiency (Factor V deficiency) causes bleeding, not clotting.

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