Case Report

A Case of Dyke-Davidoff-Masson Syndrome with Hypoplasia of the Kidney: An Unusual Association

Abstract

Dyke-Davidoff-Masson syndrome (DDMS) is a rare neuro-osteal syndrome of childhood and a constellation of cerebral hemiatrophy, facial asymmetry, seizures, osseous changes, and hemiplegia. It commonly presents with seizures and hemiplegia. The involvement of the kidney in DDMS is not known in the available literature, except in a case report that described ectopic kidney in DDMS. We present the case of a 15-year-old boy who presented with recurrent seizures, right facial palsy, left hemiparesis, and advanced renal failure. The neuroimaging revealed diffuse right cerebral atrophy, dilatation of the ipsilateral lateral ventricle, and ipsilateral thickening of the calvaria. The nephrological evaluation suggested the diagnosis of chronic kidney disease stage VD, probably secondary to congenital hypoplasia of the kidney.

Keywords: *Cerebral atrophy, chronic kidney disease, Dyke-Davidoff-Masson syndrome, hemiplegia, hypoplasia of kidney, seizure*

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Introduction

Dyke-Davidoff-Masson syndrome (DDMS) is a constellation of cerebral hemiatrophy, facial asymmetry, seizures, compensatory skull hypertrophy, hyperpneumatization of frontal sinuses, and contralateral hemiplegia. It was first described in a group of children with hemiplegia and skull changes by Dyke, Davidoff, and Masson in 1933.[1] It is a rare neurological disorder of childhood and commonly presents with seizures and hemiplegia.[2-4] Since its first description in 1933, around 100 cases including children and adults have been reported in the literature.[2] The frequency of kidney involvement in DDMS is not known in the available literature, except in one case report that described the association of ectopic kidneys with this syndrome.[5] We present the case of a 15-year-old boy who presented with recurrent seizures, right facial palsy, left hemiparesis, and advanced renal failure.

Case Report

A 15-year boy presented with a recurrent episode of generalized tonic-clonic seizure and altered sensorium for 2 days. He had a history of progressive weakness of the left upper and lower limbs, difficulty in walking, and multiple episodes of seizure from 5 years of age. It was not associated with fever, headache, and visual disturbance. A history of diabetes, hypertension, or tuberculosis was negative. He did not give a history of head trauma, joint pain, skin rash, hematuria, lower urinary tract symptoms, and intake of nephrotoxic drugs. He was born of full-term pregnancy and his birth weight was about 3000 grams. Antenatal period was uneventful. He achieved all developmental milestones normally till age five, except for stunted linear growth compared to his sibling. No such family history was present. He was never evaluated and treated adequately in the past. The patient was referred to our center with advance renal failure after receiving supportive treatment at a local hospital. A general examination was unremarkable except for pallor and emaciation. His blood pressure was 160/90 mm of Hg, pulse rate 88/min, respiratory rate 24/min, and body temperature 96.8°F. Height was 140 cm. The cardiovascular, respiratory, and abdominal examination was normal. On neurological examination, higher mental function was normal and there were decreased sensations, hypertonia, brisk reflexes, and the power 3/5 on the left side of the body. We noted a left-sided

How to cite this article: Patel PS, Krishna A, Archana, Kumar O. A case of Dyke-Davidoff-Masson syndrome with hypoplasia of the kidney: An unusual association. Int J App Basic Med Res 2023;13:259-62.

Submitted: 25-Apr-2023
Revised: 15-Oct-2023
Accepted: 27-Oct-2023
Published: 08-Dec-2023

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Access this article online

Website: https://journals.lww.com/IJAB

DOI: 10.4103/ijabmr.ijabmr_189_23

Quick Response Code:

deviation of the angle of the mouth [Figure 1]. Papilledema and a sign of meningeal irritation were absent. His urine output was normal. Laboratory workup including baseline investigations revealed anemia (Hb - 6.9 gm/dl), renal failure (serum creatinine - 25.3 mg/dl), and mineral bone disease (PTH - 986 pg/mL and VIT D - 16 ng/mL). A thorough workup including an autoimmune profile for chronic kidney disease was performed. A urine dipstick albumin test was negative, and 24-h urine protein excretion was 260 mg. The autoimmune profile was within a normal range. Ultrasonography of the kidney ureter bladder showed a bilateral small kidney with insignificant postvoid residual urine. Micturating cystourethrogram was normal. Cerebrospinal fluid analysis was normal. Electroencephalogram revealed epileptic abnormalities in the right hemisphere. Magnetic resonance imaging (MRI) brain revealed diffuse right cerebral atrophy, prominence of sulci, subarachnoid space, and Sylvian fissure with dilatation of the ipsilateral lateral ventricle. MRI brain showed gliosis, hyperpneumatization of the right frontal bone, and compensatory calvarial hypertrophy [Figure 2]. All laboratory investigations are mentioned in Table 1. Thus, based on history, clinical features, neuroimaging, and neurophysiological shreds of evidence, we diagnosed DDMS with chronic kidney disease probably secondary to hypoplasia of the kidney. He was initially managed with an injection of sodium valproate, an antihypertensive, and a packed red blood cell transfusion. Hemodialysis was started (thrice per week through a temporary double-lumen jugular catheter) along with other conservative treatments for chronic kidney disease. Now, he is doing well on maintenance hemodialysis and his seizure is well controlled for 3 months.

Figure 1: (a) An asymmetrical face with a left deviation of the angle of the mouth representing right VII cranial nerve palsy. (b) Flexion deformity of the left upper limb with spastic contraction of hand and internally rotated left lower limb representing the sequelae of left hemiparesis

Discussion

This rare disorder was first described by Dyke, Davidoff, and Masson in the nine patients who presented with seizures, facial asymmetry, hemiparesis, and mental retardation in say back 1933.[1] They described the radiographic skull changes of the patients. The predilection for a particular hemisphere or age or gender predominance has not been delineated till now. It commonly manifests in childhood; however, it has been described across all ages and genders in the literature.[2-7] However, the involvement of the left hemisphere in the male gender is more prevalent.[8] The clinical manifestation of DDMS may include seizures, contralateral upper motor neuron type hemiparesis, facial asymmetry, cognitive disability, and osseous lesion. Our case also presented with recurrent seizure, right facial palsy, and left hemiparesis, and the neuroimaging revealed diffuse right cerebral atrophy, dilatation of the ipsilateral lateral ventricle, and ipsilateral thickening of the calvaria. DDMS can manifest in two forms depending upon underlying etiology. The infantile or congenital subtype, which presents in infancy, results from intrauterine vascular occlusion, leading to underdevelopment of the brain. The acquired subtype, which presents in childhood, results from various causes like birth asphyxia, prolonged febrile seizures, trauma, tumor, infection, ischemia, and hemorrhage.[9] The

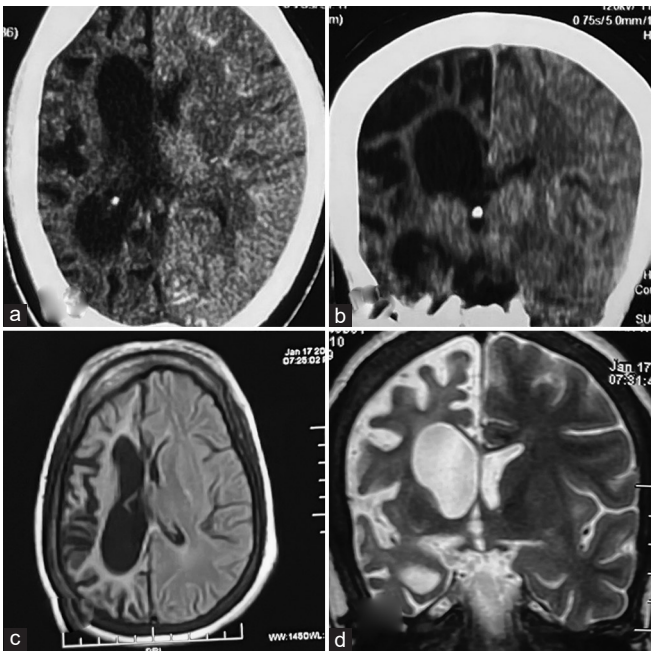

Figure 2: (a and b) Axial and coronal section of non-contrast computerized tomography of the head showing atrophy of right cerebral cortex, ex vacuo dilatation of ipsilateral lateral ventricle, calcification of right, left cerebral cortex, and bilateral basal ganglia. (c and d) Axial and coronal section of non-contrast magnetic resonance imaging of the brain showing diffuse right cerebral atrophy, prominent sulci, subarachnoid space, and Sylvian fissure with dilatation of ipsilateral lateral ventricle

Table 1: Laboratory value of the case at the time of hospitalization

Parameters	Results	Reference value
Total leukocyte count	8384	4–10×10^3/mm^3
Differential leukocyte count (%)	Neutrophils–68	40–70
	Lymphocyte–26	20–40
	Eosinophils–2	1–6
	Monocyte–4	2–8
Platelet count	17,300	150–400×10^3/mm^3
Red blood cell count	283,000	4.5–5.5×10^7/mm^3
Hemoglobin	6.9	13–17 gm/dL
Erythrocyte sedimentation rate	25	0–15 mm/h
Serum creatinine	25.3	0.6–1.3 mg/dL
Blood urea nitrogen	169	5–18 mg/dL
Serum sodium (Na^+)	133	135–145 mmol/L
Serum potassium (K^+)	4.6	3.5–5.5 mmol/L
Serum calcium (Ca^{2+})	5.5	8.6–10.3 mg/dL
Serum phosphorus	11.3	3.5–4.5 mg/dL
Uric acid	4.8	2.5–7.0 mg/dL
iPTH	986	14–72 pg/mL
25-OH Vitamin D	16	40–90 ng/mL
Serum bilirubin	0.38	0.2–1.0 mg/dL
ALP	16	<120 U/L
ALT	10	5–45 IU/L
AST	16	5–40 IU/L
Total serum protein	6.8	6–8 gm/dL
Serum albumin	3.7	3.5–5.5 gm/dL
Random blood sugar	82	<180 mg/dL
Human immunodeficiency virus	Nonreactive	Nonreactive
Hepatitis B surface antigen	Nonreactive	Nonreactive
HCV	Nonreactive	Nonreactive
Urine analysis	Albumin - Nil, RBC-2–3/hpf, Pus cell 6–8/hpf	
Urine culture	No growth	
24 h urine protein	260 mg	<150 mg
ECG	Normal sinus rhythm	
2D ECHO	Mild concentric LVH	
USG abdomen	RK 6.7X3 cm, LK 6X3 cm in size, grade III echogenic, and CMD lost	
NCCT head	Showed atrophy of right cerebral cortex, ex vacuo dilatation of ipsilateral lateral ventricle, calcification of right, left cerebral cortex, and bilateral basal ganglia	
MRI brain	Showed diffuse right cerebral atrophy, prominence of sulci, subarachnoid space, and Sylvian fissure with dilatation of ipsilateral lateral ventricle	

HCV: Hepatitis C virus; AST: Aspartate aminotransferase; ALT: Alanine aminotransferase; ALP: Alkaline phosphatase; iPTH: Intact parathyroid hormone; NCCT: Noncontrast computerized tomography; MRI: Magnetic resonance imaging; USG: Ultrasonography: ECG: Electrocardiogram; LVH: Left ventricular hypertrophy; CMD: Corticomedullary differentiation; ECHO: Echocardiogram

onset of symptoms in the present case was first noticed in childhood at the age of 5. Thus, the absence of a history of intra-uterine insult and first presentation in childhood suggests the possibility of an acquired form of DDMS. Apart from the central nervous system, the involvement of another organ in DDMS is not known in the literature. Only a case report has described the ectopic kidney in a patient with DDMS.[5] Our case had advanced renal failure with sonographic evidence of bilateral small kidney and mineral bone disease and typical neuro-osteal changes. Hence, this report could be the first report describing renal involvement in DDMS. Neuroimaging with CT and MRI are the two gold standard methods to establish the diagnosis of DDMS. The characteristic imaging features include prominent cortical sulci, dilated lateral ventricles, cerebral hemiatrophy, hyperpneumatization of the sinus, and compensatory hypertrophy of the skull. The neuroimaging of the present case also revealed similar changes in the brain and skull. The common differential diagnosis of cerebral atrophy is Sturge-Weber syndrome, brain tumors, Rasmussen encephalitis, and Fisherman syndrome. Most of these can be differentiated with the help of thorough clinical examination and by typical features of neuroimaging. The absence of cutaneous manifestation (nevus) and lipomatosis

or other manifestations easily exclude the other differential diagnosis. Thus, due to the rarity of this syndrome, it may be easily missed by the inexperienced eye. Neuroimaging is only the gold standard and powerful tool to detect the characteristic imaging features associated with DDMS. Hence, knowledge of the clinical presentation and imaging features is paramount for appropriate management.

Conclusion

DDMS is a rare neuro-osteal syndrome of childhood. We described a case who presented with recurrent seizure, right facial palsy, left hemiparesis, and advanced renal failure. Further evaluation revealed diffuse right cerebral atrophy, dilated lateral ventricle, ipsilateral thickening of the calvaria, and chronic kidney disease, probably secondary to congenital hypoplasia of the kidney.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form, the legal guardian has given his consent for images and other clinical information to be reported in the journal. The guardian understands that names and initials will not be published and due efforts will be made to conceal patient identity, but anonymity cannot be guaranteed.

Acknowledgment

We are thankful to Dr. Abhay Ranjan, Associate Professor, Department of Neurology, for their help in the management of cases. We are also thankful to the Department of Radiodiagnosis for their support in neuroimaging.

Financial support and sponsorship

Nil.

Conflicts of interest

There are no conflicts of interest.

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