Radiology Case Reports 19 (2024) 6241–6245

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Case Report

Diagnosis of Dyke-Davidoff-Masson syndrome in an adult ☆

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ARTICLE INFO

Article history:
Received 17 July 2024
Revised 25 August 2024
Accepted 26 August 2024

Keywords:
Adult diagnosis
Epilepsy
MRI imaging
Delayed diagnosis
Internal medicine

ABSTRACT

Dyke-Davidoff-Masson syndrome is a rare neurological condition characterized by intractable seizures, cerebral hemiatrophy with contralateral hemiparesis. Our patient, a 38-year-old female, presented following a left focal seizure with secondary generalization. She had a history of epilepsy, associated with left-sided hemiparesis, beginning at the age of 7. Physical examination showed increased left-sided tone and brisk reflexes, with an extensor plantar reflex on the left. The MRI brain showed features suggestive of Dyke-Davidoff-Masson syndrome: right-sided cortical atrophy, calvarial thickening and dilated frontal sinus. Additional MRI findings were of right cerebral peduncle atrophy and left cerebellar atrophy. This case report intends to emphasize the importance of Dyke-Davidoff-Masson syndrome as an unusual cause of seizures in an adult complicated by poor social determinants of health, leading to its delayed diagnosis.

Summary

Dyke-Davidoff-Masson syndrome is a rare neurological condition characterized by intractable seizures and cerebral hemiatrophy with contralateral hemiparesis. A 38-year-old female presented with a left focal seizure with secondary generalization. She had a history of epilepsy and left-sided hemiparesis, both of which began at age 7. Physical examination revealed increased left-sided tone and brisk reflexes, with an extensor plantar reflex on the left side. Brain MRI showed features consistent with Dyke-Davidoff-Masson syndrome, which included right-sided cortical atrophy, calvarial thickening, and a dilated frontal sinus. Additional MRI findings included atrophy of the right cerebral peduncle and left cerebellum. This case highlights the importance of considering Dyke-Davidoff-Masson syndrome as a potential cause of seizures in adults, particularly in the context of delayed diagnosis due to poor social determinants of health.

☆ Competing Interests: The authors declare that they have no known competing financial interests or personal relationships that could have appeared to influence the work reported in this paper.

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https://doi.org/10.1016/j.radcr.2024.08.138

Background

Dyke-Davidoff-Masson syndrome (DDMS) was first described in a case series of 9 infants by C.G. Dyke, L.M. Davidoff, and C. B. Masson in 1933. They observed a radiological finding of cranial asymmetry with cerebral atrophy, lateral ventricle dilatation, calvarial thickening, and sinus hypertrophy [1]. Since its initial description, fewer than 250 cases have been documented, of which only 21 have been reported in adults [2,3].

Clinical features include multiple seizure semiologies, hemiparesis, facial asymmetry, and neuropsychiatric manifestations [3]. The timing of cerebral insult may help in the further classification of congenital or acquired subtypes based on the history of presenting complaints. The diagnosis is based on clinical examination and classical neuroimaging findings, as seen on brain MRI [4]. The treatment is based on a multidisciplinary approach to reduce morbidity and mortality, as early diagnosis can help prevent the disease's sequelae, especially if it is of an acquired subtype [5]. Although diagnosis is usually made in childhood, the following case highlights how socio-economic factors, among other social determinants of health, can lead to a delayed diagnosis of such a rare neurological condition in an adult.

Case presentation

A 38-year-old female was referred to our emergency department. According to her relative, the patient had a seizure at home 8 hours earlier. The seizure began with spontaneous jerky movements of the left upper and lower limbs, followed by eye rolling and generalized rhythmic jerking of all extremities, lasting less than 5 minutes. The relative reported that his event occurred while the patient was reading a book, without any noted triggers. The seizure stopped without intervention and was accompanied by a drowsy postictal phase. Owing to a prolonged history of multiple seizures over the past few months without any apparent triggers, the patient was brought to a primary care clinic and was subsequently referred to our hospital.

A review of the patient's medical history and social background revealed that she came from a socioeconomically disadvantaged rural community and belonged to a large family with more than 10 members. Her birth history indicated a nonconsanguineous marriage and a full-term normal vaginal delivery in a hospital, with no perinatal complications. She was diagnosed with epilepsy and left-sided hemiparesis at the age of 7, for which she was administered phenytoin and carbamazepine. Most of her medical records were from rural primary care institutions with large gaps in follow-up care. Although she had been referred to a neurologist several times since her initial diagnosis, financial constraints prevented these consultations.

The patient and the family reported that she had been taking the same dosage of anti-seizure medications since childhood. While her seizures were well-controlled, they began to recur annually in her early 20s and have since increased in frequency. At presentation, the patient was having 1-2 seizure episodes every 2-3 months. She was a nonsmoker with no history of substance abuse. There was no history of recent infections, sleep disturbances, or stressors.

Social history was significant for the patient, being semi-dependent on her relatives. She was able to complete her activities of daily living and ambulate with the help of a walker, which she needed due to weakness on her left side.

Upon evaluation, the patient appeared to be at her baseline with stable vital signs. The post-ictal period, lasting 3 hours, was uneventful prior to her arrival. Physical examination revealed normal higher mental function and cranial nerve function, with intact sensory function. Her speech was noted to have a baseline staccato. Motor system examination revealed reduced muscle bulk and power on the left side, along with hypertonia. Brisk deep tendon reflexes and an extensor plantar response were noted on the right side, with minimal hemifacial atrophy on the right side. These findings were consistent with the patient's baseline neurological status. Psychological evaluation using the Revised Slosson Intelligence Test (RSIT) and Wechsler Adult Intelligence Scale (WAIS) indicated no intellectual disability. Other systemic examinations were unremarkable, and no neurocutaneous markers were observed.

Investigations

Routine laboratory investigations, including those for glucose and serum electrolytes, were within normal limits. The EEG performed upon arrival was also normal. Owing to the absence of previous imaging, a neurologist was consulted. Through shared decision-making, considering the initial normal EEG and the patient's return to baseline, the patient and her relatives opted against continuous epilepsy monitoring because to concerns about mounting costs. Based on the neurologist's recommendation, an MRI funded by the institution was conducted.

The brain MRI revealed right cerebral hemiatrophy, a dilated right ventricle, calvarial thickening, and a dilated right frontal sinus. Additional findings included atrophy of the right cerebral peduncle and contralateral cerebellar atrophy (Figs. 1 and 2). Based on clinical evaluation and characteristic neuroimaging findings, a unifying diagnosis of DDMS was made.

Differential diagnosis

Given the presentation of seizures with contralateral hemiparesis, Rasmussen syndrome was initially considered as a diagnosis. However, imaging findings and the absence of progressive neurological deficits made this diagnosis less likely [5]. Additionally, the patient's normal intellectual profile with hemicalvarial thickening made unilateral cerebral polymicrogyria with hemiatrophy (UPCH) unlikely [6]. The absence of neurocutaneous findings helped to exclude Sturge-Weber syndrome, linear nevus syndrome, and Fishman syndrome as potential diagnoses [5,7]. Ultimately, the clinical and radiographic findings, along with the patient's history of seizures since childhood, were consistent with the diagnosis of DDMS.

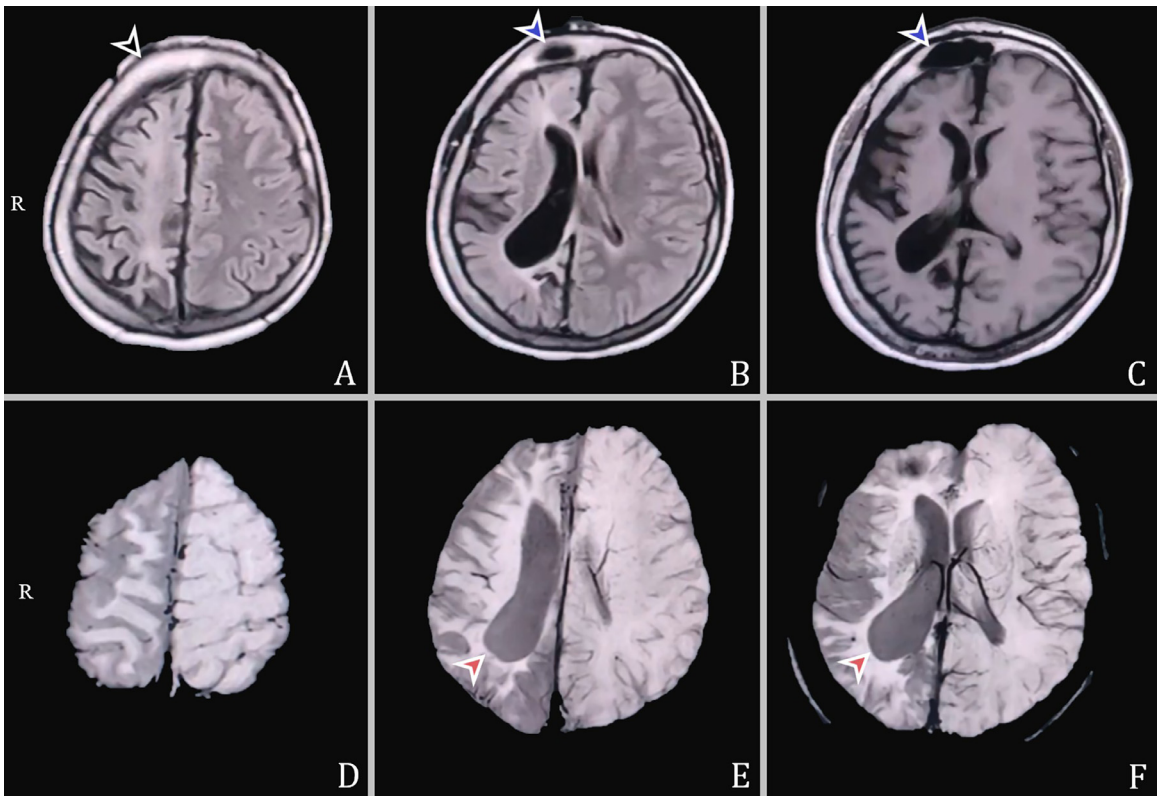

Fig. 1 – MRI image of the brain. (A-C) FLAIR-weighted images showing calvarial thickening (black) with unilateral hyperpneumatization of the right frontal sinus (blue) and hemiatrophy of the right cerebral hemisphere (C image limited by motion artifacts). (D-F) SWI images showing hemiatrophy in the right cerebral hemisphere with unilateral enlargement of the right lateral ventricle (red).
Widespread changes in white matter were also observed.

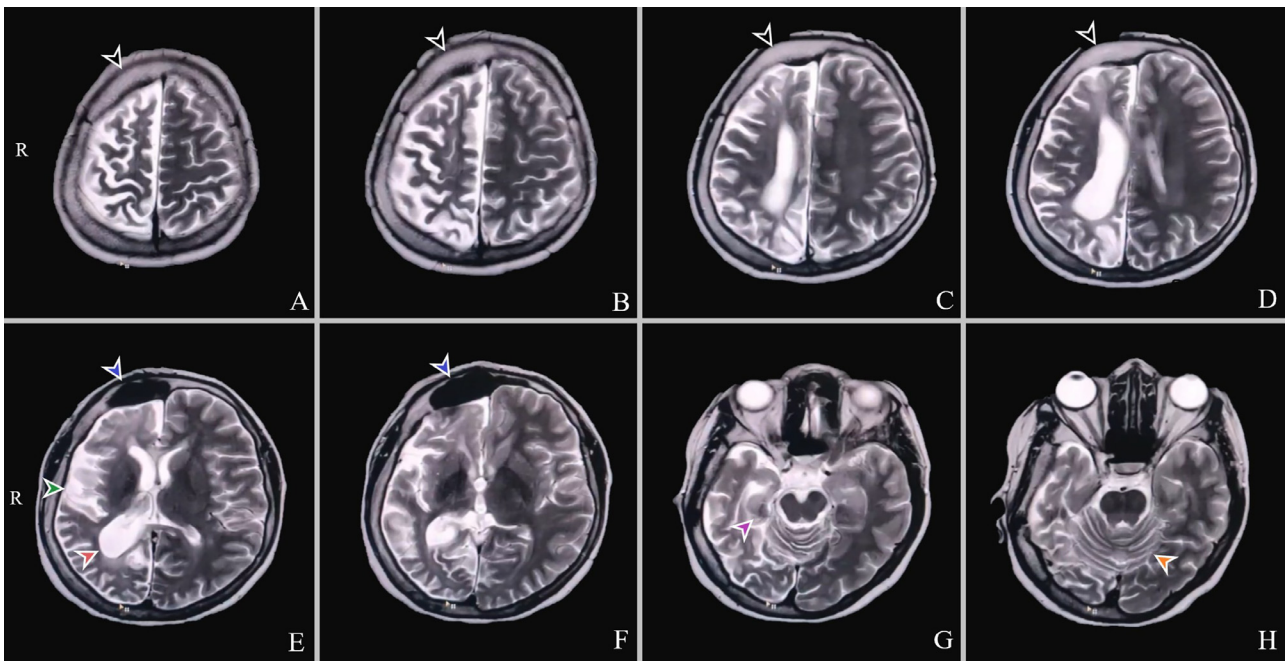

Fig. 2 – Brain magnetic resonance imaging. (A-H) T2-weighted images showing calvarial thickening (black), unilateral hyperpneumatization of the right frontal sinus (blue), frontal lobe atrophy (green), hemiatrophy of the right cerebral hemisphere (red), right cerebral peduncle (magenta), and contralateral (left) cerebellum (orange).

Treatment

The management involved careful and strategic adjustment of the patient's anti-seizure medications. Being on pediatric doses of phenytoin and carbamazepine, the persistence of breakthrough episodes, and the lack of a definite diagnosis prompted a neurologist review. A decision was made to stop phenytoin and start levetiracetam 500 mg twice daily, which was preferred for its spectrum efficacy. Additionally, the dosage of carbamazepine was titrated to that of an adult (100 mg/day). This was performed to optimize seizure control by leveraging the synergistic effects of these medications.

The patient underwent daily physiotherapy sessions to treat the hypertonia secondary to hemiplegia. These sessions encompassed a range of exercises that were designed to improve mobility. Vocational therapy was provided to address her staccato speech.

Following the diagnosis, the patient was provided with a detailed explanation of her diagnosis and the underlying disease process of the acquired DDMS. It was explained that she likely had the condition since childhood, but because of the absence of a formal diagnosis at that time, it was only identified in adulthood. Upon further counselling, the patient and her family members were informed about seizure precautions. Educational materials were handed to the patient's family for the following steps in the event of breakthrough seizures.

The importance of regular follow-ups at a government primary care center was emphasized, with reassurance that these would incur no additional costs. The patient was provided with coupons for free travel to the healthcare center and medications for the next 3 years.

Outcome and follow-up

The patient did not experience any recurrence of seizures following optimization of her treatment plan and was discharged after 4 days of inpatient care. Over the next 3 months, follow-up showed significant progress: she no longer required the use of a walker for activities of daily living, and her speech patterns improved markedly due to the combined efforts of physiotherapy and vocational therapy. Initially, the patient continued follow-up with the neurologist for 6 months, after which her care was transitioned to her primary care provider. She was advised to return to the neurologist if breakthrough seizures occurred, but none have been reported since her discharge.

Discussion

DDMS is a rare neurological disorder characterized by a broad spectrum of symptoms. The classical features include varying degrees of cerebral atrophy, hyperpneumatization of the sinuses, and compensatory calvarial thickening [2,5,7]. DDMS predominantly affects the pediatric population because of congenital or early infantile neuronal insult, and adult-onset as an acquired condition is uncommon [2,5]. Although our patient was diagnosed in adulthood, the history of seizures beginning at age 7 suggests a delayed diagnosis, likely complicated by financial constraints and caregiver fatigue. There was no evidence to suggest perinatal neuronal insult in our patient. The usual presentation of DDMS consists of the triad of epilepsy (most commonly intractable seizures), intellectual disability, and contralateral hemiparesis. Although hemiparesis is a part of this triad, it is seen in only 16% of cases and was also observed in our patient [7]. Differences have been noted in male presentations, which could be attributed to a higher concentration of circulating androgens, which could create a hyperplastic state in the developing brain, causing wider neuronal remodeling after injury in comparison to females [8].

The etiopathology of DDMS is centered on cerebral hypoxia resulting from infarction, hemorrhage, CNS infections, or traumatic brain injury [5,6]. Cortical involvement is more common, with congenital cases predominantly affecting males and acquired cases more frequently observed in females [3,6,8–10]. Cerebral hemorrhage observed in DDMS is hypothesized to result from neurodegeneration as a result of increased blood flow to the contralateral hemisphere during the initial years (1-3) of age following the insult, indicating a relationship between the development of cerebral lateralization and regional susceptibility to neurodegenerative disease.

The diagnosis is primarily based on neurological findings supported by neuroimaging findings. MRI typically reveals ipsilateral calvarial thickening, loss of convolutional markings on the inner table of the skull, overdevelopment of the paranasal sinuses (especially the frontal sinus), mastoid air cells, elevation of surrounding bony structures (petrous ridge, sphenoid wing, orbital roof), displacement of the falx attachment, and a reduced size of the middle or anterior cranial fossa [11]. Notably, there is no correlation between the degree of cerebral atrophy and extent of calvarial thickening [9]. Most of the changes described above occur because of a vacuum phenomenon and reflect adaptation to atrophied brain tissue [8]. Additionally, neuronal insult reduces the production of brain-derived neurotrophic factor (BNF) [6,12]. This reduced BNF and frequent excessive excitatory input during seizures (via glutaminergic corticopontine cerebellar pathways) induces further cortical atrophy [12].

The radiological features described above become more pronounced with age [13]. Based on MRI findings, 3 DDMS patterns have been recognized: Pattern I involve diffuse cortical and subcortical atrophy; pattern II is characterized by diffuse cortical atrophy with a porencephalic cyst; and pattern III shows atrophy following previous infarction with gliosis in the middle cerebral artery (MCA) territory [12]. In our case, the imaging features and clinical profile of our patient were indicative of an acquired DDMS pattern I.

The treatment of DDMS includes stewardship of anti-seizure medications (ASM) [12]. However, this task presented challenges in our case. The economic burden of seizures in our region stems from the relatively high cost of ASM, poor compliance, and the social and cultural stigma surrounding epilepsy. The involvement of nonmedical, pseudoscience practitioners in the early care of the patient led to significant challenges for the patient and her family in understanding the

disease process [14]. Moreover, in our case, the lack of local pharmaceutical support leading to medication refills of pediatric doses of ASM in an adult highlighted much-needed future improvements in the system and was representative of the pharmaceutical processes present in satellite rural impoverished communities.

In cases where seizure control is not achieved despite maximal ASM doses, hemispherectomy may be considered in children, with success rates of approximately 85% [11]. However, such surgical interventions are complicated by cost, availability of specialized care, and need for ongoing surveillance. The prognosis is unfavorable when the age of onset is less than 2 years and if the patient has recurrent or prolonged seizures despite maximally tolerated ASM, implying that onset after 2 years of age results in less severe seizures [6,15]. Irrespective of the modality of approach, long-term treatment includes physiotherapy and vocational therapy [5,12]. Ultimately, a multidisciplinary approach involving neurologists, physicians, physiotherapists, and neurosurgeons is essential to manage DDMS. Early diagnosis through knowledge of this rare disease coupled with proper evaluation could lead to early intervention and prevention of future disease sequelae [5].

Conclusion

Dyke-Davidoff-Masson syndrome is a rare neurological disorder that can be diagnosed in adults with variations in classically described findings. The multitude of clinical manifestations depends on the interplay between the age of onset, sex, severity of the neuronal injury, and degree of compensatory response. MRI findings include ipsilateral calvarial thickening, loss of convolutional markings of the inner table of the skull, overdevelopment of the paranasal sinuses and mastoid air cells, and alteration of the surrounding bony structures. Diagnosis may be delayed, leading to adult diagnosis in cases where there may be a relatively high cost of medications, poor compliance, and social and cultural stigma surrounding epilepsy. As reported in our case, the lack of local pharmaceutical support leading to medication refills of pediatric doses in an adult highlighted the need for improvement in the system and was representative of pharmaceutical processes in satellite rural impoverished communities. Ultimately, DDMS treatment includes stewardship of anti-seizure medications with sequential up-titrations, if needed, to achieve seizure control.

Patient consent

Consent for publication material and text was obtained directly from the patient. The patient did not receive any financial benefits from the publication of this case and did not involve any additional costs to the patient. There are no physical images of the patient nor individual determining factors present in the case description.

REFERENCES

[1] Aldhaleei WA, Bhagavathula AS, Alshehhi F. Dyke-Davidoff-Masson syndrome presenting as recurrent chronic headache in the late adult life. Brain Circ 2020;6(2):123–5.
[2] Al-Smair A, Hafez SA, Saadeh A, Al-Ali A. An adult with Dyke–Davidoff–Masson syndrome: a case report. Cureus 2022;14(3).
[3] Rondão MB, Hsu BR, Centeno RS, de Aguiar PHP. Dyke-Davidoff-Masson syndrome: main clinical and radiological findings-systematic literature review. Seizure 2023;110:58–68.
[4] Khan MZ, Mahapatra SS, Patel T, Razzaq W, Khawaja UA. Presentation of Dyke-Davidoff-Masson syndrome in a 32-year-old female: report of a rare case with a literature review. Cureus 2023;15(6).
[5] Al-Attas AA, Alwazna EO. Presentation of Dyke-Davidoff-Masson syndrome in adult male. Neurosciences J 2023;28(2):143–7.
[6] Diestro JD, Dorotan MK, Camacho AC, Perez-Gosiengfiao KT, Cabral-Lim LI. Clinical spectrum of Dyke-Davidoff-Masson syndrome in the adult: an atypical presentation and review of literature. Case Rep 2018;2018.
[7] Wang B, Jiang W, Yan W, Tian J, Xu J, Li Y, et al. Clinical characteristics and neuroimaging findings of seven patients with Dyke Davidoff Masson syndrome. BMC Neurol 2021;21(1):1–9.
[8] Ayas ZÖ, Asil K, Öcal R. The clinico-radiological spectrum of Dyke-Davidoff-Masson syndrome in adults. Neurolog Sci 2017;38:1823–8.
[9] Atalar MH, Icagasioglu D, Tas F. Cerebral hemiatrophy (Dyke–Davidoff–Masson syndrome) in childhood: clinicoradiological analysis of 19 cases. Pediatr Int 2007;49(1):70–5.
[10] Sia CC, Astejada MN. Dyke-Davidoff-Masson syndrome in the Philippines: a case series. Acta Med Philipp 2015;49(1).
[11] Roy U, Panwar A, Mukherjee A, Biswas D. Adult presentation of Dyke-Davidoff-Masson syndrome: a case report. Case Rep Neurol 2016;8(1):20–6.
[12] Adebayo PB, Bakare A, Bello MM, Olaewe OD, Wahab KW. Dyke-Davidoff-Masson syndrome in a Nigerian. Epilepsy Behav Case Rep 2016;7:10–12.
[13] Abdul Rashid AM, Md Noh MS. Dyke-Davidoff-Masson syndrome: a case report. BMC Neurol 2018;18(1):1–3.
[14] Thomas SV, Sarma PS, Alexander M, Pandit L, Shekhar L, Trivedi C, et al. Economic burden of epilepsy in India. Epilepsia 2001;42(8):1052–60.
[15] Arora R, Rani JY. Dyke-Davidoff-Masson syndrome: imaging features with illustration of two cases. Quant Imaging Med Surg 2015;5(3):469.