An unusual case of Dyke-Davidoff-Masson syndrome revealed by status epilepticus in a Malian patient

Samba Djimdé[1], Abdoulaye Yalcouyé[2], Abdou Koita[2], Hassana Samir [2], Pofinet Kebkiba[2], Awovi Gueli[2], Alassane Maiga[2], Adama Sissoko[3], and Guida Landouré[3]

[1] Service de Neurologie, Centre Hospitalier Universitaire du Point G, Bamako, Mali
[2]Faculté de Médecine et d'Odontostomatologie, USTTB, Bamako, Mali
[3]Service de Neurologie, Centre Hospitalier Universitaire du Point G, Bamako, Mali

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Abstract

Duke-Davidoff-Masson syndrome (DDMS) is a rare neurological condition with unknown global prevalence. It typically manifests with body asymmetry, drugs resistant epilepsy, mental retardation, cerebral atrophy, skull bone thickening and hyperpneumatization of the frontal sinuses. In this report, we present an unusual case of DDMS revealed by status epilepticus.

Introduction

Duke-Davidoff-Masson syndrome (DDMS) is a rare neurological condition with unknown global prevalence and incidence. This disease was first described by three physicians, Cornelius G. Dyke, Leo M. Davidoff and Clement M. Masson in 1933.[1] DDMS is a clinico-radiological recognized entity characterized by typical manifestations including drugs resistant epilepsy, intellectual disability, hemiparesis, skull bone thickening associated with cerebral hemiatrophy and hyperpneumatization of paranasal and frontal sinuses on neuroimaging.[2] Some patients may present with additional symptoms including cerebellar and basal ganglia atrophy, ear malformations and neuropsychiatric disorders.[3, 4]DDMS' etiologies are broadly categorized into congenital and acquired.[5] Congenital causes of DDMS include intrauterine vascular injury and cerebral hemispheric hypoperfusion and acquired causes mostly derive from birth trauma, periventricular leukomalacia, cerebral hemorrhage, cerebral infarction, cerebral infection, radiation, postictal cerebral hemiatrophy, and prolonged febrile seizures.[6, 7] Almost a century after the first description, the pathogenic mechanism remains unclear and is subject to controversy. To date, less than 100 cases have been reported worldwide with only four cases in the African population.[8-11] We report here the case of an 18-year-old adolescent who was admitted in our neurology clinic for convulsive status epilepticus and in which clinical and laboratory findings were in favor of DDMS, the first case in Mali.

Case report

An 18-year-old male was admitted in our neurology clinic for recurring tonic clonic seizures that started three days prior to his admission. Medical history was consistent with a full-term infant, born from healthy and non-consanguineous parents after an uneventful pregnancy. However, delivery was done by caesarean section due to dystocia. Family history was unremarkable. He had mental and motor acquisition delay because he could not seat after one year of age and walked after the age of two. Parents reported that he presented focal tonic clonic seizures in the right half of his body at one year of age, at least once a day that worsened over the time to more than ten episodes daily. He was first treated with Valproic Acid, Phenobarbital and Carbamazepine at different times which could poorly control the seizures, despite having received the correct doses. He also presented frequent falls and injuries due to seizures. Later, they

noticed that the right side of his body was becoming smaller and weaker than the left one. In addition, he presented a progressive intellectual disability that led him to drop out of school at the age of nine. On examination, the body temperature, blood pressure, heart and breathing rates, and the oxygen saturation were normal. Neurological examination found status epilepticus with right-sided focal tonic clonic seizures followed by secondary generalization. He had severe cognitive decline and couldn't count from one to ten. Mini Mental State Examination (MMSE) was not possible. Body asymmetry and hemiparesis was confirmed in addition to ankylosis in the right wrist and elbow and a prominent forehead (Figure 1A&B&C). Abdominal, cardiovascular, and pulmonary examination were unremarkable. He did not present any cutaneous lesions other than scars caused by seizure-related injuries. Blood chemistries including total blood cell count, liver enzymes, serum creatinine and blood ions were normal. EEG performed two weeks after admission found a diffuse slow background with delta wave activities in hyperpnea and a continued focal to diffuse pseudo periodic activities (Figure 2A&B). Brain CT-scan revealed a pronounced atrophy of left cerebral hemisphere with *ex vacuo* of the ipsilateral ventricle. Furthermore, a bilateral abnormal thickening of the skull bone with an hyperpneumatization of the frontal sinuses were seen (Figure 2C&D). His clinical and imaging features were consistent with DDMS. He was initially put on intravenous Levetiracetam (1500mg/day), and continuous infusion of Clonazepam (2mg/day). The disease course remarkably changed to fewer seizures (down to 10 a day). Then, we added oral Valproic Acid (500mg twice a day) that drastically improved the seizures with a maximum of two seizures daily and the patient was discharged on this treatment. After the discharge, parents reported that he presented three episodes of seizures in one month.

Discussion

Dyke, Davidoff and Masson recognized and reported the first cases of this syndrome in 1933.[1] Almost a century later, less than 100 cases reported worldwide but its global incidence is still unknown. DDMS falls in the category of epilepsy with brain structural abnormalities of the International League Against Epilepsy classification (ILAE).[12] The clinical manifestations encompass a focal and/or generalized drugs-resistant epilepsy, hemiparesis or hemiplegia, facial or body asymmetry with atrophy and mental retardation as seen in our patient.[2, 3] Besides these manifestations, rare cases may include cerebellar atrophy, neuropsychiatric disorders and ear malformations.[3]However, the disease phenotype can vary from one patient to another, and some may not present epilepsy, mental retardation or body asymmetry.[13] The patient was referred for convulsive status epilepticus that led to the diagnosis as seen in previous reports.[14] DDMS is usually diagnosed in childhood, mostly in the first decade, but cases of late diagnosis or adults cases were also reported.[2] Although the syndrome is easily recognizable by clinical and brain imaging findings, the diagnosis delay was eighteen years in our patient. This is likely due to the limited access to specialists who could further investigate with brain imaging as this was not performed before we saw the patient. A similar case with a long diagnosis odyssey was reported in a patient from Nigeria.[11] Brain CT-scan or MRI have contributed to facilitate the diagnosis by typically showing hemi cerebral atrophy, ipsilateral thickening of the skull bone and ipsilateral hyper pneumatization of the frontal and paranasal sinuses.[15]In our case, the hyper pneumatization of the frontal and paranasal sinuses and the thickening of the skull bone were bilateral. To the best of our knowledge, these brain imaging findings have not been previously reported. This case could be another phenotypic variant of DDMS that might be due to some specific genetic factors as suggested by previous studies[2] or to recurring brain. Although our patient fulfilled the diagnosis criteria of DDMS, other diseases such as Rasmussen's encephalitis (RE), hemiplegia-hemi convulsion epilepsy (HHE) syndrome and Sturge Weber syndrome (SWS) are also possible.[15, 16] However, in RE and HHE there is no skull bone thickening or a hyper pneumatization of the sinuses that are pathognomonic radiological findings in DDMS as seen in the patient presented here. In addition, in HHE syndrome, the seizures are in the hemiplegia/hemiparesis side without secondary generalization. SWS is a neurocutaneous syndrome characterized by facial birthmark also called port-wine birthmark and cerebral vascular calcifications which were absent in our patient. The management of DDMS is challenging, especially in resource-limited settings due to the resistance to several antiepileptic drugs (AED). The treatment is based on the use of a combination of AEDs and the surgery with hemispherectomy is an option in the case of refractory epilepsy.[14]

Conclusion:

This is the first report of DDMS in a Malian patient. It highlights the challenges for an early diagnosis and the treatment of this rare condition. We suggest that in the presence of pharmaco-resistant epilepsy in childhood and body deformities, further investigations should be undertaken to establish evidence-based diagnosis for an informed disease management strategy, particularly in low resources setting where access to the surgery of epilepsy is highly limited or unavailable.

Author contributions

Samba Ogomaly Djimdé , data acquisition and interpretation;**Abdoulaye Yalcouyé** , data analysis and interpretation, and drafting the first version of the manuscript; **Abdou Koita** , clinical management and data acquisition; **Hassana Samir** , clinical management and data acquisition; **Pofinet Kebkiba** , clinical management and data acquisition; **Awovi Chrystelle Gueli** , clinical management and data acquisition; **Alassane Baneye Maïga** , revision of the manuscript; **Adama Seydou Sissoko** , revision of the manuscript; **Guida Landouré** , editing and supervising of the study.

Ethical approval

None

Consent

Informed consent was obtained form the parents including publishing photographs

Data availability statement: All the data included in this study is available from the corresponding author upon reasonable request

Conflict of interest: authors declare no conflict of interest.

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Figure legends

Figure 1: Clinical findings in patient with Duke-Davidoff-Masson syndrome. A): Images of the patient showing right side hemi atrophy of the body more pronounced in upper limb. B,C): ankylosis of the right wrist and elbow.

Figure 2: Laboratory findings in patient with Duke-Davidoff-Masson syndrome. A,B): EEG showing slowed background with delta wave activities in hyperpnea and a continued focal to diffuses pseudo periodic activities (red squares). C,D): Brain CT-Scan in axial cut showing a left hemisphere atrophy (blue arrow), bilateral calvarial thickening (black arrow) and hyperpneumatization of the frontal sinuses (red arrow).