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Case Report

Isolated unilateral medial rectus palsy: a rare presentation of midbrain infarction in a child

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ABSTRACT

This case study describes a rare instance of isolated unilateral medial rectus palsy in a young patient with Fabry disease, an X-linked lysosomal storage disease. A five-and-a-half-year-old child did not exhibit any other symptoms associated with a stroke, other than abrupt onset diplopia and a pronounced left-eye squint. Isolated medial rectus palsy as a sole presentation of midbrain infarct is a rare entity. Fabry disease is associated with an increased risk of ischemic strokes in patients because of the build-up of glycosphingolipids in the vascular endothelium. This case emphasizes how crucial it is to conduct early neuroimaging and to have a high index of suspicion when dealing with uncommon neurological presentations, particularly in individuals who have underlying genetic conditions like Fabry disease.

Keywords: Fabry diseases, Isolated medial rectus palsy, Midbrain infarction, Diplopia

INTRODUCTION

Medial rectus palsy is characterized by a dysfunction in the medial rectus muscle, which is responsible for adduction of the eye. It is often caused by orbital lesions or muscle-related disorders.1 Its incidence owing to a midbrain infarction is extremely unusual, especially in youngsters. Midbrain infarctions are frequently coupled with widespread brainstem dysfunction, affecting other cranial nerves and causing motor deficits. In this case, the solitary involvement of the medial rectus muscle, which caused a squint and diplopia, made diagnosis difficult.

Fabry disease, characterized by GLA gene mutations that result in a deficiency of the enzyme alpha-galactosidase A, is a well-known risk factor for recurrent strokes. This condition is characterized by the gradual accumulation of glycosphingolipids in many tissues, including blood vessels, the heart, kidneys, and the nervous system.2 Strokes in Fabry disease patients are typically ischemic, affecting tiny vessels in the brain. While stroke is acknowledged as a possible Fabry disease consequence, isolated medial rectus palsy caused by a midbrain infarction is extremely rare, especially in juvenile patients. This article describes the case of a five-and-a-half-year-old child with Fabry disease who had isolated medial rectus palsy due to a midbrain infarction. It emphasizes the significance of early detection and intervention in preventing long-term sequelae as well as being cautious when confronted with rare neurological presentations.

CASE REPORT

A five-and-a-half-year-old child, who had been diagnosed with Fabry disease at the age of four, came to the emergency department with acute onset of diplopia and left-eye squint. The symptoms appeared suddenly the day before, and there was no history of trauma, fever, or

other systemic issues. Notably, there was no evidence of weakening, sensory impairments, or involvement of other cranial nerves, ruling out more widespread neurological dysfunction. Upon physical assessment, the child appeared alert and responsive. His vital signs remained normal, including blood pressure in the 50th percentile for his age. The neurological exam demonstrated exotropia of left eye in the primary gaze, as well as limited adduction in the left eye, which indicated medial rectus palsy. The pupils were bilaterally equal and reactive to light with no ptosis. The rest of the cranial nerves and neurological examination revealed no deficit and other systems were normal.

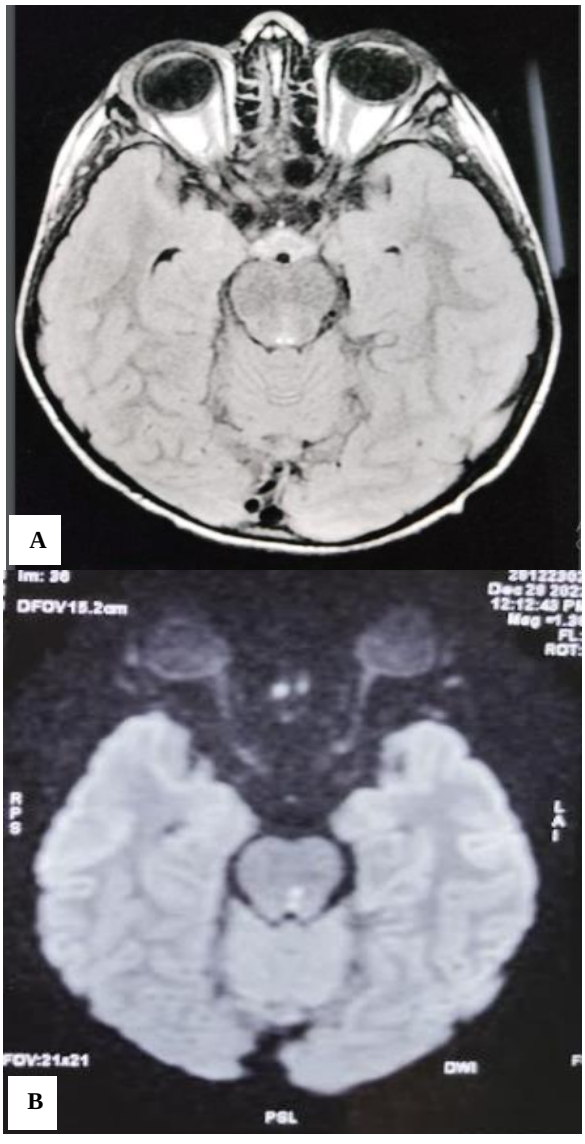

Figure 1 (A and B): T2 FLAIR and DWI images revealed tiny focus of restricted diffusion involving the posterior left para-median portion of the mid-brain).

The initial differential diagnoses were traumatic injury, internuclear ophthalmoplegia, refractive error, and ischemia stroke was considered possible. Urgent neuroimaging was performed to further investigate the cause. Magnetic resonance imaging (MRI) revealed a tiny area of restricted diffusion in the left paramedian region of the midbrain on diffusion-weighted imaging (DWI) suggestive of an acute infarct. Routine blood tests, such as a complete blood count, liver function tests, and coagulation profiles, all returned normal results, ruling out any additional systemic explanations. Given the findings, the child was started on aspirin at 3 mg/kg/day. After two weeks, the dose was decreased to 1 mg/kg daily, as per the protocol and the patient was regularly monitored. Recovery at follow-up was complete with no residual deficits.

DISCUSSION

Isolated medial rectus palsy with midbrain infarction is a rare diagnosis, particularly in pediatric patients, with the youngest case being reported in a 22-year-old. Isolated medial rectus palsy is typically linked to orbital lesions or muscular disorders, but when associated with other extra ocular muscle involvement and other signs of brainstem dysfunction, it can indicate midbrain infarctions.[1] In this case, the patient's known diagnosis of Fabry disease increased the risk of stroke and led us to suspect a midbrain infarction, which was confirmed through MRI findings.

Fabry disease is an X-linked lysosomal storage disorder caused by mutation in the alpha-galactosidase A (GLA) gene, resulting in a deficiency of the enzyme. Young males are nearly 12 times more likely to experience cerebrovascular events, like stroke and TIA than the overall population. In Fabry disease, strokes can be ischemic or hemorrhagic, affecting the carotid or vertebrobasilar vasculature and tiny vessels. The progressive GL-3 accumulation within the endothelium of intracranial blood vessels is linked to play a role in vasculopathy and risk of ischemic stroke.[2]

Due to its effect on the vascular system, Fabry disease raises the risk of ischemic strokes dramatically, even in young patients. Although stroke is a known consequence of Fabry disease, the majority of strokes in Fabry patients are associated with hemiparesis, dysarthria, or other generalized neurological symptoms, making this instance with solitary ocular involvement extremely rare. Midbrain infarction in pediatric Fabry patients has only been observed in a small number of cases, and isolated ocular palsy is rarer. Early neuroimaging is crucial in young patients with unexplained diplopia or eye movement abnormalities, especially if there is a known underlying disorder such as Fabry disease.[1]

Diffusion-weighted imaging (DWI) is extremely sensitive at detecting ischemic lesions in the brainstem, even in the absence of other stroke symptoms. This instance emphasizes the necessity of investigating vascular lesions, such as ischemic stroke, in pediatric patients with Fabry disease who exhibit neurological symptoms. Early

detection and management, as seen in this patient, can lead to complete recovery without long-term consequences.[4,5] The presentation is unique due to the localized nature of the lesion, affecting only the lateral subnuclei of the oculomotor nerve complex. The lateral subnucleus of the oculomotor nuclear complex is composed of three subnuclei that supply the inferior rectus, inferior oblique, and medial rectus muscles, arranged from dorsal to ventral respectively.[3] The specific involvement of this lateral subnuclei results in isolated dysfunction of the medial rectus muscle, causing acute onset of diplopia in one eye, unlike bilateral palsy, which involves more extensive brain stem damage and more severe visual impairment. The patient's diagnosis, confirmed by MRI, highlights the importance of early neuroimaging to identify ischemic lesions, with prompt treatment leading to complete recovery.

CONCLUSION

This case presents a rare instinct of isolated unilateral medial rectus nuclear palsy which can be the only manifestation of midbrain infarction. Particularly notable in the context of a child diagnosed with Fabry disease. Early neuroimaging is crucial for determining the causative lesion. A systematic clinical approach, including a thorough history and physical examination, and a high index of suspicion, is necessary to determine the underlying cause. This example further emphasizes the importance of a multidisciplinary approach to managing unusual and complex manifestations of juvenile stroke.

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