Section: Ophthalmology

Case Report

STURGE-WEBER SYNDROME WITH CONCURRENT DYKE-DAVIDOFF-MASSON SYNDROME: A CASE REPORT

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Received : 01/03/2024
Received in revised form : 23/04/2024
Accepted : 10/05/2024

Keywords:
Sturge-Weber syndrome, Dyke-Davidoff-Masson syndrome, glaucoma, Infant.

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DOI: 10.47009/jamp.2024.6.3.29

Source of Support: Nil,
Conflict of Interest: None declared

Int J Acad Med Pharm
2024; 6 (3); 134-136

Abstract

Purpose: To report a rare case of Sturge-Weber Syndrome (SWS) with concurrent Dyke-Davidoff-Masson syndrome (DDMS) in a neonate and highlight the importance of early diagnosis and management.

Observations: A 1-month-old infant presented with a left facial port-wine stain, break through seizures and ocular abnormalities including left eye stromal Corneal haze, flat eyelids, conjunctival congestion, deep anterior chamber, round and regular mid-Dilated pupil, and bluish sclera.

Investigations on MRI brain with Contrast revealed Left loss of parenchyma of left hemisphere suggestive of cerebral atrophy, leptomeningeal Enhancement-tram track calcification and Enlarged left choroid plexus suggesting of choroid Papilloma. Hematological investigations shows elevated inflammatory markers – CRP-80 mg/L And WBC slightly increased to 11.These findings confirmed co-existing SWS and DDMS. The infant Exhibited developmental delay, achieving neck Holding at 7 months and sitting with support at 8 Months (typically achieved by 4-6 months). The Infant underwent glaucoma surgery-Trabeculotomy and trabeculectomy on the left eye.

Conclusions: This case presents the potential for combined SWS and DDMS presentation in neonates. Early diagnosis through clinical features and imaging studies is crucial for prompt management of glaucoma and other associated complications. While surgical Intervention can address glaucoma, long-term management of seizures and developmental Delays remains challenging in such cases.

INTRODUCTION

This is a case report which explains complex presentation of a 1month old child which present to the OPD with forward displacement of left eye since birth associated with pale pink birthmark in face and body. Baby also had history of seizure right after birth. On examination patient had bupthalmos, IOP was digitally hard and 0.5 cupping on left eye. Contrast Enhanced MRI shows tram track appearance, cerebral atrophy, choroid papilloma Suggesting a diagnosis of Sturge Weber syndrome with Dyke Davidoff- Masson Syndrome. Patient underwent Trabeculotomy with Trabeculotomy in left eye. Early diagnosis and treatment will have good visual outcome.

CASE REPORT

Patient initially presented to the OPD at 1month with complain of forward displacement of left eyeball and pale pink birthmark over face and body. Patient had past history of seizure right after birth and developmental delay. Patient was examined and found to have bluish scleral, hazy cornea, Megalocornea, bupthalmos, IOP was digitally hard, port wine stain on face and on fundoscopy retina on ,0.5 cupping was found on left eye. On suspicious of Sturge Weber syndrome CEMRI was advised.

CEMRI shows prominent leptomeningeal enhancement, typical tram track calcification [Figure a and b] of Sturge Weber syndrome. Cerebral hemiatrophy [Figure 1], Choroid papilloma [Figure

ISSN (O): 2687-5365; ISSN (P): 2753-6556

2] Suggesting of Dyke Davidoff Masson syndrome. All other haematological investigations were within normal limits.

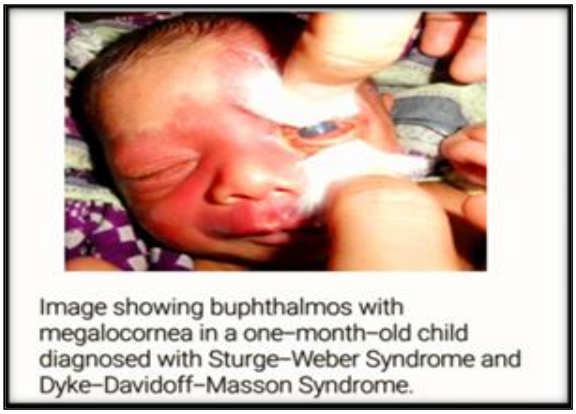

Image showing buphthalmos with megalocornea in a one–month–old child diagnosed with Sturge–Weber Syndrome and Dyke–Davidoff–Masson Syndrome.

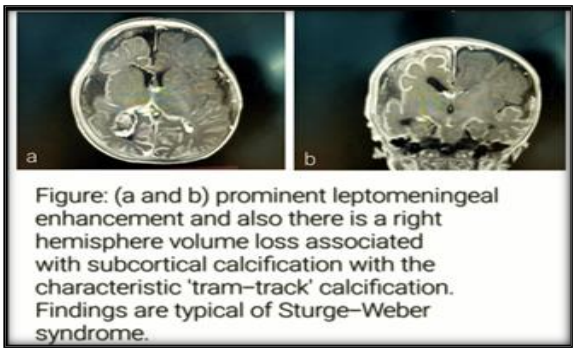

Figure: (a and b) prominent leptomeningeal enhancement and also there is a right hemisphere volume loss associated with subcortical calcification with the characteristic 'tram–track' calcification. Findings are typical of Sturge–Weber syndrome.

Patient had undergone Trabeculotomy along with Trabeculotomy under GA. Post operatively patient received topical prednisolone acetate 1%. Topical antibiotics.

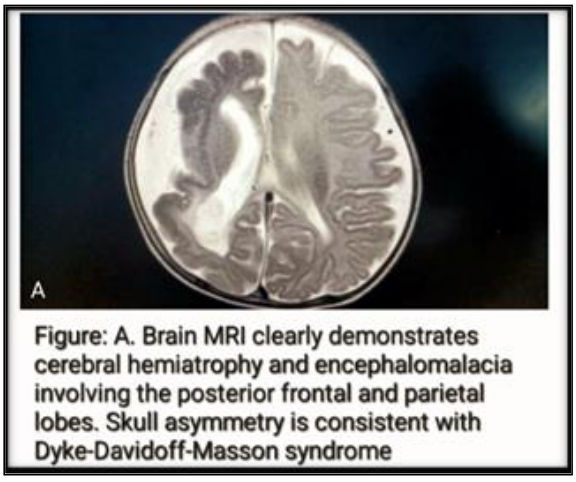

Figure: A. Brain MRI clearly demonstrates cerebral hemiatrophy and encephalomalacia involving the posterior frontal and parietal lobes. Skull asymmetry is consistent with Dyke-Davidoff-Masson syndrome

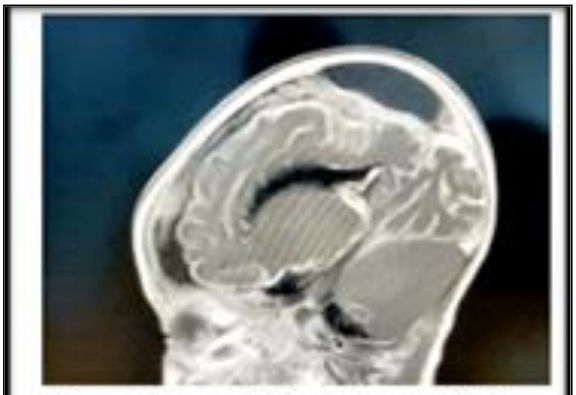

Figure 1: Loss of Parenchyma of left cerebral hemisphere suggestive of Cerebral Atrophy

Figure 2: Enlarged Left Choroid Plexus suggesting of Choroid Papilloma

DISCUSSION

DDMS is a rare syndrome characterized by left hemisphere and male sex dominant cerebral hemiatrophy, contralateral hemiplegia, mental and motor retardation, cognitive impairment, and seizures.[1] SWS is a rare neurocutaneous syndrome whose main clinical features are facial, mostly unilateral nevi, leptomeningeal angiomatosis, and congenital glaucoma affecting the cephalic venous microvasculature.[2] Glaucoma is almost always ipsilateral to the facial port-wine stain. Not all patients with port-wine stains have Sturge-Weber syndrome. Patients with Sturge-Weber syndrome may present with cerebral symptoms without facial findings. Ocular involvement in infancy may present with increased vascularity of the conjunctiva, eye enlargement, strabismus, and increased tearing. Other symptoms are intellectual disability, early handedness, and gaze preferences. Diffuse choroidal hemangioma is seen in about 20% of patients with Sturge-Weber syndrome and is usually on the same side as a facial port-wine stain.[3-5] Diagnosis of Sturge-Weber syndrome is based on typical clinical symptoms, facial appearance, and brain magnetic resonance imaging (MRI) findings.[6] An ophthalmic examination is required to rule out glaucoma. Ocular ultrasound can demonstrate diffuse choroidal thickening which suggests choroidal hemangioma. Gyriform calcifications can be seen on the skull radiographs and are classically described as "tram-

ISSN (O): 2687-5365; ISSN (P): 2753-6556

track sign.[4] Typical radiographic findings of DDMS include cerebral hemiatrophy with ipsilateral ventriculomegaly, compensatory enlargement of the skull, and significant sulcal spaces.[5] Trabeculectomy or glaucoma drainage device may be considered for resistant patients.[6] Treatment is primarily symptomatic, which includes anticonvulsants for seizures. In cases of intractable disabling seizures, hemispherectomy is an available neurosurgical option and has reported an 85% success rate. Long-term management includes physiotherapy for hemiparesis, occupational therapy, speech therapy for speech defects, psychiatric counselling, and medications if required.[7,8]

CONCLUSION

This case underscores the challenges in diagnosing and treating Seizure and congenital glaucoma in SWS with DDM syndrome. Early diagnosis and treatment will have good visual outcome. Underlying cerebral atrophy may result in persistence of episode of seizure. Mental retardation is a common association. Seizure management is often challenging in SWS with DDMS. Patient may undergone surgery for the management of the same.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. Patient father has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that the due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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ISSN (O): 2687-5365; ISSN (P): 2753-6556