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Wyburn Mason Syndrome - A Case Report

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Abstract: ***A 5 year old female child presented with swelling of left eyelid and was treated as chalazion. While routine ophthalmological examination, the child was incidentally detected to have vascular malformation of left eye. The child had no other ocular complaints or any visible malformations. All other systemic examinations were normal. Neurologist opinion was obtained and MRI Brain showed retro-orbital prominent tortuous high flow vessels on the left side adjacent to optic nerve. Digital Subtraction Angiography showed Left Intraconal Compact Nidus Arterio Venous Malformation. The child is asymptomatic and hence is now under observation and regular follow up.***

Keywords: Wyburn Mason Syndrome, Bonnet-Dechaume-Blanc syndrome, Racemose hemangioma, Retinoencephalofacial angiomatosis

1. Introduction

Wyburn-Mason syndrome is a rare non-hereditary congenital disorder of unknown etiology with less than 100 cases reported. Affected infants have arteriovenous malformations (AVM) of retina and brain due to developmental abnormalities affecting the blood vessels.

2. Case Report

A 5 year old female child presented with swelling of left eyelid and was treated as chalazion. On ophthalmological examination, the child was incidentally detected to have suspicious vascular malformation of left eye. Her visual acuity was 6/9 in both eyes not improving with glasses with normal colour vision and full visual fields. Anterior segment examination was normal in both eyes with no restriction of Extraocular movements. Right eye showed normal fundus. Left eye fundus showed tortuous and dilated vessels over the optic disc extending to the periphery as shown in figure 2. The child had no other ocular complaints or any visible malformations. All other systemic examinations were normal. Neurologist opinion was obtained and MRI brain was suggested which showed few retro-orbital prominent tortuous high flow vessels on the left side adjacent to optic nerve. Digital Subtraction Angiography showed Left Intraconal Compact Nidus AVM. The child is asymptomatic and hence is now under observation and regular follow up.

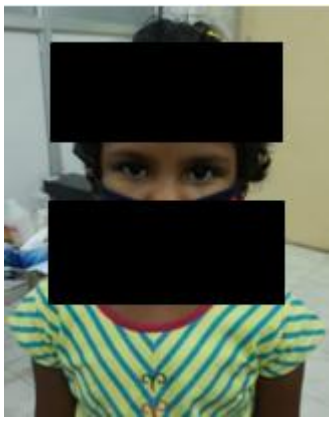

Figure 1: Picture of the Child with Wyburn Mason Syndrome

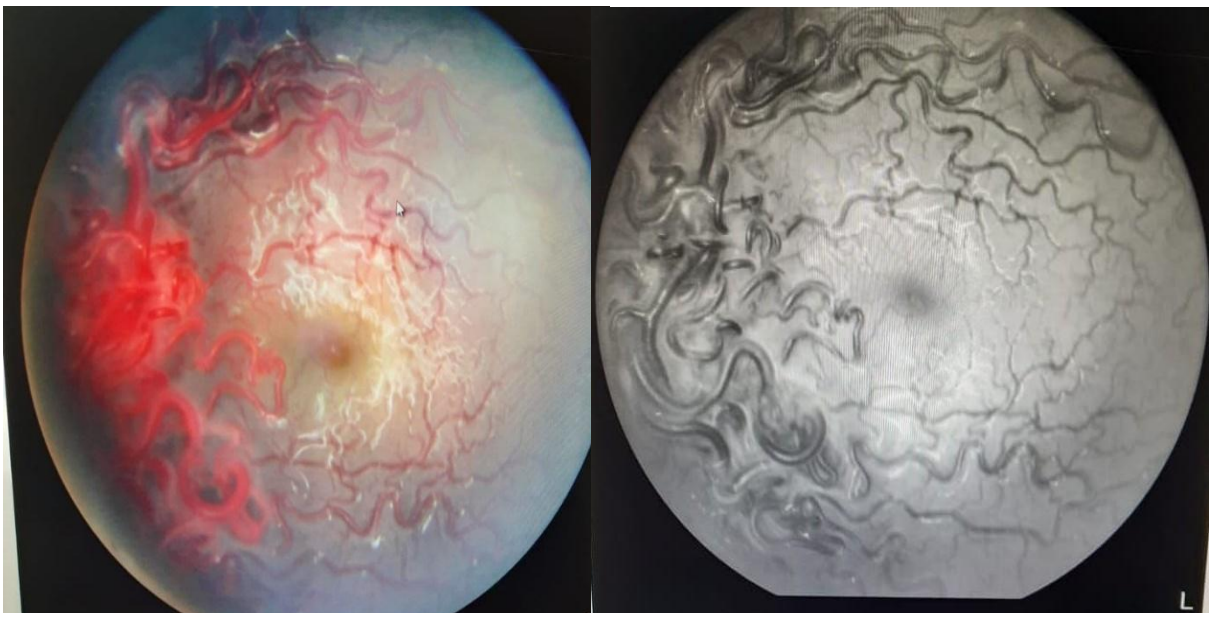

Figure 2: Fundus Image Showing Arteriovenous Malformation in Left Eye

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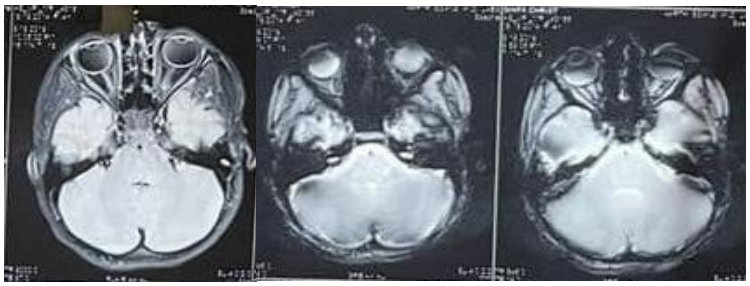

Figure 3: MRI Brain Showing Left Retro Orbital Space Prominent Tortuous Blood Vessels

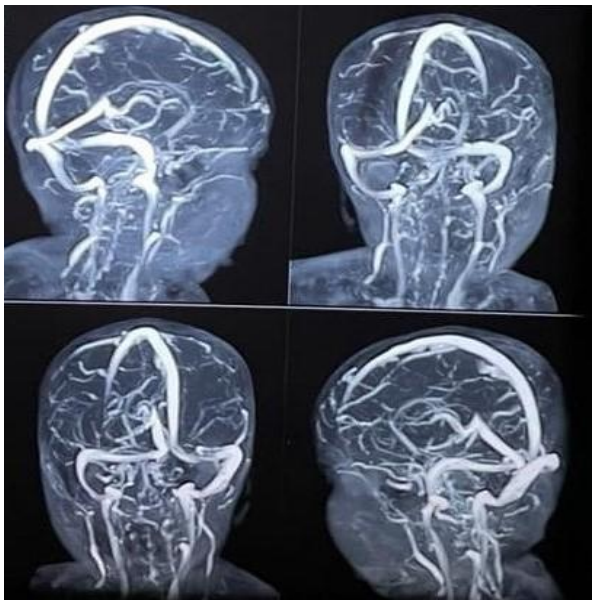

Figure 4: MRA and MRV showing normal study

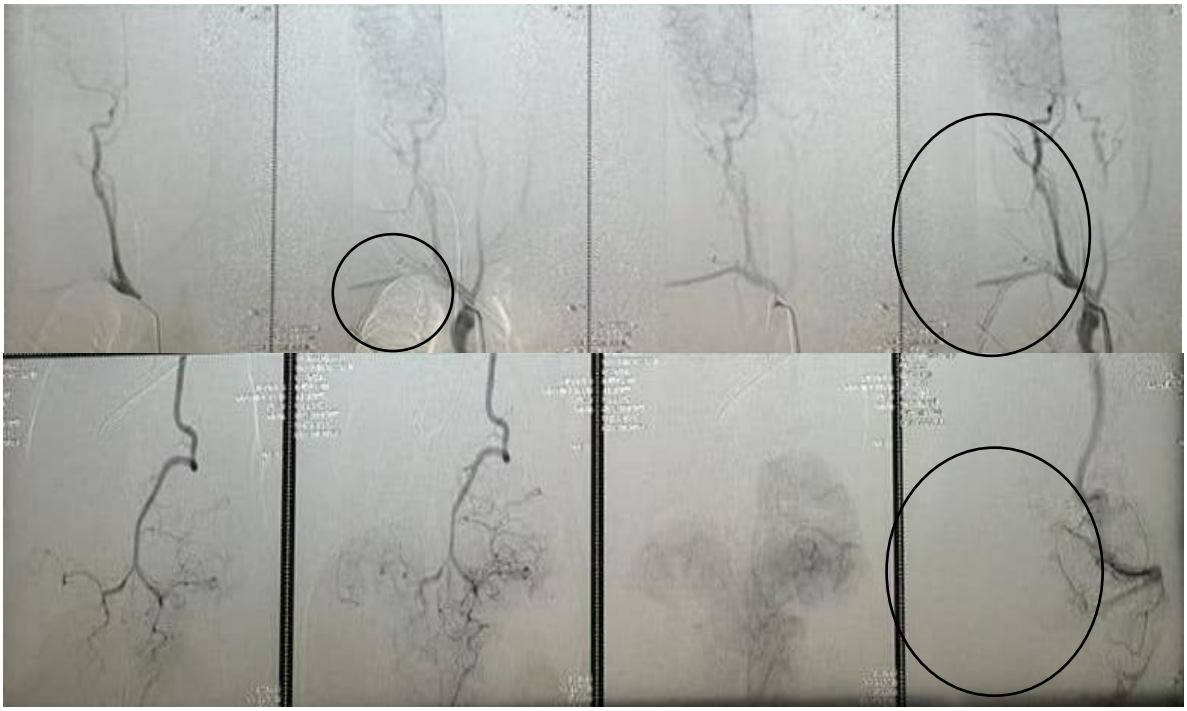

Figure 5: DSA showing compact NIDUS in Left Intra Orbital Region Posterior to Globe

Work up Investigations showed normal blood counts, coagulation profile, serum blood glucose, electrolytes, urea and creatinine.MRI Brain showed Left retro orbital space prominent tortuous blood vessels adjacent to optic nerve suggestive of vascular malformation as shown in figure 3. MRA and MRV showed Normal study as shown in figure 4. Digital Subtraction Angiography showed Compact nidus measuring 18*9.3*8.7mm in left intra orbital region posterior to the globe. Left intraconal AVM fed by left ophthalmic artery and drained by left Superior Ophthalmic vein into left cavernous sinus as shown in figure 5.

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3. Discussion

Wyburn-Mason Syndrome is an exceedingly rare, non-hereditary congenital neurocutaneous disorder with arteriovenous malformations (AVM) found within the midbrain, eyes, orbit and rarely cutaneous nevi. It is often included in the phakomatoses syndromes, which cause tumours in the brain, spinal cord and other organs.[1]

Etiology
The exact etiology, incidence and prevalence rates of Wyburn-Mason syndrome are currently unknown.[1]No specific genetic or hereditary pattern has been identified. Itis caused by a sporadic abnormality in the development of blood vessels during embryonic or fetal growth.[2] It is a very rare disease with less than 100 cases reported, with no racial or gender predilection.[2]

Pathophysiology:
The AVM are direct artery-to-vein communications without a capillary system in between to mitigate the high-flow arterial blood which causes turbulence in the vessel leading to vessel wall damage, resulting in thrombosis and occlusion, leading to ischemia downstream of the occlusion. Histologically, AVM presents with irregularly thick muscularis layer of the arterial and venous walls with or without stromal haemorrhage.

Clinical Features:
Symptoms and signs depend on the number, location, size and type of arteriovenous malformations present. Diagnosis of AVM at an early age is associated with a higher risk of systemic involvement.[1,3]

Ocular signs or symptoms include:
Proptosis, Blepharoptosis, Abnormally dilated vessels of the conjunctiva, Nerve palsies, Nystagmus, Strabismus, Decreased visual acuity or total blindness, Vitreous Hemorrhage, Vein occlusions, Retinal Detachment, Neovascular Glaucoma, Rubeosis Iridis, Optic Disc Edema, Optic Atrophy.Lesions in the eye or orbit are almost always unilateral.[3,4]

Neurological Symptoms include:
Severe Headaches, Vomiting, Seizures, Cranial Nerve Paralysis,Nuchal Rigidity, Epistaxis, Hemorrhage, Hydrocephalus, Hemiparesis, Hemiplegia or Death.AVM in the occipital lobe presents as amaurosis fugax and headache, homonymous visual field defects and visual auras.[5,6] AVM are seen in themidbrain, baso-frontal region and posterior fossa as vascular aneurysms leading to spontaneous haemorrhage, therefore requiring proper imaging.

Cutaneous and other Organ system Signs include:

Facial Angiomas, Oral Hemorrhage, Excessive Bleeding with dental procedures, Hematuria, Hemoptysis.

Classification: [7]
Group 1: Retinal AVM between a major artery and vein with an abnormal capillary plexus in between, which are usually asymptomatic.

Group 2: Retinal AVM between an artery and a vein without any capillary network in between, presenting with risk of retinal complications.

Group 3: Complex and extensive AVM with large vessels without a capillary plexus in between, presenting with high risk for retinal complications and intracranial malformations.

Investigations:
Dilated fundus examination shows abnormal vessels, Fundus Fluorescein Angiography shows vascular anomalies and leakage, MRI Brain and Orbit indicates size, location and mass effect of AVM, Cerebral Angiography shows feeding arteries and draining veins, OCT shows macular edema.

Management:
Multidisciplinary approach is needed. Majority of retinal AVM are stable and managed conservatively. Treatment is reserved for patients with visual symptoms. Brain imaging and regular ophthalmologic evaluations are required for monitoring AVM and its complications. Surgical resection, embolizationand radiation therapyof abnormal vessels of the brain may be considered.Retinal neovascularization or neovascular glaucoma can be treated with panretinal photocoagulation and anti-angiogenic agents and Pars plana vitrectomy for persistent vitreous haemorrhageand retinal detachment. [8]

Prognosis:
Patients with Wyburn- Mason syndrome may remain asymptomatic or have significantly decreased vision to total blindness due to late ocular complications. Recurrence rate for extracranial AVM after surgical resection is high. This condition is associated with high morbidity and mortality due to the high risk of spontaneous cerebral AVM haemorrhage.

4. Conclusion

Majority of the retinal AVM are stable and asymptomatic. Hence they are treated conservatively according to the clinical presentation. Regular follow up, imaging and review is the mainstay of treatment to detect complications earlier and to prevent visual loss. This case is reported for its rarity.

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