

## CONGENITAL ARTERIO-VEINUS MALFORMATION OF OPTIC NERVE HEAD- A RARE CASE

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### Abstract

**Introduction:** Retinal arterio-venous malformations (AVMs) are rare, congenital, high-flow congenital developmental vascular anomalies without intervening capillary bed also known as racemose angioma, They are unilateral, nonhereditary lesions located anywhere in retina and may involve optic nerve.

**Case report:** A 55 year old male was found to have abnormal congenital arterio-venous malformation over optic nerve head in his left eye, as an incidental clinical finding. He had reported for his routine fundus examination due to diabetes. He had no other relevant systemic findings. Further follow up was advised to see the progression of the lesion and structural damage to optic nerve.

**Discussion:** The typical clinical picture, fluorescein angiogram findings, normal MRI brain and orbits, and normal base line blood investigations all points in favour of a congenital arterio-venous malformation.

**Conclusion:** Arterio- venous malformations of Optic nerve head are very rare to find as compared to retinal malformations. This is the first such case report from India.

**Keywords:** Arterio-venous malformations (AVMs), Relative afferent pupillary defect (RAPD), Fundus Fluorescein angiography (FFA), Retinal nerve fibre layer (RNFL), Ganglion cell complex (GCC).

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### INTRODUCTION

Retinal arterio-venous malformations (AVMs), first described by Magnus in 1874,<sup>1</sup> are rare, congenital, high-flow developmental vascular anomalies. They may range from a simple arteriovenous communication to a complex anastomosis where differentiating artery from vein is very difficult.

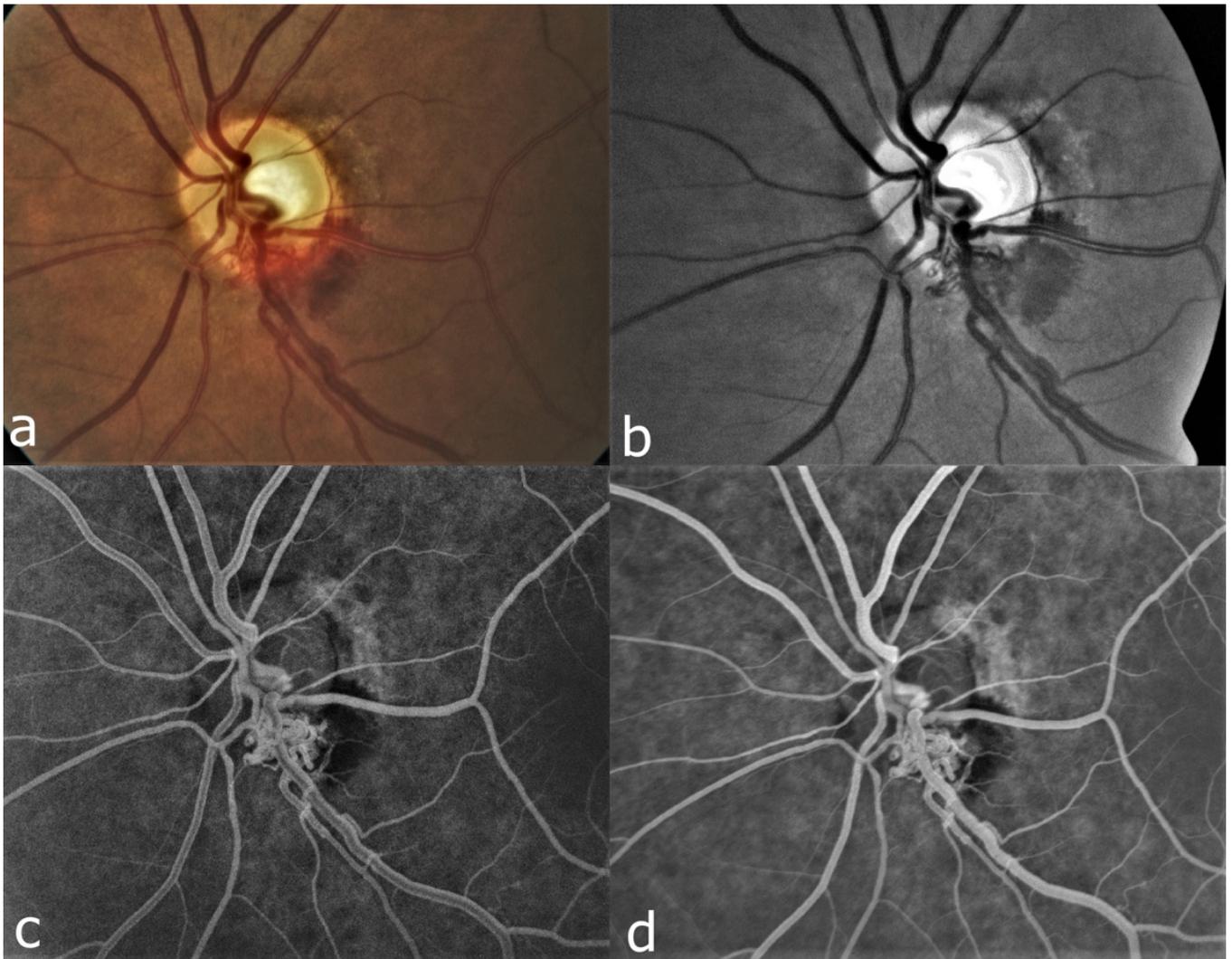
AVMs are unilateral, nonhereditary lesions located anywhere in retina and may involve optic nerve. The retinal lesions may be a part of Wyburn-Mason syndrome, which is characterised by central nervous vascular malformations along the optic tract. Majority of retinal malformations are asymptomatic.<sup>2</sup>

We present a rare case of unilateral congenital arterio-venous malformation of optic nerve head; this is the first case report from India.

**CASE PRESENTATION**

55 years old Indian male, a known diabetic since four years, reported for routine fundus examination.

clock to 6'o clock position. There was a small sub retinal haemorrhage infero-temporal to optic disc (Figure 1a & 1b). Fundus fluorescein angiography (FFA) revealed early filling of vascular channels without leakage in late phase in left eye (Figure 1c & 1d)



**Figure 1, a & b - Fundus colour and red free photo of left eye showing tuft of vessels with haemorrhage, c & d - early and late phase of FFA.**

His best corrected visual acuity was 6/6 both eye. Anterior segment examination was normal for right eye. Left eye revealed relative afferent papillary defect (RAPD). Fundus examination right eye was normal, left eye examination revealed a tuft of abnormal vessels over inferior aspect of the optic disc obscuring the disc margin extending from 5'o

Optical coherence tomography retinal nerve fibre layer (RNFL) revealed thinning in all quadrant and ganglion cell complex (GCC) thinning in all the sectors in left eye (Figure 2a & 2b respectively). Visual field examination left eye revealed blind spot enlargement with superior field defects corresponding to the location of abnormal vasculature (Figure 2c)..

examination and lab investigations were within

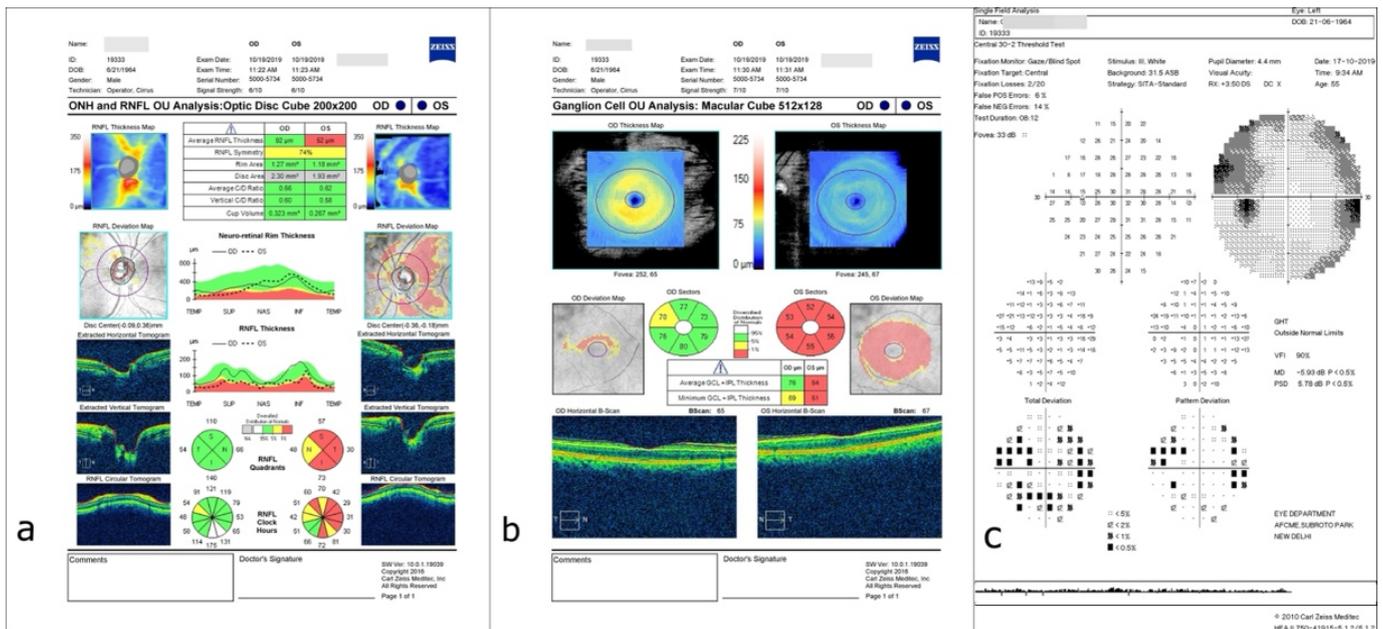


Figure 2, a- RNFL, b- Ganglion cell analysis and c- Single visual field of patient.

IOP were 10mmHg both eyes with applanation tonometry (AT). Pachymetry were 510 μ and 497 μ for right eye and left eye respectively. All systemic

normal limits. MRI brain and orbit was done to rule out any cerebral AVM and was within normal limits (Figure 3).

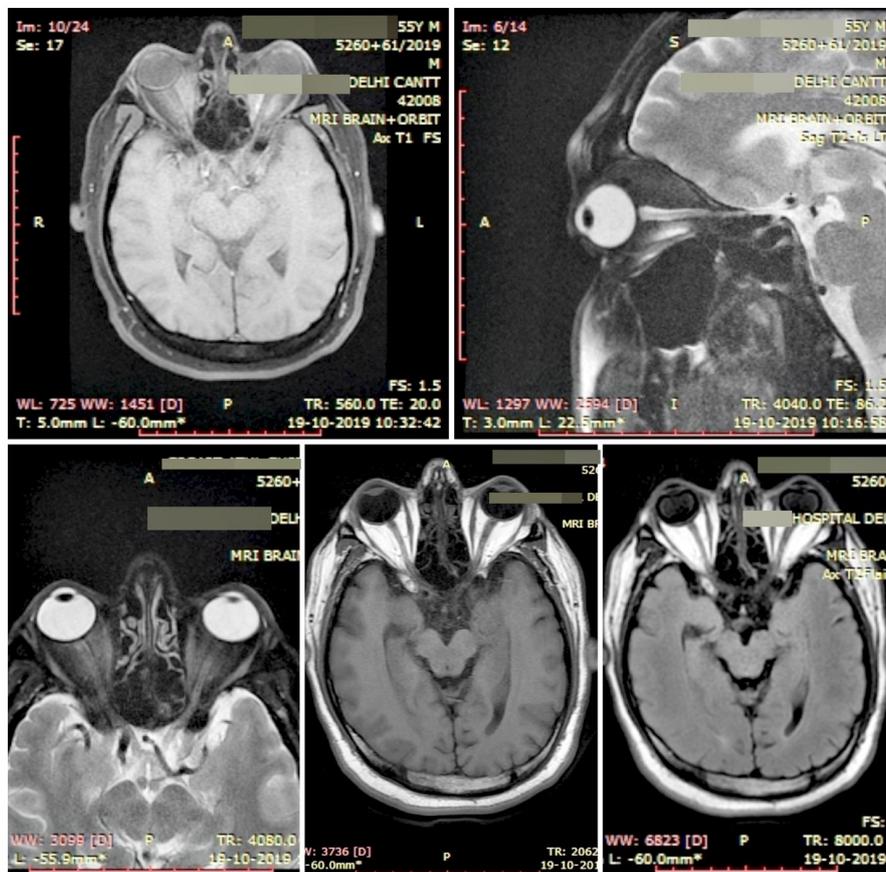


Figure 3, Magnetic resonance imaging of brain and orbit.

This confirms the diagnosis of congenital AVM of optic nerve head. The patient was apprised of his clinical condition and its congenital nature; no active intervention was indicated. Patient was counseled regarding risks associated with visual field defect in the left eye. Patient was called for regular follow up for visual fields on account of RNFL and GCC thinning noted in the left eye to detect progression and fundus examination for diabetic retinopathy.

**DISCUSSION**

Arterio-venous malformation of the retina are rare, sporadic, and nonhereditary disorders those can be associated with intracranial and systemic AVMs. [2] Usually identified accidentally during routine examination and may associated with decrease visual acuity depending on The patient presents with an unusual unilateral retinal vascular anomaly as an incidental clinical finding. The presentation, clinical features and angiographic appearance is consistent with the diagnosis of a congenital arterio-venous malformation of optic nerve head type1. [3]

Congenital AVMs or communications were classified into three types on the basis of severity. [3,4]

Congenital AVMs have rarely been reported as

close to optic disc as well. [4,5] The AVM in our case was located over optic disc inferiorly which was associated with small subretinal haemorrhage.

Association of retinal arteriovenous malformations with systemic arterio-venous malformations is known as Wyburn-Mason syndrome. [2] Various sites of involvement include the central nervous system, skin, lung, and spine, orbit, eyelid, oro-nasopharyngeal area. Hence additional investigations viz MRI brain and orbit are required.

Retinal AVMs are often seen in asymptomatic patients and are believed to be stable, with a little threat to vision. However, central retinal vein occlusion, vitreous haemorrhage, and retinal haemorrhage may occur occasionally with these lesions. Mansour et al. [6] proposed a hypothesis to explain potential complications: AVMs shows Steal phenomenon, which leads to ischemia due to decrease arterial pressure and increased venous pressure, causing the surrounding retina to be ischemic. This could be the possible explanation for structural damage to the retina (GCC thinning) leading to RAPD in our case. In addition, capillary

Table1 - Classification of AVMs. [3]

Types	Features
1	Localized, well-compensated, intervening arteriolar or abnormal capillary plexus between the communicating vessels.
2	Direct arterio-venous malformation without intervening vascular components that may decompensate, leading to leakage of fluid (from slight leakage to extensive retinal edema, exudates and haemorrhages).
3	Large and diffuse anastomosing channels, leading to difficulty in recognizing arterial and venous components; degenerative changes in the retina and poor vision.

optic disc AVM but have a predilection for the papillomacular bundle and the supero-temporal quadrant. They may be found in any quadrant and

ischemia may lead to retinal or vitreous haemorrhage. In cases of exudative maculopathy photocoagulation treatment may be required. [7]

The differential diagnosis of congenital AVMs includes peripapillary telangiectasia, retinal capillary and cavernous haemangioma, and retinovascular disease. [5]

AVMs may be confused with optic disc venous collaterals secondary to retinal venous occlusion or optic disc meningioma. Hence these needs to be differentiate. [7]

Capillary haemangioma of Retina may be sporadic or occur as a part of Von Hippel–Lindau's disease. [8] Clinically; these often present with exudative features and show arteriovenous shunting. [9] These features were absent in our patient.

Typical features of Cavernous haemangioma are its location between two retinal veins and typical appearance of a cluster of grapes.

Peripapillary vascular loops are usually arterial in origin and they appear as simple hairpin loops, figure of eight, or in a corkscrew configuration. They usually extend from the optic disc to the vitreous cavity. They may be associated with complications like vitreous haemorrhage and branch retinal arterial obstruction. [10]

The clinical picture, fluorescein angiogram findings, normal MRI brain and orbits, and normal base line blood investigations all points in favour of a congenital arterio-venous malformation.

## CONCLUSION

The rare features in this case are the location of the arterio-venous malformation, which was over the inferior part of optic disc with associated haemorrhage and no visible leakage on FA. Also this is the first case report from India showing AV malformation of Optic Disc. Clinical features usually depend on type and location of AVMs, e.g. even a small malformation over optic nerve head can lead to severe ischaemic change and ganglion cell death as noted in this case report.

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