



Keywords

Retinal Vascular Malformation,
Racemose Angiomatosis,
Retinal Macrovessel,
Prepapillary Vascular Loop

Received: June 4, 2016

Accepted: June 21, 2016

Published: February 8, 2017

An Isolated Prepapillary Arterial Looping Malformation

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Citation

Ibrahim Kocak. An Isolated Prepapillary Arterial Looping Malformation. *International Journal of Clinical Medicine Research*. Vol. 3, No. 6, 2016, pp. 95-98.

Abstract

An unusual retinal arterial malformation is presented in this case report. A 50-years-old female with no significant past medical history, presented with an unusual retinal arterial malformation without any vision loss in the right eye. Inferior temporal retinal artery had a corkscrew like looping and extension to vitreous cavity at its emerging site from optic disc. Fundus fluorescein angiography confirmed its arterial source. Optical coherence tomography and cranial magnetic resonance imaging showed normal findings. Ophthalmologic findings were not consistent with well-known retinal vascular malformations like Wyburn-Mason syndrome or retinal macrovessels. It can be classified in congenital prepapillary vascular loop disorders.

1. Introduction

Congenital retinal arteriovenous malformations (AVM) are a group of rare nonhereditary disorders involving retinal vascular structure. They comprise a spectrum of anomalies from an arteriovenous communication to retinal macrovessels (RM). Since associations with neurological and facial involvements are not rare, additional investigation may be required. Initial case series of retinal AVMs were published by Wyburn-Mason [1]. Wyburn-Mason syndrome (WMS), also known retinoencephalofacial or racemose angiomatosis (RA) is a relatively well-known type of retinal AVM because of the author's initial description. Also, there are some reports including rare disorders like hereditary retinal arteriolar tortuosity [2] inherited retinal venous beading, congenital prepapillary vascular loops [3], hereditary hemorrhagic teleangiectasis [4]

The presenting case shows an unusual retinal arteriolar malformation without systemic involvement.

2. Case Presentation

A 50-years-old female with no significant past medical history, referred to our clinic with reading difficulty that is related to her presbyopia. Best corrected visual acuity was 20/20 in both eyes. Intraocular pressures were in normal range. Slit-lamp anterior segment examination revealed normal findings except bilateral conjunctival melanosis. Dilated fundus examination disclosed an unusual retinal arterial malformation in the right eye. Inferior temporal retinal artery had a corkscrew-like looping and extension to vitreous cavity at its emerging site from optic disc (Fig. 1).



Fig. 1. Anomalous looping of inferior temporal arteriole.

Fundus fluorescein angiography confirmed its arterial source and had no leakage. (Fig. 2) Optical coherence tomography and cranial magnetic resonance imaging showed normal findings.

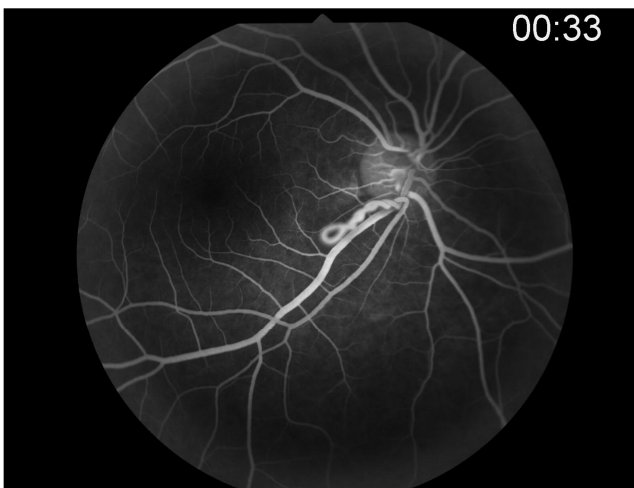


Fig. 2. Fundus fluorescein angiography during venous phase.

3. Discussion

Vascular malformations of retina are rare, sporadic, and nonhereditary disorders those can be associated with systemic involvement. They are considered to be a type of congenital nonhereditary phacomatoses. Congenital RM and RA are relatively well defined malformations.

RMs are large, anomalous blood vessels, crossing the horizontal raphe. Some authors classified RM as Type 1 RA [5]. Abnormal embryologic development during the 4th month of gestation may have a substantial role in pathogenesis. RM is generally venous, unilateral, and asymptomatic [6, 7]. Congenital RM is mostly identified during routine examination. Visual acuity decrease rarely occurs. Decreased visual acuity can be related to complications such as branch retinal artery occlusion, serous detachment, hemorrhage, macular ischemia, or angioscotoma

[6-9]. Congenital RM can involve an arteriole or a venule alone, or both of them. Usually, one or more large veins passes the central macula and have branches extending beyond the horizontal raphe [10]

RA (WMS) is characterized by anomalous arteriovenous communications. The terminology of this disorder is controversial, including nomenclature such as Wyburn–Mason syndrome, Bonnet–Dechaume–Blanc syndrome, and congenital unilateral retinocephalic vascular malformation syndrome. It is classified as Type 1 in case of abnormal capillary plexus between the major vessel arcades; Type 2 in the presence of direct arteriovenous anastomosis without interspersed capillaries in a limited area; and Type 3 in case of massive arterio-venous shunts throughout the entire fundus [6]. Most of the cases are Type 2 and 3, because Type 1 RA is usually asymptomatic and difficult to detect. Type 2 and Type 3 are generally associated with cerebral vascular malformations [11]. Massively dilated vessels can be mistaken for angiomatic lesions. According to another classification, there are ‘typical and atypical congenital unilateral retinocephalic vascular malformation syndrome groups [12]. Typical group comprises cerebral and retinal vascular malformations associated with facial vascular skin lesions. Atypical group signifies the absence of skin lesions. RA diagnosis is usually in young adulthood, but it can be diagnosed at any age. RA may cause complications such as retinal and vitreous hemorrhages, retinal exudates caused by the high blood flow in the arterio-venous communications, retinal venous occlusions, neovascular glaucoma. Retinal complications or cerebral vascular malformations involving the anterior visual pathway lead to severe visual decrease. The diagnosis is easy with the typical fundus view. In the fluorescein angiography, the high-flow arteriovenous communications and adjacent areas of retinal capillary dropout can be seen. Some patients show mild proptosis. A neurological consultation is required, because cerebral vascular malformations can cause visual loss. Vascular malformations of the jaw can cause severe hemorrhage after dental extraction. Prognosis is guarded; regression or progression of the retinal vascular lesions can develop. There is no effective treatment [12].

After Beyer’s first report of hereditary retinal artery tortuosity and macular hemorrhages, different reports came from Japan, Europe, and United States. This disorder is characterized by retinal arteriolar tortuosity, superficial macular hemorrhages, and autosomal dominant transmission. Venules are spared. Aorta coarctation can be associated with isolated retinal arteriolar tortuosity. Congenital storage diseases and Familial dysotonomia may be associated with combined arteriolar and venular tortuosity. Phacomatoses are associated with vascular tortuosity secondary to arteriovenous shunts as in the case of racemose angiomatosis [13].

Hereditary hemorrhagic telangiectasia (HHT) is a disorder which can involve retinal vessels. It is a rare genetic disorder characterized by hemorrhagic vascular lesions. Visceral,

dermal and mucosal involvement causes complications based on the localization and the severity of telangiectasia. Ocular involvement is not rare in HHT. Most common ocular manifestation of HHT is conjunctival telangiectasia. Retinal telangiectasia, retinal arteriovenous malformations, and choroidal telangiectasia are other ocular involvement types of HHT [14].

As described in *Duane's Ophthalmology* [15]; prepapillary vascular loops are blood vessels that project from the optic disc into the vitreous cavity and then return to the disc to continue their natural course. First described by Liebrich in 1871, over 90 cases have since been reported. The loops have at least one ascending and one descending limb, and 85% to 95% are arterial in origin. Occasionally, an arterial loop projects from the disc and returns to the retina, whereas its venous counterpart may arise from the retina and exit into the disc. Rarely, a loop arises from the retina and returns to the retina; this particular anomaly has been termed a preretinal vascular loop. The vessels may appear as simple hairpin loops (180° turns), in a figure eight, or in a corkscrew configuration. In about 30% of cases, the loop is surrounded by a white, glial-appearing sheath. The average arterial loop extends approximately 1.5 mm into the vitreous cavity, probably within Cloquet's canal. Venous prepapillary loops usually are less elevated. In contrast to persistent hyaloid arteries, prepapillary vascular loops have not been observed to extend as far anteriorly as the posterior lens capsule. Embryologically, prepapillary loops are thought to occur at about the 100-mm stage, when the retinal vessels are developing. For an unknown reason, a vessel probably grows into Bergmeister's papilla, which is maximally developed at about the 180-mm stage, and then returns to the retina. It has been proposed that the loop requires Bergmeister's papilla as a scaffold for growth. Thus, its growth is limited, since Bergmeister's papilla usually does not extend more than one third of the distance into the vitreous cavity. Prepapillary loops are rare; the incidence ranges from 1 in 2000 to 1 in 9000 patients. Bilaterality occurs in about 9% to 17% of cases of arterial loops, but the percentage with venous loops is uncertain. Arterial prepapillary loops most commonly supply the inferior retinal vascular system, in contrast to venous prepapillary loops, which usually drain the superior retinal vascular system. On fluorescein angiography, prepapillary loops demonstrate a rapid flow. However, there may be a sector delay in filling of the optic disc or the area of retina supplied by the loop caused by the increased distance that blood must travel through the loop. Cilioretinal arteries have been seen in up to 75% of eyes with prepapillary loops. Ocular complications associated with prepapillary vascular loops include branch retinal artery obstruction in the area of retina supplied by the loop, amaurosis fugax, and vitreous hemorrhage. Presumably, kinking of the loop and impairment of blood flow dynamics in some way contributes to the obstruction. Vitreous hemorrhage occurs in conjunction with acute posterior vitreous detachment. No associated systemic abnormalities have been found associated with prepapillary loops. The differential diagnosis of arterial prepapillary loops

includes persistent hyaloid artery. However, the latter is only a single vessel, without ascending and descending branches. Congenital venous prepapillary loops must be differentiated from the acquired variety. Congenital loops usually are single and unassociated with other ocular abnormalities, whereas acquired venous loops often are multiple and seen with disease entities such as retinal venous obstruction and optic nerve tumors [15].

4. Conclusion

The current case has a sporadic, isolated congenital retinal arterial malformation without any systemic involvement. Differential diagnosis includes congenital retinal vascular malformations and persistent hyaloid artery. Relatively frequent vascular anomalies like RM and RA often have systemic involvement and venous in origin. Persistent hyaloid artery has a different shape with only a single vessel, without ascending and descending branches. The presenting case is similar in shape with prepapillary vascular loop disorder, nevertheless with a much longer extension into vitreous and having very prominent and more numerous coils. It should be classified in prepapillary vascular loop disorder, a rare malformation, only 90 cases of which have been reported since the first description of Liebrich in 1871.

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