



Unilateral vascular abnormality: A case of peripheral retinal arteriolar tortuosity associated with a prepapillary vascular loop

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ARTICLE INFO

Keywords:

Vascular tortuosity
Prepapillary loop
Unilateral vascular abnormality
Congenital

ABSTRACT

Purpose: To report a case of unilateral prepapillary vascular loop along with peripheral third order retinal arteriolar tortuosity in the same eye, and to discuss potential diagnostic considerations.

Observations: Color fundus pictures and wide-angle fluorescein angiography (FA) demonstrated a pre-papillary vascular loop and a region of retinal arteriolar tortuosity in third order arterioles superotemporally in the left eye. The examination and ancillary testing on the right eye were normal.

Conclusion and Importance: The vascular abnormality in this case does not fit a pattern present in other disease states both hereditary or acquired. The patient's immediate family do not demonstrate a similar abnormality and the patient remains visually asymptomatic upon one-year follow-up.

1. Introduction

Retinal vascular abnormalities have been reported in a number of conditions both hereditary and congenital with varying degrees of visual impairment and differing systemic associations. Prepapillary retinal vascular loops are usually unilateral and congenital. They are far more commonly associated with an artery than a vein, typically arising from a hemiretinal artery root overlying the optic disc. They carry a small risk of occlusion but are generally benign in nature.¹ Furthermore, retinal vascular tortuosity can also be a sign of a systemic disease state. The use of high-quality fluorescein angiography imaging can help to appreciate minute details in retinal vasculature in order to guide further exploration.

2. Case report

A 16-year old visually asymptomatic female with no significant past medical or surgical history was referred to our retina group practice for the evaluation of a retinal vascular abnormality in the left eye detected on a routine eye exam. The patient did not have a history of perinatal or postnatal medical problems and was born at term.

Visual acuity was 20/20 in each eye, and intraocular pressures were 19 mmHg and 18 mmHg for OD and OS, respectively. Anterior segment examination was unremarkable OU. Posterior segment exam was

normal OD, but in the OS, a prepapillary vascular loop (Fig. 1) and focal superotemporal arteriolar tortuosity was discovered. Wide angle fluorescein angiography showed multifocal third order arteriolar tortuosity in the superotemporal periphery OS, with hairpin turns and corkscrew characteristics (Fig. 2, A-B, curved arrows). There was delayed AV transit within the tortuous superotemporal arterioles. The prepapillary vascular loop was in a different distribution, associated with the inferior hemiretinal branch artery. The distal portions of the inferotemporal retinal vein crossed well over the horizontal meridian (straight arrow). This is likely of less significance, and similar crossing of the superotemporal arcade artery was noted OD (Fig. 3). There was otherwise normal microcirculation without vascular leakage or occlusion.

Examination and imaging of the patient's two male siblings as well as both parents were without notable retinal findings. The patient underwent magnetic resonance imaging of the head and neck with and without contrast, which was unremarkable.

3. Discussion

The patient's clinical appearance did not fit neatly into established criteria for a number of hereditary and non-hereditary syndromes. As the patient's siblings and parents did not have a similar abnormality or other notable conditions, a hereditary disorder seems less likely.

Prepapillary vascular loops are usually unilateral and congenital. As

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<https://doi.org/10.1016/j.ajoc.2020.100869>

Received 18 May 2020; Received in revised form 14 July 2020; Accepted 9 August 2020

Available online 13 August 2020

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Fig. 1. Prepapillary vascular loop in inferior hemiretinal branch artery OS.

in our case, they are far more commonly associated with an artery than a vein, typically arising from a hemiretinal artery root overlying the optic disc. They carry a small risk of occlusion but are generally benign in nature. They may be a component of persistent hyperplastic primary vitreous.¹ Additionally there is a report of prepapillary vascular loops in a case of Williams-Beuren syndrome,² however the affected patient presented with bilateral visual decline, CME and mild foveal dysplasia which were not present in our patient.

Familial retinal arteriolar tortuosity is characterized by tortuosity of second or third order retinal arteries with inheritance typically being autosomal dominant. Furthermore, familial retinal arteriolar tortuosity is generally seen bilaterally with involvement of the posterior pole and not solely in the periphery or in a segmental pattern.³ Mutations in the COL4A1 gene, encoding type IV collagen within the basement membrane, have been reported in this condition.⁴

Retinal tortuosity has also been reported in Wyburn-Mason Syndrome.⁵ This is a rare nonhereditary disorder characterized by multiple arteriovenous malformations predominantly affecting the face and brain. Visual symptoms at presentation range from normal vision to absence of light perception with headaches, decline in vision, proptosis and other neurological symptoms which were not present in our patient. In particular, the multifocal pattern and lack of venous involvement make Wyburn-Mason syndrome unlikely.

Retinal arterial tortuosity has been previously described in a case of Moyamoya syndrome.⁶ In this case, the tortuosity was diffuse and bilateral, and also appears to have been associated with some venous involvement. Because of the potential serious cerebrovascular abnormalities associated with these conditions, our patient underwent CNS imaging, which was negative.

Other causes of retinal vascular tortuosity include neurofibromatosis, congenital retinal venous tortuosity, Fabry disease, and branch retinal vein occlusion, all of which are associated with venous involvement. Neurofibromatosis is a neurogenetic disorder classically displaying café-au-lait macules, bony abnormalities and neurofibromas. While

a correlation has been shown between the presence of retinal microvascular abnormalities with age and the presence of neurofibromas,⁷ our patient does not display the characteristic findings of neurofibromatosis. Subsequent to the patient's presentation, her father was diagnosed with glioblastoma multiforme, which may also be associated with neurofibromatosis, but in his case there is also no other systemic manifestation.

In summary, we present a case of unilateral multifocal retinal arterial abnormalities in a 16 year old female. The pattern does not resemble previously described syndromes, and there is no apparent systemic disease association. All immediate relatives are visually asymptomatic with no tortuosity on imaging. Follow up at one year revealed no changes.

Patient consent

Consent to publish the case report was not obtained. This report does not contain any personal information that could lead to the identification of the patient.

Funding

No funding or grant support was received.

Authorship

All authors attest that they meet the current ICMJE criteria for Authorship.

Declaration of competing interest

JP serves as a consultant for Alcon. The following authors have no conflicting interests to disclose (PP, MM, HW).

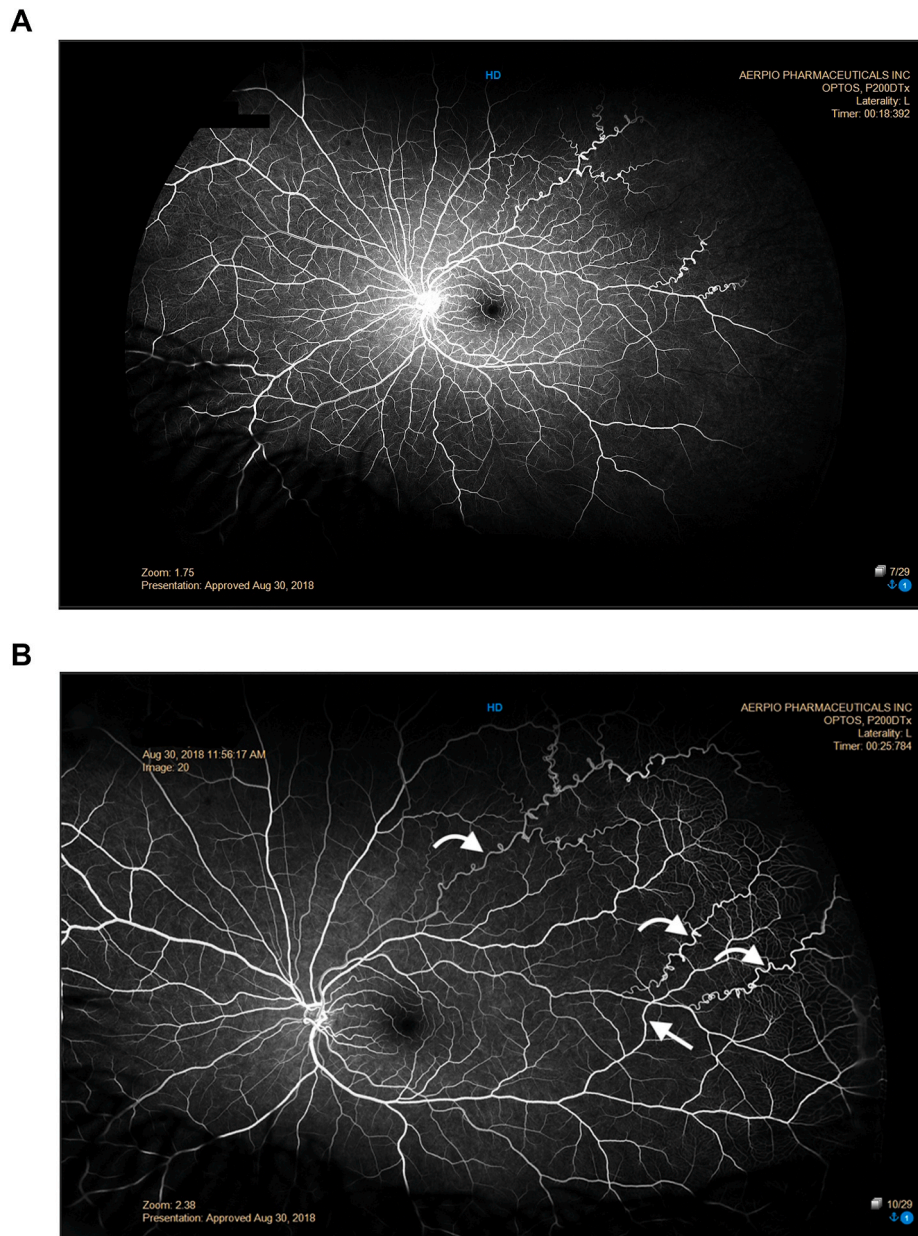


Fig. 2. A-B. Fluorescein angiography OS demonstrated arteriolar tortuosity superotemporally with delayed A-V transit (curved arrows). There is crossing of the distal inferotemporal arcade vein over the horizontal meridian (straight arrow).

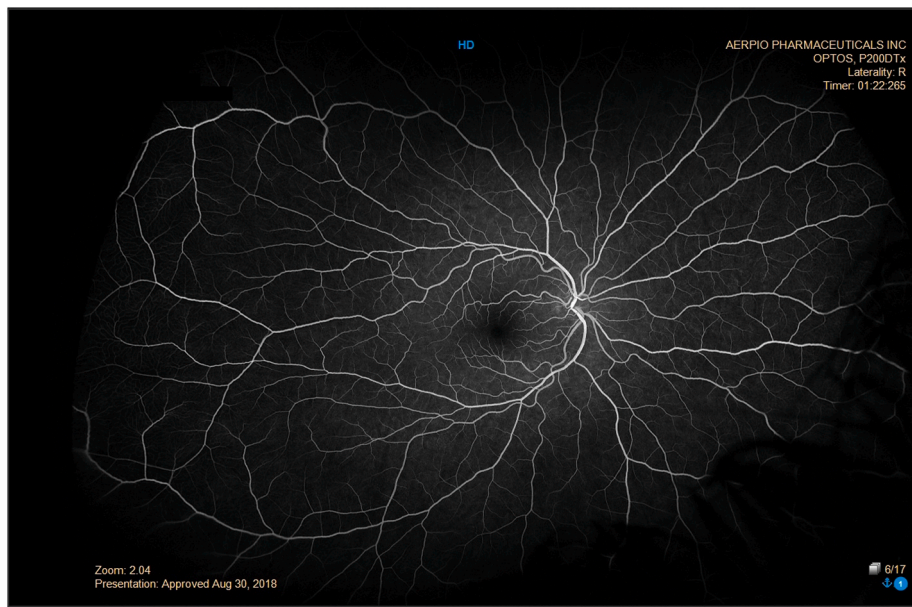


Fig. 3. Normal wide-angle fluorescein angiography OD.

Acknowledgment

None.

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