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Case Report

Congenital Retinal Macrovessel in Stargardt Disease: A Case Report

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Abstract

Purpose: To describe a presentation of Congenital retinal macro vessels (CRM) with Stargardt disease.

Case presentation: A 12-year-old girl was examined with the complaint of recent visual loss in both eyes. Anterior segment examination and intraocular pressure were normal in both eyes. Fundal examination of both eyes showed multiple yellow-white retinal flecks and macular atrophy with a bull's eye appearance. In the right eye an abnormal artery was seen separating from the inferior arcade, that` turned upward and passed above the atrophic macular area. Fundus auto-florescence and Fluorescein angiography confirmed the diagnosis of Stargardt disease and an asymptomatic CRM.

Conclusions: According to the observations made in this study, CRM may occur in conjunction with macular dystrophy, the first ever observation to the best of our knowledge. CRMs are often diagnosed as an isolated abnormality and accidentally detected. However, as they have also been reported to be concurrent with systemic/retinal diseases it is warranted to do a more thorough retinal and systemic examination once CRM is diagnosed.

Keywords: *Congenital Retinal Macrovessel; Aberrant Vessels; Retinal Macrovessel; Stargardt Disease*

Introduction

CRMs are unusual unilateral vascular anomalies consisting of large aberrant branches of retinal arteries or veins that usually cross the macula [1]. Vision is generally not affected [1,2]. However, they may cause visual impairment when crossing the fovea or as a result of formation of foveolar cysts, retinal edema, or hemorrhage [1,3-6].

Prevalence of CRM has been estimated to be approximately 1/200,000 [1]. It is believed that they are formed in week 15 - 16 of gestation [3,7] and their development is independent of that of retinal nerves [5]. The first description of the retinal abnormal vessels was presented by Mauthner in 1869 [8]. They were later defined as large aberrant vessels crossing the middle horizontal raphe without affecting visual acuity in 1982 by Brown [1]. Reported cases are usually isolated vascular abnormalities. And do not coincide with other retinal diseases.

Herein we report a particular type of CRM that is also associated with Stargardt disease. To the best of our knowledge the concurrent CRM and Stargardt disease has not been reported in any other study.

Case Report

A 12-year-old girl came to our clinic with the complaint of recent visual loss in both eyes. Past medical and drug history were negative. Visual acuity was 20/200 in both eyes.

There were no pathological findings in anterior segment examination. Intraocular pressure was within normal limits.

Fundal examination of both eyes showed multiple yellow-white retinal flecks throughout the posterior pole with extension to the equatorial retinae and frank macular atrophy with a bull's eye appearance. In addition, an abnormal artery was detected in the right eye. The abnormal branch separated from the inferior arcade then turned upward and passed above the atrophic macular area, traversing a horizontal path thereafter. No evidence of hemorrhage, exudate, aneurysms or edema was seen along the path of the abnormal branch (Figure 1).

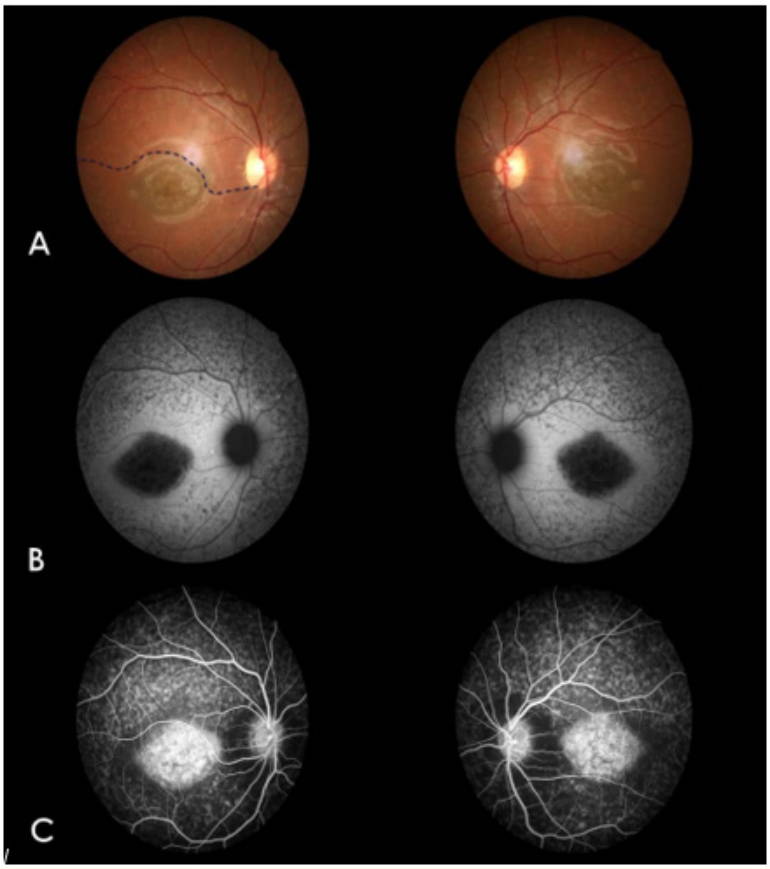

Figure 1: *A: Color fundus photograph of both eyes showing bull's eye maculopathy. The aberrant vessel has been shown as a blue dotted line. B: Fundus autofluorescence showing a hypofluorescence area in the fovea corresponding to RPE atrophy with peripapillary sparing of the RPE changes in both eyes. C: Fluorescein angiography showing "dark-choroid" sign. and window defect in the macular region corresponding to RPE atrophy in both eyes. The abnormal artery in the right eye shows no leakage or staining. The vascular pattern of the left eye is normal.*

Fundus autofluorescence demonstrated a diamond shaped hypofluorescence area in the fovea corresponding to RPE atrophy. Characteristic peripapillary sparing of the RPE changes and the outward expanding pattern of hypoautofluorescent (near posterior pole) and then hyperautofluorescent (in periphery) flecks were also observed (Figure 1).

In fluorescein angiography, both eyes showed the "dark-choroid" sign. Scattered hyper and hypo fluorescence dots with a poorly defined margin, mostly concentrated in the upper macular region and superior arcade, were seen corresponding to the flecks. A 3-disc diameter hyperfluorescent area with an irregular margin was seen in the posterior pole region corresponding to the area of pigment epithelial atrophy (window defect). The abnormal artery of the right eye filled within a normal timeframe and did not show any leakage or staining the vascular pattern of the left eye was normal. There was no arteriovenous anastomosis (Figure 1).

Discussion and Conclusion

An abnormal retinal vessel that crosses the macular region is recognized as a CRM. A rare finding that can be artery [9], vein [1,4] or both. CRMs were first reported by brown in 1982 [1]. According to studies, vascular differentiation occurs in the fourth or fifth month of gestational age from mesenchymal cells [3,7]. Cilioretinal artery is known as the most common vascular anomaly, which originates from the posterior ciliary artery [10].

These vessels often remain asymptomatic. There have been reports stating that these vessels have remained asymptomatic for as long as 14 years [2]. Some cases become symptomatic. Vessels that pass through the fovea my affect visual acuity and result in consecutive amblyopia [11]. Reduced sensitivity of the retina has also been observed in the area where the vessels pass through the macula [12]. Vessels can also be associated with bleeding, exudate, retinal edema, retinal ischemia, vascular obstruction, valsalva retinopathy and foveolar cysts [1,3,6]. As mentioned most cases have been veins [1,4] and cross the macula, however the CRM in this study was an aberrant branch from the inferior arterial arcade that had changed its path upwards and crossed above the macular region finally. This patient did not have any associated pathological findings in fundoscopy.

Angiographic findings such as: early filling and delayed evacuation of the venule, dilated surrounding capillary plexus, areas with no capillary perfusion [6], hyperfluorescence due to RPE alterations [1], nonspecific leaks or alterations in the vascular walls and vein-artery communication [1,4,6] have been reported. In our case the aberrant artery was not associated with abnormal findings in angiography.

These vessels have been observed in systemic diseases such as Wyburn-Mason syndrome [1] and neurofibromatosis [13]. It has also been reported in a case of retinal cavernous hemangioma [14]. Our case although did not associate with any systemic diseases but was unique in terms of its association with Stargardt retinal degeneration. Stargardt disease is the most common form of inherited juvenile macular degeneration with progressive vision loss. The prevalence of Stargardt disease is reported to be between 1:8,000 and 1:10,000 live births [15]. To the extent of our literature review the concomitant presence of these two has not been previously reported. In conclusion CRMs are often isolated and remain asymptomatic. However, the physician should keep in mind that some cases can be affected by complications such as bleeding, exudate, retinal edema, retinal ischemia, vascular obstruction and etc. which indicates the need for follow-up in these patients. Additionally, CRMs can also be associated with systemic/retinal diseases which requires a more detailed examination in suspicious cases.

Disclosure

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