Congenital retinal macrovessel with intracranial venous malformation in a pediatric patient: a case report

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Received: 2021-06-17        Accepted: 2022-02-21

DOI:10.18240/ijo.2022.07.26

Citation: Park HJ, Yoon SH, Kim SY, Lee D. Congenital retinal macrovessel with intracranial venous malformation in a pediatric patient: a case report. Int J Ophthalmol 2022;15(7):1214-1216

Dear Editor,

Congenital retinal macrovessel (CRM) is defined as an abnormal large retinal vessel that encircles the foveal avascular zone (FAZ) [1]. Since the first description of a large aberrant vessel crossing the macula by Mauthner [2] in 1868, there have been several case reports about CRM [3-10]. And recently, Pichi et al [1] reported clinical manifestations and multimodal retinal imaging of 49 eyes with CRM. In their study, they had identified an association between macrovessel in the retina and venous anomalies of the brain and they emphasized the importance of systemic workups including brain magnetic resonance imaging (MRI). Meanwhile, previous reported cases were almost adults and abnormal macular vessel in pediatric patient was rarely reported [7-11]. Souissi et al [7] reported a case of 15-year-old girl with congenital hamartoma associated with CRM that resulted in the mild visual impairment. However, systemic evaluation including brain imaging was not performed.

Here, we report a case of CRM in a pediatric patient aged 4y with cerebrovascular analysis through brain MRI with magnetic resonance angiography (MRA) and retinal vessel analysis using optical coherence tomographic angiography (OCTA).

Case Report A girl aged 4y presented to the Ophthalmic Department with the complaint of frequent eye blinking for a month. The patient had no other history of ocular trauma and no known ocular or systemic disease. Her mother also had no medical history during the perinatal period. The visual acuity was 20/40 in both eyes, and there was no afferent pupillary defect. On slit-lamp examination, mild punctate epithelial erosions on her left eye were observed. For revealing her refractive errors, cycloplegic refraction was performed and +3.00 diopters of hyperopia with +1.00 diopters of astigmatism in both eyes was noted.

There were no remarkable findings on right fundus examination (Figure 1A); however, the left fundus revealed multiple branches of retinal vessels and a large aberrant central retinal vein originating from the inferotemporal disc cup that transverses the macula (Figure 1B). On OCTA images (AngioVue; Optovue, Fermont, CA, USA), the aberrant macrovessel crossed the foveal area beyond the horizontal raphe (Figure 2A), and thin retinal vessels branching from the macrovessel ran through the foveal avascular zone (FAZ; Figure 2B). Moreover, spectral domain OCTA revealed an irregular contour of the inner retinal layers owing to multiple superficial retinal vessels (Figure 2C). There was no exudate, hemorrhage, cyst, or leakage on the retina.

For excluding other cerebrovascular malformations, MRI with MRA of the brain was performed and there was no significant abnormality in the brain parenchyma. However, in the axial three-dimensional time-of-flight image of the MRI, a slightly dilated left superior ophthalmic vein was noted (Figure 3A). Additionally, MRA revealed a small fenestration of the proximal basilar artery (Figure 3B).

Since there were no other symptoms associated with CRM on the left eye, the patient was managed conservatively using glasses to correct hyperopic compound astigmatism. Use of artificial tears for treating corneal erosion was recommended. After 6-month of observation while wearing hyperopic glasses, her best-corrected visual acuity improved to normal range without any evidence of amblyopia. Her eye blinking also improved. On fundus examinations, the CRM remained stable, and complications associated with macrovessels were not observed.

Ethical Approval The study was conducted in accordance with the Declaration of Helsinki. Informed consent for publication was obtained from the family of the patient in this case report.
Previous studies on retinal macrovessels are mostly in the form of case reports\[7-8,10-13\]; therefore, the accurate prevalence of CRM is unclear; however, Hayasaka et al\[9\] reported that unusual retinal vessel patterns were found in 21 of 3506 eyes in their retrospective fundus photographs analysis. Meanwhile, among previous case reports of CRM, pediatric cases have only rarely been reported\[7-8,11\]. de Crecchio et al\[8\] described a patient aged 5y with an anomalous retinal vein crossing the macula horizontally and crossed the fovea. Visual impairment occurred by the mere presence of a foveal lesion. However, there is a limitation that a systemic examination including brain imaging was not performed to reveal any other lesions related to ophthalmic findings.

The current report is meaningful since the patient was the youngest ever, compared to previously reported cases, and intracranial vascular malformation was also noted. Morphologically, CRM can be venous, arterial\[2\], and sometimes arteriovenous communications\[10\]. In Pichi et al’s\[1\] reports with a large series of eyes with CRM, early phase frames of fluorescein angiography (FAG) further confirmed the venous nature of the CRM. However, in this case, FAG could not be performed since the patient was young. Instead, OCTA was performed, and as previously described, microvascular capillary abnormalities around the CRM were evident in the deep capillary plexus image. Moreover, they described that 24% of enrolled patients with CRM had potential cerebral associations. Likewise, this pediatric patient also had asymptomatic cerebrovascular abnormalities, dilated superior ophthalmic vein, and fenestration of the basilar artery.

There have been several hypotheses regarding the presence of aberrant retinal vessels in the FAZ. Embryologically, the development of FAZ is debatable. Some authors propose that the FAZ develops by “apoptotic remodeling” after 36wk of gestation\[14\], while others report that retinal vasculature developed towards the fovea and the foveal region was normally avascular during all gestational periods\[15\]. CRM refers to the presence of aberrant vasculature where capillaries are usually absent, and Bhatia et al\[11\] reported that vascular proliferation may reach the foveola if foveal hypoxia occurs and CRM develops. Meanwhile, Ashton\[16\] explained that CRM develops around 15-16wk of gestation when differentiation of retinal arteries and veins occurs. Moreover, the origin of CRM is probably related to hemodynamic variation. In our

Figure 1 Color fundus photographs in both eyes Comparing to the normal fundus of her right eye (A), fundus photo of the left eye (B) shows a hyperemic disc with more dilated central retinal vessels. There are multiple branches of retinal vessels and a large aberrant retinal vein originating from the inferotemporal disc cup transverses the macula horizontally and crossed the fovea.

Figure 2 Optical coherence tomography angiography (OCTA) images of the left eye using the Angiovue OCTA software of the RTVue XR Avanti (AngioVue; Optovue, Fermont, CA, USA) En-face image of the superficial capillary plexus (A) illustrates an aberrant macrovessel that crosses the horizontal raphe. Deep capillary plexus en-face image (B) shows retinal branches running through the foveal avascular zone. Spectral-domain OCT B-scan image (C) show the irregular contour of inner retinal layers owing to multiple superficial retinal vessels. Shadowing of macrovessel is also noted.

Figure 3 Brain magnetic resonance imaging (MRI) and magnetic resonance angiography (MRA) A: In the axial three-dimensional time-of-flight image, the left superior ophthalmic vein is slightly dilated (arrow). Further, there is no significant abnormality in the brain parenchyma. B: The MRA shows a small fenestration of the proximal basilar artery (arrow).
patient, there was no previous history of ischemic risk, such as maternal gestational diabetes; therefore, so it is unlikely that any systemic disease that could cause foveal hypoxia and development of CRM. Using Ashton’s[16] embryologic concept, it is possible that large interindividual variation in cerebrovascular and retinal vascular anatomy occurred during the gestational period. Most studies have found that CRMs are asymptomatic and visual acuity is preserved[17]. Likewise, our case did not have any discomfort associated with CRM. Her mild visual impairment at the initial visit in the ophthalmologic clinic had improved after wearing glasses; therefore, it was assumed to be owing to her refractive errors rather than the macular lesion. However, visual impairment may be caused by preretinal hemorrhage or by the mere presence of the macrovessel in the foveal area[3]. de Crecchio et al[3] described that 5 of 13 cases complained of visual impairment caused by the CRM in their study. Vitreous hemorrhage, macroaneurysms, retinal ischemia, or macular edema originating from the CRM have also been reported. Therefore, even if there is no visual discomfort in patients with CRM, regular follow-up in an ophthalmology clinic should be considered to check changes in the macular lesion.

CRM can be detected in asymptomatic pediatric patients. In addition to ophthalmic examinations, careful neuroimaging should be considered to reveal other cerebrovascular malformations. For pediatric patients, regular follow-up is necessary considering the complications that are associated with CRM, such as aneurysms, retinal hemorrhage, or amblyopia.

ACKNOWLEDGEMENTS

Conflicts of Interest: Park HJ, None; Yoon SH, None; Kim SY, None; Lee D, None.

REFERENCES


