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OPTICAL COHERENCE TOMOGRAPHY ANGIOGRAPHY FINDINGS IN A CASE OF CONGENITAL RETINAL MACROVESSEL WITH ANOMALOUS RETINAL ANASTOMOSIS ASSOCIATED WITH CONTRALATERAL MYELINATED NERVE FIBERS AND RETINAL VASCULAR ABNORMALITIES

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Purpose: To describe a case of congenital retinal macrovessel complicated by cystoid macular edema associated with contralateral myelinated retinal nerve fibers and retinal vascular abnormalities studied with optical coherence tomography angiography (OCTA).

Methods: Case report.

Results: A healthy 25-year-old woman with decreased vision in her right eye was found to have a congenital retinal venous macrovessel in the macula associated with cystoid edema. In the contralateral amblyopic eye, the examination revealed a tuft of myelinated retinal nerve fibers along the superotemporal vascular arcade associated with superficial vascular abnormalities. A complete multi-imaging examination was obtained, including fundus color photography, fluorescein angiography, indocyanine green angiography, optical coherence tomography (OCT), and optical coherence tomography angiography. At 1-week follow-up, the optical coherence tomography displayed spontaneous resolution of the edema that remained stable at consecutive 1-month follow-up.

Conclusion: Congenital retinal macrovessels can be associated with other ocular developmental anomalies. Vascular complications can occur, leading to macular edema and retinal ischemia. Optical coherence tomography angiography can be useful for the diagnosis and follow-up of this condition.

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Congenital retinal macrovessels (CRMs) are aberrant blood vessels, typically unilateral large retinal veins, which cross the horizontal raphe in the macular region.[1] They seem to originate from abnormal embryologic development during gestation; they are typically asymptomatic and discovered incidentally. Reduction of vision in the involved eye is rare and can be due to macular hemorrhages, foveal cysts, serous macular detachment, or the course of the vessel itself through the foveal avascular zone.[2–5] Congenital retinal macrovessels have been previously described by

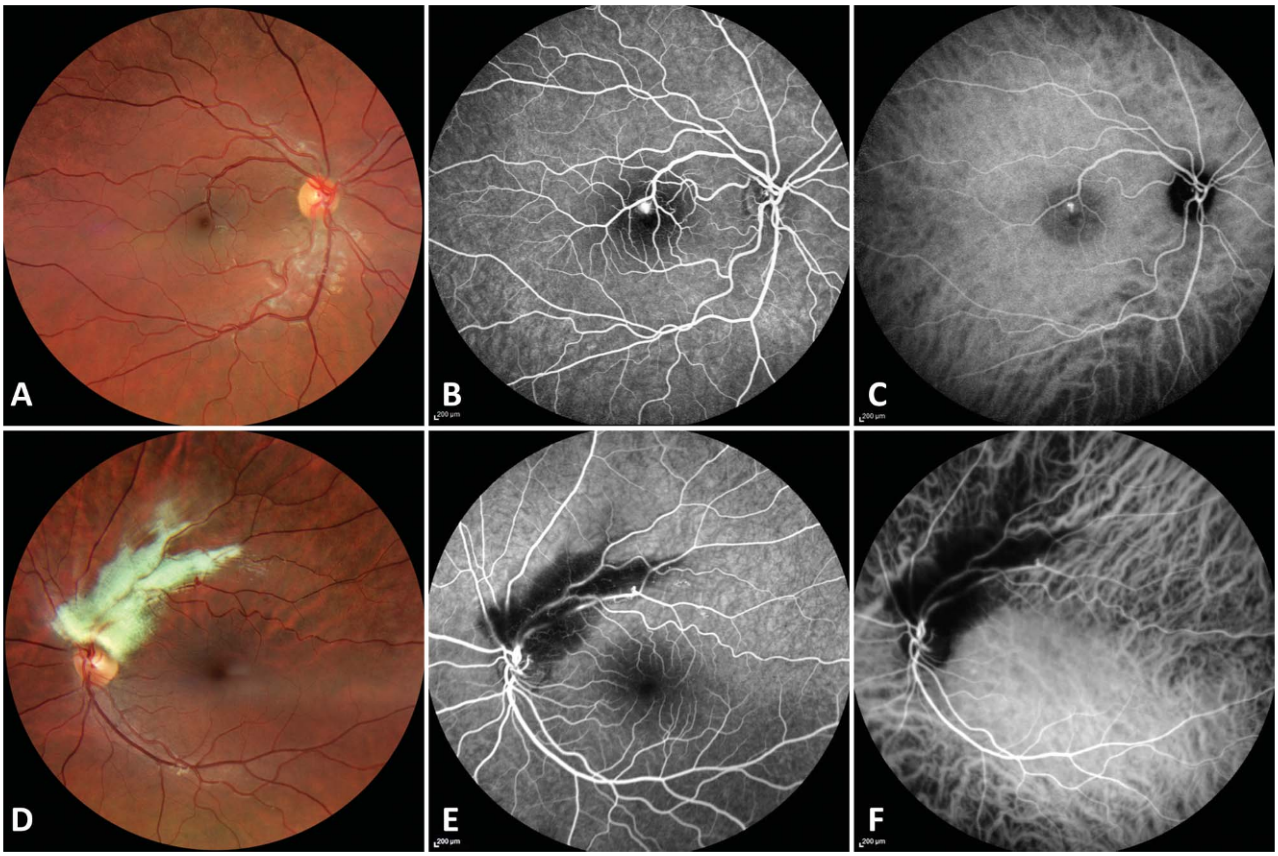

Fig. 1. Multimodal imaging of the patient at baseline. Color fundus photography (**A** and **D**), fluorescein angiography (FA, **B** and **E**), and indocyanine green angiography (ICGA, **G** and **F**) of the right eye (RE, **A–C**) and the left eye (LE, **D–F**) at first visit. Color image of RE (**A**) shows the CRM as a large retinal vein that cross the macular region branching immediately above the fovea. In the LE (**D**), a tuft of MRNF originates from the optic nerve running along the superiotemporal vascular arcade. Fluorescein angiography of RE (**B**) showing early and late dye leakage from the arterovenous communicating vessel located nearby the fovea. In the LE (**E**), small vascular alterations are visible along the superior vascular arcade. Both FA and ICGA images display a hypofluorescent area located respectively in the macular region in the RE (**B** and **C**) and along the temporal arcade in the LE (**E** and **F**) due to the masking effect of CME and MRNF, respectively.

means of fluorescein angiography (FA),[1] but optical coherence tomography angiography (OCTA) currently allows noninvasive, safe, and rapid assessment of retinal vessels[6] and has already been applied to examine vasculature in patients with CRMs.[7–9]

Myelinated retinal nerve fibers (MRNFs) are developmental anomalies in which myelin sheaths extend to a group of retinal nerve fibers along their intraocular portion. Most patients with MRNF are asymptomatic; however, visual function can be affected resulting in axial myopia, amblyopia, and strabismus in the affected eye.[10] Vascular abnormalities have been described in association with MRNF, and they include telangiectasia, branch artery or vein occlusions, retinal neovascularizations, and vitreous hemorrhages.[11]

Herein, we describe a case of CRM with abnormal vascular anastomosis complicated by foveal cysts and associated with vascular and retinal developmental anomalies in the contralateral eye examined using OCTA.

Case Report

A 25-year-old woman presented referring vision loss since 3 days in her right eye (RE). She had no history of ocular trauma or surgery, and she reported known amblyopia in her left eye (LE). Her medical history was unremarkable, and she had no family history of ophthalmic diseases. Her best-corrected visual acuity was 20/40 in the RE and 20/200 in the LE. Dilated fundus examination showed a large superotemporal macrovessel crossing the horizontal raphe near the fovea in the RE and myelinated retinal nerve fiber layer in the LE.

Color fundus photography, near infrared reflectance, fundus autofluorescence, FA, indocyanine green angiography, and optical coherence tomography (OCT) (Spectralis SD-OCT; Heidelberg Engineering, Heidelberg, Germany) were performed and confirmed

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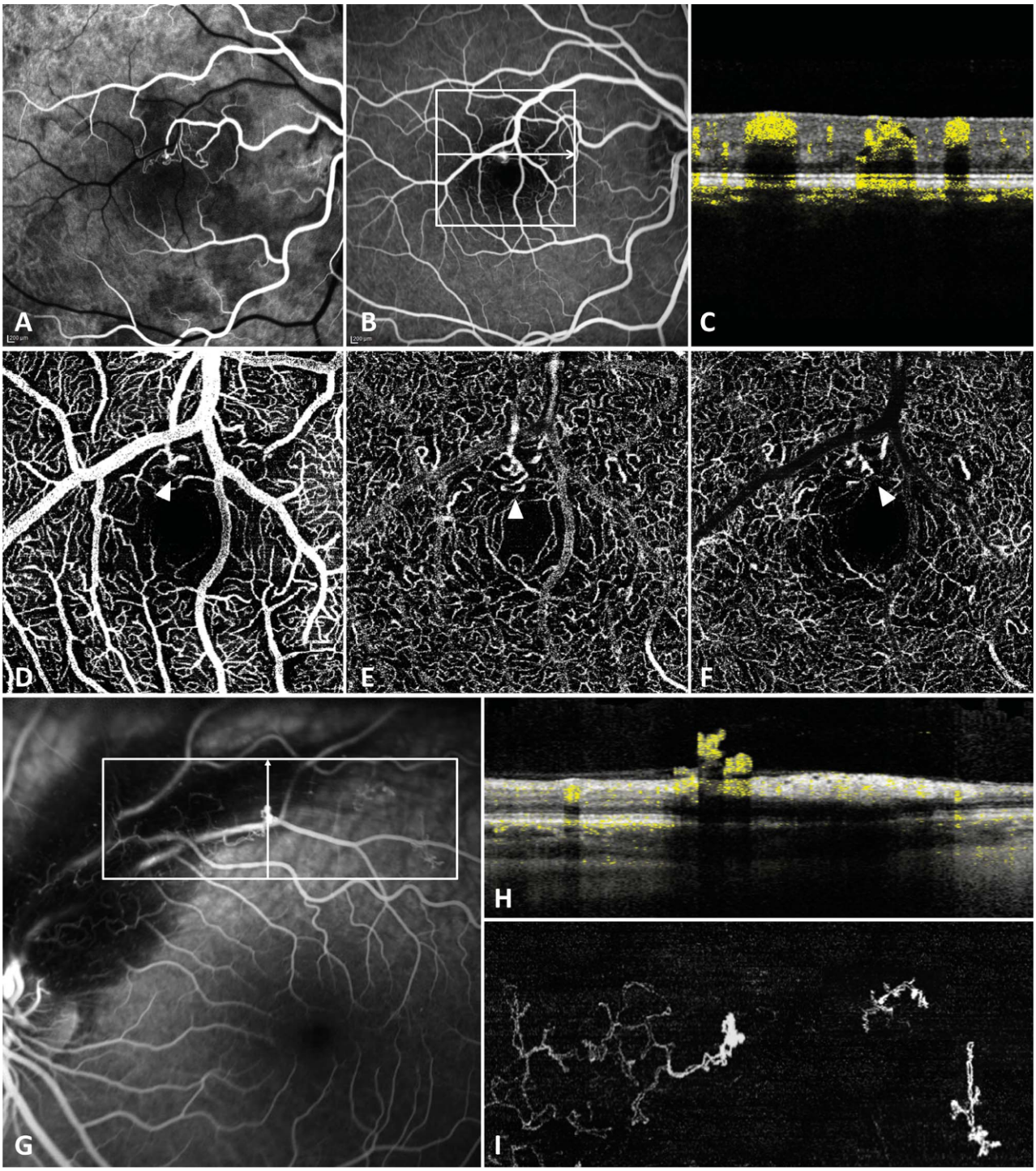

Fig. 2. Optical coherence tomography angiography (OCTA) findings of vascular abnormalities. Fluorescein angiography (**A**, **B**, and **G**), OCTA B-scan with superimposed blood flow signal (**C** and **H**), and OCTA en face images (**D**–**F**, and **I**) of the right eye (RE, **A**–**F**) and the left eye (LE, **G**–**I**). White squares and arrows indicate where exactly the OCTA scans were taken. Fluorescein angiography of the RE (**A** and **B**) shows a large aberrant dilated retinal vein and anomalous arteriovenous communication at posterior pole. The venous nature of the macrovessel is confirmed by the early-phase frame of FA. The vascular anastomosis is visible on the OCTA B-scan (**C**), and the en face images allow to study its distribution through the retinal thickness (arrowheads, **D**–**F**). In detail, the vessel originates from the superficial capillary plexus (SCP, **D**) and extends through the intermediate capillary plexus (ICP, **E**) and deep capillary plexus (DCP, **F**). Dilation and microvascular abnormalities are identified predominantly in the ICP and DCP. Fluorescein angiography of the left eye (**G**) reveals the presence of small telangiectatic vessels along the superotemporal vascular arcade. The B-scan (**H**) and en face (**I**) OCTA pictures permit to localize these vascular alterations in the vitreoretinal interface and to follow their course above the retinal surface.

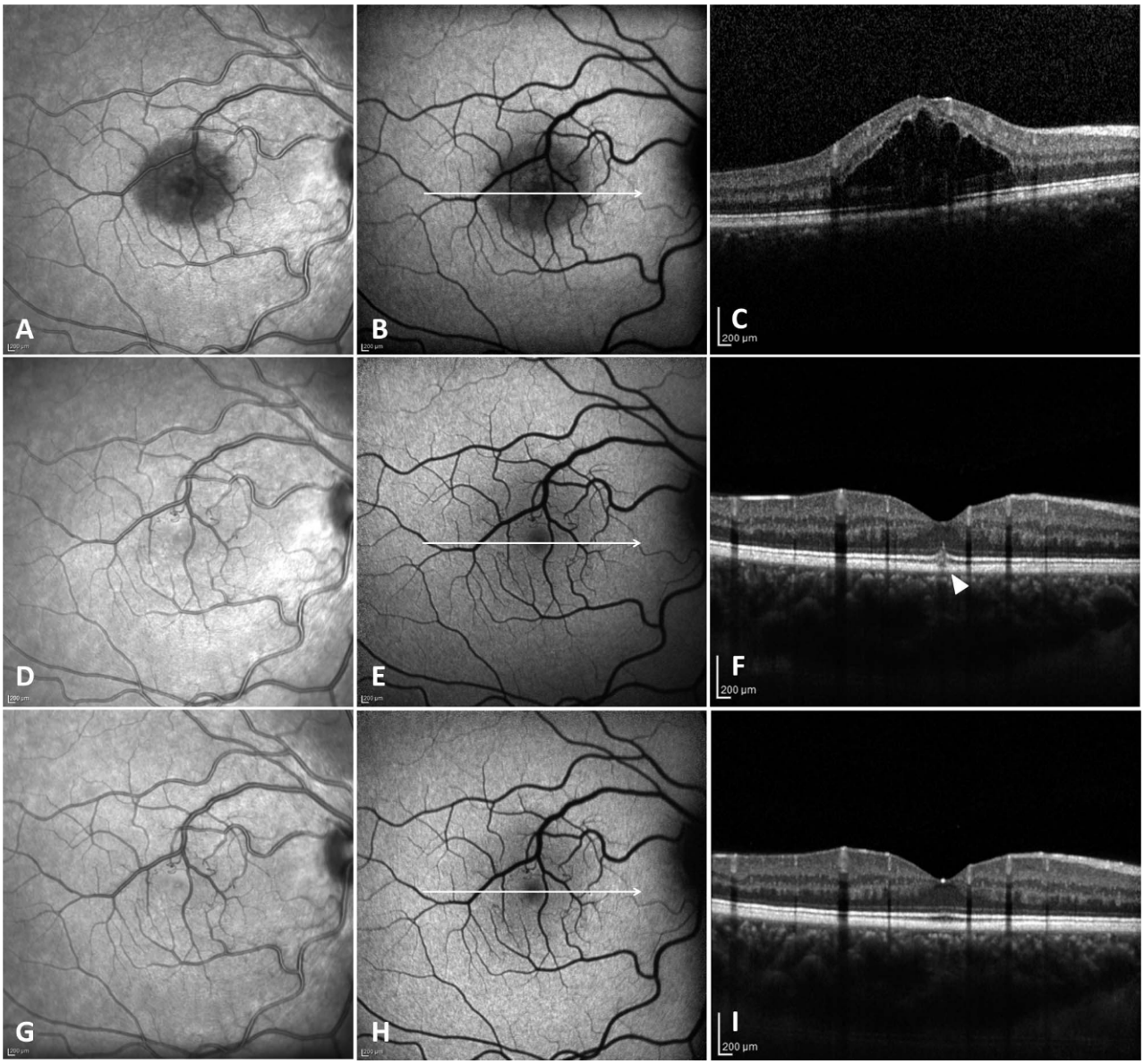

Fig. 3. Follow-up of cystoid macular edema (CME) complicating CRMs. Near infrared reflectance (NIR, **A**, **D**, and **G**), fundus autofluorescence (FAF, **B**, **E**, and **H**), and OCT (**C**, **F**, and **I**) at baseline (**A**–**C**), at 1-week (**D**–**F**) and 1-month (**G**–**I**) follow-up (white arrows showing the location where the OCT scans were performed). Baseline NIR and FAF images (**A** and **B**) reveal a round macular iporeflective area due to the presence of CME, corresponding to retinal thickening and intraretinal cystic areas of low reflectivity on OCT (**C**). In our case, the CME resolved without any treatment. At first week follow-up, just a small subfoveal defect was visible on the OCT (arrowhead, **F**), with complete resolution after 1 month (**I**).

the presence of macular retinal arteriovenous malformation in the RE, with dye leakage from the anastomotic vessel in FA (Figure 1). In detail, the anastomotic vessel filled up in the early arterial phase in FA (Figure 2), and the retinal macrovessel was completely filled during the early venous stage. In the LE, MRNF was associated with vascular abnormalities located along the superio-temporal vascular arcade (Figure 1). Wide-field FA showed the presence of small telangiectatic changes in the peripheral retina of both eyes, especially in the LE. Optical coherence tomography detected a cystoid macular edema in the RE, whereas no intraretinal cysts were found in the LE.

The patient was monitored and received no treatment. Seven days later, the best-corrected visual acuity was improved to 20/20 in the RE. Optical coherence tomography angiography (Spectralis OCTA; Heidelberg Engineering) was performed and clearly showed the retinal vascular alterations in both eyes (Figure 2). In detail, the large macrovessel was better identified in the superficial capillary plexus slab, whereas dilation and microvascular abnormalities were predominantly visualized in the intermediate capillary plexus and deep capillary plexus (Figure 2). The abnormal vascular anastomosis connected with the CRM was clearly visible with the OCTA (Figure 2).

In the LE, the OCTA showed a fine microvascular network of vessels located along the vascular arcade (Figure 2). Optical coherence tomography scan revealed the resolution of macular edema with a small residual subfoveal defect of the external retinal layers (Figure 3). At 1-month follow-up, the best-corrected visual acuity returned to 20/20 and the OCT showed a normal retinal profile (Figure 3).

Discussion

Congenital retinal macrovessels and MRNFs are rare and typically asymptomatic developmental anomalies.[1,10] They are generally isolated, but CRMs have been found to be occasionally associated with other retinal pathologies, including macroaneurysm,[12] branch retinal artery occlusion,[3] telangiectasia,[13] cavernous hemangioma,[14] serous retinal detachment,[2] and vitreous hemorrhage.[15] Retinal vascular abnormalities have also been described in patients with MRNF, ranging from mild telangiectasia to neovascularization.[11] We reported a case of CRM with vascular anastomosis complicated by cystoid macular edema associated with MRNF in the contralateral eye and peripheral vascular alterations, studied with OCTA.

Optical coherence tomography angiography is a recent imaging tool widely used for the study of retinal and choroidal vasculature. It provides noninvasive, high-resolution, depth-resolved images or retinal vessels without IV injection of dye, and, differently from FA, it allows to separately analyze the superficial and deep retinal vascular plexus.[6] In this case, OCTA allowed for an improved visualization of the anomalous vessels connecting the retinal arteries to venous macrovessel, monitoring their course through the retina and to locate the CRM in relation to the foveal avascular zone, which seemed uninvolved. Optical coherence tomography angiography also permitted to visualize the microvascular capillary abnormalities present around the venous macrovessels in the RE and above the retinal surface in the area with myelinated fibers in the LE.

Congenital retinal macrovessels mostly affect visual acuity because of the development of macular edema and retinal ischemia.[2,12–15] The edema is generally secondary to a blood–retinal barrier alteration occurring in correspondence of the dilated and tortuous vessels, and it frequently resolves spontaneously, unlike the ischemic and neovascular complications typically leading to hemorrhages or persistent damage to retinal tissues. In our case, the cystoid macular edema was due to the presence of an abnormal vascular complex connecting the CRM to the normal retinal vasculature and the intraretinal cysts resolved without any treatment. Optical coherence tomography angiography may be useful in these patients for early identification of small areas of capillary dropout and retinal ischemia and should be performed at baseline and during each follow-up.

Historically, CRMs have never been associated with systemic vascular abnormalities; however, a recent study reported a greater prevalence of venous malformations of the brain in patients with retinal venous macrovessels studied with cerebral magnetic resonance (24% vs. 0.4–6% in normal population).[9] We performed a systemic workup with brain magnetic resonance imaging with contrast in our patient that excluded the presence of venous malformation of the brain.

Conclusions

To the best of our knowledge, this is the first report describing two rare congenital development anomalies, CRM and MRNF, both present in the same patient. Moreover, we firstly described a case of AV malformation with an anomalous retinal anastomosis visible on OCTA. In patients with these conditions, OCTA should be used for classification of these retinal vascular anomalies and early detection of microvascular complications.

Key words: congenital retinal macrovessel, myelinated retinal nerve fibers, retinal arteriovenous malformation, retinal edema, fluorescein angiography, OCT, OCTA.

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