



Congenital Cyanogenic Cardiac Malformation Complicated with Branch Retinal Artery Occlusion: A Clinicopathological Case Report

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ABSTRACT

Background Branch retinal artery occlusions (BRAO) typically result in sudden, painless, mostly unilateral loss of vision presenting in the 6th to 8th decade and may have various presentations. Only a few major case series have been reported in patients younger than 40 years. Our paper describes one case of BRAO due to a congenital cyanogenic cardiac malformation in a young male patient.

Case presentation: we present the case of a 25-year old male patient which presented at the ophthalmology emergency unit, complaining of sudden vision loss in the inferior visual field of his left eye. A comprehensive ocular and general health investigations performed for the diagnosis and follow-up of the patient are presented. The ophthalmological examination revealed a superior BRAO in the left eye associated with an inferior altitudinal hemianopia. His physical examination revealed severe cyanosis, hippocratic fingers, enlarged jugular neck veins as well as hepatomegaly. The extensive cardiovascular investigations revealed a complex cardiac malformation associated to a tricuspid atresia, a functional single ventricle, large interatrial and interventricular septal defects, d-transposition of the great arteries and valvular pulmonary stenosis. The laboratory investigations revealed marked erythrocytosis.

Conclusion BRAO in young adults occur via multiple mechanisms, the secondary erythrocytosis due to congenital cyanogenic cardiac malformation being the cause found in our patient.

KEYWORDS

branch retinal artery occlusion, cyanogenic cardiac malformation, erythrocytosis.

BACKGROUND

The central retinal artery (CRA), a branch of the ophthalmic artery (OA), enters the eye through the optic disc and divides into a superior and an inferior retinal artery branch, which further subdivides to supply retinal vessels to perfuse the inner layers of the retina. The branch retinal artery occlusion (BRAO)

occurs when one of these branches becomes occluded. BRAO present abrupt, painless, severe loss of vision in the visual field corresponding to the territory of the obstructed artery. Overall, more men than women are affected, and the mean age at onset is about 60 years, with a range of reported ages from the first to the ninth decade of life [1-3]. Hayreh studies have

shown not only that central retinal artery occlusion (CRAO) and BRAO have different pathogeneses, clinical characteristics and management but also that BRAO is not one clinical entity. For example, cilioretinal artery occlusion (CLRAO) has usually been included in BRAO; but CLRAO is a distinct clinical entity because the cilioretinal artery arises from the posterior ciliary artery (PCR), instead of the CRA [2,3].

The BRAOs are associated with systemic diseases such as hypertension, atherosclerosis affecting internal carotid arteries (ICAs), and heart diseases; additional risk factors include diabetes mellitus, cigarette smoking, systemic clotting disorders (thrombophilias, etc), or vasculitis (giant-cell arteritis-GCA, etc) [1-3]. The first report of a CRAO belongs to the German Ophthalmologist Albrecht von Graefe in 1859, who described CRAO in a patient with endocarditis and multiple systemic emboli [4]. Information regarding risk factors in young adults remains scarce, and reports of young adults presenting RAO are rare [1-3,5,6,7]. The aim of our case-report is to highlight this rare pathology in young patients and describes the multitude of investigations needed to reach the correct diagnosis and to follow-up these patients.

CASE PRESENTATION

A 25-year old male patient presented at the ophthalmology emergency unit complaining of sudden vision deterioration (20/40) in the inferior sector in his left eye since 2 days. The patient was born with a cardiac murmur and intense cyanosis and diagnosed with a complex cardiac malformation associated with a tricuspid atresia, a functional single ventricle, large interatrial and interventricular septal defects, d-transposition of the great arteries and valvular pulmonary stenosis. A modified Blalock-Taussig shunt was performed with a 6mm ePTFE prosthesis at two years of age. He subsequently had an inconsistent follow-up for two decades, during which time no Fontan-type procedure to address his single ventricle morphology was proposed to him. He was suggested to be listed for a cardiac transplant which the patient refused. The initial ophthalmoscopy revealed a superior BRAO in the left eye, presenting a grayish whitening of the retina surrounding the occluded artery and a sluggish blood flow; an increased retinal vascular tortuosity was also observed (Fig.1A). The fundus Fluorescein angiography (FFA) showed a marked delay in the arm-to-retina circulation time (ARCT) of 5 seconds, having a ARCT of 17 seconds on both eyes and delayed choroidal filling. Regarding the retinal circulation time (RCT), a supplementary delay of 2 seconds was observed at the superior branch of the retinal artery at the left eye and hypofluorescence in the surrounding superior area (Fig.1B). The Optical Coherence Tomography (OCT) revealed increased thickness and hyper-reflectivity of the inner retinal layers in the superior retina, suggesting the presence of intracellular edema, and decreased reflectivity of photoreceptor and retinal pigment epithelial layers; asymmetry of optical reflectivity in the superior and inferior perifoveal regions (Fig.1 C,D). The visual field examination revealed an inferior altitudinal hemianopia at the left eye (Fig.2A). B-scan ultrasound evaluation was normal in both eyes. CDI of retrobulbar (orbital) vessels was performed with a 10 MHz linear probe for detecting and measuring orbital vessel blood flow in the OAs, the CRAs, the superior ophthalmic veins, and the PCAs (nasal and temporal). It revealed normal hemodynamic parameters, with normal orbital vessel blood flows and resistance index in all retrobulbar vessels. His physical examination revealed severe cyanosis, hippocratic fingers, enlarged jugular neck veins as well as hepatomegaly. The chest X-ray showed an enlarged cardiac silhouette, an upward tilt of the apex, a rightward deviation of the trachea as well as venous stasis in the pulmonary fields. The electrocardiogram revealed a leftward deviation of the axis, severe hypertrophy of the left ventricle and an incomplete left bundle branch block. An assessment of extracranial arteries (which was realised with a 7.5-10 MHz linear array transducer, combining B mode and Color Doppler/pulsed-wave Doppler ultrasound-Extracranial Duplex Sonography: EDS) excluded an eventual ipsilateral ICA source of emboli. Laboratory workup excluded systemic or local vasculitis (systemic lupus erythematosus, GCA, etc), or

coagulopathies (like congenital thrombophilias), and showed severe secondary polyglobulia, with markedly increased hematocrit and hemoglobin values (73.2% and 23.1 g/dl, respectively), increased creatinine (1.6 mg/dl) and INR (1.7). A cardiac MRI examination was performed, where a hypoplastic right ventricle, malposition of the great vessels, multiple ventricular septal defects and an occluded modified Blalock-Taussig shunt were apparent (Fig.2B). The fundus photography performed 4 weeks later revealed the almost complete resolution of the ischaemic edema from the affected superior retina (Fig.2C). In this month, the patient's intraocular pressure was medically decreased with eyedrops (ocular antihypertensive medication) in order to allow a better retinal arterial perfusion. The visual acuity and the abnormal visual field defect did not improve.

DISCUSSION

According to Hayreh [2], a definite diagnosis of BRAO in our male patient was based on the following findings: (1) There was a history of sudden onset of visual deterioration in the eye. (2) On initial ophthalmic evaluation, there was evidence of acute retinal ischemia in the distribution of the occluded branch retinal artery. (3) Fluorescein fundus angiography, performed soon after the onset, showed evidence of absence or marked stasis of circulation in the involved branch retinal arterial. In the present case of BRAO, one of the advantages about the documentation of these findings by OCT was the possibility of comparison of the asymmetry in the same OCT cross-section of macular area, showing both the area affected by the branch occlusion and the normal area, as previously described in the literature [4]. BRAO are either thrombotic or embolic in nature [1-3]. Embolic disease is the most common etiology of a BRAO in elderly patients; in a study of 70 patients with retinal emboli, 40 were found to have cholesterol emboli, 8 platelet-fibrin emboli, 6 calcific emboli and 1 possible myxomatous embolus [8]. In younger patients, athromatous disease of carotid origin is a rare cause of BRAO [1-3] and can be identified by EDS, which was normal in our patient. In this age group, especially those patients who have multiple or recurrent BRAO, other etiologies are more likely; associations have been noted with coagulopathies (including congenital thrombophilias, and/or antiphospholipid antibodies), systemic vasculitis or infections, local trauma (including retrobulbar injection), local ocular conditions (optic nerve drusen) or miscellaneous conditions (migraines, oral contraceptives, pregnancy) [1-3]. Frequently, in CRAO the blockage is located within the optic nerve substance and for this reason, it is generally not visible on the ophthalmoscopy [1-3]. According to Hayreh, no visible embolus in the retinal artery does not exclude CRAO caused by an embolus [2,3]. In fact, our patient had no visible embolus at ophthalmoscopy and B-scan ultrasound evaluation, and, on the other hand, CDI of retrobulbar vessels was normal in both eyes in his case, excluding GCA [9]. The BRAO in our patient can be explained by the secondary erythrocytosis due to the congenital Cyanogenic heart disease (CHD). Other authors also described acute cerebral ischemia due to CHD in a 29-year old woman, with secondary polyglobulia, successfully treated with heparin [10]. Also, a case of thrombosis of the central retinal vein in pulmonary fibrosis with and subsequent secondary polyglobulia has previously been described in the literature [11]. Cardiac causes for the occurrence of BRAO have been described; in a study regarding retinal arterial occlusions in the young, cardiac abnormalities were found in 18.7% of patients; among them mitral regurgitation was found in 15.6% and mitral valve prolapse in 9.4% patients [7]. Rheumatic heart disease (RHD) has also been described as an etiological factor for retinal vascular occlusions [12]. The ischemia of the inner layers of the retina leads to intracellular (retinal ganglion cells) edema as a result of cellular injury and necrosis. It is an anoxic retinal edema which develops in the acute stage of the retinal artery occlusion, due to the sudden retinal ischemia; in many cases it is followed by a regressive phase, giving the way, to retinal atrophy. This intracellular edema has the ophthalmoscopic appearance of grayish whitening of the superficial retina, like observed in our case; the whitening of the retina generally lasts 4-6 weeks, as in our case [1-3]. Also, the increased

retinal vascular tortuosity appears to be prevalent in adults with cyanotic congenital heart disease and is likely to be in response to hypoxemia and erythrocytosis [13]. Because prognosis for BRAO is good, no interventions usually are taken [1-3]. Intraarterial thrombolysis with recombinant tissue-type plasminogen activator (rt-PA) via a guiding catheter inserted into the femoral artery, placed into the ICA, and advanced into the OA has been used for CRAO with variable success. This procedure has also been applied, with limited benefit compared to conventional forms of therapy or observations, to patients with BRAO [1-3]. Also, a paracentesis with a 30G needle can be used to remove 0.1 to 0.3 ml of aqueous fluid within the first 24 hours; the paracentesis lowers the intraocular pressure and may allow the embolus (if any) to move further down the vessel and away from the central retina, limiting the extent of damaged retina [1]. In addition, the intraocular pressure may be medically decreased with eyedrops, what we did in our patient [1-3]. Because the polycythemia is in reaction to hypoxia (reactive polycythemia), we considered that any therapeutic approach to correct it would be unsuccessful [1-3]. Regarding the shunt obstruction, there isn't any medical or surgical procedure for desobstruction; the only solution would be the creation of a new shunt and, as this solution is linked to an increased vital risk [1], in our case was refused by our patient.

CONCLUSIONS

BRAOs in young adults occur via multiple mechanisms, the secondary erythrocytosis due to congenital cyanogenic cardiac malformation being the cause found in our patient.

CONFLICT OF INTEREST

The authors declare no conflict of interest.

FIGURE LEGENDS

Figure 1

(A) Left eye fundus photography: superior branch retinal artery occlusion (BRAO); grayish whitening of the retina surrounding the occluded artery; sluggish bloodflow; increased retinal vascular tortuosity.

(B) Left eye fluorescein angiogram (FFA): delayed choroidal filling, delayed filling of the affected superior retinal artery by the fluorescein, hypofluorescence in the surrounding superior area.

(C) Left eye OCT: cross-section through superior retina, increased thickness and hyper-reflectivity of the inner retinal layers denoting the presence of intracellular edema and decreased reflectivity of photoreceptor and retinal pigment epi-

thelial layers.

(D) Left eye OCT: asymmetry of optical reflectivity in the superior and inferior perifoveal regions.

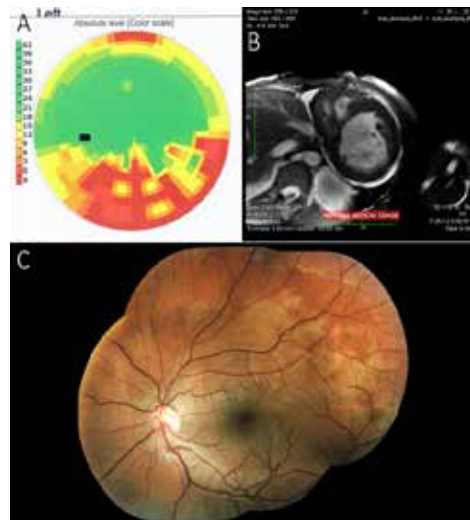
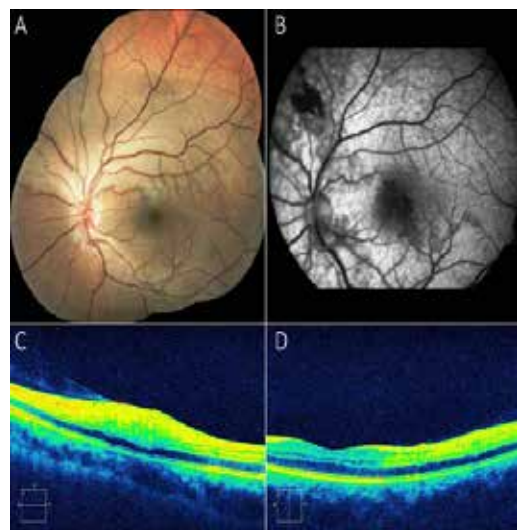


Figure 2

(A) Visual field examination of the left eye: inferior altitudinal hemianopia.

(B) Cardiac MRI examination: hypoplastic right ventricle, malposition of the great vessels, multiple ventricular septal defects, occluded modified Blalock-Taussig shunt.

(C) Fundus photography of the left eye after 4 weeks: disappearance of the ischemic edema from the superior retinal sector.



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