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RETINAL BRANCH VEIN OCCLUSION BY ARTERIAL WALL PLAQUE IN HEREDITARY AMYLOIDOSIS

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SUMMARY – We report on ocular finding that was the first sign of hereditary amyloidosis. This case report describes a 34-year-old man with a sudden loss of visual acuity, positive family history, and systemic manifestations of amyloidosis that included peripheral neuropathy, restrictive cardiomyopathy and amyloid deposits detected in colon biopsy specimen. The main outcome measures were visual acuity, fundus photography and fluorescein angiography (FA). In the early stage of hereditary amyloidosis, fundus photography of the right eye demonstrated subretinal macular hemorrhage, retinal hemorrhages and staining in the lower temporal branch wall of the central retinal artery. FA showed vascular occlusion and focal staining of the artery, and in the late-stage fluorescein leakage of the inframacular branch vein. The site of "leakage" was treated with laser photocoagulation. After 3 weeks fundus photography showed that subretinal and retinal hemorrhages were mainly reabsorbed, but focal staining of the lower temporal artery wall was still present, and new focal staining of the upper temporal artery wall appeared. FA showed two vascular occlusions, with focal staining of arterial walls, and in late stage the sites of hyperfluorescence revealed that these vascular changes continued to progress. Focal retinal arterial amyloid deposits may be a rare ocular finding of hereditary amyloidosis, causing clinical finding of vein branch occlusion.

Key words: *Retinal vein occlusion; Arterial wall deposits; Amyloidosis*

Introduction

Amyloidosis is a clinical disorder caused by extracellular deposition of insoluble abnormal fibrils, derived from aggregation of misfolded, normally soluble protein, most commonly located in the viscera, blood vessel walls and connective tissue. This rarity and the variable involvement of different organs and tissues are often responsible for missed or delayed diagnosis[1].

Amyloidosis may infiltrate any part of the orbit or the eye[2]. Ocular amyloidosis may be a component of systemic diseases involving multiple organ systems, or may occur as localized forms involving specific structures of the eye[3].

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Case Report

This report describes a 34-year-old man with positive family history and uncertain systemic manifestation of amyloidosis. His medical history was notable for peripheral neuropathy (vertiginous syndrome) and restrictive cardiomypathy resulting in pericardial effusions. Colon biopsy showed positive amyloid deposits when stained with Congo red.

Results

The patient had sudden loss of visual acuity on the right eye, which was 0.1, and ocular examination was normal except for mid-retina. Fundus photography demonstrated subretinal macular hemorrhage, retinal hemorrhages and focal staining in the lower temporal branch wall of the central retinal artery (Fig. 1a). Fluorescein angiography (FA) showed hypofluorescence at the site

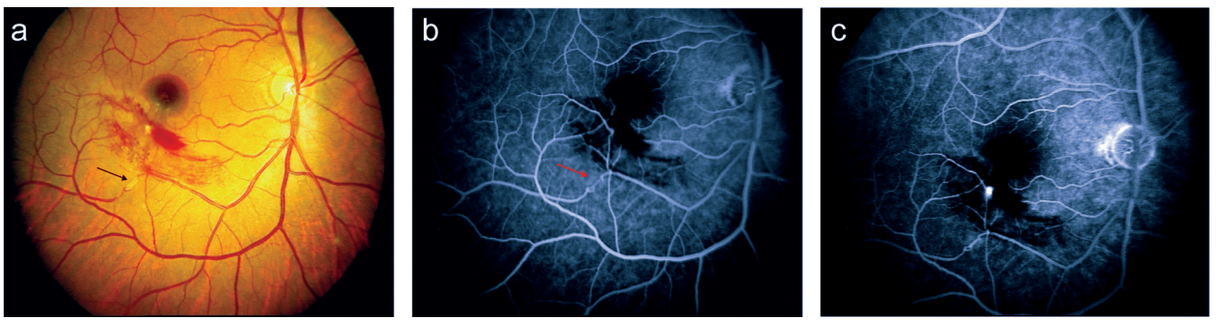

Fig. 1. Fundus photography and fluorescein angiography at first examination: (a) color fundus photography demonstrated subretinal macular hemorrhage, retinal hemorrhages and focal deposits (arrow) in the lower temporal branch wall of the central retinal artery. Early stage fluorescein angiography showed hypofluorescence corresponding to subretinal and retinal hemorrhages, vascular occlusion and focal staining in a part of lower temporal artery, vein "loop" at AV junction (b); and fluorescein leakage (arrow) of inframacular branch vein in late stage (c).

of subretinal and retinal hemorrhages, vascular occlusion and focal staining in the part of lower temporal artery, vein "loop" at AV junction (Fig. 1b), and in late-stage FA fluorescein leakage of inframacular branch vein (Fig. 1c). The site of "leakage" was treated with laser photocoagulation. On follow up examination 3 weeks later, fundus photography showed mostly reabsorbed subretinal and retinal hemorrhages, laser photocoagulation scars, amyloid deposit in lower temporal artery wall was still present, with a new focal amyloid deposit of upper temporal artery wall (Fig. 2a), indicating that these vascular changes continued to progress. His visual acuity was 0.8. Macular OCT was normal. Control FA showed no clinically significant vein obstruction or leakage (Fig. 2b).

Discussion

The most likely diagnosis is familial amyloidosis, which is associated with a nucleotide substitution in the gene for the prealbumin (transthyretin) molecule, resulting in the amino acid substitution of methionine for valine at position 30[2]. The retina may show evidence of vascular occlusion as well as focal perivascular sheath-

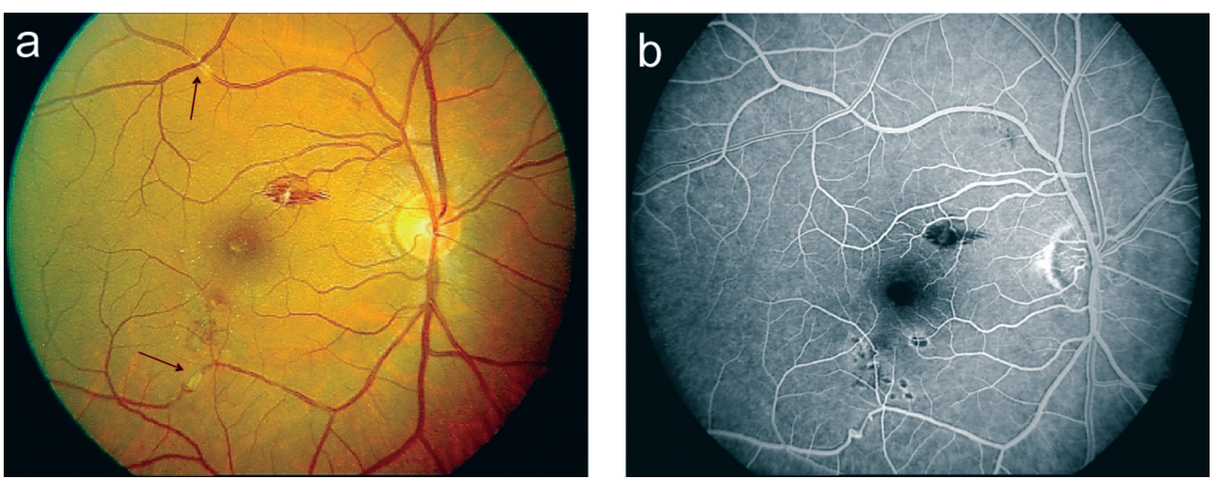

Fig. 2. Follow up at 3 weeks: (a) color fundus photography showed mostly reabsorbed subretinal and retinal hemorrhages, laser photocoagulation scars, and amyloid deposits of old lower temporal artery wall deposits (lower arrow) and a new focal amyloid deposit in the upper temporal artery wall (upper arrow); (b) fluorescein angiography.

ing of arteries and veins[4]. Focal arteriolar sheathing has been noted as the initial ocular finding in systemic amyloidosis[5]. Hereditary amyloidosis may induce ocular amyloid angiopathy. Severe and progressive retinal and choroidal vascular lesions, which caused visual disturbances, were seen in these patients[6]. This case report shows that focal retinal arterial amyloid plaques may be a rare and early ocular finding in hereditary amyloidosis, and can cause clinical finding of branch vein occlusion.

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Sažetak

OKLUZIJA OGRANKA RETINALNE VENE UZROKOVANA AMILOIDNOM NAKUPINOM U ARTERIJSKOJ STIJENCI KOD NASLJEDNE AMILOIDOZE

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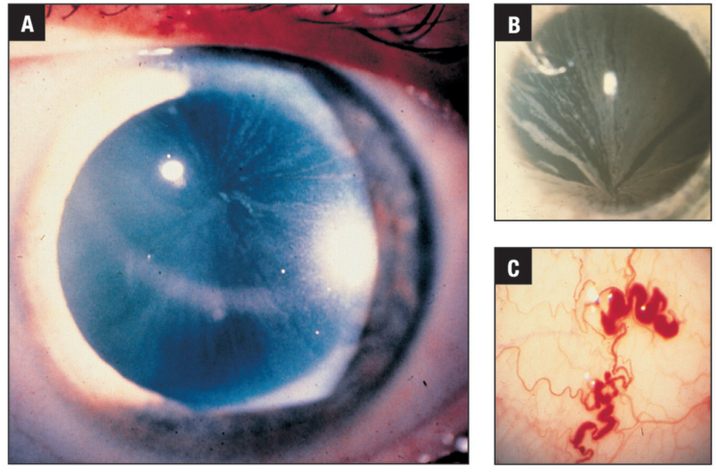
Opisuje se očni nalaz kao prvi znak nasljedne amiloidoze. Opisuje se slučaj 34-godišnjeg muškarca s iznenadnim gubitkom vidne oštrine, pozitivnom obiteljskom anamnezom i sistemskim manifestacijama amiloidoze uključujući perifenu neuropatiju, restriktivnu kardiomiopatiju i nakupine amiloida u uzorku dobivenom biopsijom kolona. Glavne mjere ishoda bile su vidna oštrina, fotografija fundusa i fluoresceinska angiografija (FA). U ranom stadiju nasljedne amiloidoze fotografija fundusa desnoga oka pokazala je subretinalno makularno krvarenje, retinalna krvarenja i bojenje u stijenci donjeg temporalnog ogranka središnje retinalne arterije. FA je pokazala vaskularnu okluziju i žarišno bojenje arterije, a u kasnijem stadiju istjecanje fluoresceina u inframakularnom venskom ogranku. Mjesto "istjecanja" liječeno je laserskom fotokoagulacijom. Fotografija fundusa provedena nakon 3 tjedna pokazala je da su se subretinalna i retinalna krvarenja uglavnom iznova apsorbirala, ali je žarišno bojenje donje temporalne arterije još uvijek bilo prisutno, dok se na gornjoj temporalnoj arteriji pojavilo novo žarišno bojenje. FA je pokazala dvije vaskularne okluzije uz žarišno bojenje arterijskih stijenka, dok su u kasnijem stadiju mjesta hiperfluorescencije ukazala na daljnje napredovanje ovih vaskularnih promjena. Žarišne nakupine amiloida u retinalnoj arteriji mogle bi biti rijedak očni nalaz kod nasljedne amiloidoze, koji uzrokuje klinički nalaz okluzije venskih ogranaka.

Ključne riječi: *Okluzija retinalne vene; Nakupine arterijske stijenke; Amiloidoza*

Ukoliko primjetite kod bolesnika vrtložne opacitete na rožnici

Možda se radi o Fabryjevoj bolesti



Fotografije A i B: Karakteristični opaciteti na rožnici (cornea vericillata ili vrtložasta keratopatija). Obratite pažnju na crvolike tračke koji polaze iz zajedničkog ishodišta poput žica na kotaču bicikla. Fotografija B je dobivena ljubaznošću dr. RL Abott. **Fotografija C:** Konjunktivalne promjene kod Fabryjeve bolesti. Obratite pažnju na značajno proširene, kobasičaste krvne žile. Ovi znakovi Fabryjeve bolesti u pravilu ne ometaju vid bolesnika.

Specijalisti za očne bolesti imaju priliku otkriti bolesnika sa ovom progresivnom bolesti koja često ugrožava život.

Rana dijagnoza i intervencija su ključni
– specijalista za očne bolesti može imati važnu ulogu

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