

## Bilateral retinal venous occlusion in Fabry disease

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Fabry disease is an X-linked lysosomal storage disorder. Due to a deficiency of alpha galactosidase A (alpha Gal A) there is a deposition of sphingo lipids within the vascular endothelium. This is associated with vascular dysfunction, alteration in vascular perfusion and a prothrombotic state [1]. Here we report of a 21 year-old male patient with acute or chronic renal failure. Kidney biopsy revealed storage material consistent with Fabry disease. Diagnosis was confirmed by deficient alpha-Gal A-activity. Genetic test revealed a previously undescribed homozygous mutation (Exon 5: c.689\_697del9pb (p.Ala230\_Ile232del) in the GLA gene. The patient started hemodialysis as well as enzyme replacement therapy with a galsidase beta (Fabrazyme; Genzyme). Two months later the patient was admitted to hospital due to an acute myocardial insufficiency with pericardial effusion. Additionally at admission to the hospital the patient reported of a predominantly left-sided visual impairment.

At ophthalmic exam, best-corrected visual acuity at 1 meter was 20/63 in the right eye and 1/40 in the left eye. The anterior eye segment examination revealed bilateral conjunctival vessel tortuosity in all four quadrants as well as a bilateral grade 1 cornea verticillata. A clear lens status and an otherwise quite anterior chamber were recorded. Fundus examination exhibited a bilateral optic disc edema with pararetinal flame-shaped retinal hemorrhages, peripheral cotton wool spots and a macular edema with ring-shaped extra foveal hard exudates on the left side. (Figure 1) An Optical Coherence Tomography (OCT) examination (Heidelberg Spectral is; Heidelberg Engineering) exposed a left-sided cystoid macular edema (Figure 2) and a right-sided regular foveal impression (not shown) besides bilateral parapapillary subretinal fluid accumulation. We were not able to perform a fundus angiography due to the terminal renal failure and the necessity to perform daily hemodialysis as well as the reduced general health of the patient.

We proposed the diagnosis of a right-sided partial and a left side complete Central Retinal Venous Occlusion (CRVO) and started a monthly intravitreal therapy regime with Bevacizumab (Avastin; Roche) in the left eye.

Ocular manifestations of Fabry disease are cornea verticillata, a whorl-like corneal pattern, cataract, as well as conjunctival and retinal vessel tortuosity. Retinal vascular tortuosity is as

present in approximately 20% of patients with Fabry disease and correlates well with disease severity [2, 3]. Ocular complications have rarely been described. Single case reports on retinal arterial occlusion in one patient as well as a case of retinal venous occlusion have been published so far [4, 5].

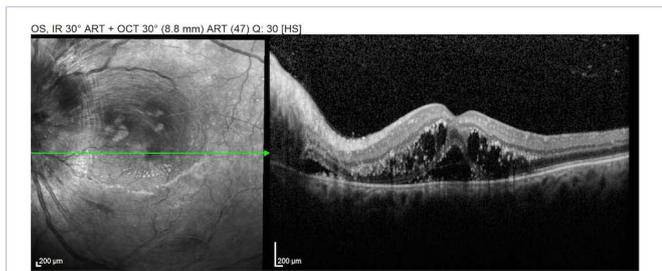
An increased risk for systemic thromboembolic complications including an increased stroke risk have been described in Fabry disease [6]. Nevertheless, ocular thromboembolic events are rather uncommon, but a sight-threatening complication. This case of a bilateral CRVO in young men with Fabry disease is most likely due to Fabry specific retinal vascular abnormalities and highlights Fabry disease as a differential diagnosis of juvenile and presenile CRVO.



**Figure 1:** Fundus photography (left eye) - Optic disc edema with pararetinal flame-shaped retinal hemorrhages extending along the vascular arches and parafoveal hard exudates.



**Figure 2:** Fundus photography (right eye) - Optic disc edema with vessel narrowing and superior parapapillary flame-shaped haemorrhages.



**Figure 3:** OCT imaging (left eye) - Intraretinal cystoid macular edema along with subfoveal fluid accumulation.

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