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Case Report

Coats-like Response in Facioscapulohumeral Muscular Dystrophy

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Abstract

Background: Facioscapulohumeral muscular dystrophy (FSHD) is an autosomal dominant disease with facial muscles weakness and scapular weakness. Ophthalmic manifestations may include retinal vasculopathy similar to Coats' disease. We report a case of Coats-like response in FSHD which has remained stable with no exudation.

Case Presentation: A 7-year old female with tortuous posterior pole vasculature and Coats-like response associated with FSHD. Fundus angiography shows avascularity of peripheral temporal retina with telangiectatic vessels and aneurysmal dilatations of vessels at the edge of avascular retina without neovascularization or exudates.

Discussion and Conclusion: This case demonstrates a Coats-like response in a patient with FSHD that has remained stable and has not required any treatment. Other reports have demonstrated peripheral/macular exudative retinopathy, peripheral neovascularization and neovascular glaucoma.

Keywords: Coats' Disease; Facioscapulohumeral Muscular Dystrophy

Abbreviation

FSHD: Facioscapulohumeral Muscular Dystrophy

Background

Facioscapulohumeral muscular dystrophy (FSHD) is an autosomal dominant disease that presents before the age of 20 with progressive weakness of the facial muscles, stabilizer of the scapula or dorsiflexors of the foot. Diagnostics include a normal creatine kinase level, mild myopathic changes on electromyography, and nonspecific chronic inflammatory changes on muscle biopsy. Genetic testing shows a deletion in 4q35 [1]. Other non-muscular manifestations include retinal vasculopathy, sensorineural hearing loss, and/or atrial tachyarrhythmias [1,2].

Few reports have characterized the retinal clinical features of FSHD including: telangiectasia, microaneurysms, exudative retinopathy, neovascular glaucoma, and peripheral retinal neovascularization [3,4].

This reports includes a case of Coats-like response associated with FSHD with fundus imaging and angiography; a review of the literature of ophthalmic manifestations associated with FSHD is described.

Case Presentation

A 7-years-old female with FSHD with bilateral facial palsy and progressive sensorineural hearing loss was referred for retinal evaluation. She was a full-term baby with no medical abnormality at birth. Her best corrected Snellen visual acuity was 20/25 in the right eye and 20/20- in the left eye. Extraocular movements were full, intraocular pressure was normal, and pupils were dilated pharmacologically. External examination revealed mild lagophthalmos in both eyes with weakness of the orbicularis oculi and without exposure keratopathy. Anterior segment examination was unremarkable in both eyes. Dilated fundus examination of both eyes revealed clear medias, with pink and sharp optic nerves. Posterior pole vessels looked slightly tortuous; temporal peripheral retinal examination revealed telangiectatic vessels without retinal edema, hard exudates, or retinal neovascularization. A small crescent area of avascular retina anterior to the telangiectatic vessels was noted (Figure 1a and 1b). Fundus angiography showed avascularity of peripheral temporal retina with telangiectatic vessels and aneurysmal dilatations of vessels at the edge of avascular retina without neovascularization or exudates (Figure 1c and 1d). Multiple follow up visits every 3

months over one year revealed stable fundus examination without active neovascularization, exudates, or retinal edema (Figure 2).

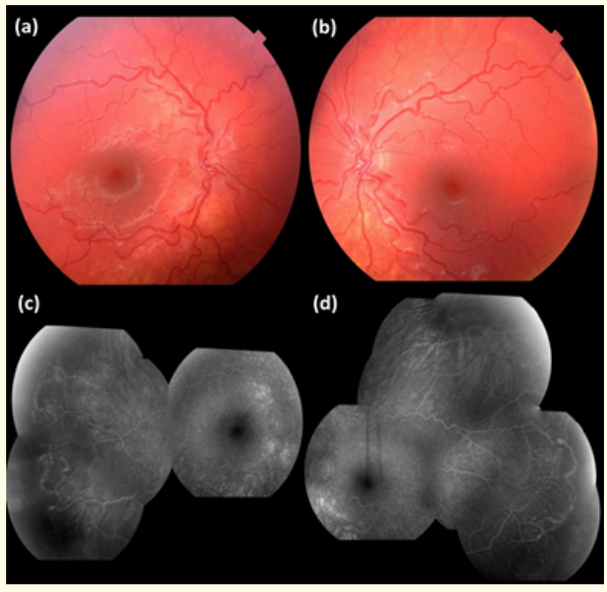

Figure 1: a-b: Fundus photos of both eyes; 1c-d: Fundus angiography collage of both eyes.

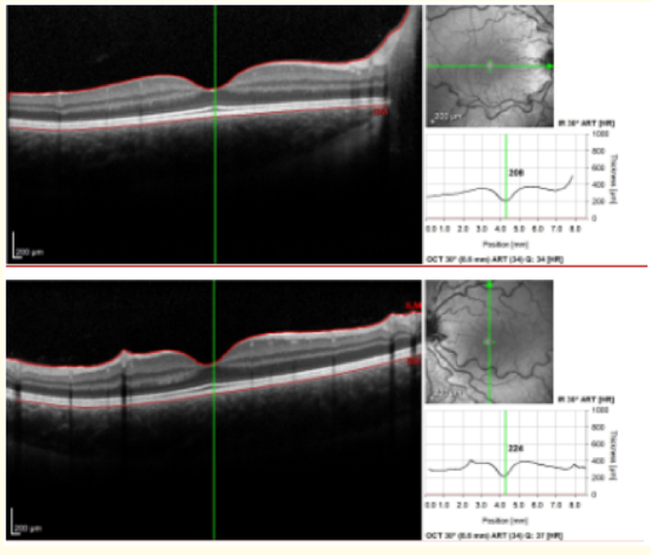

Figure 2: OCT Macula of Both eyes

Discussion and Conclusion

Facioscapulohumeral muscular dystrophy is an autosomal dominant inherited disorder with a prevalence of 1:20,000.The genetic defect involves a deletion in the repetitive element of 4q35 (D4Z4). Disease severity depends on the number of residual repeats (the less the residual repeats, the later the onset and severity of disease) [5,6].

In terms of ocular manifestations, there has been one report of three patients with atypical FSHD with chronic progressive external ophthalmoplegia, presenting with ptosis and ocular movement disorder. These patients had muscle biopsy to rule out ragged red fibers on histopathology and genetic workup to exclude coincidental mitochondrial DNA deletions [7]. In our case, we did not find any extraocular movement abnormalities. However, we did find the patient to have mild lagophthalmos with decreased orbicularis oculi strength without exposure keratopathy. FSHD patients should be carefully monitor for any corneal exposure.

Previous known retinal presentations include telangiectasia, microaneurysms and peripheral neovascularization with exudates [1,4]. Vascular changes in FSHD is hypothesized to be related to abnormal signaling pathway required during retinal angiogenesis [8]. Retinal vascularization occurs from disc to peripheral retina with temporal retina last to fully vascularize; thus, an insult in the signaling of vascularization leads to mostly temporal non-vascularization [8]. In our case, we did find temporal avascular retina. We also found tortuosity of retinal arteries and veins in the posterior pole. Longmuir., et al. have reported retinal arterial tortuosity with FSHD hypothesizing that this disease may affect the connective tissue and smooth muscles in the arterial vessel walls [9]. The cause of tortuosity has been hypothesized to be VEGF induced remodeling of vessels that occurs with increased blood flow in the premature retina causing vessel dilation and tortuosity, similar to the mechanism described for retinopathy of prematurity [10].

In our case, we did not detect any peripheral neovascularization. In terms of management, eyes should be routinely monitored for neovascularization and if detected treated with peripheral sectoral photocoagulation. There has been one report of rapid development of neovascular glaucoma in FSHD with Coats' disease [3]. Our case demonstrates a mild form of retinal vasculopathy, which we categorized Coats-like response; both eyes have vessel tortuosity, avascular peripheral retina, aneurysmal dilatation and microaneurysms.

Despite reports of these some retinal findings in FSHD, ophthalmic findings are not indicative or a clinical diagnosis criteria for FSHD [1]. Furthermore, Fitzsimons., *et al.* demonstrated that there is no correlation between severity of FSHD disease with severity or presence of retinal vascular abnormalities [11].

Declarations

- Ethics approval and consent to participate: Not applicable.
- Consent for publication: Consented obtained.
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