Neovascular Glaucoma From Advanced Coats Disease as the Initial Manifestation of Facioscapulohumeral Dystrophy in a 2-Year-Old Child

Facioscapulohumeral dystrophy (FSHD) is an autosomal dominant muscular dystrophy estimated to affect 1 in 20,000 white persons. The clinical features of this condition range from minimally detectable myopathy to severe disability. There is a characteristic pattern of weakness that affects predominantly the face (facio) and shoulder (scapulohumeral) muscles and later descends inferiorly to the abdomen and the legs.1,2 Symptoms become manifest in the teen years to early adulthood and progress slowly. Recent studies have shown a characteristic deletion in the long arm of chromosome 4 (4q35),2,3 but the exact mechanism of this disease remains unknown.

Classic Coats disease is a congenital, idiopathic retinal telangiectasia that can progress to severe retinal exudation and detachment.4 Retinal telangiectasia compatible with Coats disease can be an extraocular manifestation of FSHD, but most affected patients have asymptomatic retinal telangiectasia found at ocular screening after diagnosis of FSHD.3 The ocular findings rarely progress to advanced Coats disease.3 We describe a young child who had advanced eye findings of unilateral neovascular glaucoma from bilateral retinal telangiectasia 3 years before FSHD became apparent.

Report of a Case. A 23-month-old healthy girl had sudden onset of redness and pain in her right eye and was found to have leukokoria and neovascular glaucoma. At examination, the right eye could not fix or follow, and the left eye could fix and follow small objects. Intraocular pressure was 42 mm Hg OD and 18 mm Hg OS, and oral acetazolamide therapy was started. The right eye had diffuse neovascularization of the iris. Ophthalmoscopy showed total retinal detachment, aneurysmal dilation of the retinal vessels, and subretinal exudation. The left eye showed shallow temporal retinal detachment with peripheral telangiectasia and mild retinal exudation (Figure 1). These findings were consistent with Coats disease in both eyes: stage 4 in the right eye and stage 3 in the left eye.4 Management included enucleation of the right eye and laser photocoagulation and cryotherapy of the telangiectasia in the left eye (Figure 2). Within 6 months, the left eye was stable without exudation and the fovea remained intact.

At age 26 months, the child developed myoclonic seizures and atypical absence seizures. Subsequently, generalized hypotonia with protruding abdomen (Figure 3), flat facial appearance, open drooling mouth, and protuberant tongue became apparent. At age 30 months, hearing loss and speech delay were confirmed and hearing aids were placed. Genetic and metabolic evaluation revealed no specific syn-
drome, normal female karyotype, and normal genomic microarray analysis at 230 studied loci. Histopathologic analysis of a muscle biopsy specimen revealed no abnormalities.

A repeat systemic evaluation was performed at age 5 years after tiptoe walking was noted in the patient. The spectrum of myopathic findings led to suspicion of FSHD. Genetic testing disclosed a deletion at 4q35, consistent with FSHD.

At pathologic analysis, the globe showed florid iris neovascularization, peripheral anterior synechiae, iris pigment epithelial ectropion, and total exudative retinal detachment. The retina was detached by densely proteinaceous subretinal fluid rich in cholesterol clefts and foamy lipid-laden histiocytes, and parts of the

Figure 2. Pathologic analysis of the enucleated right eye showed features consistent with Coats disease including retinal detachment with subretinal cholesterol and foamy macrophages. A, Gross pathologic analysis showed total retinal detachment. B, Histopathologic analysis showed subretinal cholesterol clefts and foamy macrophages (hematoxylin-eosin; original magnification ×250). C, Histopathologic analysis showed dilated retinal vessels without inflammation (hematoxylin-eosin; original magnification ×25). D, Histopathologic analysis showed foci of perivascular inflammation (hematoxylin-eosin; original magnification ×100).

Figure 3. Flaccid abdominal musculature is demonstrated.
retina were thickened by intraretinal exudates. Abnormal, ectatic retinal vessels were noted anteriorly. There was severe photoreceptor atrophy, consistent with chronic retinal detachment. Several small perivascular foci of lymphocytic infiltration were observed.

Comment. Facioscapulohumeral dystrophy is a slowly progressive muscular dystrophy that follows a descending course. This condition classically is manifested by muscle weakness, fatigue, or pain in the facial and scapular regions during the second to fourth decades of life; however, onset can occur at any age and infantile presentation is most severe. The facial muscles, particularly the orbicularis oculi and orbicularis oris, show weakness, often asymmetrically, leading to difficulty in smiling, whistling, and sucking. Shoulder girdle involvement results in a sloped, anteriorly rotated shoulder with concomitant posterior winging of the scapula. The biceps are more severely affected than the deltoid and distal forearm muscles. Abdominal wall muscles can show weakness and manifest abdominal protrusion while the patient is in the upright position. Later, the lower extremities display weakness with a typical footdrop and an unstable gait. Life span is not shortened, but 20% of patients are eventually wheelchair confined. Management is limited to supportive care. Extramuscular manifestations of FSHD include hearing loss and retinal findings compatible with Coats disease. High-frequency hearing loss occurs in approximately 50% of cases. The term retinal telangiectasia has been preferred to Coats disease because of the bilateral findings in FSHD compared with the unilateral involvement in classic Coats disease. In a review of 75 patients with FSHD in which 64 were studied with fluorescein angiography, 48 (75%) demonstrated peripheral retinal telangiectasia. Of the 75 patients, 3 (4%) showed posterior pole abnormalities and 1 (1%) had related visual acuity loss. More advanced retinopathy with total retinal detachment has been rarely documented, and our case is unique because of advanced neovascular glaucoma. Our patient showed bilateral retinal telangiectasia, suggestive of a systemic or hereditary condition. In a review of 150 consecutive cases of classic Coats disease, bilateral involvement was noted in 8 patients (5%) and no patients had FSHD.

Most patients are known to have FSHD when the retinopathy is discovered, and rarely does the inverse occur, as in our patient. Our patient had markedly advanced retinopathy and neovascular glaucoma years before the features of FSHD became apparent.

The histopathologic findings of FSHD include prominent perivascular inflammation in the muscles, endomyosial and perimysial inflammation, and small angular fibers suggestive of vascular insults. It was hypothesized that there might be a primary endothelial abnormality in both the muscular and retinal disease. In our patient, histopathologic findings were consistent with Coats disease with retinal detachment, subretinal cholesterol clefts, and foamy macrophages. Several small perivascular foci of chronic inflammation were identified, but the importance of this observation, if any, is uncertain. To our knowledge, there has been one other report describing an enucleated globe with FSHD, and the findings of exudative retinopathy were suggestive of Coats disease.

Facioscapulohumeral dystrophy is a slowly progressive muscular dystrophy associated with later high-frequency hearing loss and generally mild retinal telangiectasia. Our patient demonstrated advanced retinal telangiectasia producing total retinal detachment and neovascular glaucoma years before FSHD was apparent. The clinician should be aware of the relationship of these conditions, particularly in the atypical cases of bilateral Coats disease.

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