Graefe's Arch Clin Exp Ophthalmol
(2004) 242:265–267

SHORT COMMUNICATION

DOI 10.1007/s00417-003-0818-2

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Coats-like lesions in Usher syndrome type II

Received: 8 July 2003
Revised: 6 November 2003
Accepted: 7 November 2003
Published online: 3 December 2003

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Abstract *Background:* An unusual case of Usher syndrome type II associated with bilateral Coats-like exudative retinopathy is described. *Methods:* A 14-year-old boy with congenital sensorineural deafness and normal vestibular functions presented with a recent history of night blindness. He was followed for 3 years with fundus photography, intravenous fluorescein angiography, electroretinography and audiometric testings. His parents refused any form of treatment. *Results:* Fundoscopy showed bilateral retinitis pigmentosa and a single focus of subretinal exudation and overlying telangiectatic retinal vessels inferotemporal to the vascular arcade in the right eye. He had bilateral mild macular edema. A year later, a similar lesion developed inferotemporally in the left fundus. Electroretinography responses, particularly the rod-mediated signals, were significantly reduced. Audiometric studies documented hearing loss in high frequencies. His visual acuity declined from 20/40 to 20/80 RE and from 20/80 to 20/100 LE during follow-up. No new lesions developed. *Conclusions:* Coats-type exudative lesions may develop in patients with Usher syndrome type II. Although left untreated, only a minimal increase in exudation occurred over 3 years.

Introduction

Usher syndrome is the most common retinitis pigmentosa (RP) syndrome, representing between 6 and 10% of all RP patients [3]. Three types of Usher syndrome can be distinguished based on clinical findings [1, 9]. Usher type II syndrome, characterized by a progressive visual loss combined with a congenital sensorineural hearing deficit, normal vestibular functions and intelligible speech, is the most common form of Usher syndrome and may account for more than 50% of all Usher syndrome patients [1, 4]. Recent developments in molecular genetics now allow further classification of Usher syndrome into subtypes such as types IIa, IIb and IIc. In fact, type IIa may represent up to 85% of all Usher syndrome type II cases [11]. Coats-type exudation is a well-recognized complication of RP and can occur in up to 3.6% of patients [5]. Despite the relatively high frequency of Usher syndrome among RP patients and continuous elucidation of phenotype-genotype correlations, we were unable to find any case associated with Coats-type exudative retinopathy using MEDLINE search. However, a few unpublished cases were alluded to in a review article [5]. In this observational case report, we describe a patient with Usher syndrome type II complicated by bilateral, almost symmetric Coats-type lesions.

Case report

A 14-year-old boy presented with the complaint of night blindness that gradually had been deteriorating for the past 2 years. He was discovered to have sensori-neural deafness at the age of 3 years. The family history was non-contributory. On our ocular examination, his visual acuity was 20/40 RE and 20/80 LE. Anterior segments were normal with no evidence for any cataractous changes in the lenses. Fundus examination showed bilateral pale optic discs, mildly attenuated retinal vessels and mottling at the retinal pigment epithelial layer. There was no "bone spicule"-type pigmentation. There was a single area of subretinal exudation infero-

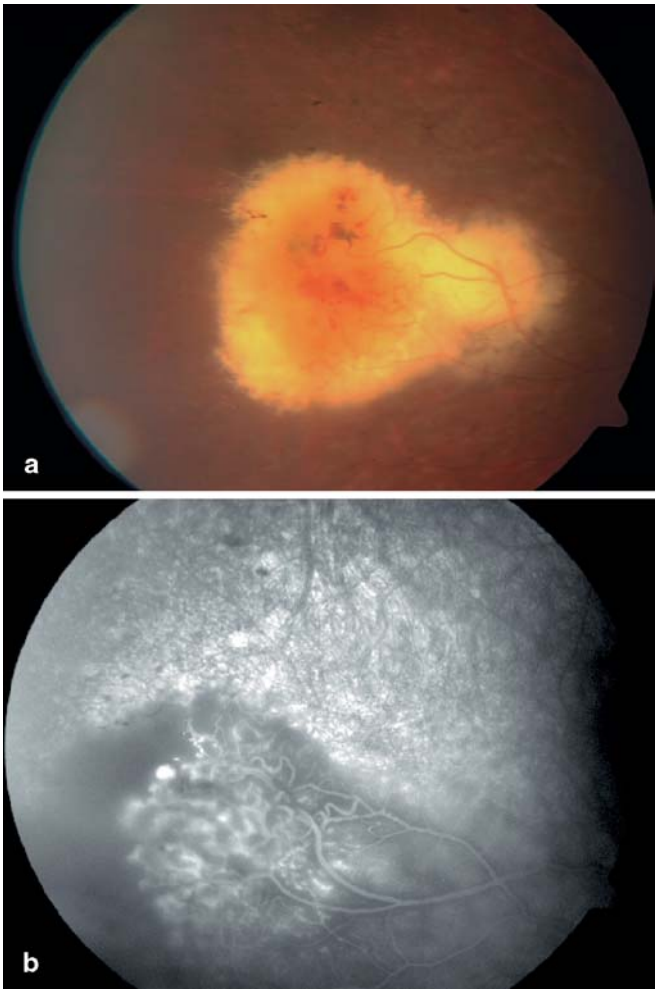

Fig. 1 **a** Right fundus view at initial presentation showing the well-delineated sub- and intraretinal exudation with overlying telangiectatic vessels approximately 7-disc diameters inferotemporal to the fovea. **b** Late venous phase fluorescein angiogram of the same area

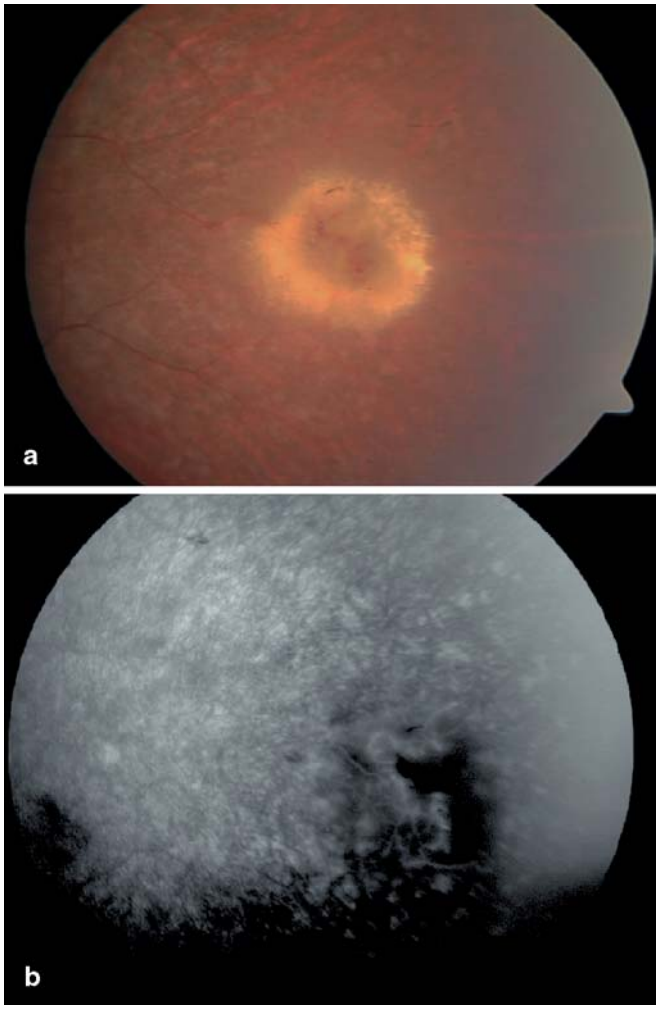

Fig. 2 **a** Fundus photograph of the left eye a year later showing the subsequently developed subretinal exudation surrounding retinal telangiectasias at the temporal and slightly inferior periphery. **b** Late venous phase fluorescein angiogram depicts the irregularly dilated retinal vessels involving a limited area

temporal to the vascular arcade in the right fundus (Fig. 1A), with accompanying 1+ cells in the vitreous. There was no similar lesion in the left eye. Early phase intravenous fluorescein angiography showed diffuse and irregular granular choroidal hyperfluorescence because of retinal pigment epithelial alterations. In the mid-venous and late phases, the previously fine granular hyperfluorescent spots became coarser. The macular areas appeared hypofluorescent, but as the study progressed, bilateral macular edema became evident. In the areas of exudation, there were irregular, tortuous and telangiectatic retinal vessels with leakage of the dye into the subretinal space and vitreous (Fig. 1B). Ganzfeld electroretinography was performed observing the revised standards proposed by the International Society for Clinical Electrophysiology of Vision (ISCEV) [7]. Rod responses in the fully dark adapted state and cone responses in light adapted eyes were reduced, but rod-mediated responses were more significantly affected. Goldmann visual field testing using V4e isopter was suggestive of bilateral midperipheral field loss. Ear, nose and throat examination showed normal tympanic membranes and middle ear structures. Audiometric tests revealed mild hearing loss in low frequencies and a profound loss in high frequencies. Neurologic examination demonstrated normal vestibular functions. The parents refused treatment in any form. A year later, a solitary area of retinal telangiectasia with exudation developed in the left inferotemporal fundus (Fig. 2) while the right eye remained stable. The patient was followed for 3 years without any treatment. At the final visit, his visual acuity was 20/80 RE and 20/100 LE. There was further decrease in electroretinographic responses. There was minimal increase in the area of lipid exudation in both eyes, but no new lesions developed.

Discussion

Usher syndrome, first described by Von Graefe [10] in 1858, is the most common genetic cause of deafness and blindness among school-age children. The signs and

symptoms of retinitis pigmentosa (RP) usually become fully expressed in early teens [9]. Vascular manifestations of RP include peripheral microaneurysms, retinal hemorrhages, telangiectasias, arteriolar narrowing and sheating, and abnormal circulation times [8]. Sometimes angiomatous proliferations of retinal capillaries may occur [2]. Coats-type changes are rare and are usually found in the inferior and/or temporal quadrants with typical dilated, aneurysmal or telangiectatic veins [5]. Although the age of diagnosis is double that of Coats disease, Coats-type changes were reported as early as age 4 years even before the signs of RP became evident [6]. Coats-type retinopathy may result from chronic microvascular leakage followed by secondary flat inferior retinal detachment with subsequent retinal hypoxia because of separation from the choroid and later development of telangiectatic abnormalities [8]. Current treatment options include cryotherapy, scleral buckling with subretinal fluid drainage and photocoagulation, the last being the least effective [5].

The mandatory observation as management in our patient demonstrated that the telangiectasias did not have an aggressive course leading to frank retinal detachment. In fact, previous observations support the sometimes self-limiting nature of the exudation [2]. This may be due to greater than normal adhesions between retinal pigment epithelia and sensory retina seen in RP [5]. We attribute the deterioration of the visual function to the progression of RP rather than the exudative process. This patient modestly enlarges the clinical spectrum of Usher syndrome type II and puts this disease into the list of RP syndromes associated with Coats-type changes including Leber's congenital amaurosis and Senior-Loken syndrome [5].

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