Bilateral Astrocytic Hamartoma of Peripapillary Retina with Optic Nerve Drusen in Leber's Congenital Amaurosis

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ABSTRACT

A 24-year old female patient presented to our clinic with bilateral visual impairment since birth. Best corrected visual acuity was light perception in both eyes. At fundus examination there were mulberry-like, yellowish-white nodules along the peripapillary margins. There was attenuation of retinal vasculature, bone-spicule-like pigment deposition in the general fundus, macular edema in the right eye and macular atrophy in the left eye. Fundus autofluorescence (FAF) imaging of the bilateral peripapillary retina and optic nerve head showed increased autofluorescence. It is important to clinically detect retinal astrocytic hamartoma (RAH). It may be with Leber's Congenital Amaurosis and optic nerve drusen.

Keywords: Astrocytic hamartoma, Leber's Congenital Amaurosis, Optic nerve drusen.

INTRODUCTION

RAH is a benign tumor. It originates from astrocytes in the nerve fiber layer at any location in the retina or from the optic nerve head. RAH can be unilateral, bilateral, solitary, multifocal, flat, elevated, opaque or transparent, mulberry-like lesion with intralesional calcification or without calcification.[1] Solitary and unilateral ones can be seen sporadically in normal individuals. Bilateral and multifocal lesions are often associated with tuberous sclerosis and neurofibromatosis.[2] Leber's congenital amaurosis is an autosomal recessive retinal dystrophy which causes severe visual impairment and macular atrophy beginning in infancy.[3] Optic nerve drusens are acellular deposits in the optic nerve head which are thought to develop from axonal metabolism products. Optic nerve drusens often coexist with retinal dystrophies.[4] In this case report, we present a patient with bilateral peripapillary RAH with optic nerve drusen in Leber's Congenital Amaurosis.

CASE PRESENTATION

A 24-year old female patient presented to our clinic with bilateral visual impairment since birth. Best corrected visual acuity was light perception in both eyes. She had bilateral nystagmus. The anterior segments and intraocular pressure of both eyes were normal. At fundus examination there were calcified, mulberry-like, yellowish-white and multilobulated nodules along the peripapillary margins in both of the eyes. There was attenuation of retinal vasculature, generalised atrophy of the retinal pigment epithelium, generalized bone-spicule-like pigment deposition in the fundus, macular edema in the rigth eye and macular atrophy in the left eye (Figure 1). FAF imaging of the both peripapillary retina and optic

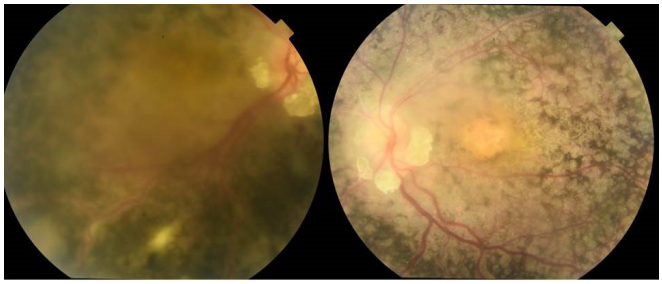

Figure 1: *Clinical photograph of the right and left eye at 24-year old female. Calcified, mulberry-like, yellowish-white, multilobulated and elevated noduler lesions along the peripapillary margins. Lesions are masking the retinal vessels and retinal pigment epithelium. There was attenuation of retinal vasculature, bone-spicule-like pigment deposition in the general fundus, macular edema in the rigth eye and macular atrophy in the left eye.*

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nerve head showed increased autofluorescence (Figure 2). Optical coherence tomography (OCT) imaging of the right and left peripapillary nodular lesions showed optically empty spaces in the lesion, thickening of the nerve fiber layer with posterior shadowing, cystic spaces and irregularities in the retinal inner layers (Figure 3 and figure 4). Combined with clinical history, ophthalmoscopic apperance and multimodal imaging of the lesions the patient was diagnosed as bilateral RAH of peripapillary retina and Leber's Congenital Amaurosis. The patient had no family history. Systemic diseases associated with RAH investigated but no systemic features of tuberous sclerosis and neurofibromatosis were found on physical examination. We followed the patient periodically with photodocumentation in our clinic since there is no effective treatment. Patient informed constent was obtained for case report.

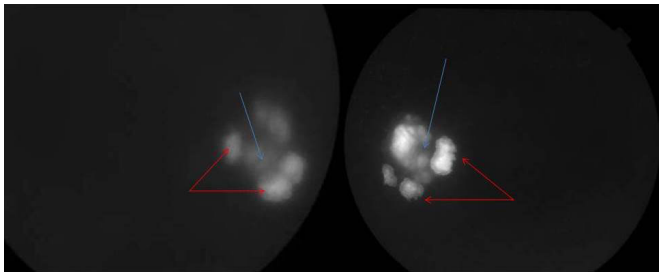

Figure 2: *Fundus autofluorescence images. There was astrocitoma related strong autofluorescence due to highly intralesional calcification in areas with peripapillary RAH (red arrows). On the optic nerve there was drusen related less autofluorescence (blue arrow).*

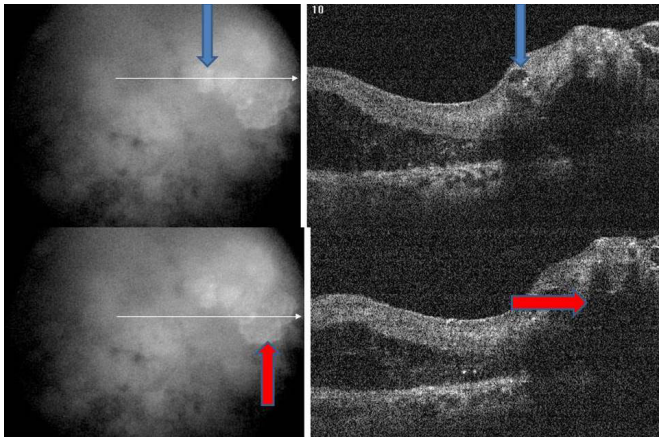

Figure 3: *Optical coherence tomography (OCT) images of the right eye. Optically empty spaces and acoustic shadows due to intralesional calcifications in the temporal peripapilary lesion (OCT figure at above, Blue arrow). Acoustic shadows due to intralesional calcifications in the inferior peripapilary lesion (OCT figure at the bottom, red arrow). Elevated mass in the nerve fiber layer and impaired retinal architecture. There is edema in the macular region.*

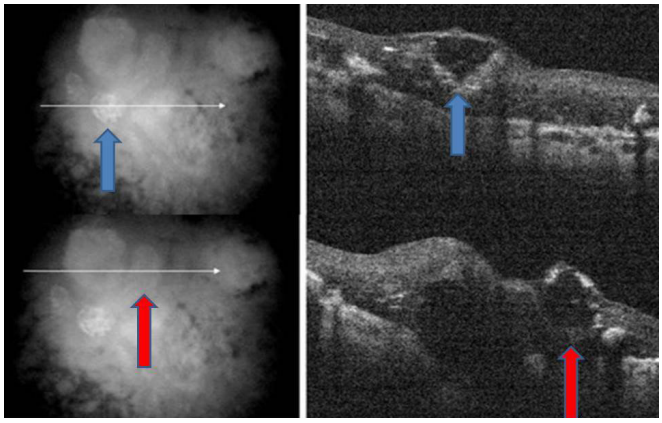

Figure 4: *Optical coherence tomography images of the left eye. Cavitation and acoustic shadows due to intralesional calcifications in the inferior and temporal peripapilary lesions (Blue arrow and red arrow).*

DISCUSSION

The diagnosis of a RAH can be made by fundus examination, OCT, FAF and ocular utrasonography. In fundus examination, RAH is categorized into 3 types. Type-1 is flat, circular shaped, semitransparent and solitary lesion that lies within the nerve fiber layer without calcification. Type-2 is elevated, opaque, nodular and mulberry-like lesion that has multiple calcification. Type-3 lesion is mixed features of both types 1 and 2. Pichi et al.[5] categorized RAH into 4 types with OCT. Type-1 shows circular shaped, flat and semitransparent lesions, involving the nerve fiber layer mainly, outer retinal layers are normal. Type-2 shows elevated and dome-shaped lesions, retinal disorganization in the inner retina, posterior shadowing. Type-3 shows elevated and mushroom shaped lesions, optically empty spaces and posterior shadowing. Type-4 shows elevated, dome-shaped and posterior shadowing lesions with large single cavity within the mass. In our case, RAHs are similar to the type-2 lesion in the clinical classification and similar to the type-3 and type-4 lesion in the OCT classification.

FAF imaging of RAHs shows hyperautofluorescence in areas with calcification. In our patient's FAF in areas with peripapillary RAH, there was astrocitoma related strong autofluorescence due to highly intralesional calcification and on the optic nerve there was drusen related less autofluorescence. The autofluorescence of the RAH depends on the range of calcification. Ocular utrasonography can detect multiple hyperechogenic lesions and calcification. Although bilateral and multiple RAHs are frequently accompained with tuberous sclerosis or neurofibromatosis type-1 in the literatüre, our patient was an isolated case because of no systemic findings.[6]

RAH are benign and often asymptomatic. Lesions do not tend to grow and cause complications. At first examination most patients do not have any visual complaints. Rarely they can cause visual symptoms with forming exudation, hemorrhage, cystoid macular edema, serous retinal detachment, localised vitreo-retinal traction and choroidal neovascularization. Rarely RAH shows dissemination and malignant transformation. Peripapillary and multiple lesions that appear at an early age tend to be aggressive. Treatment is often observation. Spontaneous regression of the RAH has also been observed.[7] Treatment of growing or symptomatic tumors are surgery, conventional laser photocoagulation, cryotherapy, intravitreal anti-vascular growth factor (anti-VEGF), photodynamic therapy and everolimus treatment. Rarely, neovascular glaucoma develops and enucleation may be required.[8] We thought that the reason for our patient's low visual acuity since birth was photoreceptor degeneration due to Leber's Congenital Amaurosis. The differences of Leber's Congenital Amaurosis from retinitis pigmentosa are congenital low vision, early developing fundus findings and macular atrophy (coloboma). We observed our patient without any treatment.

Ambiya et al.[9] reported a case of RAH in Leber's congenital amaurosis. In their case there was attenuation of retinal vasculature, generalised atrophy of the retinal pigment epithelium and bone-spicule-like pigment deposition in the general fundus. Bilateral astrocytic hamartomas of the optic nerve head, seen as mulberry-like yellowish-white nodules on the nerve head and along the peripapillary margins. These nodules showed hyperautofluorescence with an even greater autofluorescence of their calcified centres. Similar to our case, there were no systemic features of phakomatoses in their case.

Differential diagnoses should include presumed solitary circumscribed retinal astrocytic progiferation (PSCRAP), myelination of the nerve fiber layer, papilloedema, optic disc hemangiomas, retinoblastomas, meningiomas, metastatic tumours, combined hamartomas of the retina and retinal pigment epithelium and granulomas of different etiologies. [10]

CONCLUSION

It is important for an ophthalmologist to recognise an RAH clinically and know its benign prognosis and its relationship with systemic diseases, as eyes with this lesion not to undergo unnecessary examination and treatment. Retinal astrocytic hamartoma can be with retinal dystrophies (Leber's Congenital Amaurosis) and optic nerve drusen. FAF and OCT imaging can be helpful for differentiating the RAH. Close observation is important for rare complications and malignant transformation.

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