Optical Coherence Tomography Findings in a Case of Cone-Rod Dystrophy

Ioannis Emfietzoglou, MD, FEBO
Vlassis Grigoropoulos, MD
Pandelis Nikolaïdis, MD
George Theodossiadis, MD
Alexandros Rouvas, MD
Panagiotis Theodossiadis, MD

ABSTRACT
A patient with cone-rod dystrophy, who was examined thoroughly with biomicroscopy, fluorescein angiography, optical coherence tomography, and electrophysiology, is presented. Although fluorescein angiography depicted only window-defect type of hyperfluorescence, optical coherence tomography disclosed detachment of the neurosensory retina, focal absence of the outer parts of the photoreceptors’ layer in the fovea, and cystoid edema in the macula. To the authors’ knowledge, these findings have not been described in cone-rod dystrophy so far.

INTRODUCTION
Cone-rod dystrophy is a variant of retinitis pigmentosa in which the cone system is more severely affected compared to the rod system. Initially, the most common fundus finding in cone-rod dystrophy is retinal pigment epithelium (RPE) atrophy that often progresses to a bull’s eye pattern in the macula. Temporal disc pallor has also been observed in certain patients. Later, narrowing of the retinal vessels and peripheral extension of RPE atrophy with or without bone spicule formation can appear.

CASE REPORT
A 25-year-old man who complained of progressive deterioration of his vision in both eyes was examined in our clinic. His visual acuity was 0.1 bilaterally. On biomicroscopy, the anterior segments of both eyes were normal. Funduscopy revealed a zone of depigmentation around the foveola creating a bull’s eye pattern in the right and left eye and temporal pallor of the optic discs (Figs. 1A and 2A). There was no clinical evidence of cystoid maculopathy or bone spicule formation. In the macular area, fluorescein angiography showed window-defect type of hyperfluorescence without evidence of active leakage of dye or flower-petal formation. Hyperfluorescent spots were also present around the macula and along the vascular arcades due to RPE disturbances (Figs. 1B and 2B).

In the right eye, OCT depicted a neurosensory detachment in the fovea that was accompanied nasally by small cystoid spaces located in the middle layers of the retina. The macular detachment had a diameter of 1,984 µm and a height of 107 µm. There was also disruption and a complete absence of the IS/OS junction layer in some areas in the macula. The underlying RPE–choriocapillaries layer seemed to be unaffected.
In the left eye, the fovea was mainly characterized by the presence of a shallow neurosensory detachment, large cystoid spaces, and total absence of the IS/OS junction layer. There was also diffuse retinal edema in the surrounding retina. The cystoid maculopathy was more prominent in the left eye, but the height of the neurosensory detachment of the retina was larger in the right eye (Figs. 1C and 2C).

There was marked decrease of the amplitude on both the photopic and scotopic ERG (Figs. 1D and 2D). Electro-oculogram (EOG) recording was also decreased. The color vision of the patient was severely affected when tested with Ishihara's plates. Based on the ERG results and the fundus findings, we determined that the patient had cone-rod dystrophy.

DISCUSSION

There is great variation in the degree of dysfunction of the rod and the cone systems within both the retinitis pigmentosa and the cone dystrophy entities. In cases of retinitis pigmentosa, the gradual change of the amplitude of the responses of both the scotopic and the photopic ERG and the relationship between them makes the role of electrophysiology important in distinguishing these entities. However, because the photopic and scotopic amplitudes were both affected in our case, the diagnosis of cone-rod dystrophy was mainly based on the fundus findings. The characteristic bull's eye appearance bilaterally in association with the absence of bone spicules or retinal vessel attenuation and the optic disc appearance render the diagnosis of retinitis pigmentosa less probable.
OCT findings in cone-rod dystrophy in a limited number of patients have shown thinning of the retina, blunting of the foveal pit, and disorganization of the outer retinal layers. OCT of our patient depicted the presence of new findings in both eyes: neurosensory macular detachment in association with complete absence of the IS/OS junction layer in some areas, which is in agreement with the histopathological loss of the photoreceptors cells, and cystoid macular edema.

The etiology of retinal detachment and cysts in our case is not clear. However, because cone-rod dystrophy is considered a variant of retinitis pigmentosa, the macular edema could be related to a dysfunction of the outer blood–retinal barrier or to an inflammatory, autoimmune process, as in retinitis pigmentosa.

To our knowledge, the complete absence in some areas of the IS/OS junction layer and the existence of neurosensory detachment in both eyes are new findings that became evident by OCT.

REFERENCES