Clinical Features of a Pediatric Case with Cone Dystrophy

Kon Distrofilı Pediatrik Bir Olgunun Klinik Özellikleri

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ABSTRACT
The cone dystrophy is a nonhomogenous group of inherited and progressive retinal diseases that affects chiefly the cone system. It is frequently characterized by progressive loss of visual acuity, photophobia, central scotoma, color vision disturbances, and morphologic macular changes together with low response or unresponsiveness in photopic electroretinography (ERG). A girl, 9 years of age, presented with progressive visual loss, photophobia, and falling school performance. ERG revealed severe cone dysfunction with both cone and cone flicker responses (photopic) in both eyes. However, rod and rod-cone combined responses (scotopic) were evaluated at normal limits in both eyes. Fundus photography, colour vision testing, fundus autofluorescence, optical coherence tomography, ERG, visual evoked potential (VEP), Sweep VEP were performed the patient. This case report may provide clinical and diagnostic information for clinicians and may contribute to a better understanding of cone dystrophies in clinical practice. The diagnosis of cone dystrophies should be done with careful anamnesis and detailed ophthalmologic examination. With early diagnosis, there is a chance of early rehabilitation. Low vision rehabilitation is very significant because of the progressive nature of this disease, the lack of effective treatment, and the fact that the vision of the patients during the active term of school and working life are drastically affected. This case report shows that ERG can be used as a quite beneficial clinical test in terms of early and differential diagnosis of cone dystrophies.

Key Words: Retinal degeneration, cone-rod dystrophies, visual impairment, electroretinography

ÖZET
Kon distrofisi, öncelikle kon sisteminin işlevini etkileyen, kalıtsal ve ilericiyi retinal bozuklukların heterojen bir grubudur. Fotopik elektrorretinografide (ERG) dışsık yanı veya yanı yanım vermemle ilerli bir ilericiyi görme kesikliği kaybı, renkli görme bozukluğu, fotofobi, merkezi skotom ve morfolojik maküler değişiklikler ile sıkıkkla karakterizedir. 9 yaşındaki bir hasta ilericiyi görme kaybı, fotobobi ve okul performansında azalma ile başvurdu. ERG, her iki gözün kon ve kon flicker yanitlarından (fotopik) ciddi kon işlebilmekonunun ortaya çıkardı. Bununla birlikte her iki gözün rod ve rod-4 kon kombine yanitları (kotopik) normal sınırlarda değerlendirildi. Hastaya fundus fotoğrafı, renkli görme testi, fundus autofluoresan, optik koherens tomografi, ERG, görsel uyarılmış potansiyel (VEP), Sweep VEP uygulandı. Bu vaka raporu klinisyenler için klinik ve diagnostik bilgileri sağlayabilir ve klinik uygulamadada kon distrofilelerinin daha iyi anlaşılmasına katılabilir. Kon distrofilelerin tanısı dikkati dikkati anamnez ve detaylı çiftalmlojik muayene ile yapılmalıdır. Kon distrofilelerin en önemli bir tanısı sızı içinde. Dışsık görme rehabilitasyonu, bu hastalığın ilericiyi olduğu, etkili tedavinin olması ve aktif okul ve çalışma hayatı boyunca hastaların vizyonunun büyük ölçüde etkilenmesi nedeniyle çok önemlidir. Bu olgu sunumu, ERG’nin kon distrofilelerini erken ve ayrıntı tanısında en yararlı klinik test olduğunu göstermektedir.

Anahtar Kelimeler: Retinal dejenerasyon, kon-rod distrofileleri, görme bozukluğu, elektrorretinografi

Introduction
The cone dystrophy is a nonhomogenous group of inherited and progressive retinal diseases that affects chiefly the cone system (1,2,3). However, the involvement of rod system is relatively more restricted. It is characterized by progressive impairment of visual acuity, central scotoma, photophobia, sensitivity to glare, nystagmus, color vision disturbances, and morphologic macular changes together with low response or unresponsiveness in photopic electroretinography.
Fig. 1. Color fundus examination shows retinal pigment epithelium irregularity and atrophy (bull's eye lesion) at the bilateral macula central. Fundus autofluorescence examination reveals hypofluorescence appearance in the areas corresponding to bull's eye maculopathy area in the bilateral macula central, hypofluorescent spots widely distributed in the posterior pole, and hyperfluorescent spots in the midperiphery retina. A: Right eye. B: Left eye.

(ERG) (4). Frankly, it may be also stationary as well as progressive form. Besides, the cone dystrophies oughtn’t to be confused with congenital color blindness where there are no the degeneration in retina, the loss of visual acuity, and the signs of progressive disease (5). The onset of the symptoms may vary from early childhood to fifties years old, but the vast majority of cases arise in the first twenty years of life. An important part of cone dystrophies has an autosomal dominant trait, however sometimes may have a x-linked or autosomal recessive trait (1, 5, 6). Cone-rod dystrophies (CRDs) are very rare and have approximately a prevalence of 1/40000 (4). Characteristically, fundus changes limited to the macular area include patchy macular spot, a bull's eye lesion, or more prevalent atrophic alterations. However, it is more possible that most patients do not demonstrate any retina abnormalities at the onset of symptoms. It is reported that color vision disturbances are usually red-green type in the early stages (5, 7).

We aimed to evaluate the clinical features of a pediatric case with cone dystrophy, which showed a progressive decrease in visual acuity and a drop in school achievement.

Case Report

A 9-year-old girl applied to the ophthalmology clinic due to progressive visual loss, photophobia, abnormal color vision, and falling school performance was reported. There were no night vision problem, complaints of general diseases, and drug use story in the anamnesis of the patient. Also, the family history regarding ocular diseases wasn’t existent. On the ophthalmic examination, the patient’s best corrected visual acuity was measured 20/200 with -0.25D in both eyes (OU) on Snellen charts. Ocular motility testing was normal. Pupils were equally round and reactive, with no evidence of afferent defect. The anterior segment examinations of both eyes were completely unremarkable. Intraocular pressure was

Fig. 2. Optical coherence tomography (OCT) examination shows the thinning of the central macula. Furthermore, the outer layers of the retina in the parafoveal area are markedly thinned. A: Right eye. B: Left eye
Fig. 3. Electoretinography shows that rod and rod-cone combined responses (scotopic) are bilaterally at normal limits; but, cone and cone flicker responses (photopic) are markedly depressed in the left eye and decreased in the right eye.

measured as 12 mm Hg OU. Abnormal color vision was detected in both eyes by Ishihara test. Retinal pigment epithelium irregularity and atrophy defined as bull’s eye lesion at the bilateral macula central were observed on the color fundus examination (Fig 1). Hypofluorescence appearance in the areas corresponding to bull’s eye maculopathy area in the bilateral macula central, hypofluorescent spots widely distributed in the posterior pole, and hyperfluorescent spots in the midperiphery retina were seen on the fundus autofluorescence (FAF) examination (Fig 1). On optical coherence tomography examination, the thickness of the central macula was 155 micrometer in the right eye and 115 micrometer in the left eye. Furthermore, the outer layers of the retina in the parafoveal area were markedly thinned and faded, whereas the outer layers of the retina were not observed in the foveal area. (Fig 2). On the ERG, rod and rod-cone combined responses (scotopic, ERG 25 dB, ERG 15 dB) were evaluated bilaterally at normal limits; while cone and cone flicker responses (photopic) were recorded markedly depressed in the left eye and decreased in the right eye (Fig 3). On visual evoked potential (VEP) record, with stimuli of
Fig. 4. Visual evoked potential (VEP) reveals the latency and the amplitude of p100 wave
Fig. 5. Sweep VEP examination elicits mean visual acuities in both eyes

pattern 7 and 15 minutes, normal p100 wave could not be recorded. The latency and the amplitude of p100 wave obtained from 30-minute pattern stimulus were 122 msec and 7.7 mV, respectively. Whereas, the latency and the amplitude of p100 wave obtained from 60-minute pattern stimulus were 127 msec and 14.4 mV, respectively (Fig 4).

On sweep VEP examination, mean visual acuity in the right eye was 0.13 decimal, and the left one was 0.17 decimal (Fig 5). Orbital and brain MRI were normal. In laboratory tests; blood glucose level, liver and kidney function tests, triglyceride, total cholesterol, electrolytes, thyroid function tests, and urinalysis were found to be normal. Audiologic examination showed no hearing loss. Ultrasonography and electrocardiography were normal. The case was followed by ophthalmology and pediatric clinics.

Discussion

The cone dysfunction syndromes are a genetically and clinically nonhomogenous group of retinal disorders (1,2,3). The cone dysfunction syndromes are seen quite rarely and have not been enlightened in detail yet in terms of pathophysiological. We describe a girl with a cone dystrophy by characterized loss of visual acuity, a bull's eye lesion in the macula, decrease and absent of photopic ERG, photophobia, and lack of colour vision. In this report, the most notable findings were progressive loss of visual acuity, thinning of the retina, and abnormal photopic ERG flicker function. ERG revealed that photopic response was markedly depressed in the left eye and was decreased the amplitudes in the right eye. However, scotopic response was normal in the both eyes. The symptoms of the cone dystrophy are unspecific and subjective and moreover characteristic opthalmoscopic findings for this disease don’t exist, so its diagnosis is quite difficult. Cone dystrophy should be considered in the differential diagnosis of visual loss and therefore either a full-field or multifocal ERG may be necessary in order to explain the issue in such situations. In cases with cone dystrophy, visual acuity is usually better in the early stages of life, and a progressive and dramatic decline can be observed in later ages. The progression of the cone disease symptoms occurs in a symmetrical manner and experiencing symptoms are parallel in both eyes. Besides, photophobia, color vision impairment, better visual acuity in the dark, and central scotoma may be seen. Visual acuity, central visual field, and colored vision become compromised due to the progressive degeneration of the cone photoreceptor cells in the retina. Maculopathy defined as bull's eye lesion at the macula central, retinal pigment epithelial irregularity, and atrophy appearance are usually normal at onset of life. There are other diseases that cause bull's eye maculopathy, such as chloroquine maculopathy, Stargardt's disease, Batten's disease, benign concentric anomalous dystrophy, fenestrated macular dystrophy, and other photoreceptor degenerations (6, 8).

ERG test is the most beneficial clinical test in early and differential diagnosis of cone
dystrophies (3, 9). Also, in the assessment of dystrophic retinal disorders, FAF, a non-invasive retinal imaging technique, is quite advantageous (3, 10). The prognosis varies depending on the severity of the rod involvement, and a better prognosis is seen in cases with minimal rod involvement. Unfortunately, there is no specific treatment for cone dysfunction syndrome nowadays. Nevertheless, it is of utmost importance to make a correct diagnosis in order to give accurate information about the prognosis and to provide informed genetic counseling. Although there is no particular treatment, refractive errors can be corrected with appropriate spectacle, patients with low vision may take aids such as magnifiers, software for computer screen text enlargement, and closed-circuit television devices, and special educational assistance may be given. Photophobia is frequently a major symptom in the cone dystrophy and therefore dark sunglasses, contact lenses or miotics may be used in order to enhance the quality of life and vision. Low vision rehabilitation is very important because of the progressive nature of this disease, the lack of effective treatment, and the fact that the vision of the patients during the active periods of school and working life are severely affected. Briefly, with early diagnosis, there is a chance of early rehabilitation (6, 11).

Moreover, the beta-carotenoids, omega-3 fatty acids, lutein, zeaxanthin, and foods with low glycemic index have been proved to reduce progression of advanced age-related macular degeneration, and thus the consumption of these supplements may have similar advantages for cone dystrophies. Psychological support is of great importance. Consequently, in hereditary retinal dystrophies such as cone dystrophy, the residual vision of patients should be optimally protected to facilitate their daily lives and to support their psychological health (12). In conclusion, cone dystrophies are seen as quite rare. The diagnosis of this rare disease can be difficult, so it is very important that the physicians be careful. This case report may provide clinical and diagnostic information for clinicians, and may contribute to better understanding of the cone dystrophies in clinical practice. It should not be forgotten that cone dystrophy diagnosis can be done by careful anamnèsis and detailed ophthalmologic examination as in many diseases.

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