Evaluation of Central Macular Function in Bietti Crystalline Dystrophy

Bietti Kristallin Distrofisinde Santral Makula Fonksiyonunun Değerlendirilmesi

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Case Report *Olgu Sunumu*

ABSTRACT

Evaluation of central macular function by fundus autofluorescence (FAF), microperimetry and multifocal electroretinography (MfERG) in a case with Bietti crystalline dystrophy (BCD) is described. A 43-year-old woman presented with progressive visual loss in both eyes for the last 4 years. Best corrected visual acuity was 20/125 in the right eye and 20/32 in the left eye. Fundus examination revealed diffuse and profound chorioretinal atrophy at posterior pole along with glistening intraretinal crystals. FAF imaging showed central hypoautofluorescent areas encircled by hyperautofluorescent speckles. Microperimetry revealed an absolute scotoma in the right eye in central 20° . The relative scotoma in the left eye exactly demonstrated the remaining intact foveal zone as an area of reduced retinal sensitivity corresponding to angiography. MfERG in central 20° revealed significant suppression of amplitudes, particularly more reduced in the right eye. Although BCD is a progressive and untreatable chorioretinal disorder, retinal function evaluation offers a better understanding of the severity and the stage of the dystrophy.

Key Words: Bietti crystalline dystrophy, microperimetry, fundus autofluorescence, multifocal electroretinography.

ÖZ

Bietti kristalin distrofisi (BKD) saptanan bir olguda fundus otofloresansı (FOF), mikroperimetri ve multifokal elektroretinografi (MfERG) ile santral macula fonksiyonunun değerlendirilmesi anlatılmaktadır. Kırk üç yaşındaki kadın hasta son 4 senedir her iki gözde mevcut olan ilerleyici görme kaybı nedeniyle kliniğimize başvurdu. En iyi düzeltilmiş görme keskinliği sağ gözde 20/125 ve sol gözde 20/32 idi. Fundus muayenesinde arka kutupta yaygın ve şiddetli korioretinal atrofi beraberinde parlak intraretinal kristaller olduğu izlendi. FOF görüntülemesinde santral hipootofloresan alanları çevreleyen hiperotofloresan beneklenmeler saptandı. Mikroperimetri ile santral 20°'de sağ gözde absolu skotom bulundu. Sol gözde ise anjiografide izlenen sağlam kalmış olan fovea alanı ile örtüşen rölatif skotom alanı tespit edildi. Özellikle sağ gözde daha az olmak üzere MfERG'de santral 20°'de amplitüdlerde belirgin baskılanma olduğu görüldü. BCD her ne kadar ilerleyici olan ve tedavisi bilinmeyen bir korioretinal hastalık olsa da retina fonksiyonunun değerlendirilmesi hastalığın şiddetinin ve evresinin anlaşılmasında yardımcı olmaktadır.

Anahtar Kelimeler: Bietti kristalin distrofi, mikroperimetri, fundus otofloresansı, multifokal elektroretinografi.

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INTRODUCTION

Bietti crystalline dystrophy (BCD) is a progressive disorder characterized by glistening yellow-white retinal crystals at the posterior pole associated with retina pigment epithelium atrophy, choroidal sclerosis and pigment clumping.[1,2] Visual impairment, nyctalopia and paracentral scotoma frequently develop between the 2nd and 4th decades of life. Herein, we report the evaluation of central macular function by fundus autofluorescence (FAF), microperimetry and multifocal electroretinography (MfERG) in a case with BCD.

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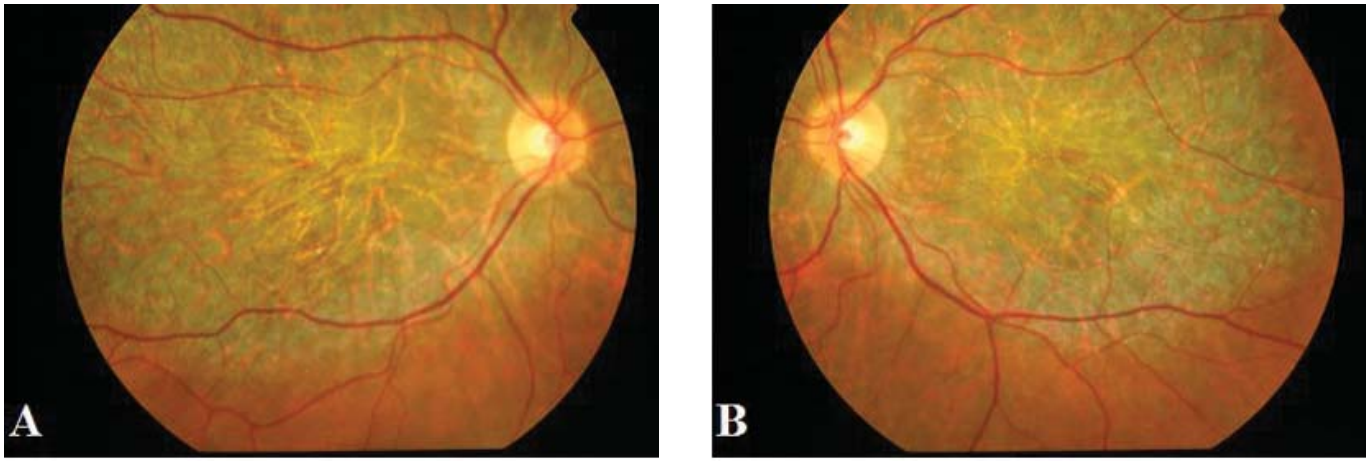

Figure 1: (A) Right and (B) Left fundus photograph. Intraretinal crystals are seen in the posterior pole.

CASE REPORT

A 43-year-old woman presented with progressive visual loss for the last 4 years. Best corrected visual acuity was 20/125 in the right eye (RE) and 20/32 in the left eye (LE). Anterior segment evaluation was unremarkable. Fundus examination revealed diffuse and profound chorioretinal atrophy at posterior pole along with glistening intraretinal crystals (Figure 1a,b). There intraretinal crystals were also apparent in confocal infrared imaging (Figure 2a,b). FAF imaging showed central hypoautofluorescent areas encircled by hyperautofluorescent speckles (Figure 2c,d). Fundus angiography demonstrated hypofluorescence with significant choroidal vessels and baring of the larger choroidal vessels except a small preserved foveal area in both eyes (Figure 1e,f). Hypofluorescent areas in the central macula secondary to choriocapillaris and retina pigment epithelium atrophy were surrounded by speckled hyperfluorescent zone due to window defect.

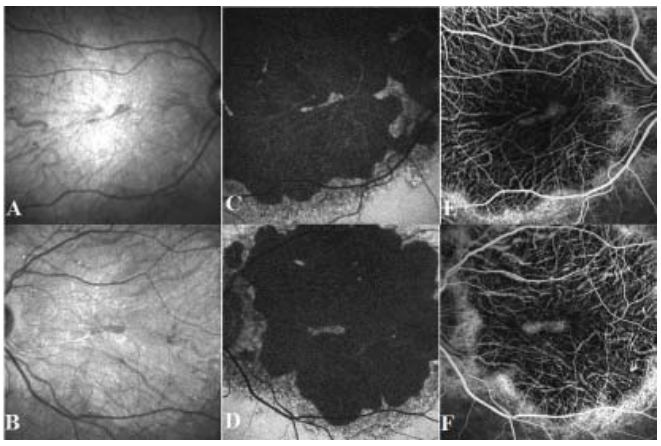

Figure 2: (A) Right and (B) Left confocal infrared fundus imaging revealing intraretinal crystals. (C) Right and (D) Left fundus autofluoresence imaging showing central hypoautofluorescent areas surrounded by hyperautofluorescent speckles. The early phase of the fluorescein angiography in the right (E) and left eye (F) reveals hypofluorescent areas in the central macula secondary to choriocapillaris and retina pigment epithelium atrophy and adjacent speckled hyperfluorescent zone due to window defect.

Central macular thickness was 111 μm in RE and 150 μm in LE by optical coherence tomography with increased reflectivity and irregularity of retinal pigment epithelium-choriocapillaris layer.

Retinal function in central 20° tested by microperimetry (MP1 Microperimeter, Nidek Technologies) was severely distorted in both eyes (Figure 3a,b). Mean sensitivity was 0.1 dB and 0.6 dB; mean defect was -11.3 dB and -18.0 dB in RE and LE respectively. Fixation was relatively unstable in RE, however a stable fixation was detected in LE. Fixation locations were predominantly central in both eyes (61% in RE and 100% in LE).

The average of retinal response density of the retinal area corresponding to ring 1 (fovea) was 43.8 nV/deg 2 and 44.5 nV/deg 2, to ring 2 (parafovea) was 24.3 nV/deg 2 and 25.6 nV/deg 2, to ring 3 (perifovea) was 11.9 nV/deg 2 and 20.7 nV/deg 2 in RE and LE respectively in mfERG (Roland-Consult RetiSCAN System), (Figure 4a,b).

DISCUSSION

In clinical practice, visual function assessment is usually performed by measuring visual acuity level. However, visual acuity level does not completely represent the state of visual functions. Assessment of retinal sensitivity with microperimetry is a rapid, safe and non-invasive diagnostic method.[3,4] It has been previously reported that in BCD a macular island is spared until later in the disease despite chorioretinal atrophy involving the whole of the posterior pole.[5]

In the present case, visual function was supplied by a small preserved foveal area despite severe chorioretinal atrophy both seen clinically and angiographically. A generalized dense scotoma was found in RE. In LE, microperimetry exactly demonstrated the remaining intact foveal zone as an area of reduced retinal sensitivity. Microperimetry also identifies the location and the stability of retinal fixation. Our case demonstrated a relatively unstable fixation in RE that had a more reduced visual acuity than LE having a stable fixation.

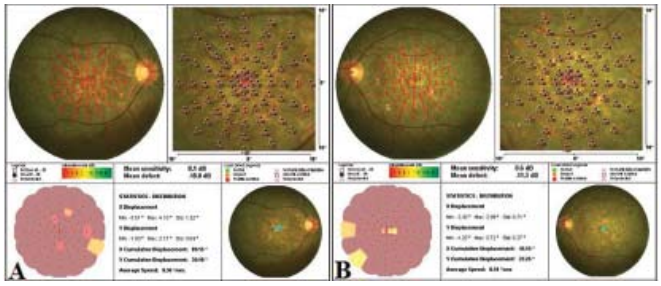

Figure 3: Microperimetry revealed (A) absolute scotoma in the right eye and (B) relative scotoma exactly demonstrated the remaining intact foveal zone as an area of reduced retinal sensitivity corresponding to angiography.

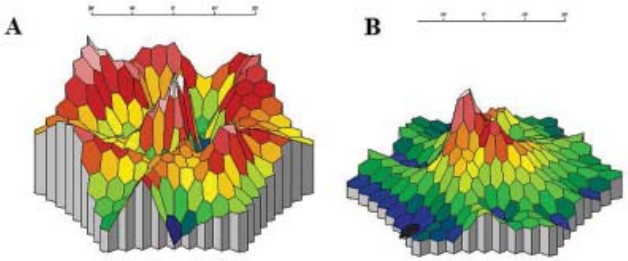

Figure 4: Multifocal electroretinography in the (A) right and (B) left eye.

MfERG provides the functional mapping of the retina and the evaluation of retinal function especially in regional disorders of the outer retinal layers. Comparison of MfERG responses with corresponding to areas of scotoma detected by microperimetry in central 20° revealed significant suppression of MfERG amplitudes. MfERG responses were more reduced in the right eye, parallel to visual acuity and microperimetric evaluation.

FAF imaging has been a valuable method that gives critical information about the retinal pigment epithelium layer and its metabolism in the current practice. In the present case, central patchy hypoautofluorescent areas were detected secondary to severe retinal pigment epithelium atrophy in the central macula that is also demonstrated by FFA. The adjacent areas of speckled hyperautofluorescence in FAF imaging which are hyperfluorescent during FFA may stand for the areas of further chorioretinal degeneration. Recently, in a report of three cases these junctional area of hyperautofluorescence were held responsible for the centrifugal degeneration pattern.[6]

MP is useful for central macular function assessment even in retinal dystrophies causing severe visual impairment, and gives supplementary information to MfERG recordings such as fixation parameters. Although BCD is a progressive and untreatable chorioretinal disorder, retinal function evaluation offers a better understanding of the severity and the stage of the dystrophy.

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