Central Areolar Choroidal Dystrophy Case

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ABSTRACT

Central areolar choroidal dystrophy is a rarely seen, hereditary retinal disease which primarily affects macula. Here, we aimed to review clinical findings, ophthalmological imaging results and electrodiagnostic test results in a 43-years old man presented with visual impairment to our clinic and diagnosed as central areolar choroidal dystrophy.

Keywords: Central areolar choroidal dystrophy, Electroretinography, Visual impairment.

INTRODUCTION

Central areolar choroidal dystrophy (CACD) is a hereditary retinal disorder which mainly affects macula. In general, it results in retinal pigment epithelium in central macula and choriocapillaris atrophy.[1-4] In patients with CACD, visual acuity is decreased between 30 and 6o years of age due to dysfunction of macular photoreceptors.[4-6] In CACD, clinical presentation was described by Noble in 1977 and the diagnosis of CACD is made based on clinical examination, fundus examination and fundus fluorescein angiography (FFA) features.[7] The diagnosis may be challenging at early stages and fine spotted depigmentation is seen in macula. Then, retinal pigment epithelium and choriocapillaris atrophy are seen, extending from perifoveal area to foveal area. Until fourth and fifth decades, progressive macular atrophy causes marked decrease in bilateral central visual acuity in most cases.[4,6] The areolar choroidal dystrophy is late-onset heterogeneous disease with variable expression. Majority of cases are familial; however, sporadic cases were also reported.[8] It generally has autosomal dominant inheritance;[9] however, autosomal recessive cases were also reported.[10] The autosomal CACD shows genetic heterogeneity.[8, 11] However, mutations in peripherin/RDS gene (Official Human Gene Nomenclature Committee gene symbol: PRPH2 or peripherin-2) are considered as most common cause.[12,13]

Her, we aimed to review clinical findings, ophthalmological imaging results and electrodiagnostic test results in a case with CACD.

CASE REPORT

A 43-years old man presented with visual impairment in both eyes. In ophthalmological examination, visual acuity was found as 0.9 (Snellen charts) in right eye and finger counting at 2 meters in left eye. The patient could bilaterally read only 2 plates correctly in Ishihara Test for Color Deficiency. In fundus examination, minimal parafoveal hypo-pigmented areas were observed in right eye while chorioretinal atrophy (approximately one disc area in size) with well-defined borders was seen in the left eye (Picture 1). On spectral domain optical coherence tomography (SD-OCT, Spectralis OCT, Heidelberg Engineering, Heidelberg, Germany), foveal disorganization in outer layers and hyper-reflective deposits over retinal pigment epithelium were striking at right eye. In left eye, all retinal layers were thinned at well-defined atrophy (Picture 2). In fundus autoflorescence (FAF), deposits seen in right eye showed hyper-autoflorescence while well-defined retinal atrophy area in left eye showed absolute hypo-autoflorescence (Picture 3). In pattern visual evoked potentials (VEP), P100 latency could not be determined since no wave was formed in both eyes. In flash VEP, waveforms were symmetrical in both eyes. The pattern electroretinography was subnormal bilaterally. In Ganzfield ERG, photopic and scotopic responses were subnormal. The scotopic

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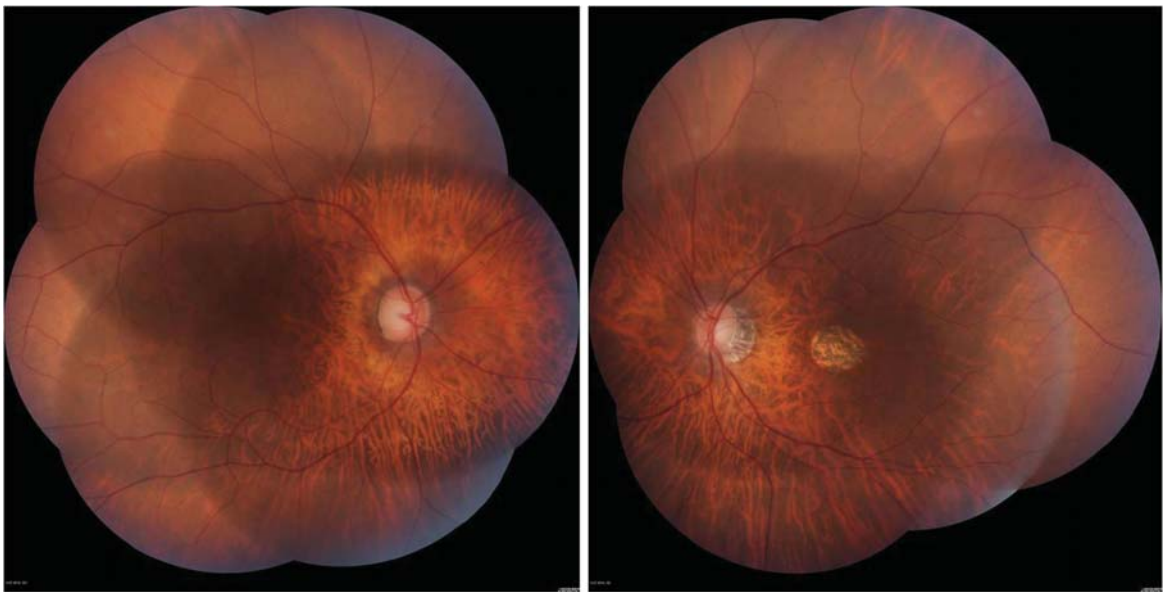
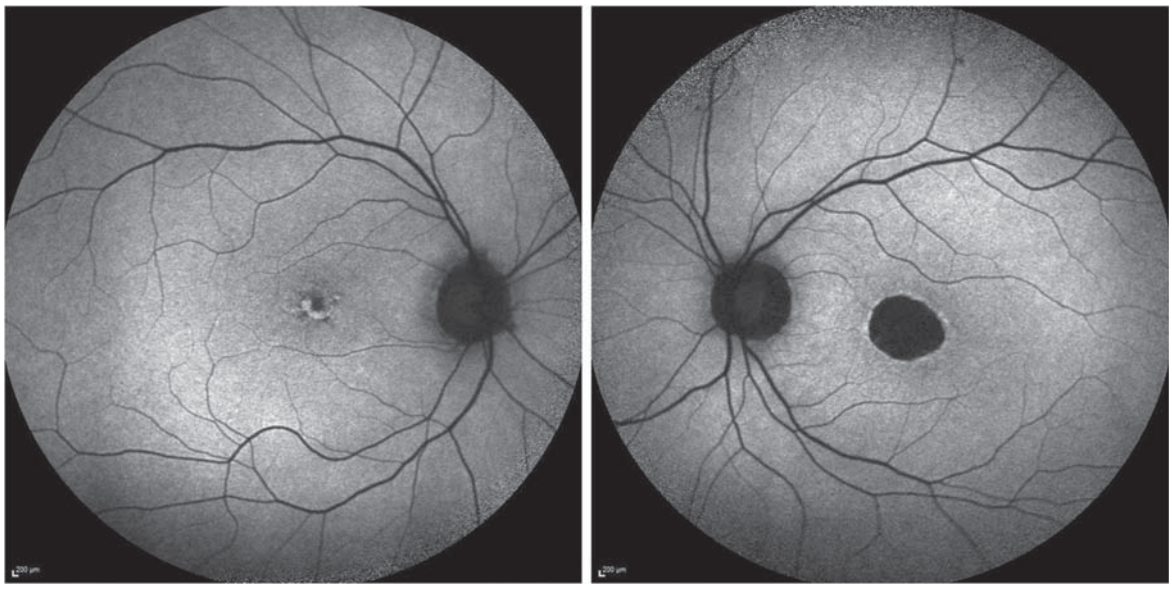


Picture 1: *On fundus images, minimal parafoveal hypo-pigmented areas were observed in right eye while there was chorioretinal atrophy (approximately one disc area in size) with well-defined borders in the left eye.*

Picture 2: *On optical coherence tomography, foveal disorganization in outer layers and hyper-reflective deposits over retinal pigment epithelium are seen in right eye while thinning in all retinal layers at well-defined atrophy area are observed in left eye.*



Picture 3: *In fundus autoflorescence, deposits seen in right eye showed hyper-autoflorescence while well-defined retinal atrophy area in left eye showed absolute hypo-autoflorescence.*

maximum response b/a ratio was determined as 1.2 in both eyes. The Arden ratio was 2.3 in right eye and 2.1 in left eye on electrooculography. In multifocal electroretinography (mERG), decreased P1 amplitude was observed in all loops in both eyes (Picture 4). In computed visual field analysis, central scotoma was observed in both eyes. These findings were considered as compatible with stage 4 CACD in left eye and stage 1 CACD in right eye.

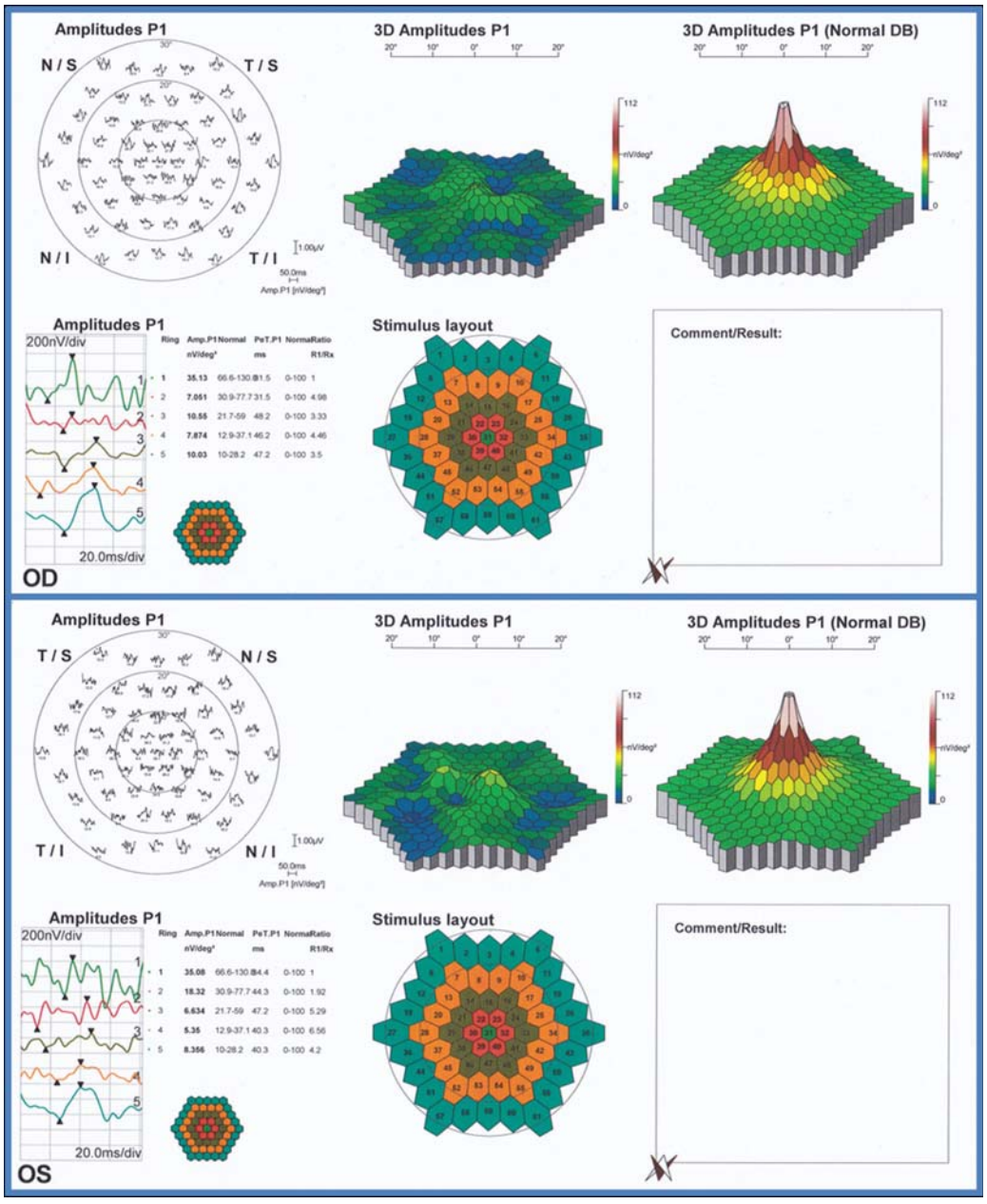
DISCUSSION

The central areolar choroidal dystrophy is a rare macular disease. This dystrophy is characterized by bilateral, symmetrical loss of oval-round choroid and retinal tissue areas. The CACD only involves macula; optic nerve and peripheral retina are normal in these patients.[14]

Four clinical stages have been identified in the disease. Fine, focal parafoveal pigmentary RPE changes are observed in stage 1 CACD. Oval-to-round, slightly atrophic, hypo-pigmented area is typical in stage 2. On fundus auto-florescence image, the area is associated with spotted auto-florescence pattern and decreased reflectivity. Stage 3 is characterized by patchy, well-defined RPE atrophy in one or more fields out of fovea. In stage 4, atrophic area involves fovea as well, resulting in marked decrease in visual acuity.[4, 15] Based on these criteria, we considered that there was stage 1 CACD in right eye and stage 4 CACD in left eye.

In CACD, differential diagnosis includes eye bull's maculopathy and cone dystrophy in early stages[16] while atrophic age-related macular degeneration (AMD) should have to be considered at late stages.[17] Currently, AMD is a one of the common diseases seen in middle-aged or elder individuals. In clinical practice, the CACD must be kept in mind in middle-aged patients with atrophic AMD.

The clinical relevance of CACD is that it is among differential diagnoses in atrophic AMD. Detailed history, clinical examination, ocular imaging modalities (OCT and FAF) and electrodiagnostic test will be helpful in differential diagnosis. In a study, Smailhodzic et al. described the differences on SD-OCT and FAF images between CACD and atrophic AMD in details.[17] In the study, authors emphasized that patients with CACD were rather younger than those with AMD with higher rate of positive family history. In a case report, Dinc et al. presented a detailed analysis of FAF findings in a patient with CACD [18]. The differences observed in SD-OCT and FAF images are guiding in the differential diagnosis between CACD and atrophic AMD. In cases with early stage CACD, spotted pattern and increased or decreased autoflorescence areas on FOF images are more prominent in CACD when compared to AMD. Although spotted pattern is rarely seen AMD, demarcation lines are not as prominent as those in CACD. Focal deposits beneath retinal pigment epithelium are more prominent in AMD on SD-OCT images. In addition, dome-like RPE elevation and related soft drusens are highly typical in these cases. However, such deposits are rather rare in cases with CACD. No reticular drusen can be seen in early CACD cases while they are observed in 50% of AMD cases. Diffuse FAF pattern (in granular, branching or drop forms) are generally observed around macular atrophic area in advanced AMD. In fact, well-defined, oval or round demarcation line observed in atrophic area



Picture 4: *In multifocal electroretinography, decreased P1 amplitude is observed in all loops in both eyes.*

is highly typical in CACD. On SD-OCT images obtained at advanced stage, deposits formed beneath RPE around atrophic area are more prominent in AMD cases when compared to CACD. At this stage, reticular drusens are often seen in AMD cases while no such pattern is observed in CACD cases. In addition, irregular retinal layers are more prominent due to deposits in Bruch's membrane in AMD [17, 18].

In our case, we observed that, in addition to clinical findings, SD-OCT and FAF images are very helpful in the diagnosis of CACD (Picture 2 and 3). In the left eye of our case, thinning in all retinal layers at atrophic area was striking on SD-OCT image.

The mERG is one of the valuable tests which can be used in the diagnosis of central areolar choroidal dystrophy. In a case report, Hartley et al. suggested that full-field ERG is a test that can gather data from retina; thus, cases with small, localized and early disease may be overlooked and mERG can provide better results [14]. Similarly, the mERG study showed that P1 amplitude was decreased in all loops, particularly in central. We think that this finding supports CACD diagnosis (Picture 4).

Although central areolar choroidal dystrophy is a rare disease, it is a clinical entity that should be kept in mind in differential diagnosis by all ophthalmologists. In addition to detailed history, evaluation of ocular imaging modalities and electrodiagnostic test together will be helpful in order

to support diagnosis and distinguish from other retinal dystrophies and AMD.

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