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Case Report

Multimodal Imaging in Central Areolar Choroidal Dystrophy

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Abstract

Central Areolar Choroidal Dystrophy (CACD), a rare hereditary retinal disorder affecting the macular area, typically presents as an autosomal dominant inherited condition. It results from mutations in the PRPH2 gene and has a prevalence of 1 to 9 cases per million. CACD progresses through four clinical stages, ultimately causing profound central vision loss. This article describes a case of a 68-year-old male initially misdiagnosed with age-related macular degeneration (ARMD) due to a 25-year history of gradual vision loss. Examination revealed symmetrical lesions with RPE and choroidal atrophy in both eyes. Optical coherence tomography (OCT) confirmed outer retinal layer atrophy without drusen-like deposits seen in ARMD. The patient received low vision aids and genetic counseling due to CACD's hereditary nature, highlighting the importance of early diagnosis and management.

Keywords: Central Areolar Choroidal Dystrophy (CACD); Optical Coherence Tomography (OCT); Age-Related Macular Degeneration (ARMD)

Central areolar choroidal dystrophy (CACD) also called as central choroidal sclerosis [1] is a rare retinal hereditary dystrophy involving the macular area. It commonly presents as an autosomal dominant inherited disorder (rarely autosomal recessive and sporadic pattern also have been seen). It occurs due to mutation of peripherin-2 (*PRPH2*) gene located on chromosome 17p13 [2]. The prevalence is 1 to 9 in 1,000,000 [3].

It is a well-defined, bilateral and symmetrical lesion causing atrophy of neurosensory layer, retinal pigment epithelium (RPE) and choroidal tissue along with choriocapillaries over the macular region, resulting in progressive and profound loss of central vision. It usually presents between the third and fourth decade of life and gradually progresses to severe vision loss over the seventh decade. It has been described in four clinical stages [4].

- Stage 1 – subtle parafoveal pigmentary RPE changes.
- Stage 2 – poorly defined round to oval area of atrophic hypopigmented area of around 1.5 disc diameter or more.
- Stage 3 – large patch of well circumscribed area of RPE and choriocapillaries atrophy in the parafoveal region.
- Stage 4 – includes stage 3 involving fovea also, which leads to profound loss of vision.

This is a case of a 68-year-old male, who was referred to us for retinal evaluation, as he was diagnosed to have age-related macular degeneration (ARMD) elsewhere. He presented with the chief complaint of gradual progressive diminution of vision in both eyes for 25 years. Systemic history was non-contributory.

On examination, his best-corrected visual acuity was 20/200 in both eyes. Fundus examination revealed round and symmetric lesions of 2-3 disc diameter size with the atrophy of underlying RPE and choroidal vessels on the macula of both eyes (Figure 1a and 1b showing fundus photo of right and left eye respectively, with stage 4 CACD). There was a loss of foveal reflex, and the optic disc, retinal vessels, and periphery were normal in both eyes. Optical coherence tomography (OCT) (Figure 2a and 2b) showed atrophy of all

outer retinal layers with loss of ellipsoid layer and thinning of RPE-Bruch's membrane complex in the atrophic area but no drusen-like deposits were seen in the sub pigment epithelial space as would be seen in geographic atrophy. Also in contrast to CACD where atrophic areas are smooth and homogenous, they are irregular due to outer retinal tubulations and show basal linear deposits between RPE and Bruch's membrane in ARMD. The data have described 3% of prevalence of late-stage ARMD in population of above 65 years of age. In central areolar dystrophy (CAD) of the RPE, atrophic changes occur in RPE and choroid. It is usually unilateral affecting mainly white females in the 5th to 6th decade of life. OCT Angiography showed visible choriocapillaries and choroidal vessels in both eyes (Figure 3, 4) due to overlying thinning of RPE-Bruch's complex. Hence, with these features, the patient was diagnosed with CACD.

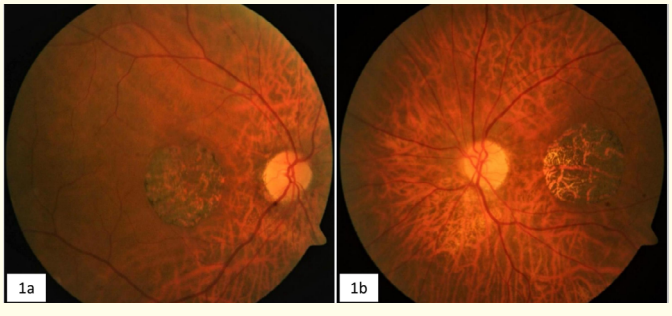

Figure 1

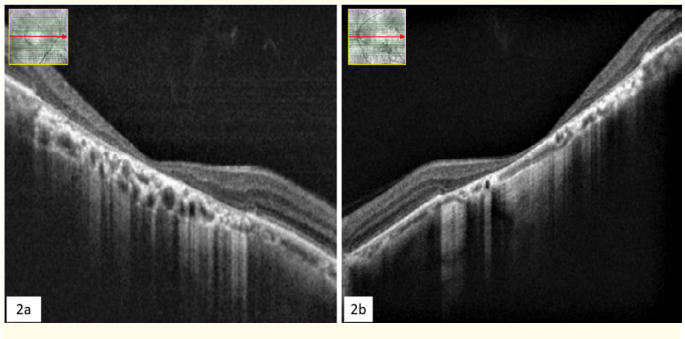

Figure 2

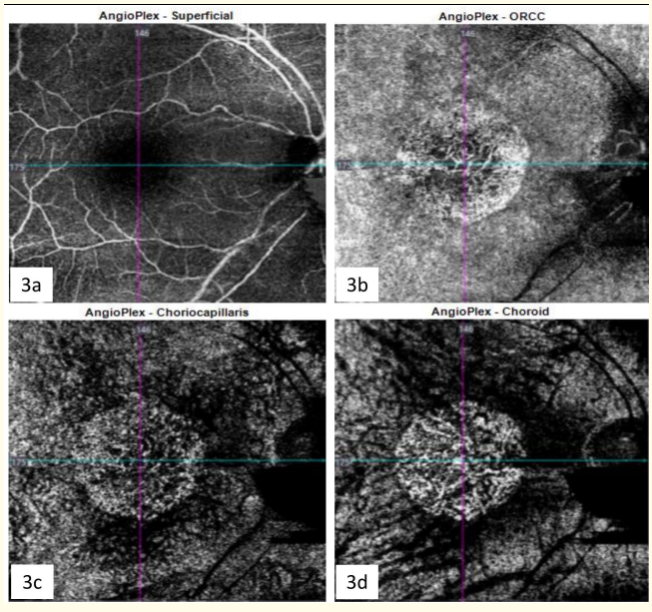

Figure 3

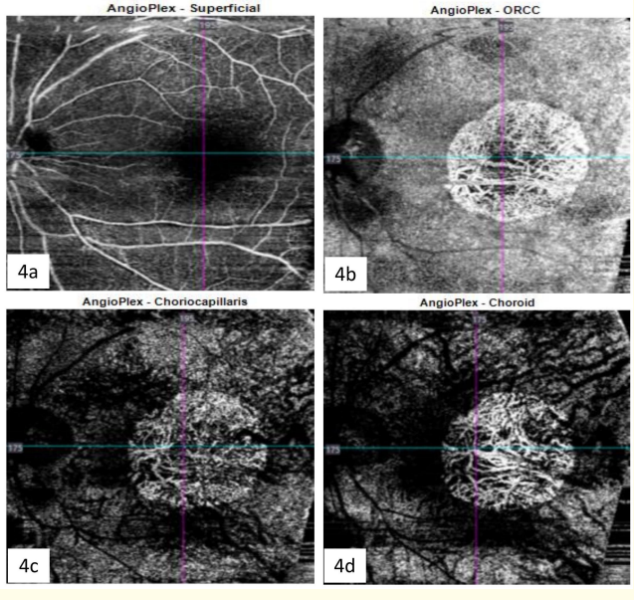

Figure 4

Several other macular dystrophies can also mimic similar macular lesions as seen in early stage of CACD like cone dystrophy and Stargardt's disease. Both present with RPE mottling and bull's eye maculopathy. In cone dystrophy the lesion appears as central reddish circular area surrounded by de-pigmented areas. It is presented with moderate to severe loss of vision, usually in the first to second decade of life and can be inherited as sporadic, autosomal dominant or X-linked recessive. In Stargardt's disease non-specific macular mottling occurs, giving 'snail slime' or 'beaten-bronze' appearance surrounded by fish-tail flecks which are not seen in CACD [1,4].

The patient was given low vision aids and was counseled about the guarded visual prognosis. He was educated about the hereditary nature of the disease and also counseled to undergo family screening and genetic counseling.

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