DOI: 10.14260/jemds/2014/2789

# CASE REPORT

## CENTRAL AREOLAR MACULAR DYSTROPHY: A CASE REPORT AND REVIEW OF LITERATURE

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**HOW TO CITE THIS ARTICLE:**
Anurag Narula, Shilpa Singh, Sunil Chakravarty, Sapna Chanana. "Central Areolar Macular Dystrophy: A Case Report and Review of Literature". Journal of Evolution of Medical and Dental Sciences 2014; Vol. 3, Issue 24, June 16; Page: 6610-6612, DOI: 10.14260/jemds/2014/2789

**ABSTRACT**: **PURPOSE:** To report a case of Central Macular Areolar Dystrophy and to discuss the literature pertaining to it. **METHODS:** Clinical data including medical history, findings on physical examination and local examination were recorded for a 35years old male, case of Central Macular Areolar Dystrophy. No treatment was advised and patient was called for regular follow up to cater for low vision aids if required in the future. **RESULTS:** Best corrected Visual acuity of the patient remained constant at 6/36 for a period of one year with the patient being on 3monthly follow up. **CONCLUSION:** No particular or specific treatment is available for this rare macular dystrophy. The only modality available in the form of low vision aids for progressive disease.
**KEYWORDS:** Central Areolar Macular Dystrophy.

**CASE HISTORY**: A 35years old male, presented to us with headache, eye ache and loss of vision progressive deterioration since birth to the present status of vision being 6/36 (best corrected in both eyes).

General physical examination and systemic examination was unremarkable. On examination best corrected visual acuity was 6/36 both eyes, with normal anterior segment examination. On fundus examination an oval-to-round, atrophic, hypo pigmented area and area of well demarcated RPE atrophy was seen (figures 1 to 4). This area, on a fundus auto fluorescence (FAF) image, shows increased as well as decreased reflectivity resulting in a speckled FAF pattern.

Provisional Diagnosis: Central Macular Areolar Dystrophy.

No treatment was advised and patient was called for regular follow up to cater for low vision aids if required in the future.

Best corrected Visual acuity of the patient remained constant at 6/36 for a period of one year with the patient being on 3monthly follow up.

**DISCUSSION:**

**Central Macular Areolar Dystrophy:** Central areolar choroidal dystrophy (CACD) is a hereditary retinal disorder that affects the macula, resulting in progressive and usually profound visual loss. The hallmark feature of the disorder is a well-defined atrophy of the retinal pigment epithelium (RPE) and the choriocapillaris.[1] Four clinical stages of the diseases have been described.[2] In stage 1 CACD, subtle focal parafoveal pigmentary RPE changes can be observed on ophthalmoscopy. A typical stage 2 finding in the color image is an oval-to-round, mildly atrophic, hypopigmented area. This area, on a fundus autofluorescence (FAF) image, shows increased as well as decreased reflectivity resulting in a speckled FAF pattern. Stage 3 is characterized by one or more patches of well-demarcated RPE atrophy outside the fovea. In stage 4, the atrophic area involves the fovea, resulting in a markedly decreased visual acuity.[2, 3]

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Autosomal dominant CACD is most commonly caused by mutations in the peripherin-2 (PRPH2) gene (formerly known as peripherin/RDS).[4, 5] More than 90 different PRPH2 mutations associated with a wide spectrum of fundus alterations have been reported. To date, seven different mutations in the PRPH2 gene have been identified to cause the CACD phenotype.[6–12] It may be challenging to diagnose CACD in the early stages of the disorder because of the relative nonspecific RPE abnormalities. Also, the late-onset variant may easily be confused with age-related macular degeneration (AMD) and thus be misdiagnosed.

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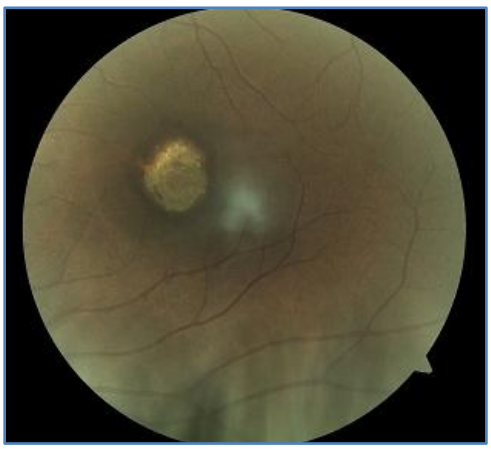
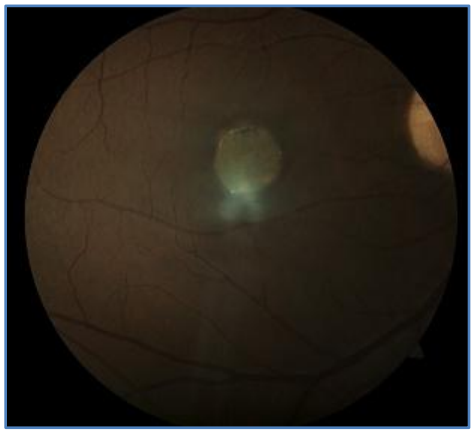


**FIGURE 1 AND 2 FUNDUS LESION OF RIGHT EYE**



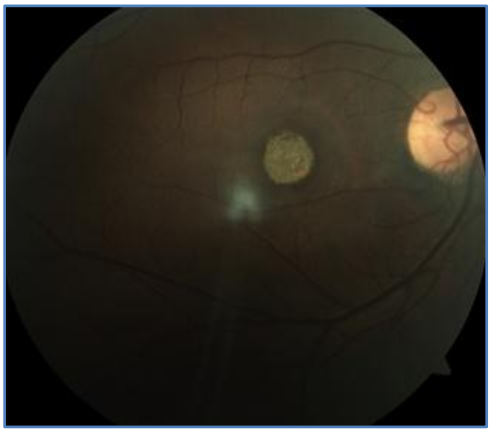


**FIGURE 3 AND 4 FUNDUS LESION OF LEFT EYE**

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Date of Submission: 10/05/2014.
Date of Peer Review: 12/05/2014.
Date of Acceptance: 02/06/2014.
Date of Publishing: 11/06/2014.