Central Areolar Choroidal Dystrophy with Dominantly Inherited Drusen

Abstract
A 59-year-old male is diagnosed with central areolar choroidal dystrophy (CACD) with dominantly inherited drusen. Clinical examination found macular changes consistent with CACD. Diagnosis was confirmed with optical coherence tomography and fundus autofluorescence.

Case History
59-year old African American male
CC: constant blur at distance and near in both eyes for two years; wears readers only
LEE: Memphis VA, 1 month prior
OHx: CE OU, 4 years prior
Ocular meds: none
MHx: Type 2 DM with neuropathy, hypertension, arthritis
FOHx: none

Pertinent findings
VA sc: 20/20 OD, 20/20 OS at distance
20/100 OD, 20/80 OS at near
Pupils, EOMs, CVF: unremarkable OU
Manifest refraction: +0.50 sph OD, +0.25 sph OS with +2.25 ADD and BCVA 20/20 OD, OS at distance and near
Slit lamp: moderate palpebral and bulbar conjunctival hyperemia OU
Lens: clear and well-centered PCIOL OU
Goldmann applanation: 14mmHg OD, 15mmHg OS
DFE:
- ONH: C/D 0.30, round with moderate depth and distinct rim margins OU
- Macula: flat, intact OU
- Posterior pole: scattered soft drusen around posterior pole, greater temporal and superionasal to macula OU and an area of circumscribed hypopigmentation superionasal to macula OU
- Periphery: retinoschisis superiotemporally OS

Fundus photos taken
Macular OCT: area of atrophy of choriocapillaris and RPE superionasal to macula OU, corresponding with clinical findings

Differential diagnosis
Atrophic AMD
Cone dystrophy
Stargardt’s disease
Central areolar choroidal dystrophy (CACD) with dominantly inherited drusen

Diagnosis and discussion
CACD is an autosomal dominant hereditary retinal disease that primarily affects the choriocapillaris. It is caused by a mutation in the peripherin/retinal degeneration slow (RDS) gene, and a specific mutation of Arg142Trp in this gene is thought to manifest the combined presentation of CACD and dominant drusen. CACD presents with subtle, mottled depigmentation in the early stages, and patients are usually...
asymptomatic with normal visual acuity. The RPE alterations are hardly detectable early on; they’re more easily appreciated with fluorescein angiography. Vision starts to decrease in the 3rd to 4th decade and patients can have absolute central scotomas. Clinically, it may resemble a macular coloboma because of its excavated look. Well-defined atrophy of the RPE and choriocapillaris is the hallmark sign in late stages. This late-onset CACD can easily be confused with geographic atrophy in AMD. However, unlike AMD, CNVM in CACD is rare since there is no functional choriocapillaris or choroidal vascular system.

Four clinical stages of CACD:
- **Stage 1:** subtle focal parafoveal pigmentary RPE changes. Fundus autofluorescence (FAF) will show increased reflectivity of these changes and OCT will reveal RPE and photoreceptor disruption.
- **Stage 2:** oval or round hypopigmented area of mild atrophy. FAF will show increased and decreased reflectivity resulting in a speckled pattern. OCT will show reflectivity changes and interruptions of photoreceptor layer and thinning of ONL.
- **Stage 3:** single or multiple well-demarcated areas of RPE atrophy outside of fovea. FAF will show typical speckled pattern with an absent FAF signal in the area of atrophy. OCT will reveal a total absence of outer retinal layers in the atrophic area.
- **Stage 4:** atrophy of fovea, resulting in decreased vision. FAF will show typical speckled pattern with outer retinal changes on OCT.

**Treatment, management**
- Educate on progressive nature of disease.
- Visual prognosis is poor, although peripheral vision is preserved so mobility isn’t affected.
- Refer for genetic counseling.
- Low vision services if vision significantly affected.

**Conclusion**
CACD normally presents without drusen. However, in patients manifesting a specific mutation, CACD may present in conjunction with drusen. One study showed that 95% of individuals with CACD and dominant drusen carried an Arg142Trp mutation in peripherin/RDS. Additionally, there were no individuals with dominant drusen in the absence of the Arg142Trp mutation, suggesting that the Arg142Trp mutation is one of the factors predisposing to drusen formation. However, recently, a type of autosomal dominant CACD has been described in members of a large Northern Irish family to be linked to a GUCY2D mutation in chromosome 17p13.

**References**


