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# Patient Presentation

CC: Decreased VA OU x several years

HPI: 45 yo and 53 yo WF (sisters) are referred to the retina clinic for evaluation of similar visual and clinical findings. Both experienced gradual loss of vision OU in their mid 20's. The vision loss stabilized in their mid-thirties. Also, complained of photopsia and nyctalopia.

No one else in the family has had similar visual problems.

# Sister A (45 yo)

POH: as described in HPI

PMH: osteoarthritis

Meds: glucosamine and chondroitin

All: PCN

ROS: (-)

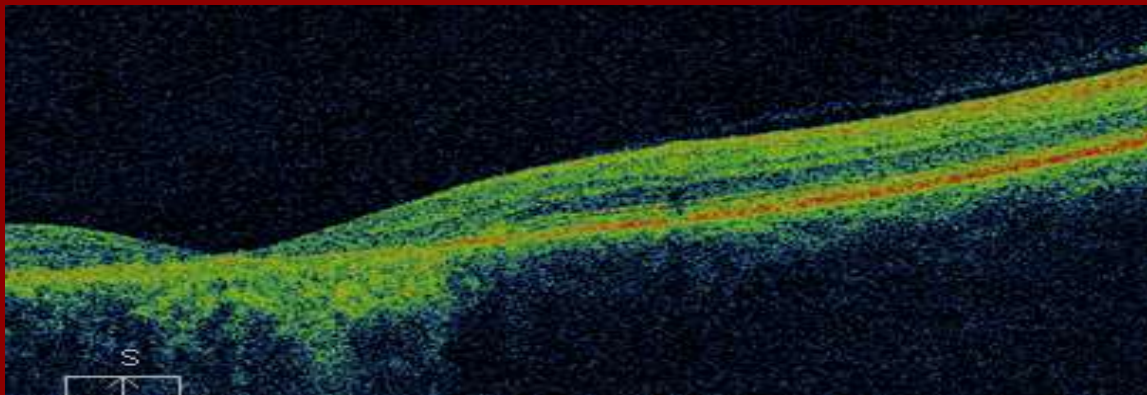
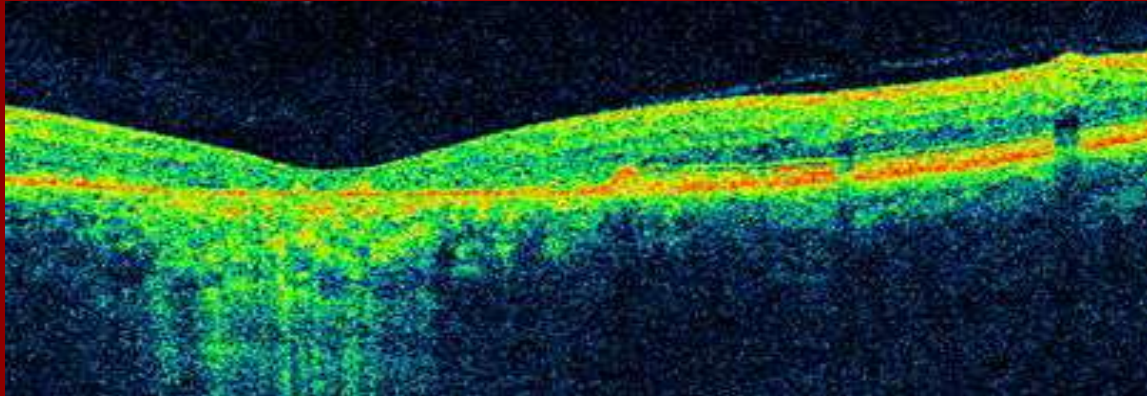
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# Fundus Photo



Color fundus photos of right and left eyes demonstrating  
of RPE atrophy and flecks in the macula of both eyes

# OCT



OCTs of right and left eyes:  
Foveal atrophy and subfoveal deposits of highly  
reflective material

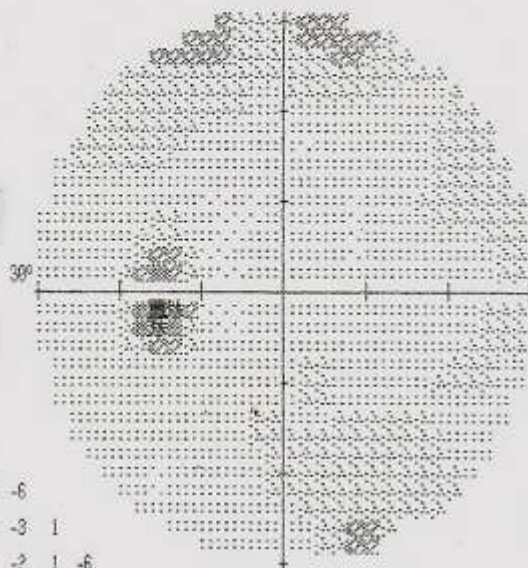
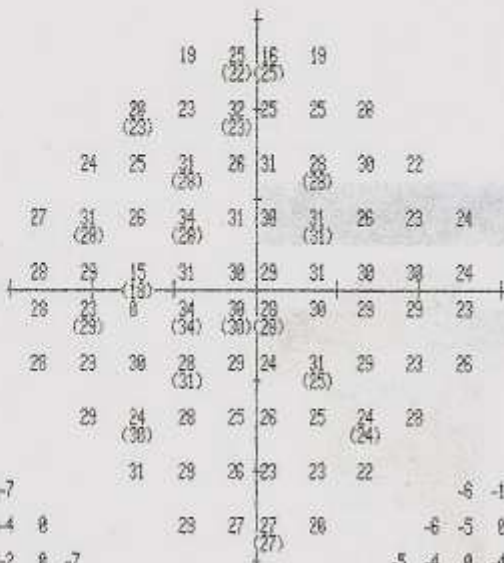


NAME  
 STIMULUS III, WHITE, BCKGND 31.5 ASB BLIND SPOT CHECK SIZE III  
 STRATEGY FULL THRESHOLD  
**FASTPAC**  
 LOW PATIENT RELIABILITY

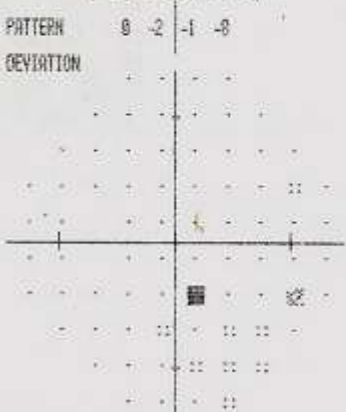
BIRTHDATE 7-20-65 DATE 04-23-96  
 FIXATION TARGET CENTRAL IO TIME 07:50:33 AM  
 RX USED + 0.00 DS DCX DEG PUPIL DIAMETER 7.0 MM VA 20/80

**LEFT**

AGE 31  
 FIXATION LOSSES 3/15 \*\*  
 FALSE POS ERRORS 8/4  
 FALSE NEG ERRORS 1/8  
 QUESTIONS ASKED 273  
 FOMER+ 21 DB ■  
 TEST TIME 00:03  
 NFA S/M 630-5732



*SB  
 good copy*



PROBABILITY SYMBOLS  
 ● P < 5%  
 \* P < 2%  
 ■ P < 1%  
 ■ P < 0.5%

MD -3.04 DB P < 5%  
 PSD 2.64 DB  
 SF 2.43 DB  
 OPSD 0.00 DB

# Sister B (53 yo)

POH: as described in HPI

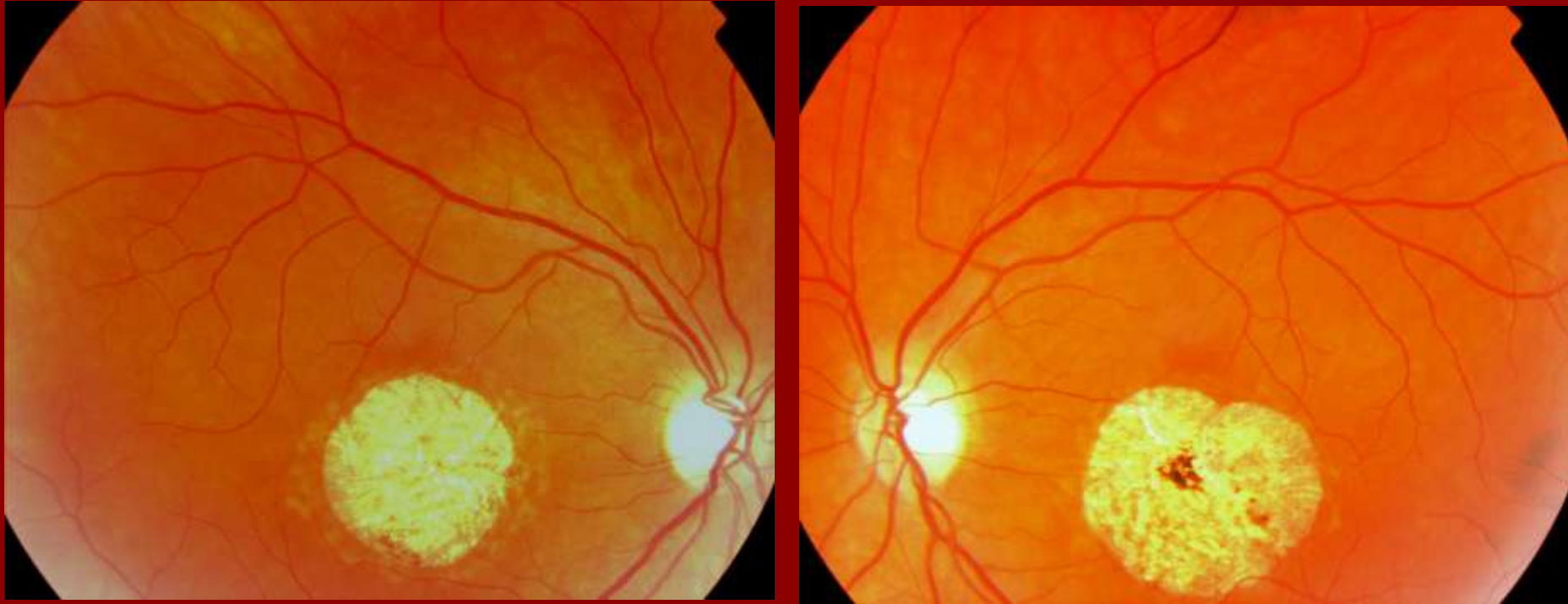
PMH: Depression

Meds/All: Zoloft

ROS: (-)

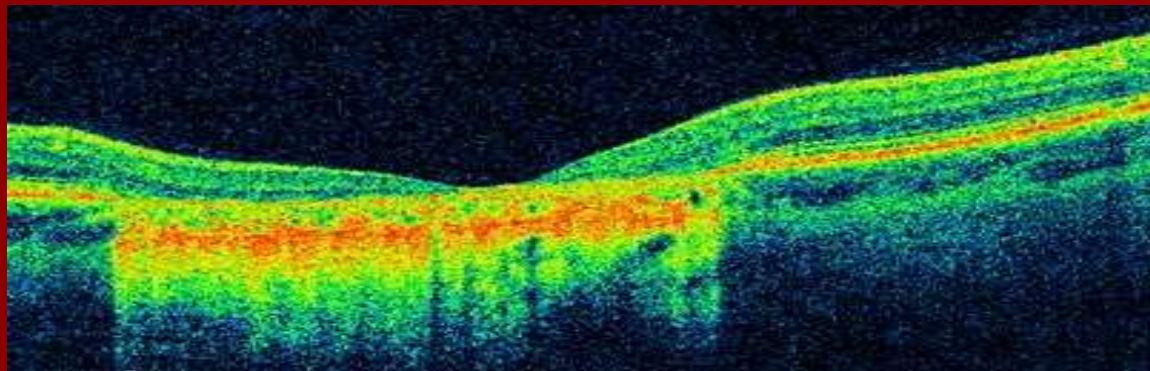
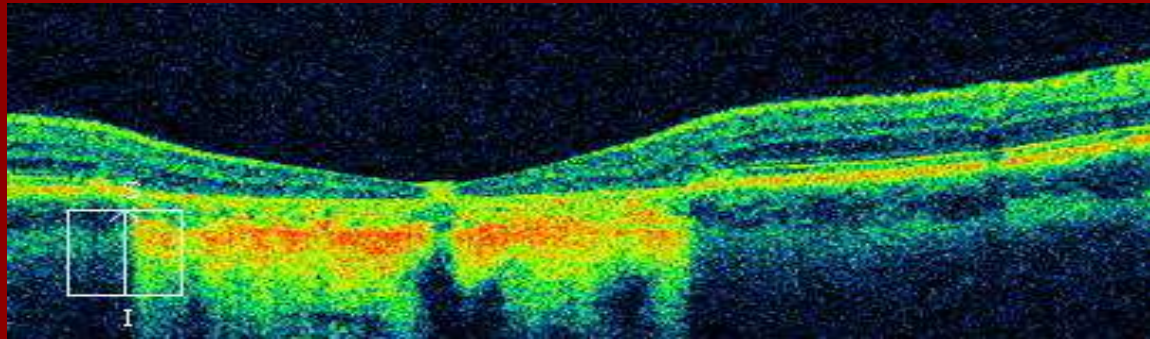
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# Fundus Photo



Color fundus photos of right and left eyes demonstrating round, well-demarcated areas of chorioretinal atrophy with surrounding flecks

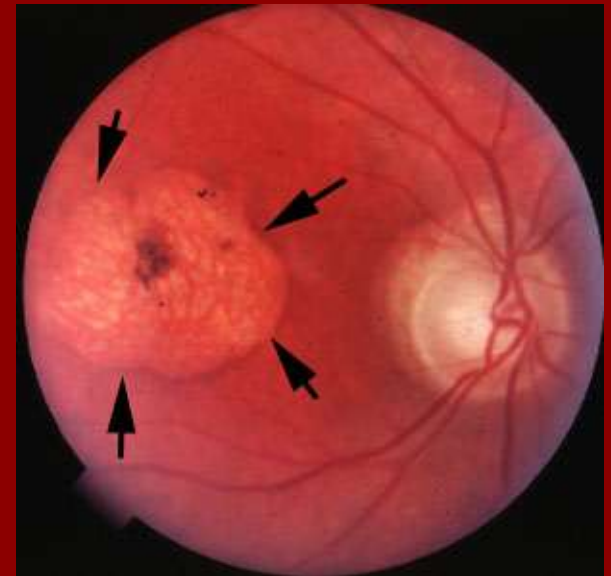
# OCT



OCT images of right and left eye:  
NSR atrophy and collection of highly reflective deposits under RPE

# Impression/DDx

- 45 and 53 yo WF sisters who present with gradual loss of vision and nyctalopia are found to have similar pattern of bilateral progressive macular dystrophy
- Differential Diagnosis:
  - Central areolar choroidal dystrophy
  - End stages of Stargardt's disease
  - Sorsby dystrophy
  - Cone-rod dystrophy



# Plan

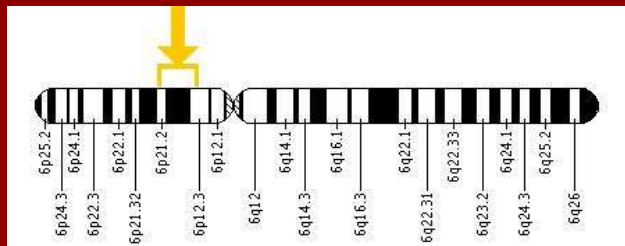
- ERG
  - mfERG
  - Scotopic and photopic ERG
- HVF
- FA

# Central Areolar Choroidal Dystrophy (CACD)

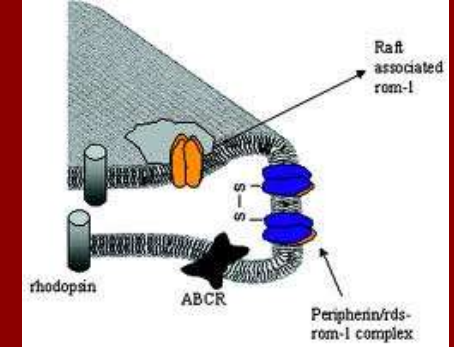
- Hereditary disorder that affects the macula, resulting in a well-defined area of atrophy of RPE and choriocapillaries in the center of macula
- Most cases are autosomal dominant although autosomal recessive and sporadic have also been reported
- Mutations in peripherin/RDS gene seem to be the most common cause

# Pathophysiology

- All studies have shown an absence of photoreceptor, RPE and choriocapillaries in the area of atrophy
- Mutation in peripherin/RDS → positively charged arginine substitutes nonpolar and hydrophobic tryptophan → change in protein structure → dysmorphic cone and possible rod outer segment → increase in phagocytosis of abnormal outer segments → ↑ lipofuscin and toxic byproducts in RPE → apoptosis of photoreceptors and RPE



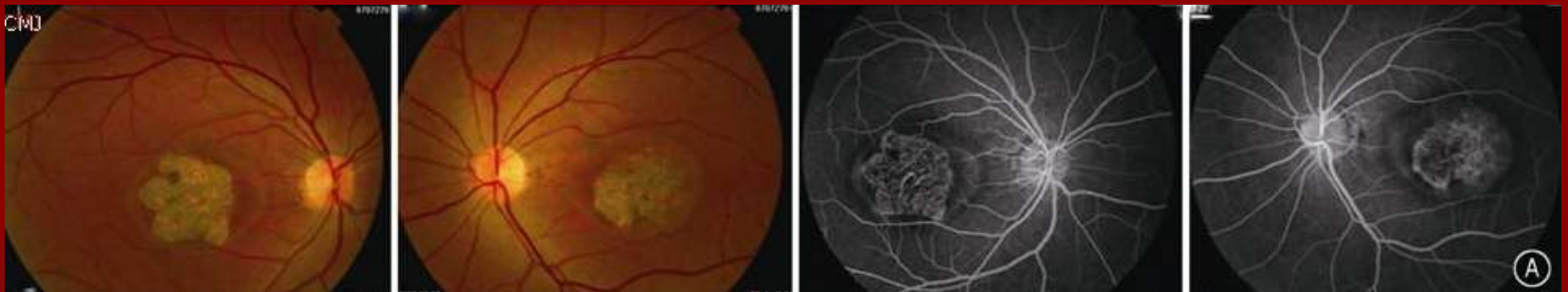
# Peripherin



- Peripherin is a surface glycoprotein that in humans is encoded by *PRPH2* gene (on the short arm of chromosome 6)
- A member of transmembrane 4 superfamily (tetraspanin)
- Found in the outer segment of both rod and cone photoreceptors
- It may function as an adhesion molecule involved in stabilization and compaction of outer segment discs (essential for disc morphogenesis)
- Involved in multiple retinal degenerations:
  - CACD
  - Adult vitelliform dystrophy (at least in 20% of cases)
  - AD retinitis pigmentosa

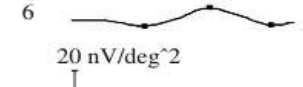
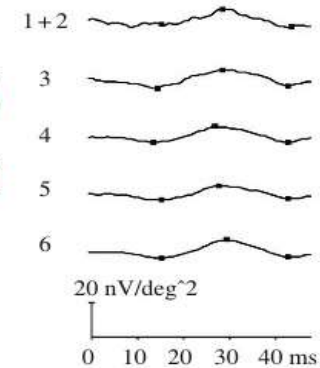
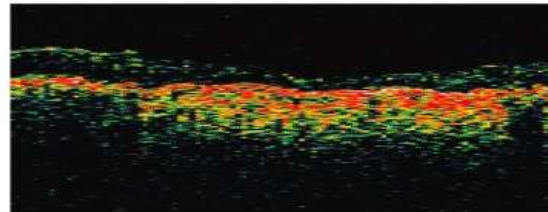
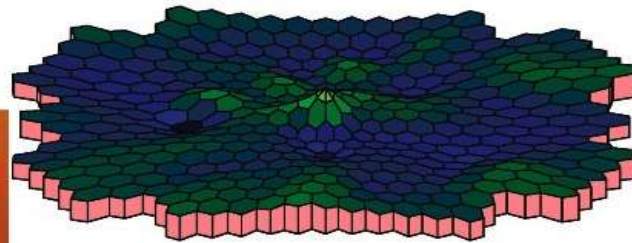
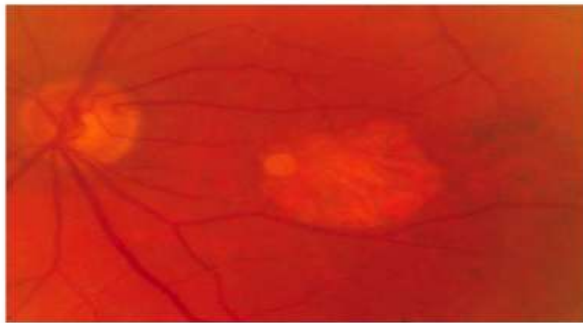
# CACD

- Clinical findings: RPE stippling → atrophy affects the whole macular area sparing the fovea (around age 40) → atrophy of choriocapillaries → around age 60, fovea starts to be affected → VA can drop to CF within few years
- FA findings:
  - Early cases: window defect
  - Longer standing: disappearance of background fluorescence



# CACD

- VF: Central or paracentral scotoma
- Color Vision: moderately affected
- Dark adaptation can be abnormal
- ERG: normal in many patients



# CACD vs AMD

- Shared clinical characteristics:
  - Geographic atrophy
  - Drusen-like deposits
- In an elderly patient, CACD may be confused with AMD; Distinguishing factors:
  - Early AMD displays dispersed hard or soft drusen; oval area of hypopigmentation is observed in early CACD
  - In end-stage AMD, geographic atrophy is usually bordered by a band of hyperpigmentation
  - Rate of progress of geographic atrophy in AMD is usually higher than CACD
  - Drusen caused by CACD shows more intense autofluorescence (difference in composition)

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Thank You!