

UNIQUE PHENOTYPIC EXPRESSION OF PACHYDRUSEN IN TWO BANGLADESHI-AMERICANS WITH PACHYCHOROID AND CENTRAL SEROUS CHORIORETINOPATHY

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DISCLOSURE

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SUMMARY

We present 2 young Bangladeshi-Americans who had a unique phenotypic expression of pachydrusen. The age of these patients was significantly younger than previously reported cases. Also the number of drusen was more than usually reported and remarkable symmetry between eyes was noted. Given the similar ethnic background we hypothesize an environmental or genetic predisposition to this particular phenotypic expression

CASE 1

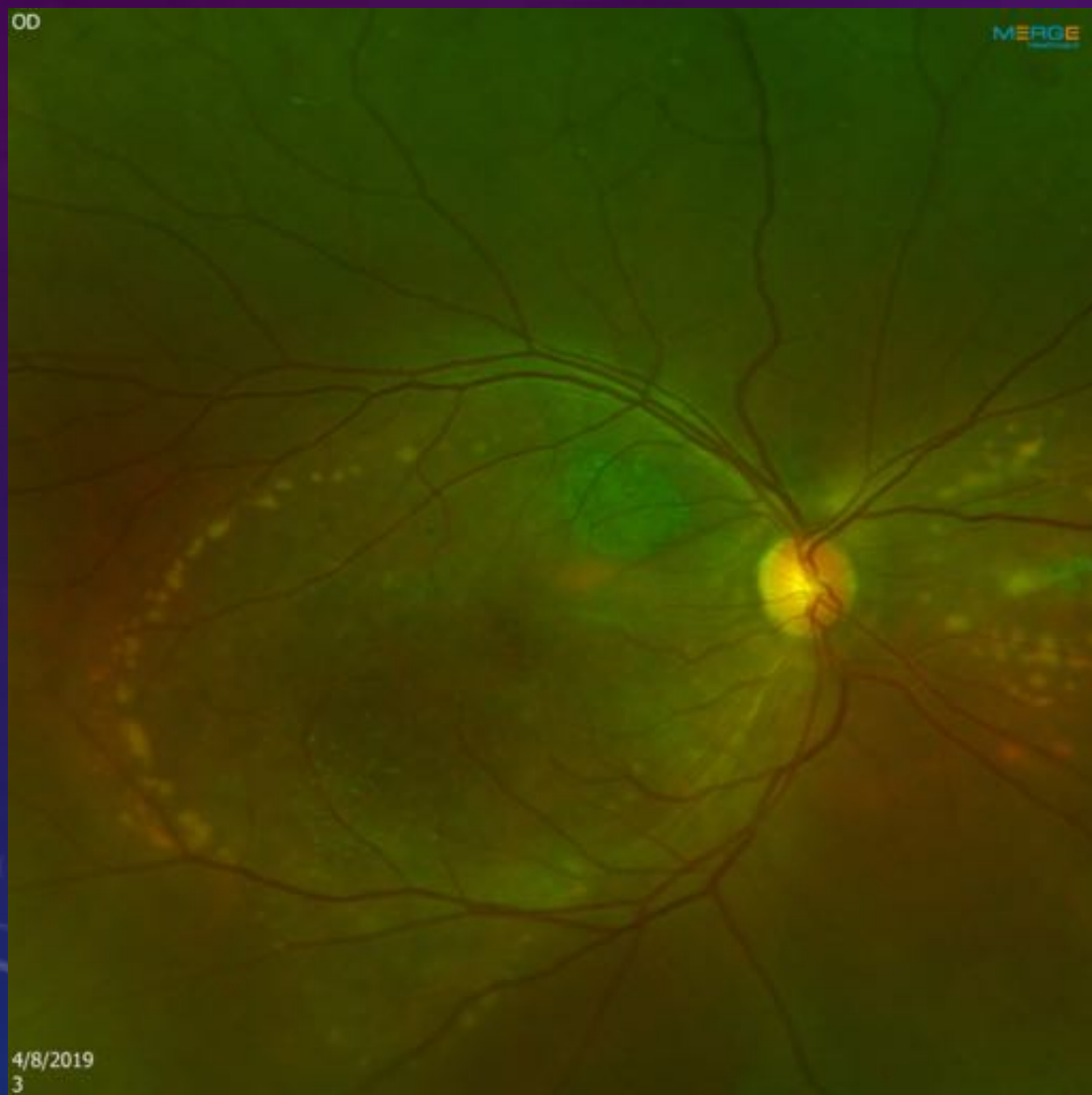
- 32 year-old Bangladeshi-American man
- **Chief complaint** blurry vision in both eyes for many years
- **Past medical history** Diabetes
- **Medications** Metformin
- **Past ocular history** negative
- No history of corticosteroid exposure

CASE 1

- Vision 20/30 OD 20/50 OS
- Anterior segment normal

OD

MERGE



4/8/2019
3

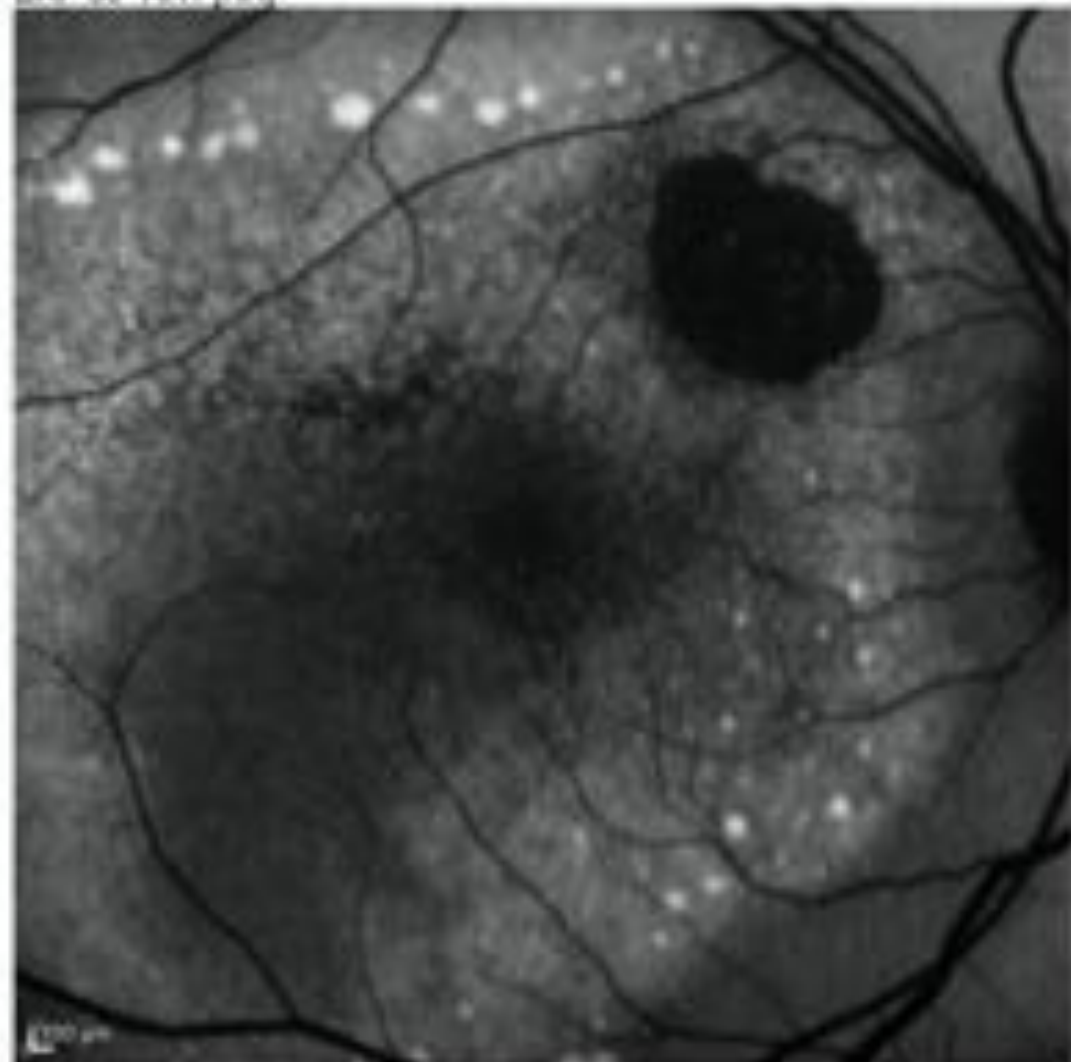
OS

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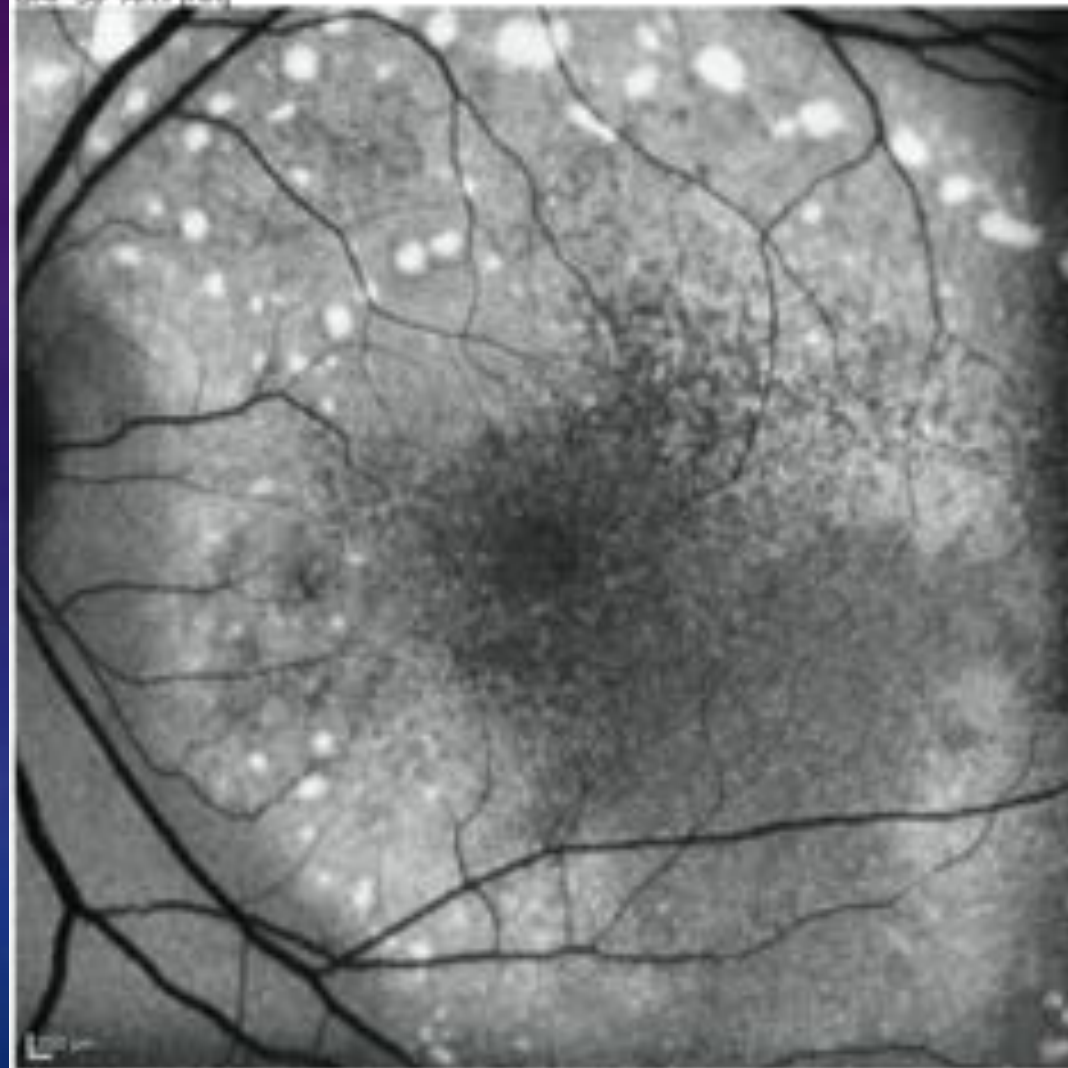


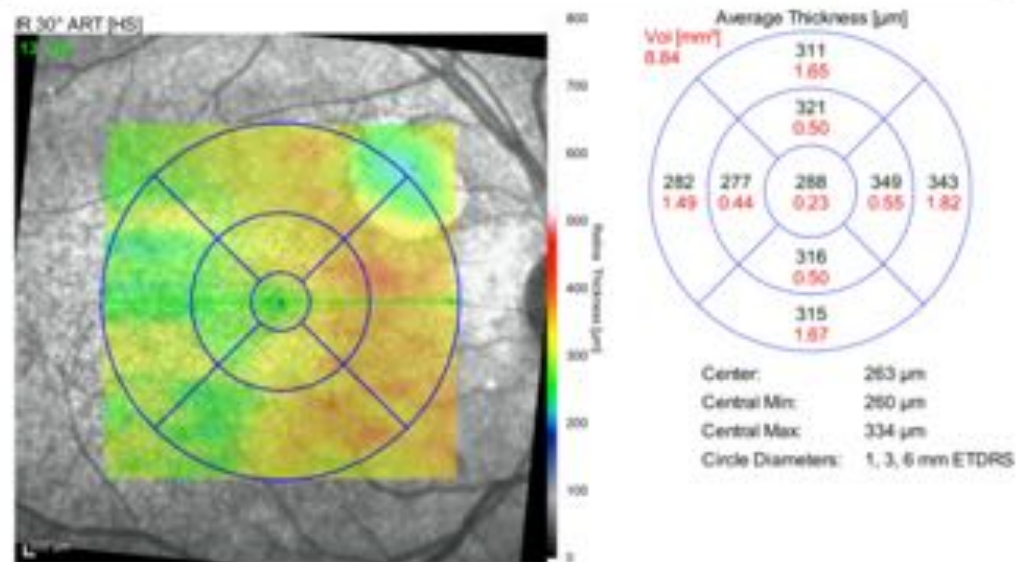
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BAF 30° ART (HR)

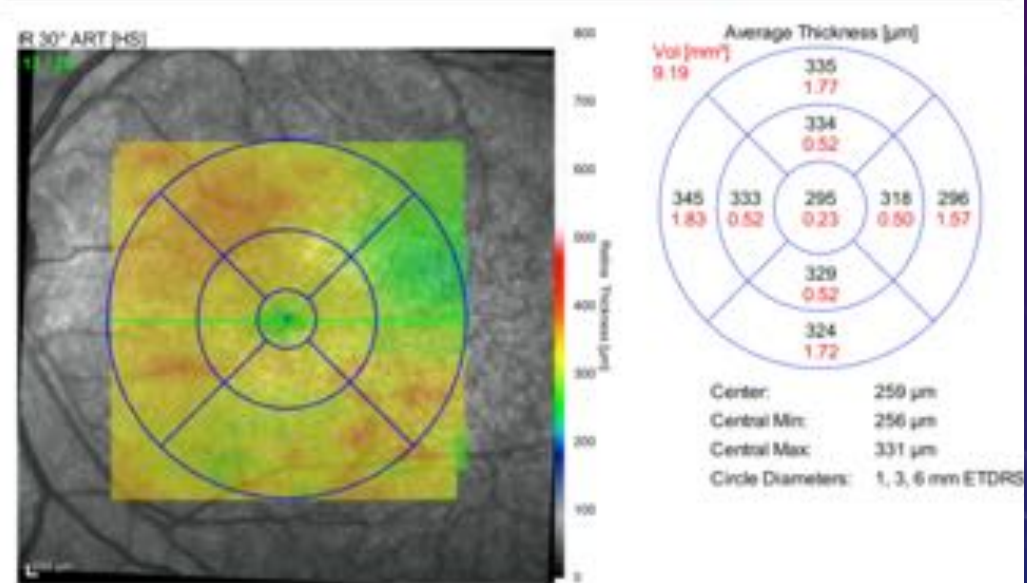
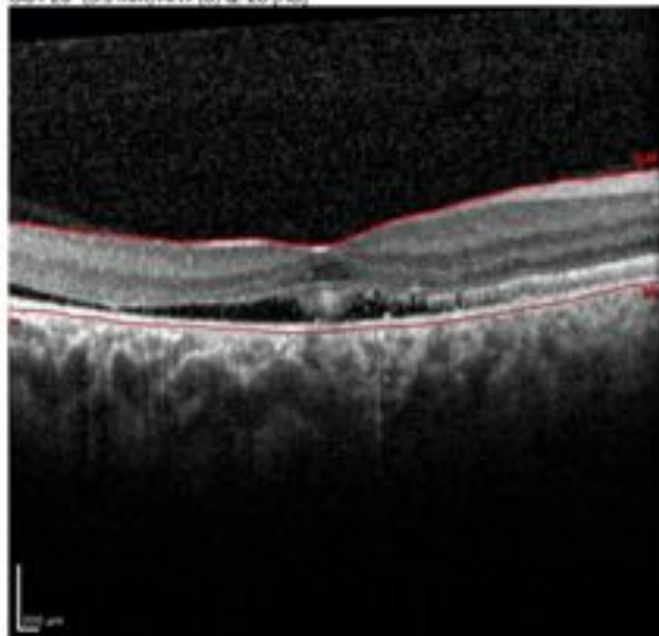


BAF 30° ART (HR)

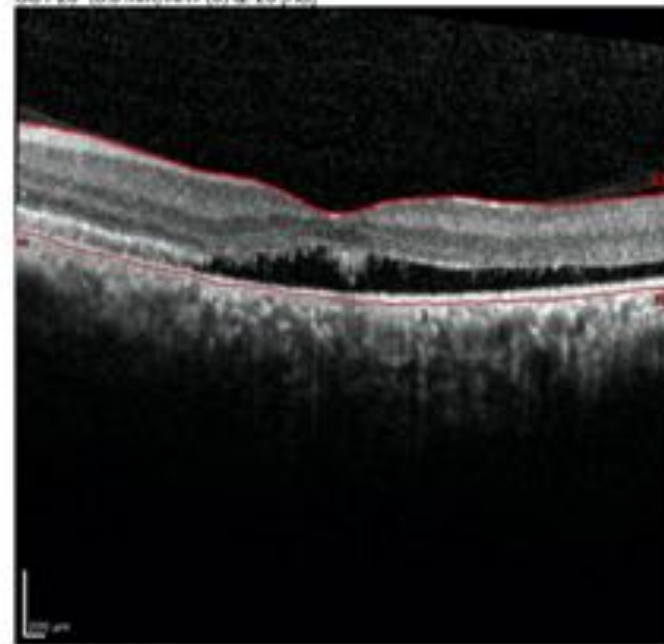




OCT 20° (5.9 mm) ART (B) Q: 28 (HS)



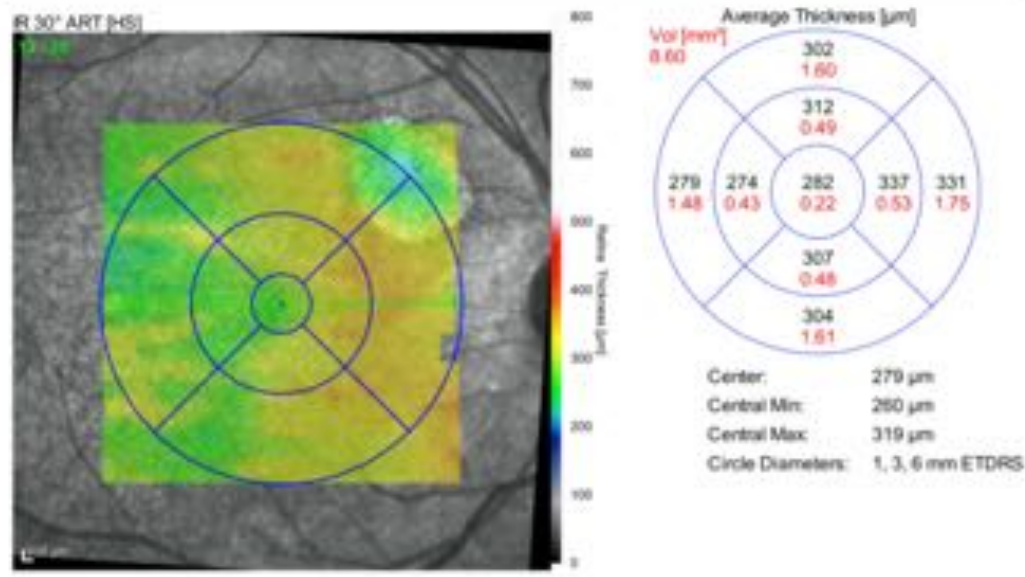
OCT 20° (6.0 mm) ART (B) Q: 29 (HS)



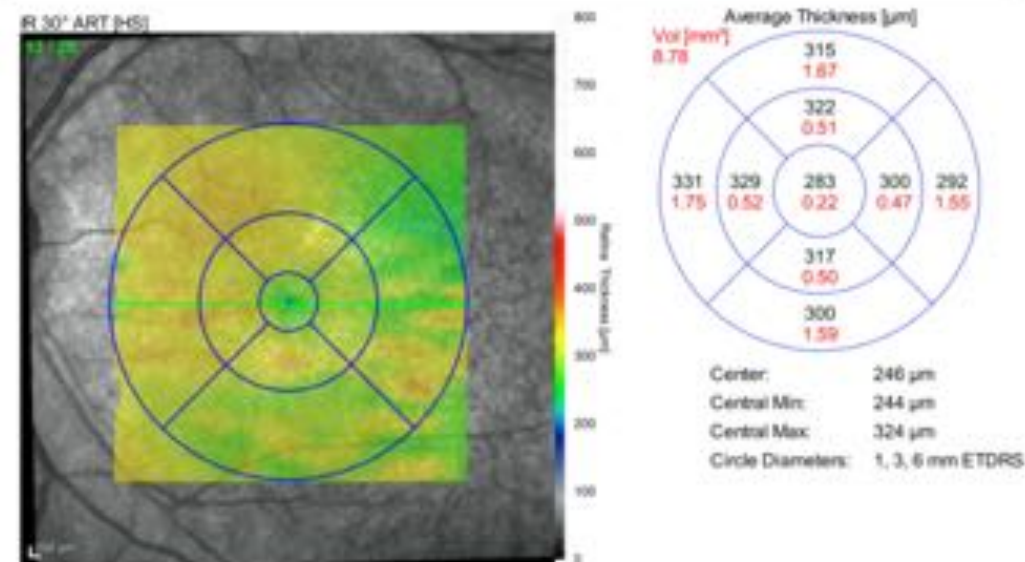
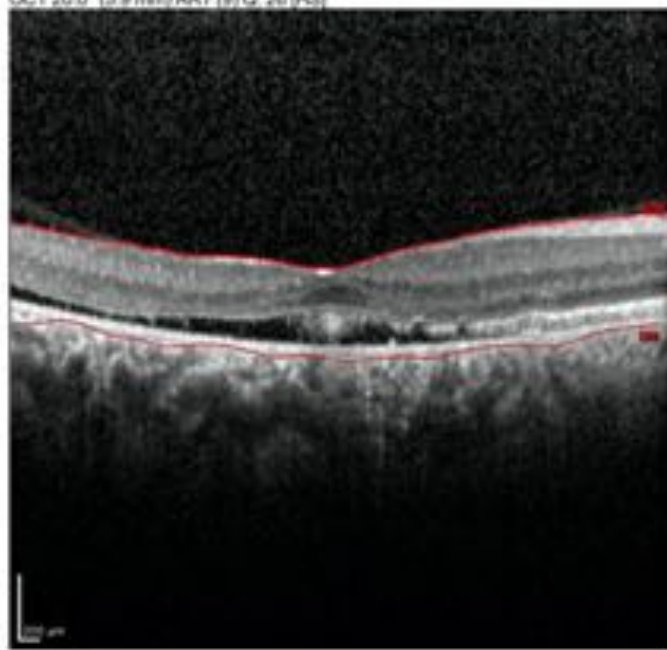




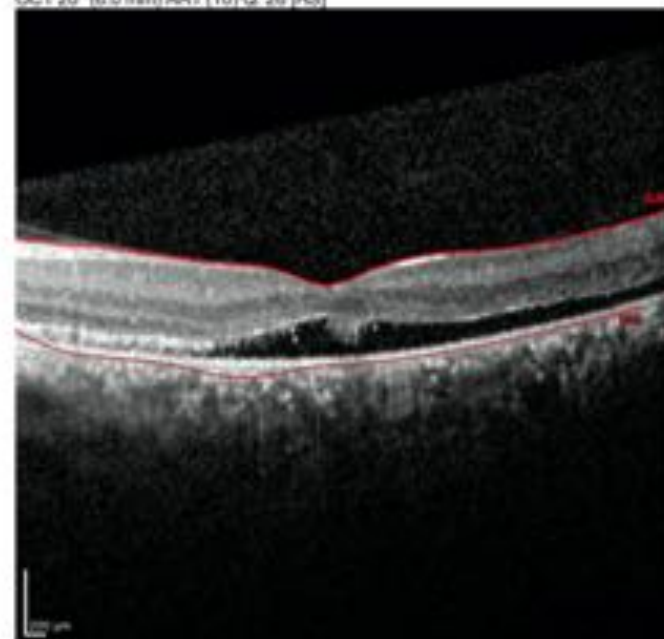
-
- Patient was treated with Spironolactone 25 mg BID
 - Little response so was increased to 50 mg BID
 - Lost to follow up for a few months
 - Had not used Spironolactone and did not see any benefit with or without
 - 10 months later VA 20/25 OD 20/30 OS



OCT 20.0° (5.9 mm) ART (9) Q: 26 (HS)



OCT 20° (6.0 mm) ART (10) Q: 26 (HS)



CASE 2

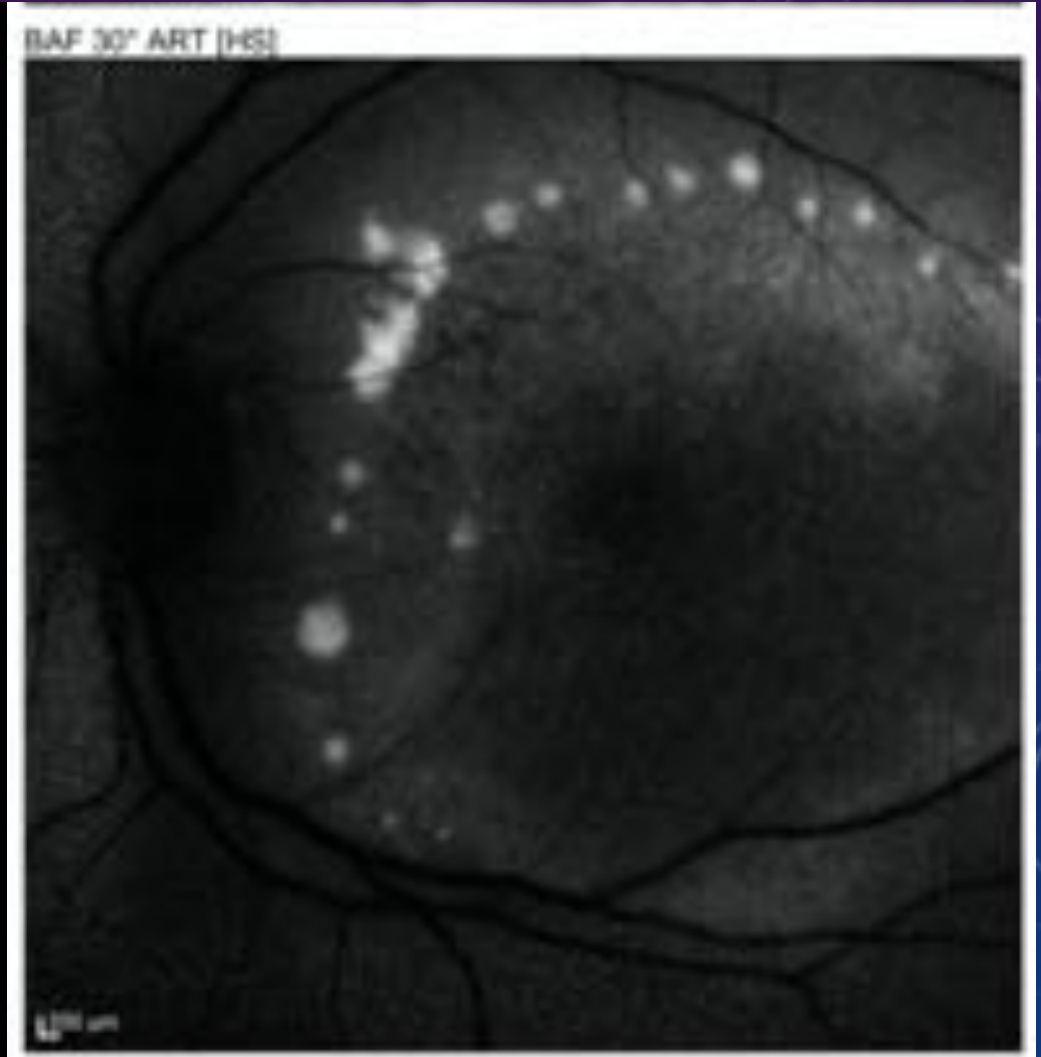
- 28 year-old Bangladeshi-American woman
- **Chief Complaint** poor vision in the left eye
- **Past medical history** negative
- **Medications** none
- **Past ocular history**
 - Trauma to right eye with NLP vision
 - Cataract extraction left eye
- No history of corticosteroid exposure

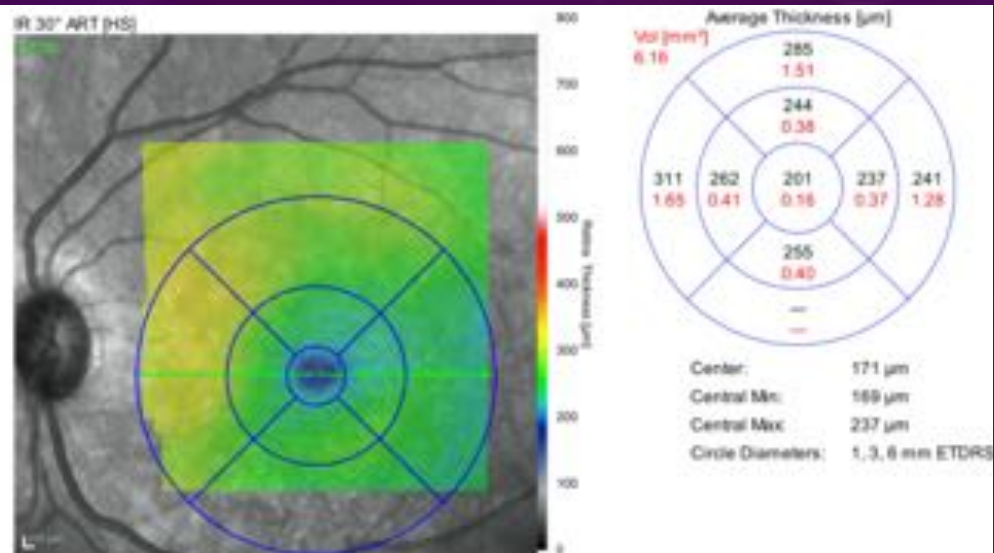
CASE 2

- Vision NLP OD 20/80 OS
- Anterior segment exam OD
 - Band keratopathy
 - Aphakia
 - IOP 46
- Anterior segment exam OS
 - Normal with well-placed PCIOL
- Posterior segment exam OD
 - Silicon oil
 - Attached retina with PRP scars and atrophic nerve

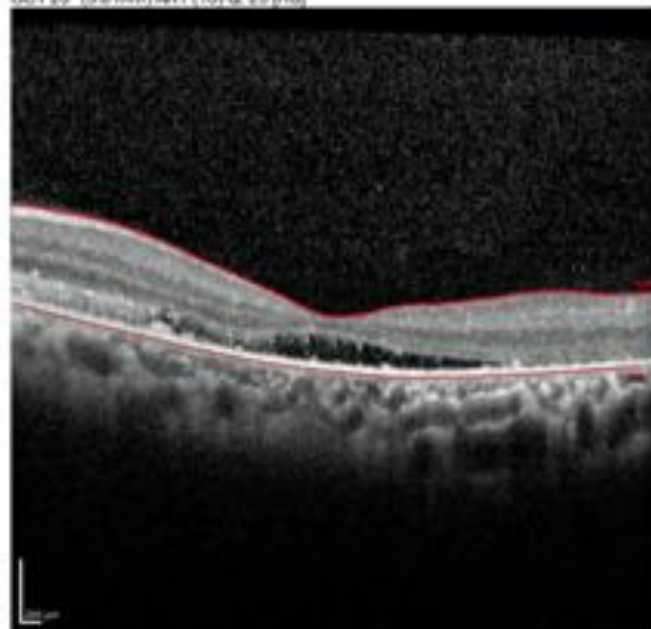


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2

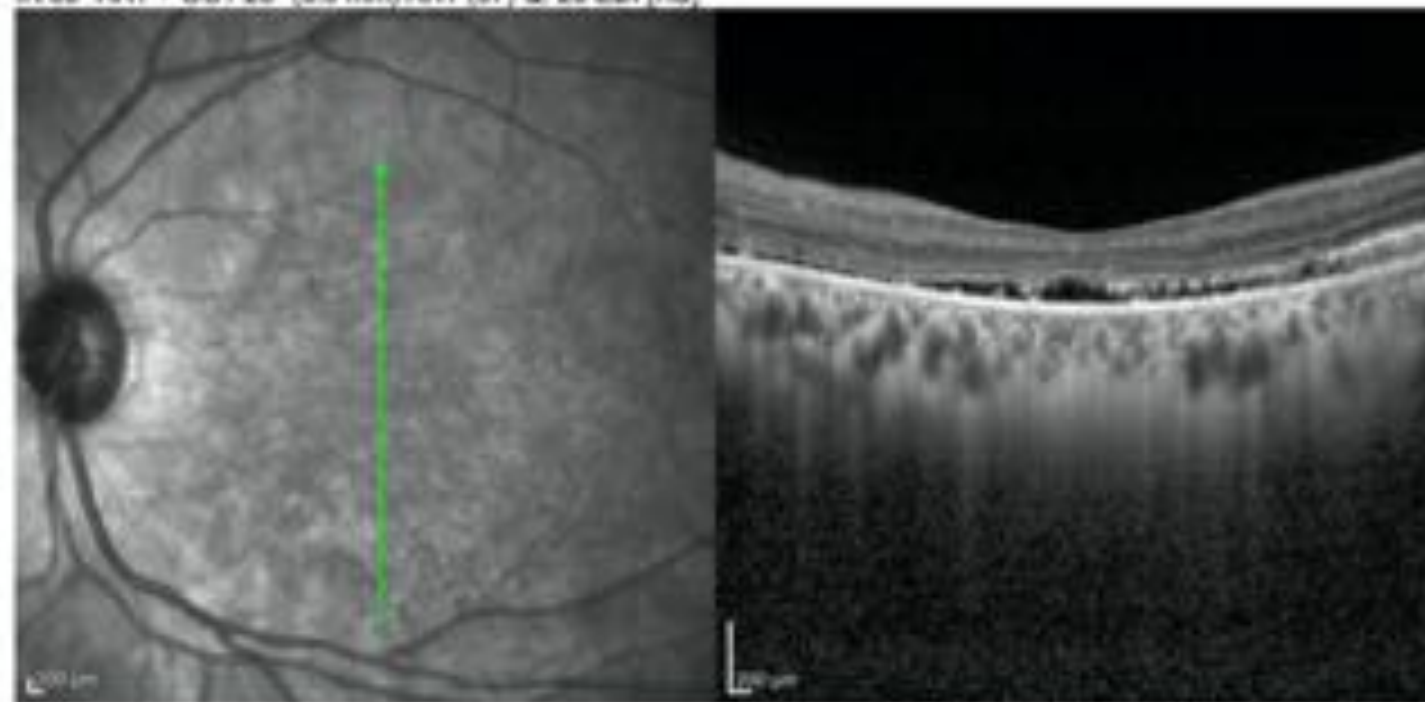




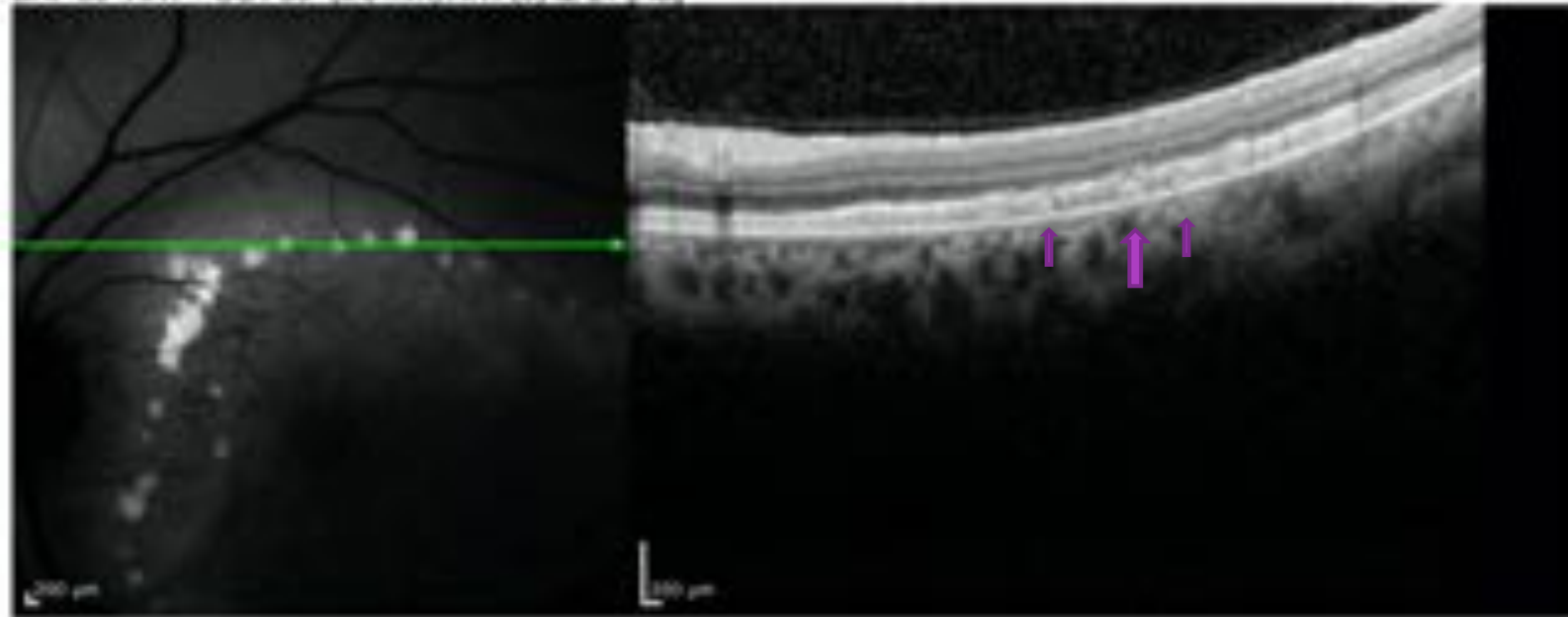
OCT 20° (5.8 mm) ART (10) Q: 25 [HS]



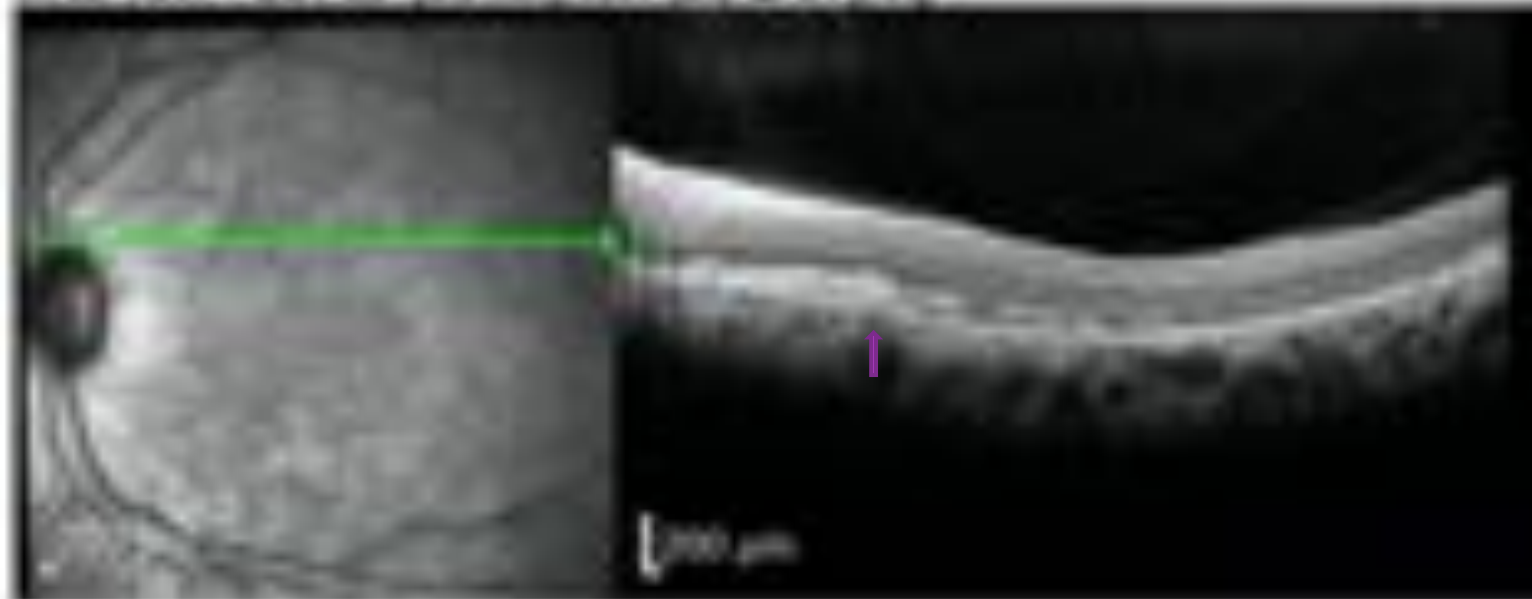
IR 30° ART + OCT 20° (5.9 mm) ART (97) Q: 28 EDI [HS]



BAF 30° ART + OCT 30° (9.0 mm) ART (6) Q: 27 [H5]



BAF 30° ART + OCT 30° (9.0 mm) ART (14) Q: 31 [H5]



00:25.646

MERGE



01:17.392

11/4/2019
11

early



mid

07:07.489

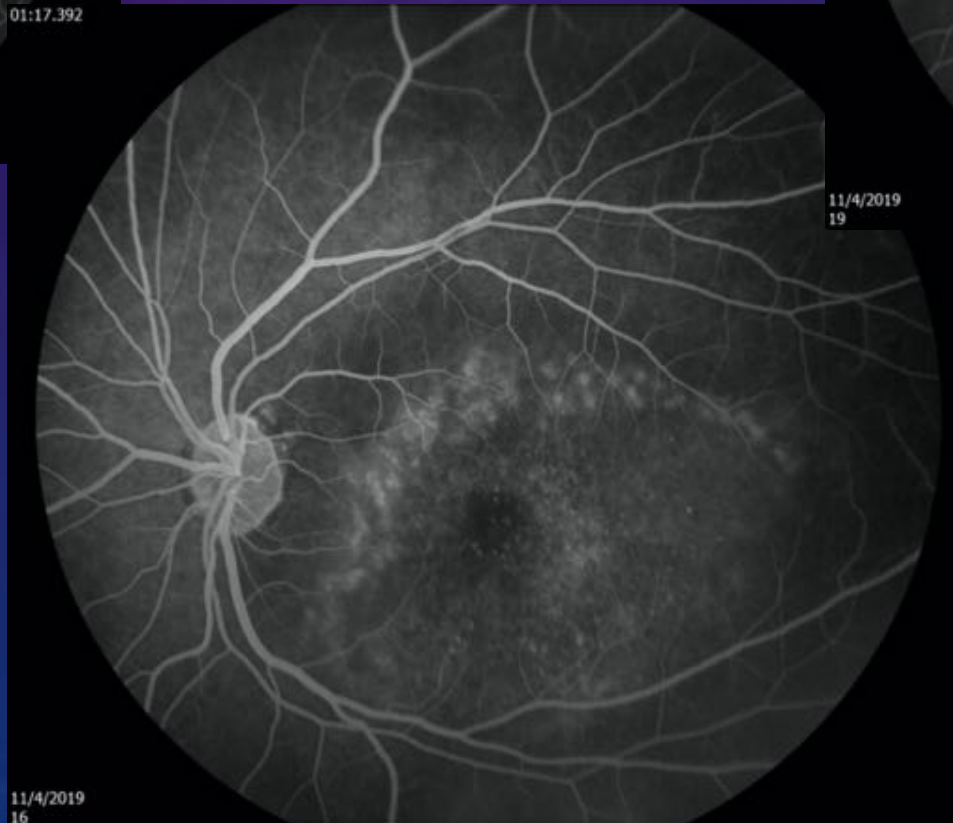
MERGE



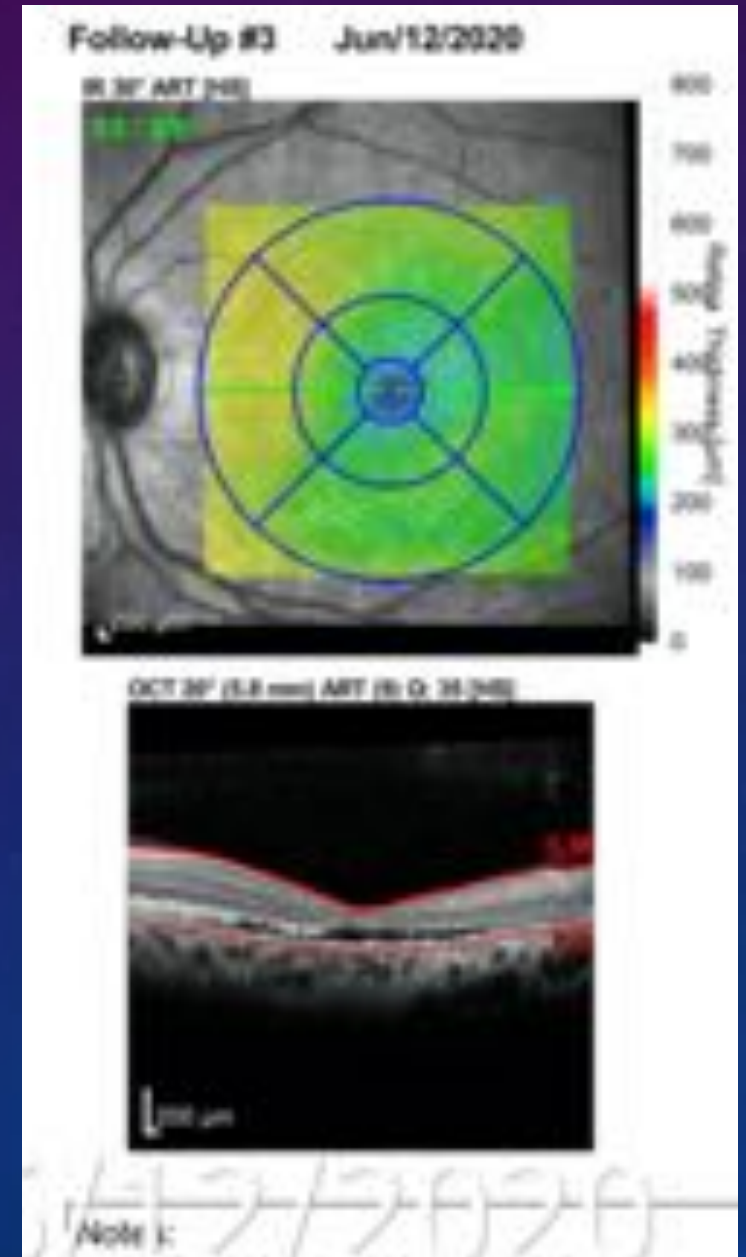
11/4/2019
19

late

11/4/2019
16



- Patient was treated with Spironolactone 25 mg BID
- Little response so was increased to 50 mg BID
- She did not notice much difference in vision
- 11 months later VA NLP OD 20/60 OS



PACHYDRUSEN

- Described by Spaide Retina 2018
- Greater than 125 microns in size
- Single or groups of few drusen scattered in posterior pole
- More likely to be seen in the peripheral macula or next to optic nerve
- Well defined borders with more complex shapes than soft drusen
- No focal hyperpigmentation on surface
- Associated with thicker choroids

PACHYCHOROID

- Possibly an absolute value of > 300 microns subfoveal choroidal thickness
- Attenuation of the choriocapillaris and Sattler's layer with the Haller's layer making up a higher than normal percentage of the choroidal thickness
- Enlargement of the luminal diameter of the vessels in Haller's layer with formation of "pachyvessels"

PACHYCHOROID SPECTRUM

- Pachychoroid pigment epitheliopathy (PPE)
- Central serous chorioretinopathy (CSC)
- Pachychoroid neovascularopathy (PNV)
- Polyploidal choroidal vasculopathy (PCV)

Fung Retina 2012

Baek Clin Exp Ophthalmol 2018

Bousquet Retina 2018

PACHYDRUSEN PER EYE

- Baek Clin Exp Ophthalmol 2018 **1.65 +/- 1.07**
- Singh Indian J Oph 2020 **2.1 +/- 1.5**
- Spaide Retina 2018 **“occur in isolation or in groups of only a few drusen”**

PACHYDRUSEN SEEN IN OLDER PATIENTS

- Baek Clin Exp Ophthalmol 2019 mean age 61.9
- Lee Retina 2019 mean age 66.7
- Singh Indian J Opth 2020 mean age 55.4
- Matsumoto Graefes 2019 61.4 +/- 11.7
- Fukuda Scientific Rep 2019 mean age 71

MORE PACHYDRUSEN SEEN LATER IN PACHYCHOROID SPECTRUM

- Singh IJO 2020 6.8% in CSC 14.1 % in PCV
- Baek Clin Exp Ophth 2018 Total number of pachydrusen significantly more in PCV patients than in any of the earlier pachychoroid spectrum disorders
- Sheth Eye 2020 41.67% of PPE, 60% CSC, 56% PCV

DIFFERENCES IN OUR PATIENTS

- At 28 year old and 32 year old much younger than usual pachydrusen patients
- The number of pachydrusen more numerous than usually reported
- Bilateral patient had remarkable symmetry between eyes
- Both patients had appearance of chronic CSC but without the gravitational tracts
- CSC can have pachydrusen but less likely

GENETIC MAY AFFECT VARYING PREVALENCE RATES OF PACHYDRUSEN

- Singh Indian J Ophth 2020
- Sheth Indian J Ophth 2020
- Fukuda Sci Rep 2019
- Saurabh Indian J Ophth 2020

VARIABLE CHOROIDAL THICKNESS WITH VARIABLE DISEASE EXPRESSION MAY BE HERITABLE

- Spaide Retina 2018
- Sardell Ophthalmology 2016
- Sohn Invest Ophthalmol Vis Sci 2014

CONCLUSION

Given the similar ethnic background in our two patients we hypothesize an environmental or genetic predisposition may have resulted in this particular phenotypic expression of pachydrusen

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