SUNY DOWNSTATE Medical Center

Grand Rounds

Matthew Gorski, MD
SUNY Downstate Medical Center
January 10, 2013

History

- 48 yo Hispanic man presents complaining of blurry vision OD>OS for 1 month

- Patient denies pain, nyctalopia, photophobia, metamorphopsia, flashes, floaters, trauma. No previous similar incidents. Review of systems unremarkable.

- PMH:
  - Systemic Lupus since childhood
  - ESRD s/p Renal Transplant 1993- 2003, 2012- present
  - History of hemodialysis 80s - '93, '03- 5/2012
  - Hypertension
  - Gout

History

- POHx: Cataract Extraction with PCIOL OU, 2003
  Gtts: none
- Fam Hx: denies glaucoma/blindness/ocular disease
- Social: denies EtOH, smoking, drugs
- All: NKDA

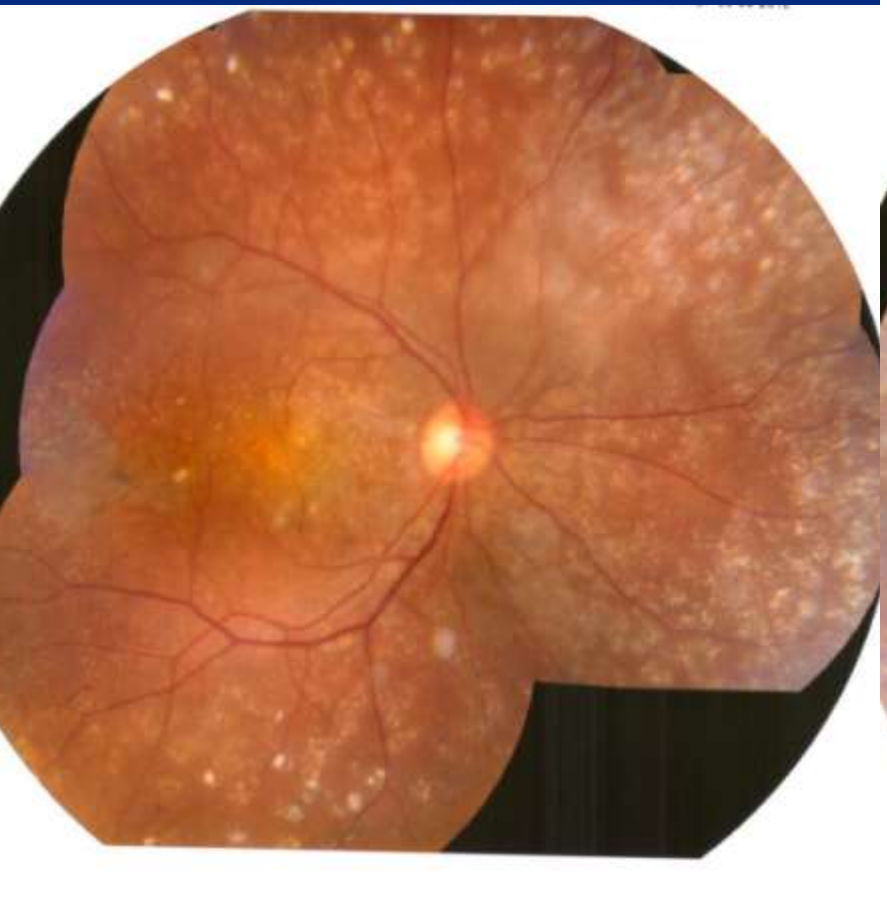
- Medications
  - Prednisone, Prograf, Sensipar, Metoprolol, Norvasc, Valcyte, Bactrim, Cellcept

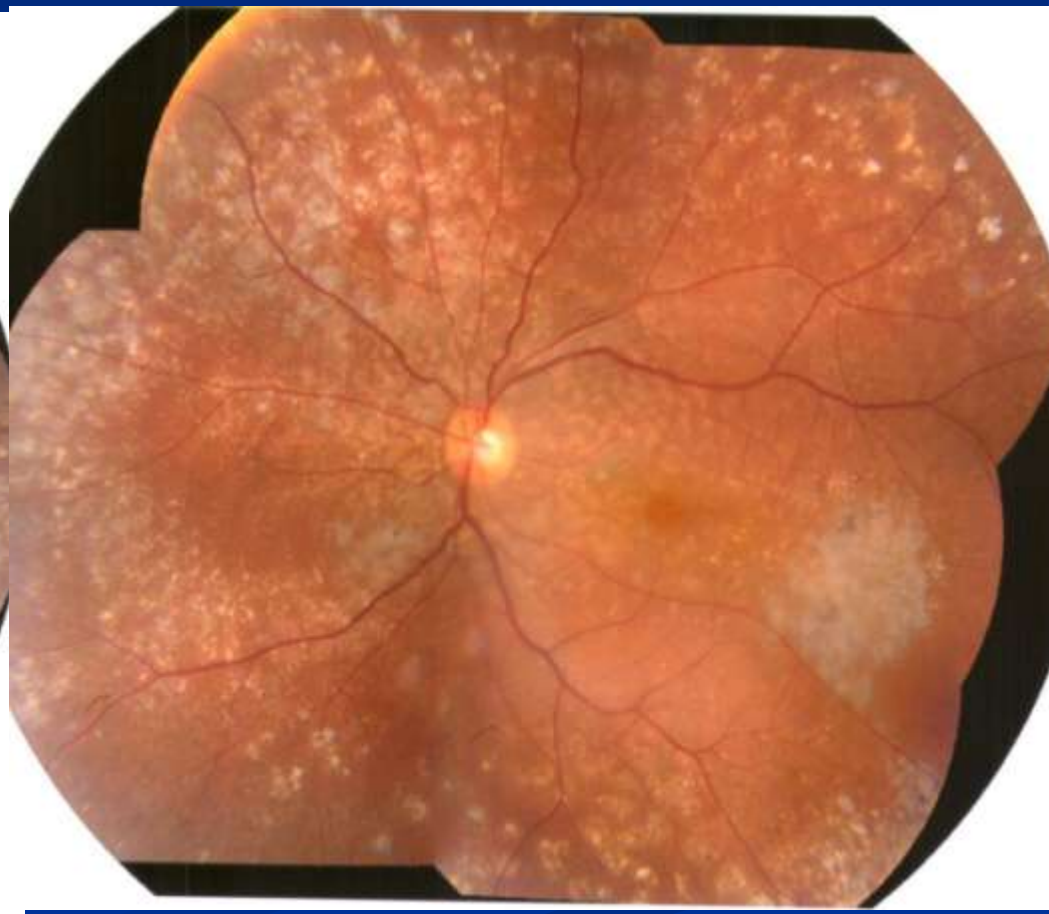
Examination

- VAcc: CF OD, 20/50 OS
- EOM: Full OU, no nystagmus OU
- P: 5→3 OU, no APD OU
- Tapp: 16 OU
- Color: 4/14 OD, 14/14 OS

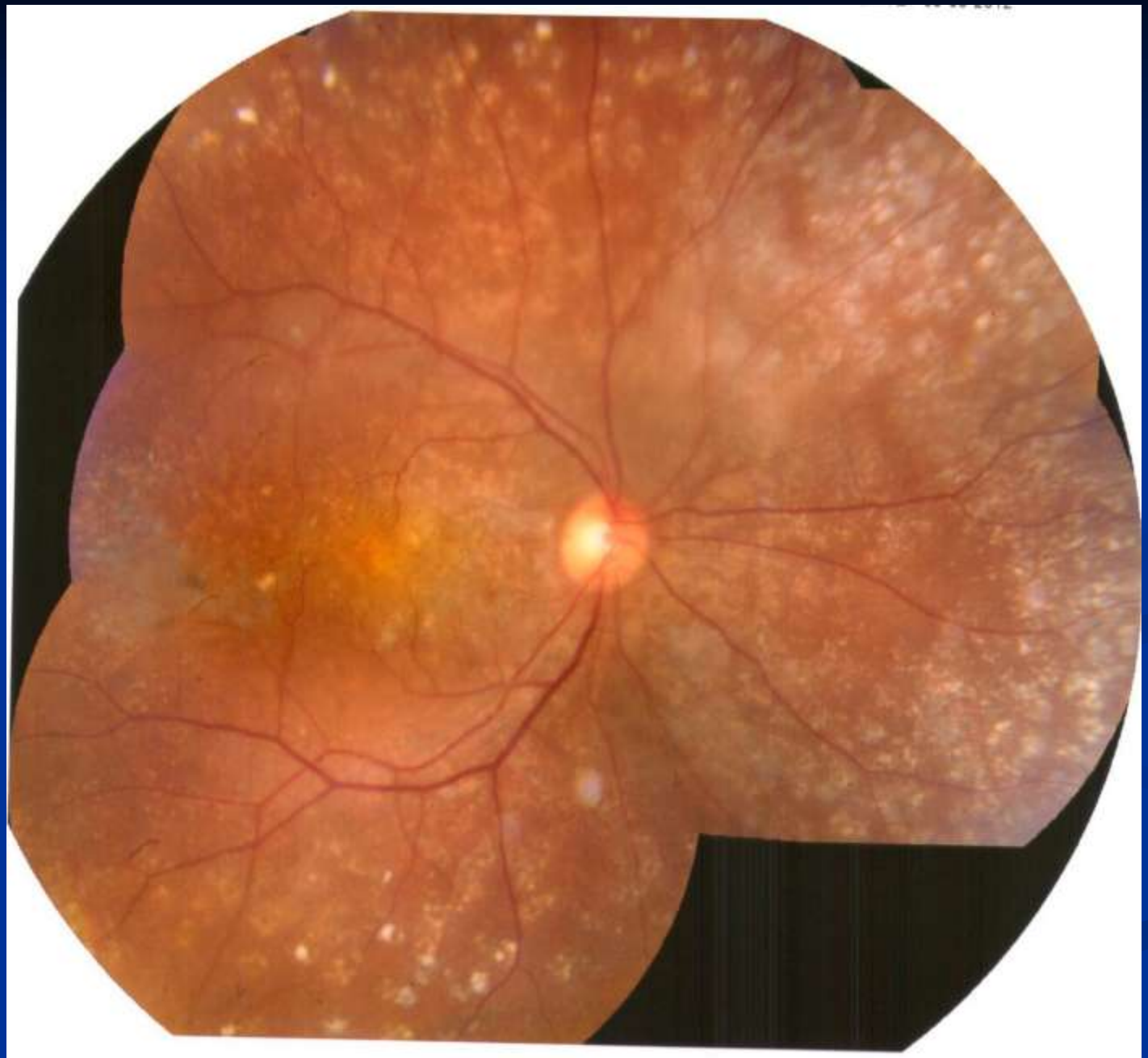
SLE

- Unremarkable

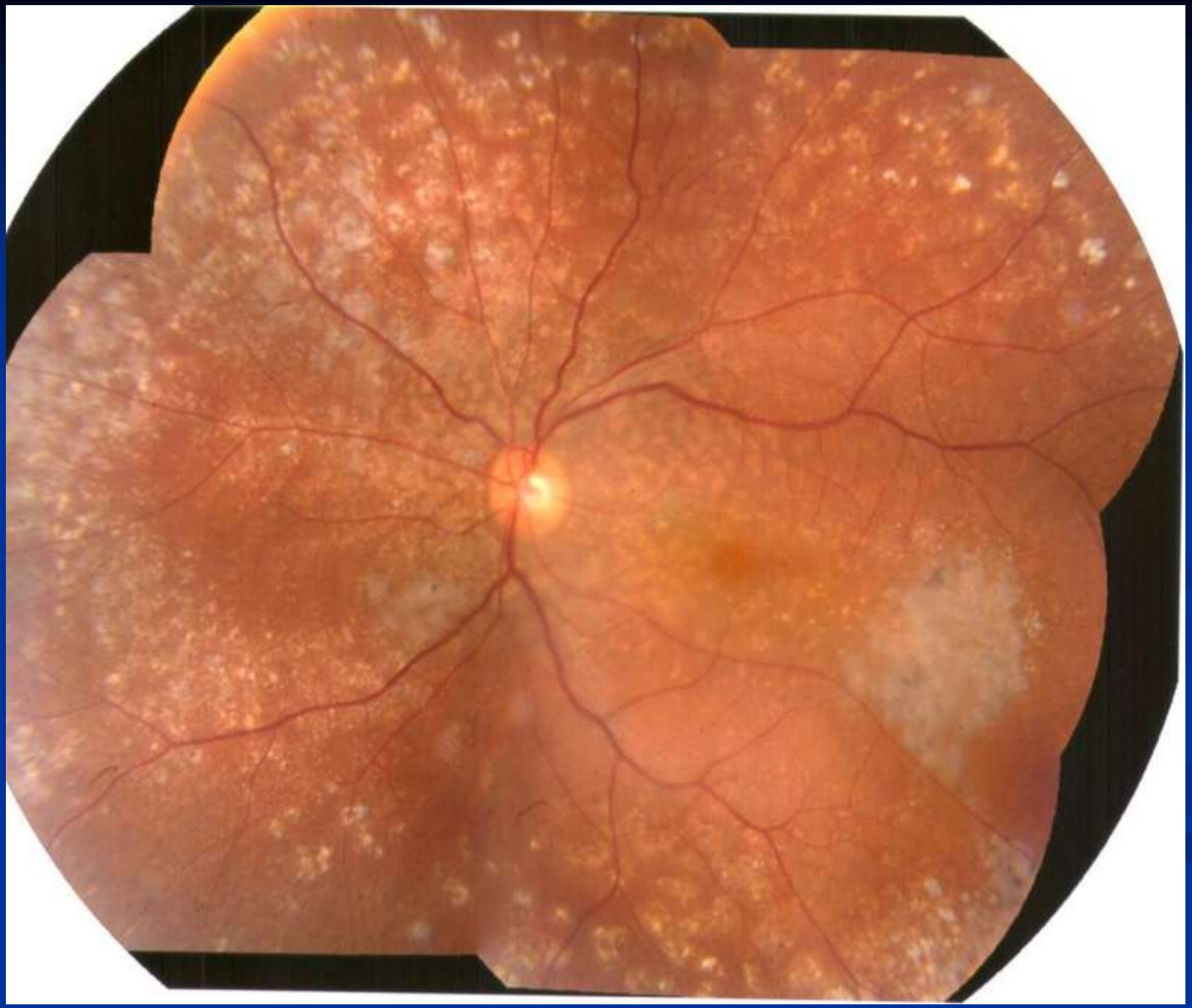


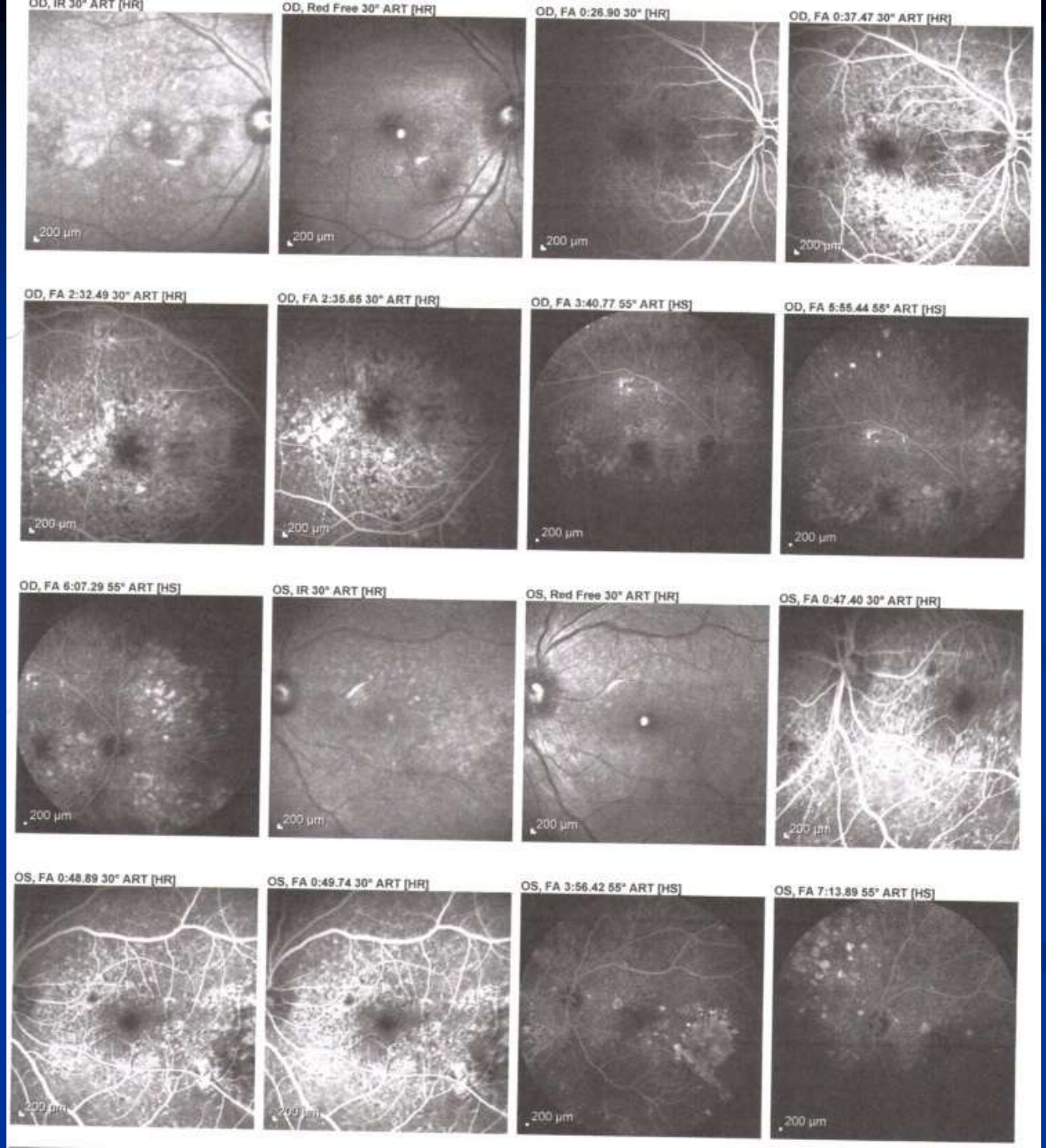


Patient care



Patient care





Patient care

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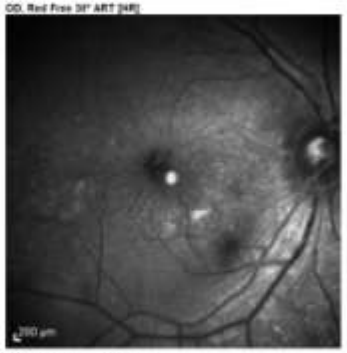
HEIDELBERG ENGINEERING

Sex: M

OD, Red Free 30° ART [HR]



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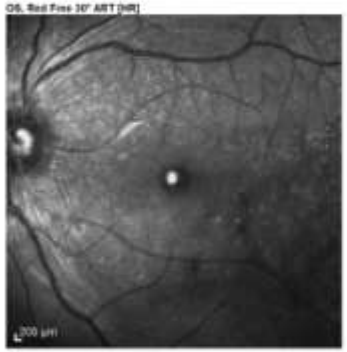

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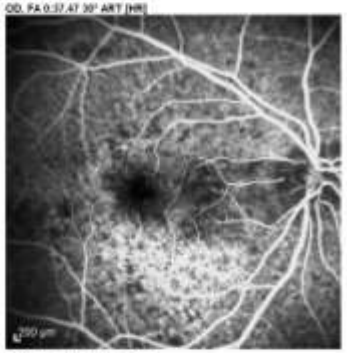

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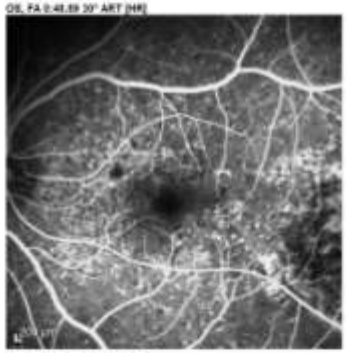

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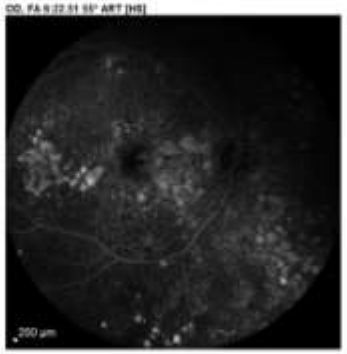

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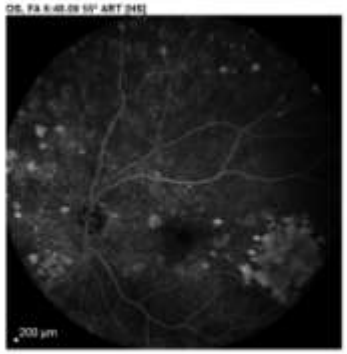

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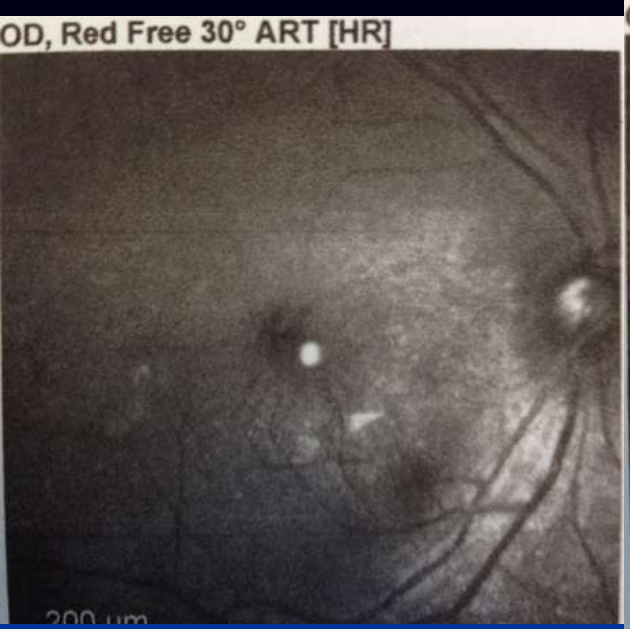
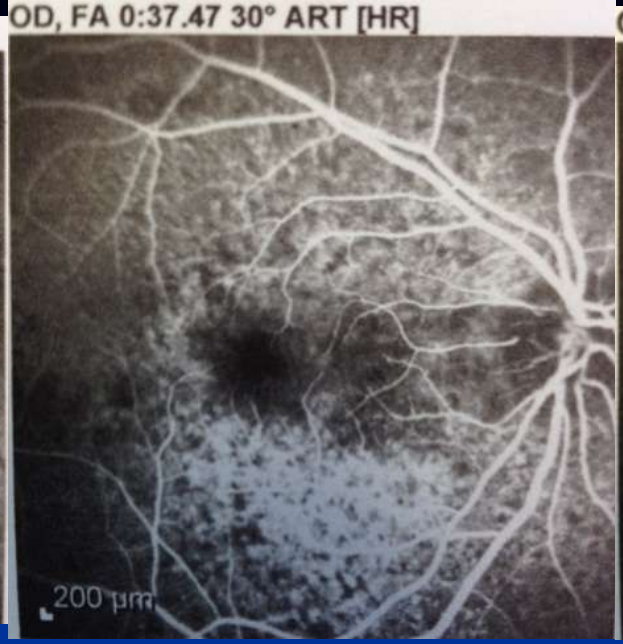
Date: 9/5/2012 Signature:


OD, Red Free 30° ART [HR]
200 µm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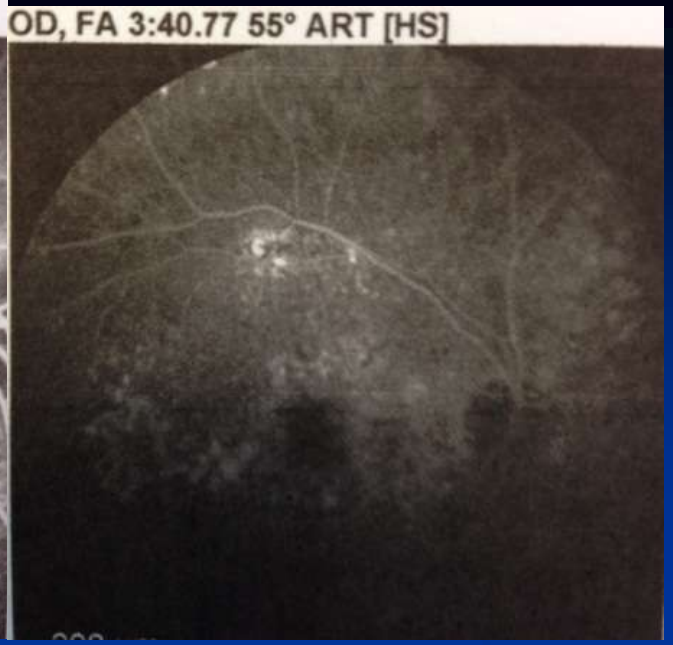
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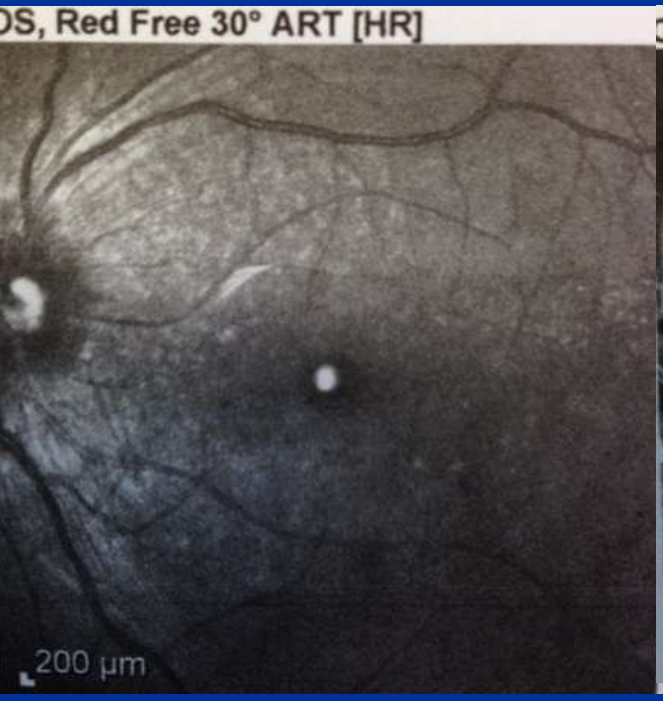
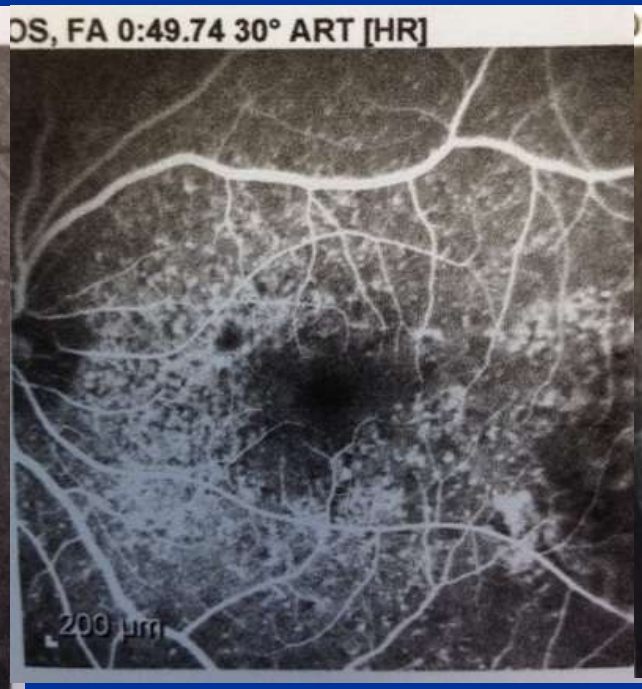

OS, Red Free 30° ART [HR]
200 µm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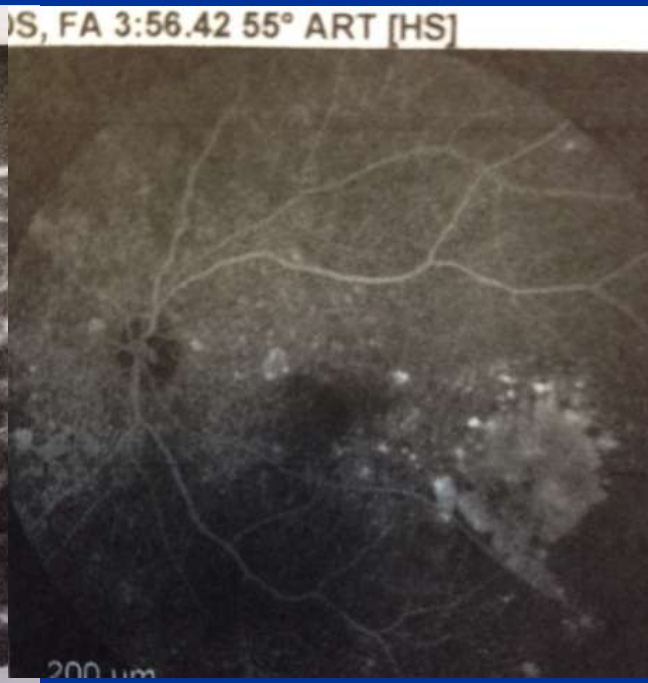
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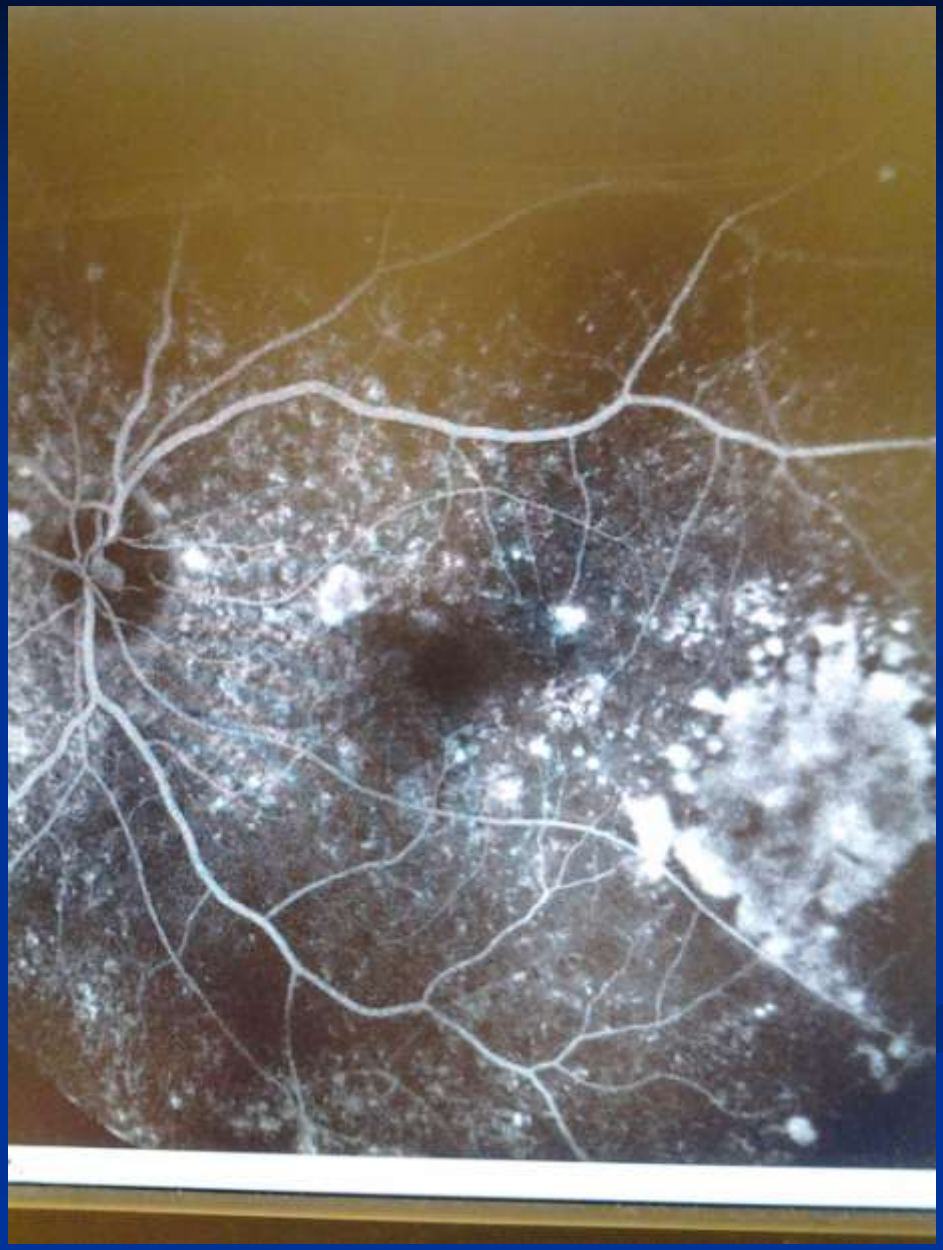
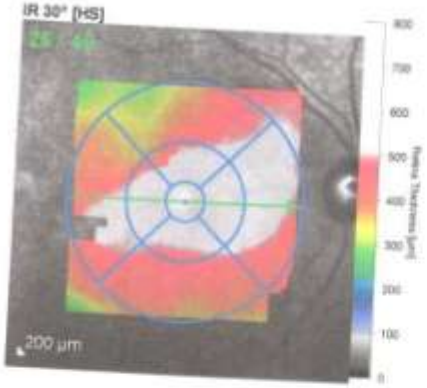


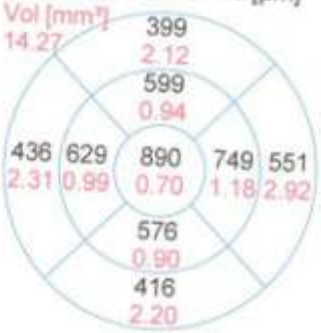
Patient care

Comment: ---

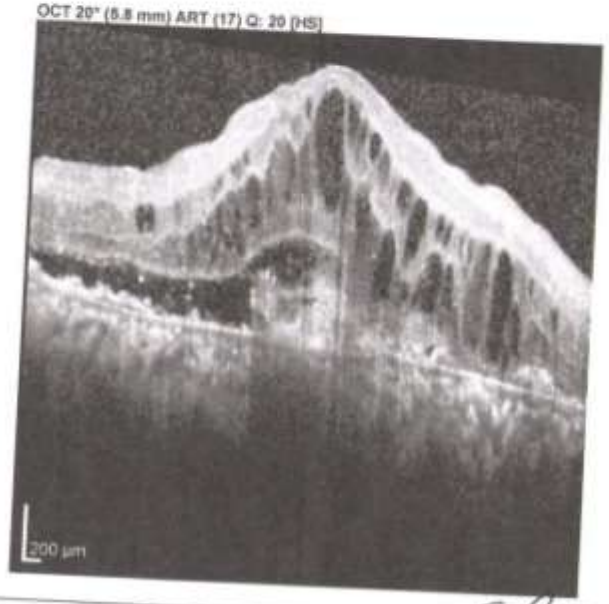
OD



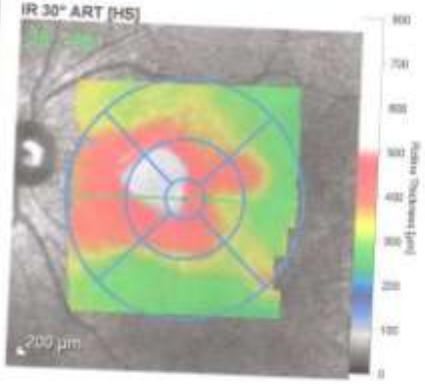
Average Thickness [µm]



Center:	945 µm
Central Min:	754 µm
Central Max:	972 µm
Circle Diameters:	1, 3, 6 mm ETDRS



OS



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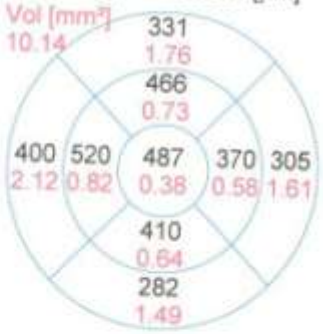
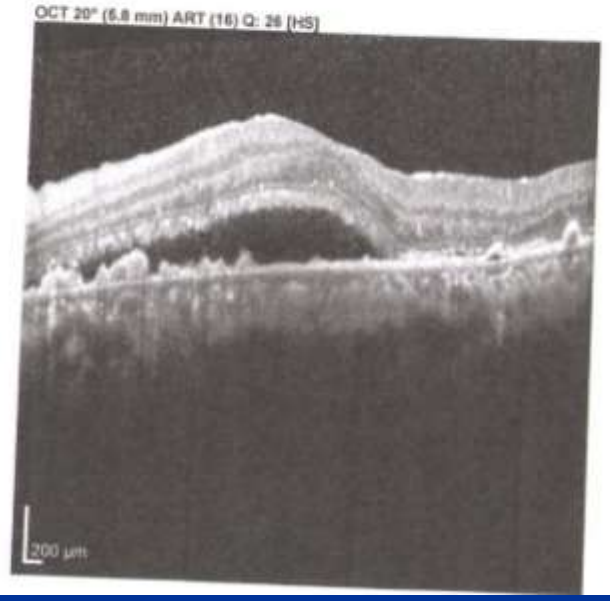


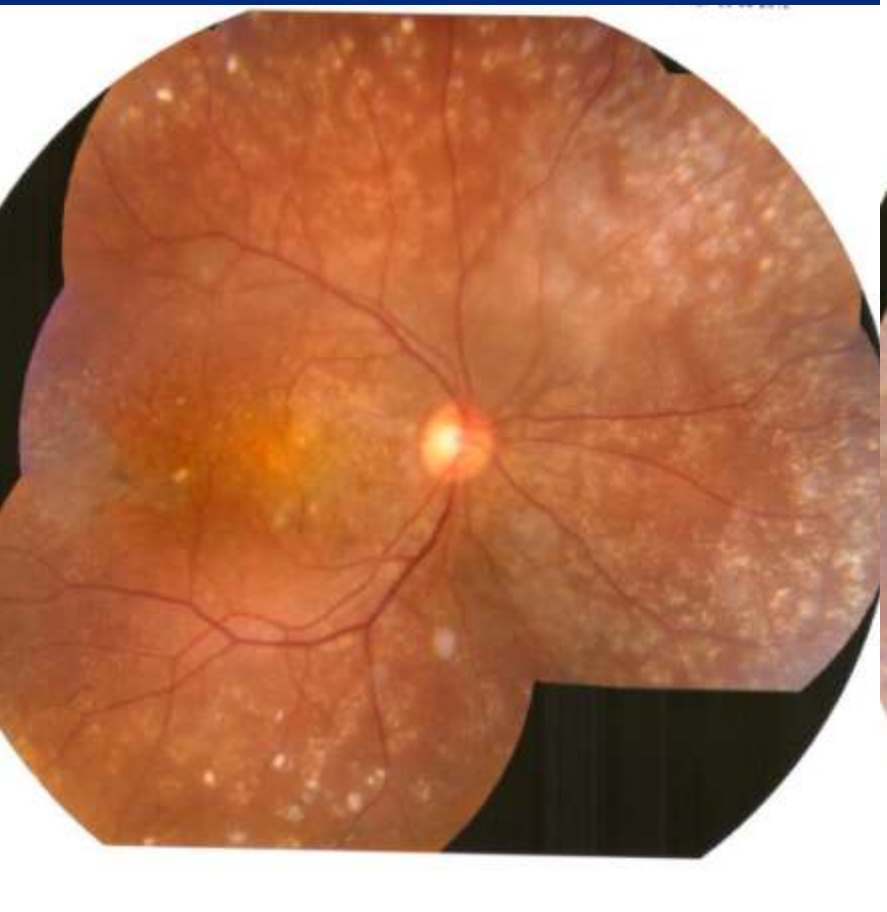
Center:	489 µm
Central Min:	393 µm
Central Max:	578 µm
Circle Diameters:	1, 3, 6 mm ETDRS

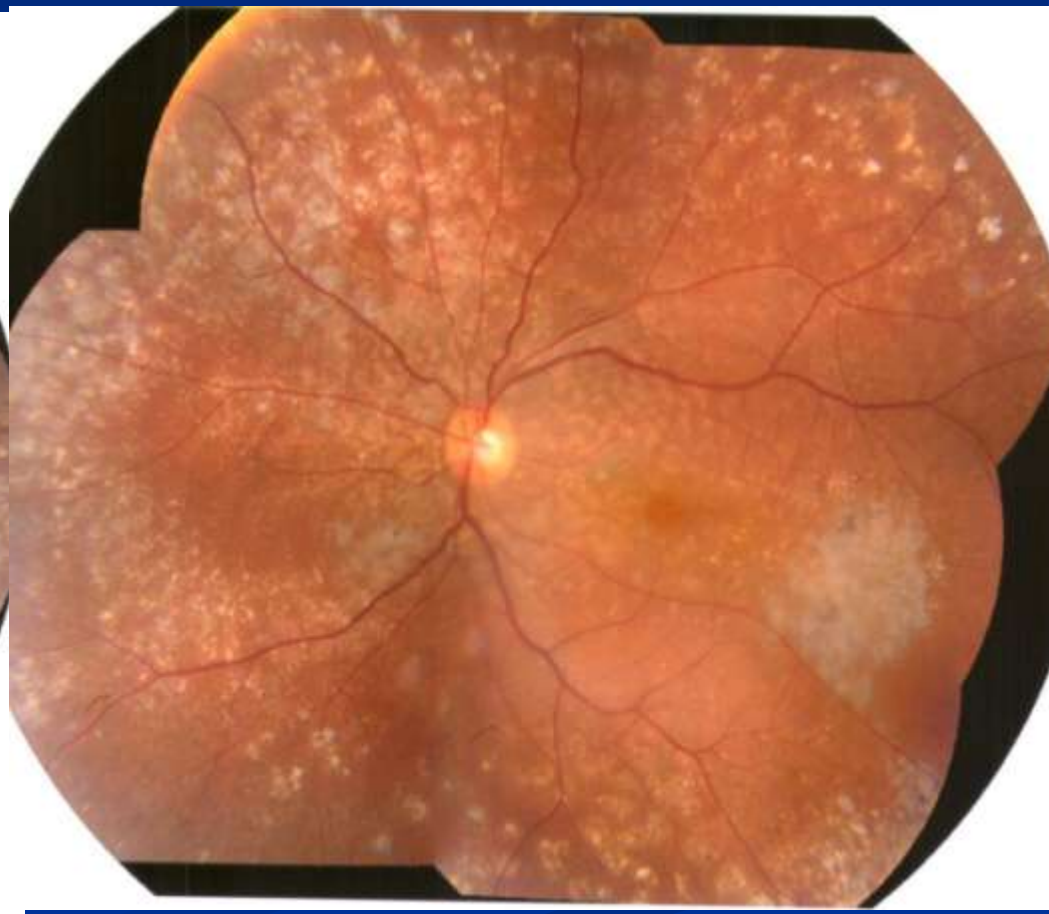


otes:

More History…

- In 2003, dilated exam pre-cataract extraction, reveals, " diffuse subretinal drusen OU…follow-up retina after cataract surgery"
- s/p CE/PCIOL OU→ VAsc 20/20 OU in 2004.
- No renal biopsy performed
- No neurological symptoms
- No nyctalopia





Patient care

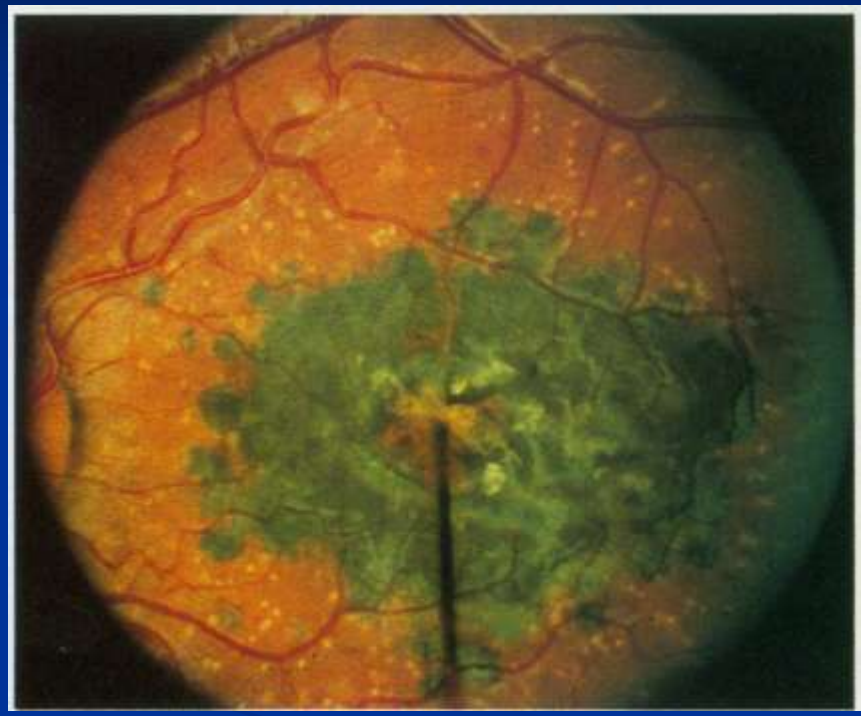
Differential Diagnosis

48 yo M history of Lupus, multiple renal transplants, most recently in May 2012, c/o bilateral blurry vision X 1 month, with bilateral CME and chronic peripheral sub-retinal changes

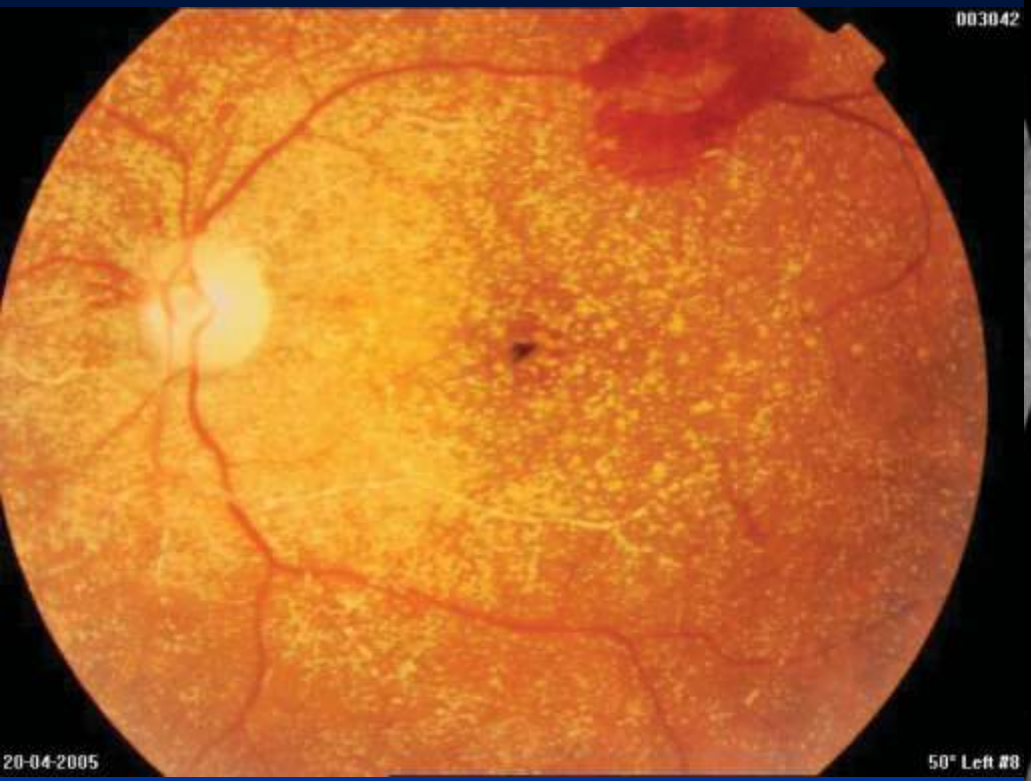
- Crystalline retinopathy
  - Oxalosis
  - Medication related (tamoxifen, canthaxanthine, etc…)
  - Bietti crystalline dystrophy
- ARMD (atypical)
- Renal-related Retinopathy (cilioretinopathies, Alport's, cystinosis, medication)
- Retinitis Pigmentosa/CSNB spectrum disorder (Retinitis punctata albescens, Kandori)
- White Dot Syndromes
- Fleck Dystrophies
- Basal Laminar Drusen vs Calcified Drusen
- Hypertensive retinopathy
- Familial Drusen (Malattia Leventinese)

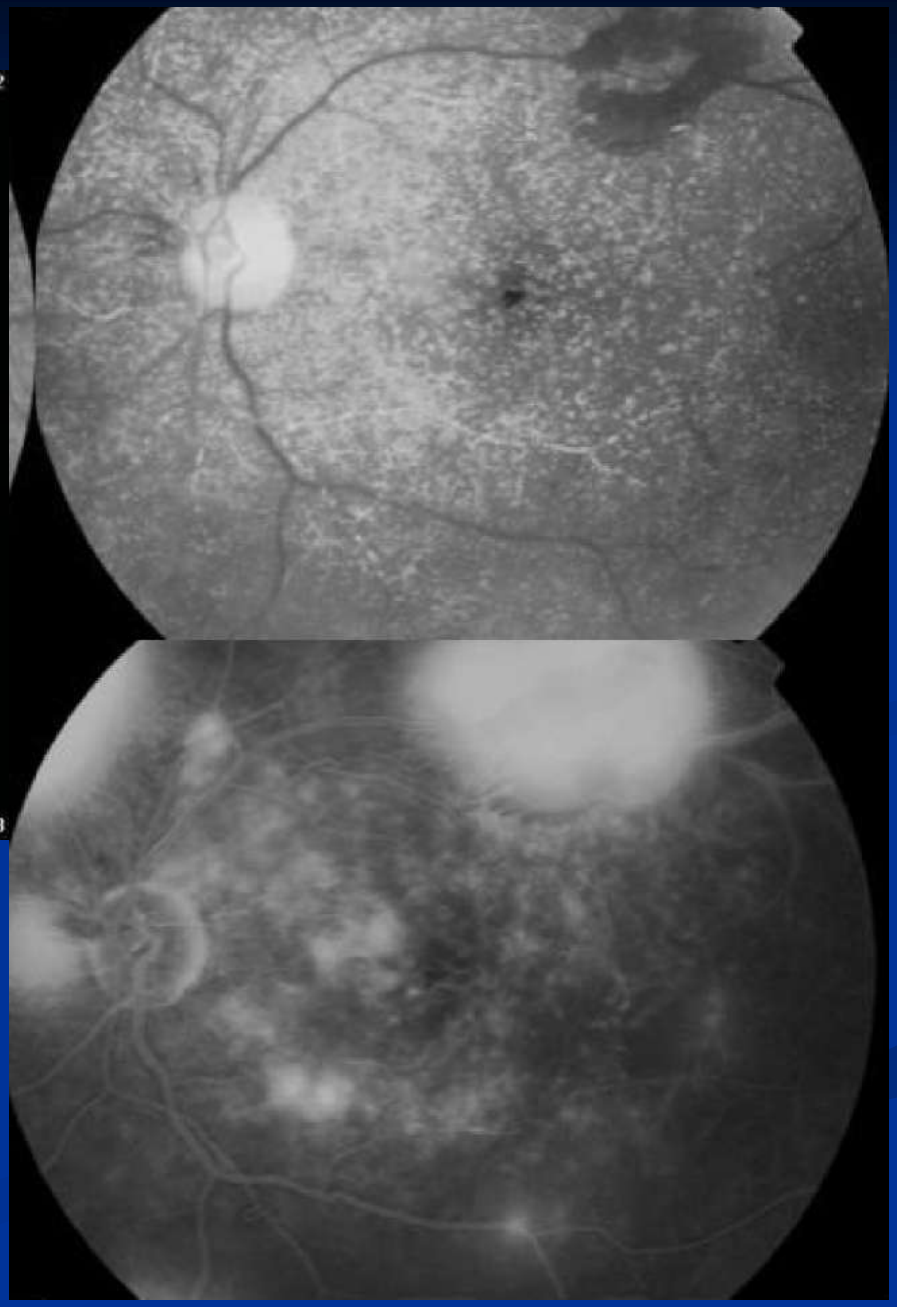
Primary Hyperoxaluria

- Incidence:1 in one million; AR
- Multiple types
- Presents age 2 months to 18 years
- Enzyme deficiency of alanine glyoxylate-Aminotransferase
- Deposition of oxalate crystals in RPE, ON (30% incidence)
- Signs: yellow-white iridescent flecks, large geographic atrophy, ON atrophy, RPE clumps, small, subretinal black ringlets, rare macular edema, ? CNV, ERG: diminished
- Prognosis: VA: 20/20 to CF
- Renal Transplant as child (usually)



Primary Hyperoxaluria in 31 yo VA 20/20
(Small, KW, 1990)





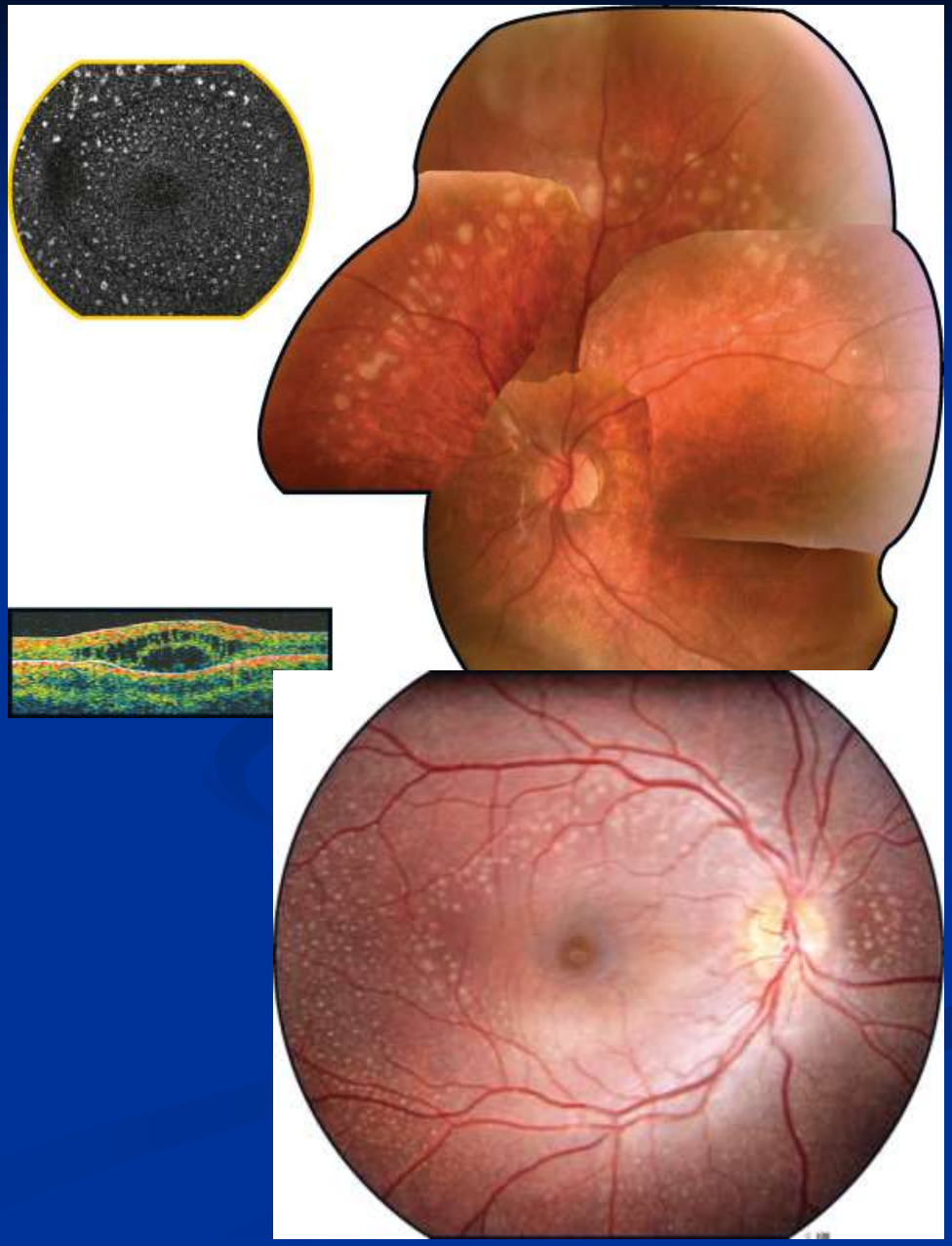
Regressive course of oxalate deposition in primary hyperoxaluria after kidney transplantation

- **+ CNV**

(Celik, G et al, *Renal Failure*. 2010)

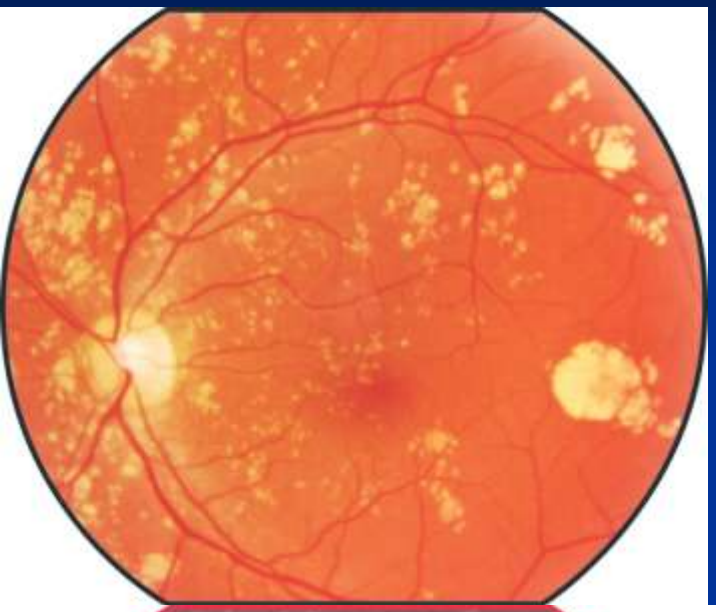
Retinitis Punctata Albescens

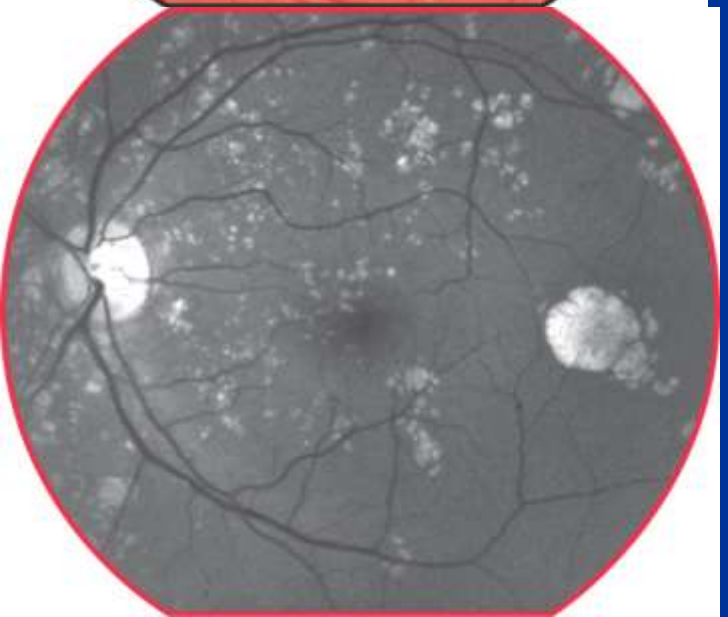
- Form of Retinitis Pigmentosa
- Autosomal Recessive
- Progressive, severe nyctalopia and peripheral visual field loss (as opposed to fundus albipunctatus)
- Narrowed retinal vessels
- Small to large grey-white flecks not reaching periphery
- +/- RPE pigment clumping
- ERG: Severely Depressed



(Genead MA, Ophthalmic Genetics, 2010) Medical Knowledge (Yannuzzi, The Retinal Atlas, 2010)

Flecked Retina of Kandori





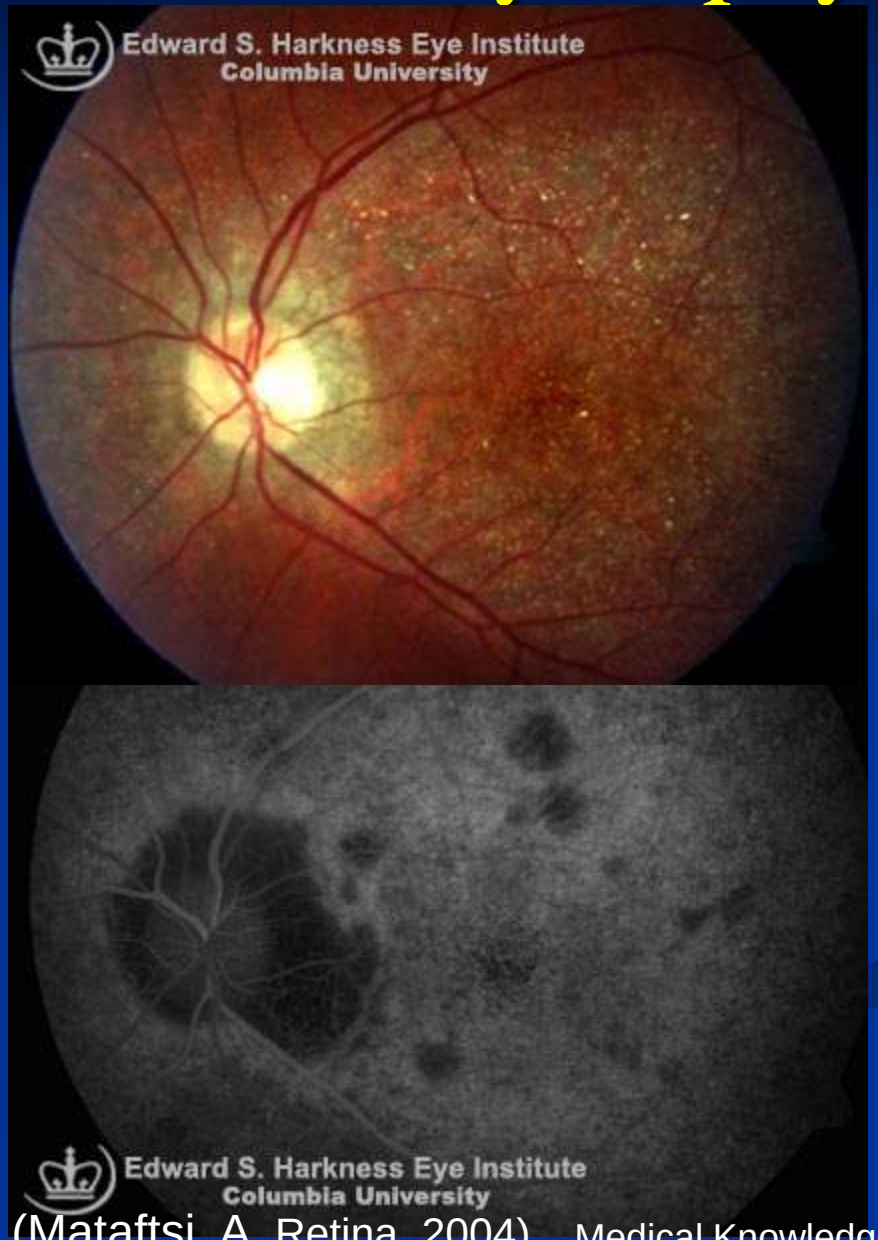
-Rare, AR
- less severe, often asymptomatic, but non-progressive night blindness;
- mild when compared to other forms of congenital stationary night blindness
- sharply defined irregularly shaped, often large flecks, that largely spare macular region; associated with RPE atrophy
- EOG/ERG: WNL

(Agarwal, Gass' Atlas of Macular Diseases, 2012)

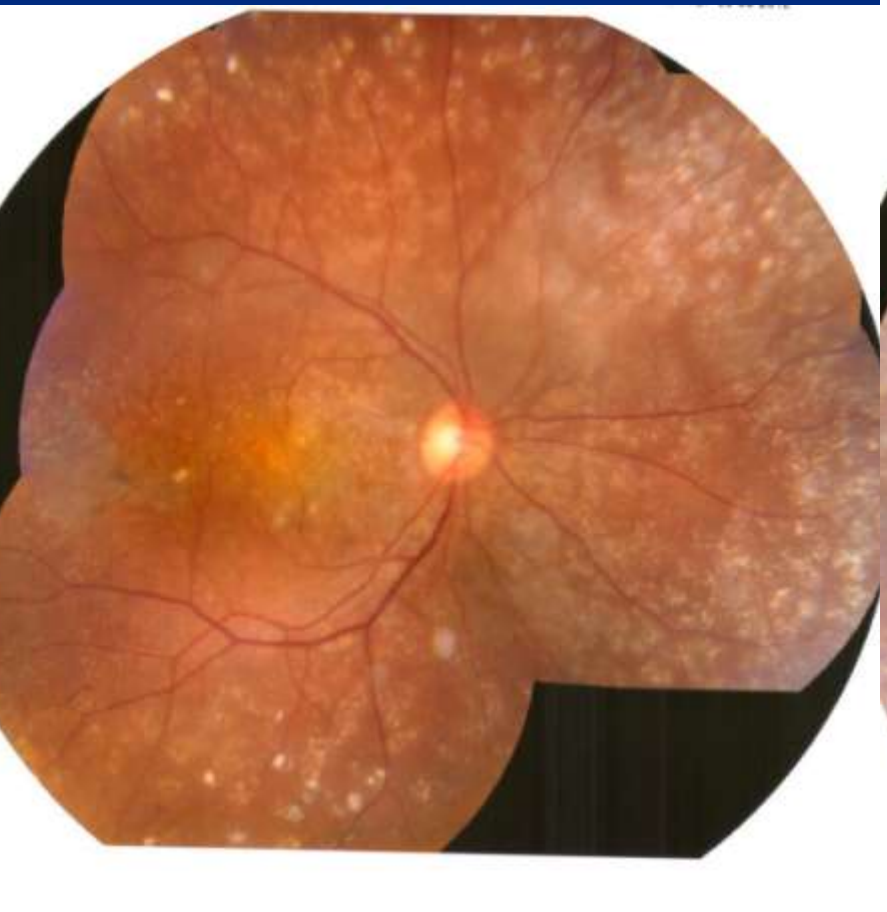
Yannuzzi, The Retinal Atlas, 2010 Medical Knowledge

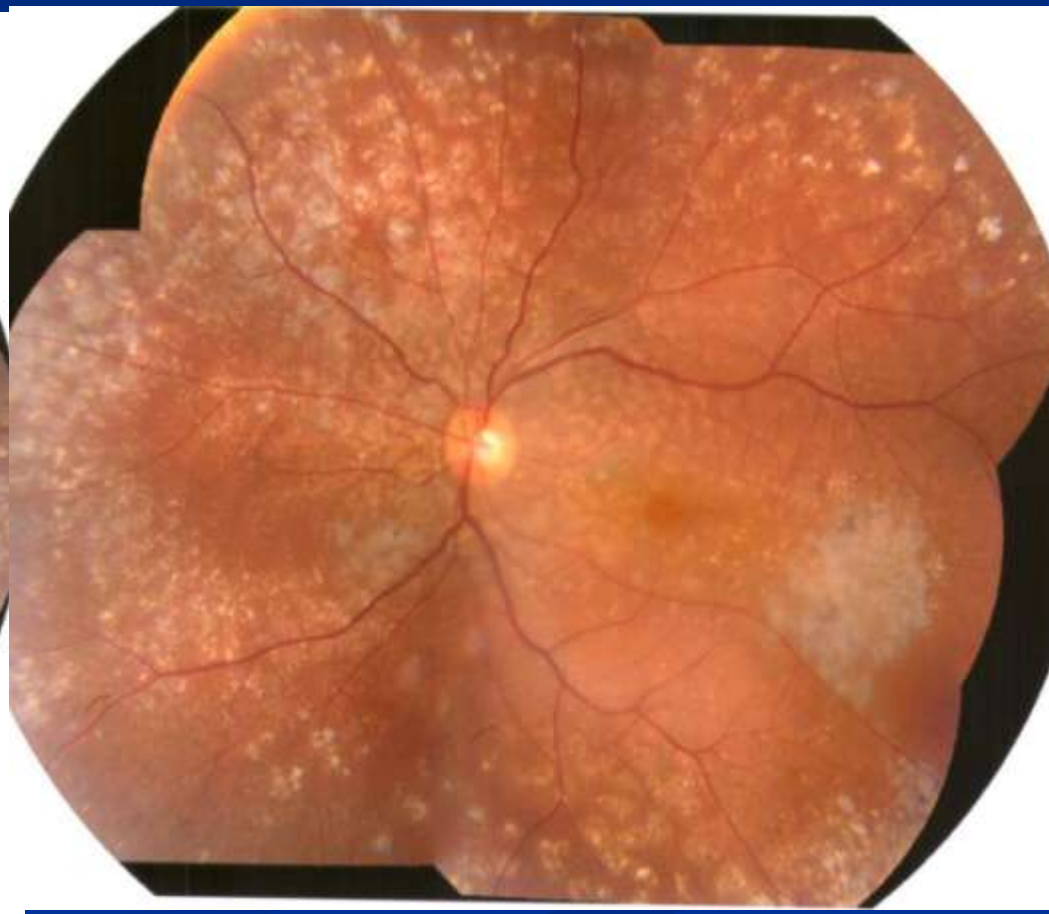
Bietti Crystalline Corneoretinal Dystrophy

- Rare, AR, most common in Asians
- Symptoms: nyctalopia, paracentral scotomas, blurry vision
- Avg. age of onset of symptoms: 33
- Signs: Diffuse, intraretinal, glittering, yellow, crystalline deposits
  - Conjunctival-keratic deposits near limbus
  - Geographic atrophy, RPE clumps
- FA: chorioretinal atrophy with patch preservation of RPE and choriocapillaris; blockage by crystals
- ERG: diminished, EOG: very diminished (preceding symptoms)

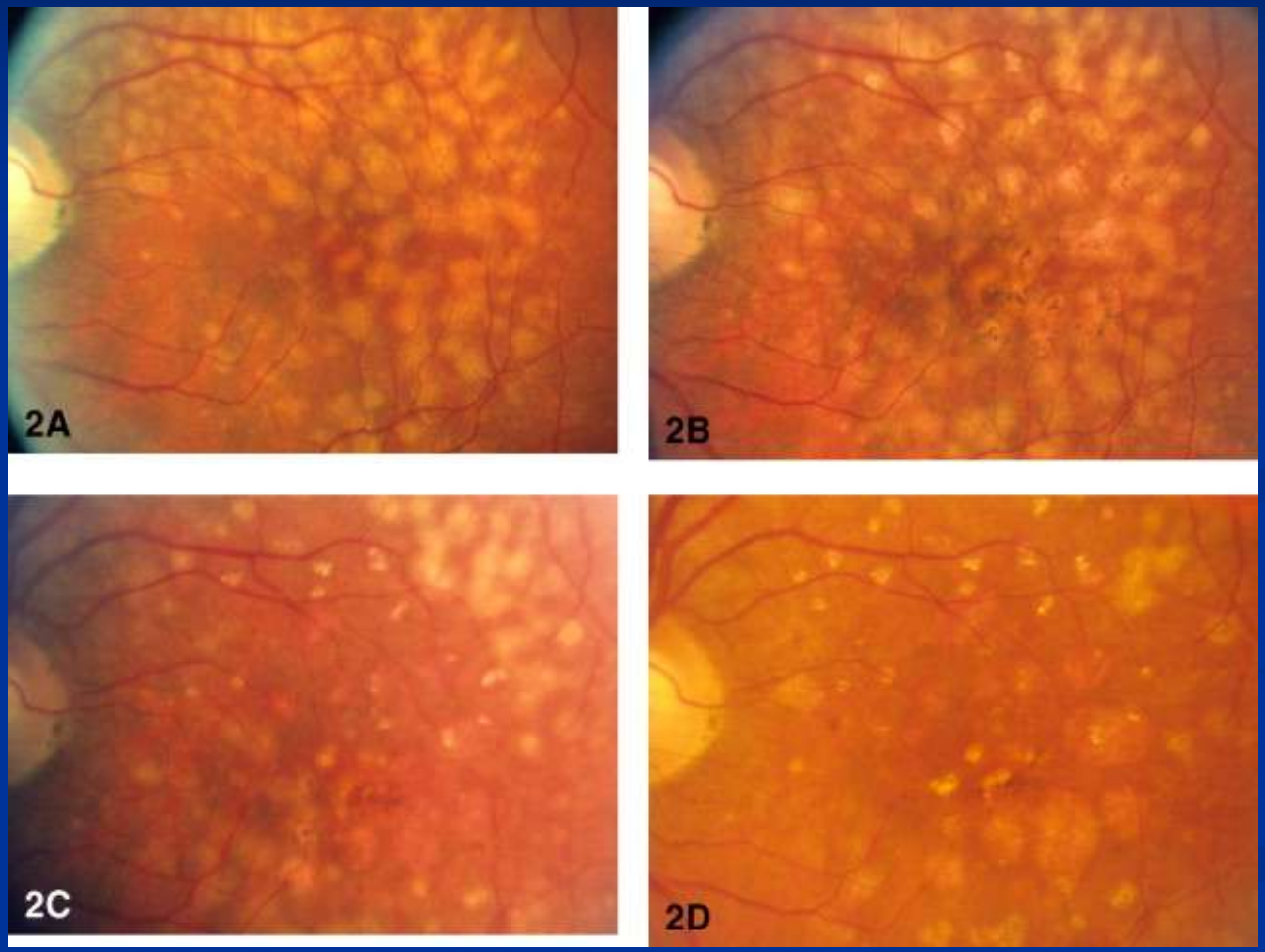


(Mataftsi, A, Retina, 2004)



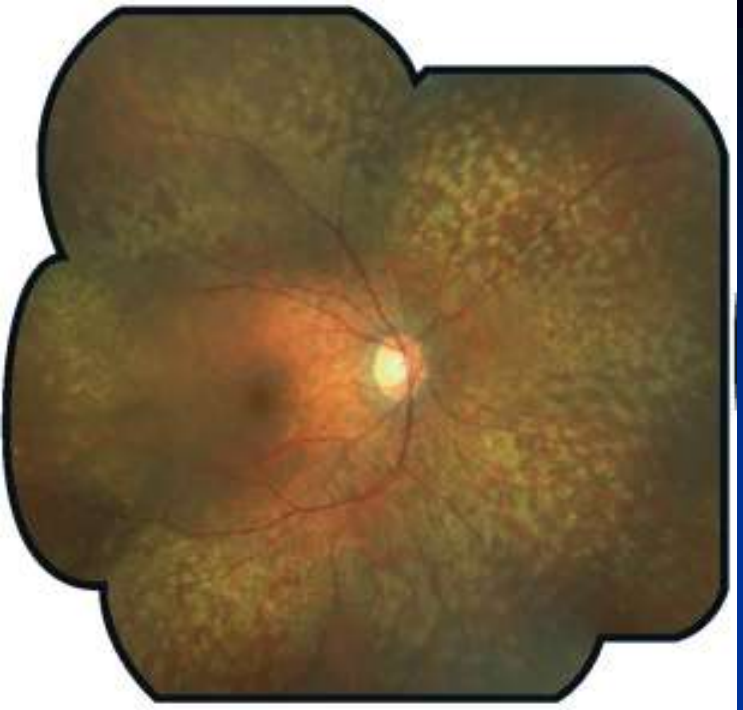


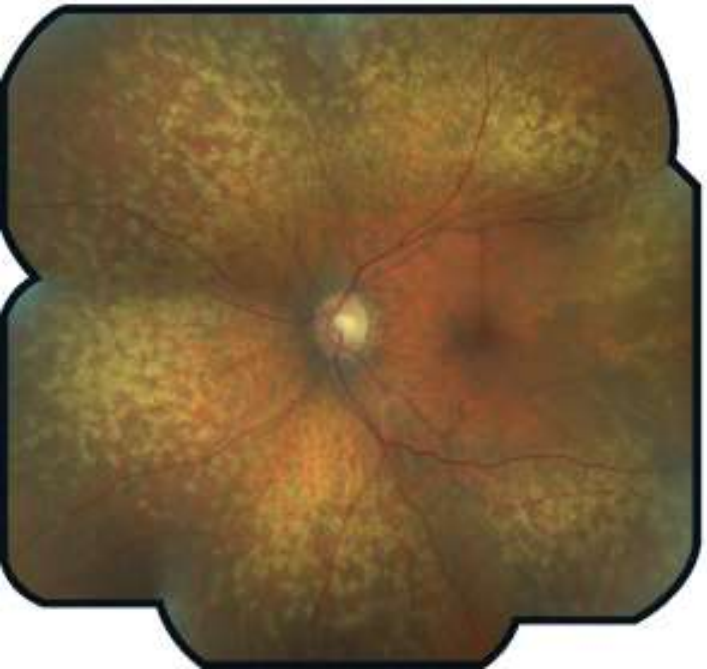
Calcified Drusen



Klein, ML, Ophthalmology 2008

Medical Knowledge



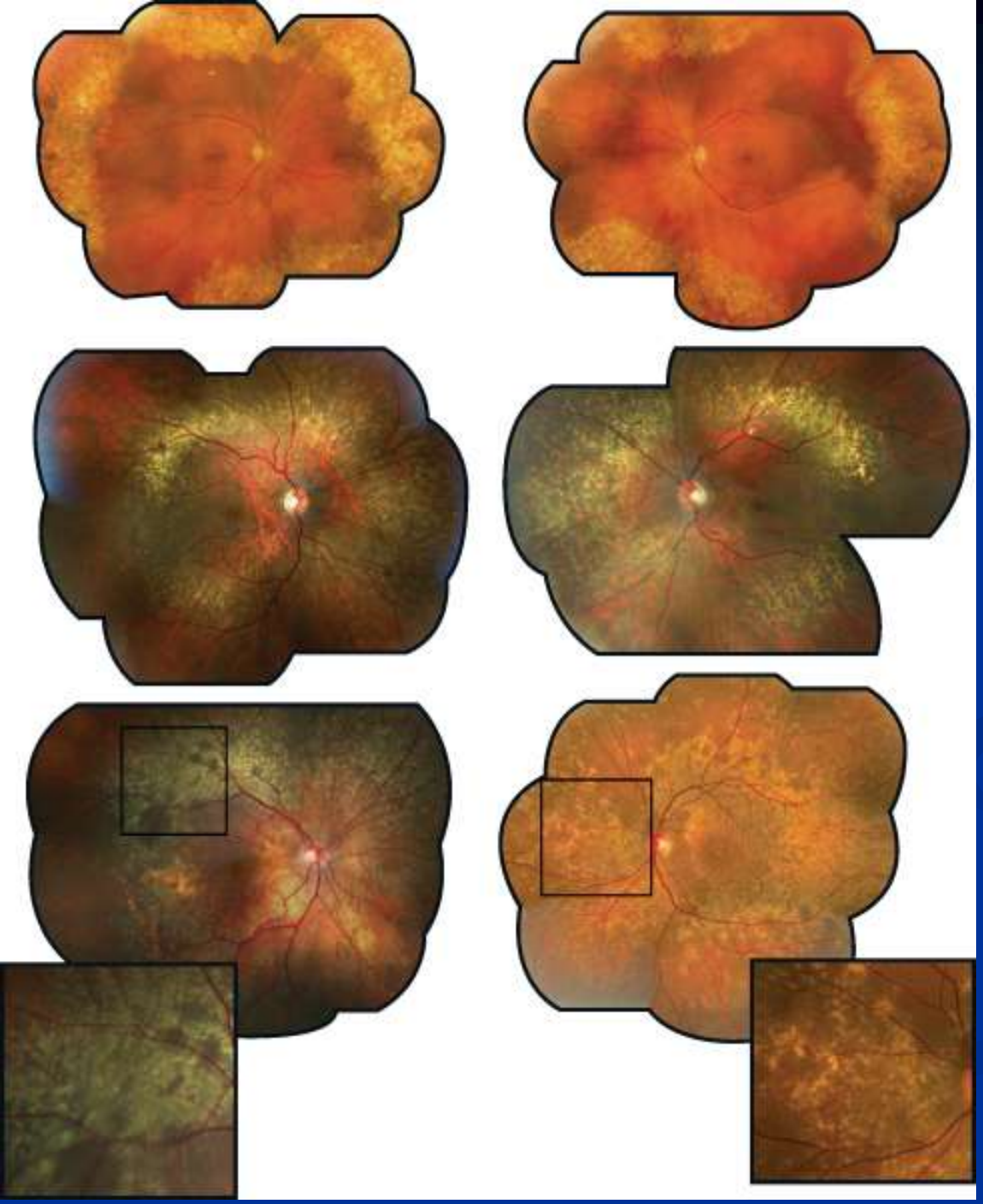


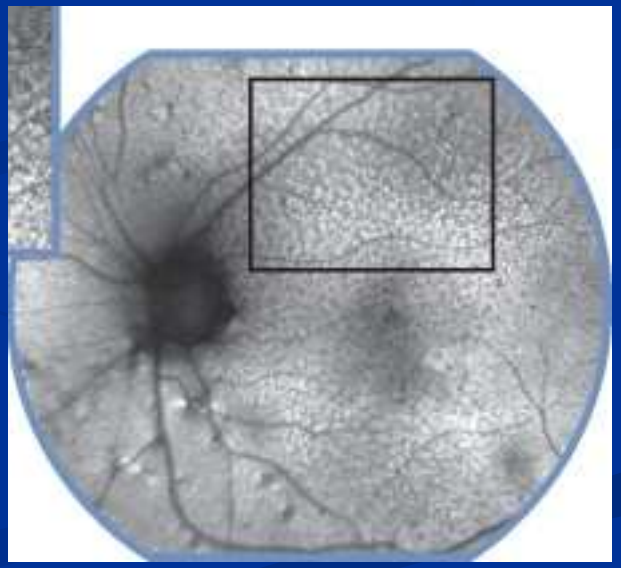
Peripheral Drusen

Medical Knowledge

Yannuzzi, The Retinal Atlas, 2010

Peripheral reticular Pseudodrusen

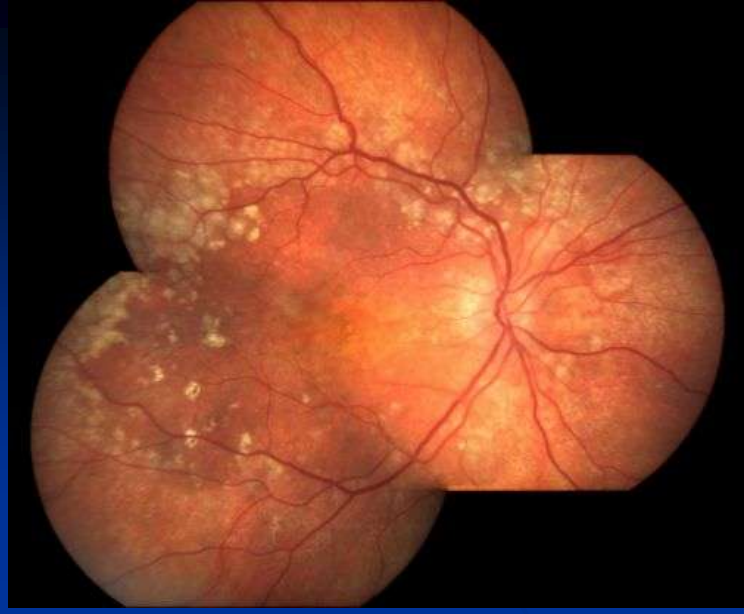




Yannuzzi, The Retinal Atlas, 2010

Medical Knowledge

Basal Laminar Drusen

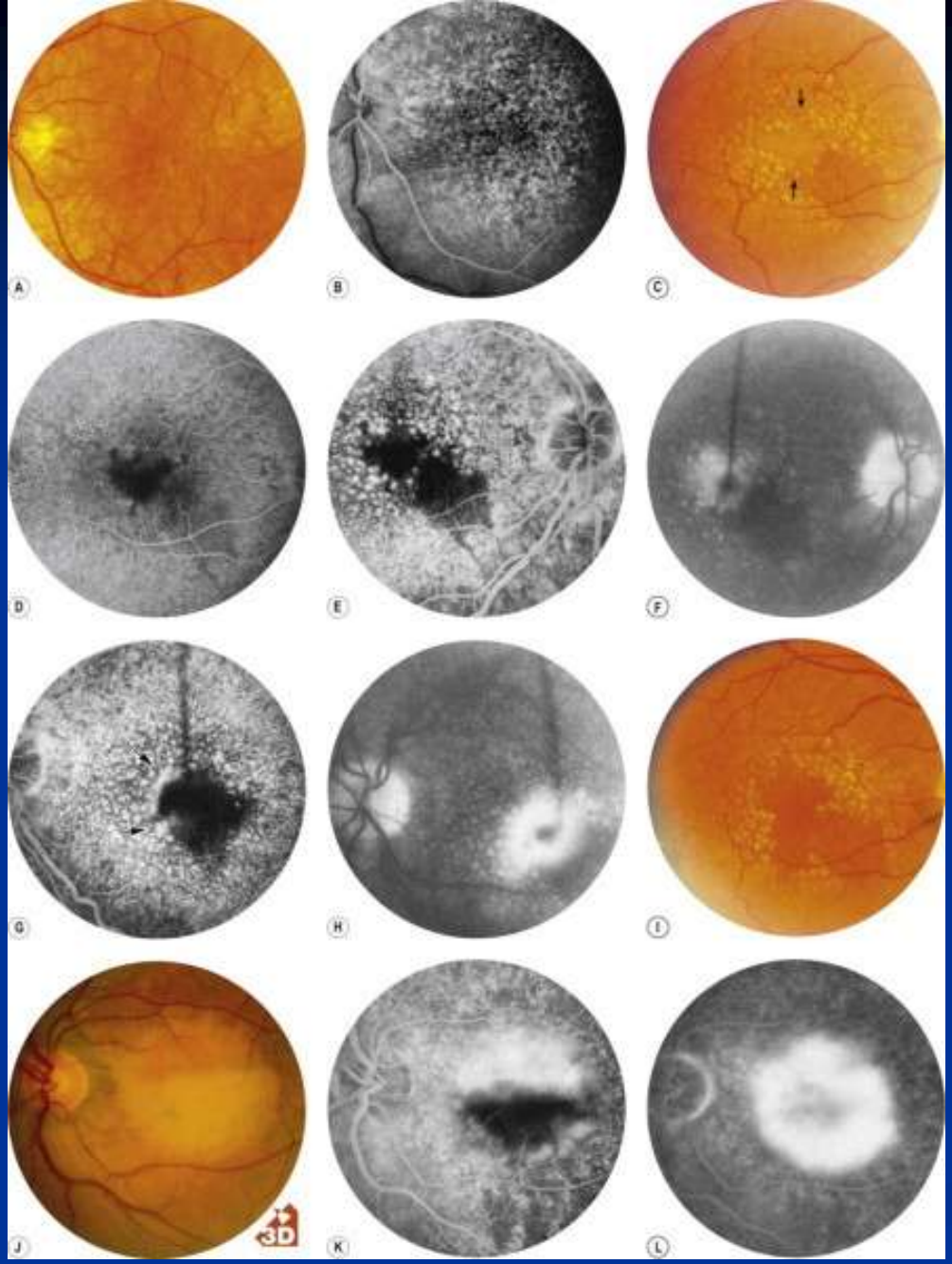


- AKA cuticular drusen
- seen in younger patients than ARMD
- Nodular thickening of RPE BM

- Small (25-75 um), round, slightly raised, yellow nodules, scattered randomly in macula, then coalesce later in life giving a course orange-peel like appearance

- FA: hyperfluoresence discretely during the early arteriovenous phase, "starry sky." fades from view earlier with less intense staining than in the case of exudative drusen

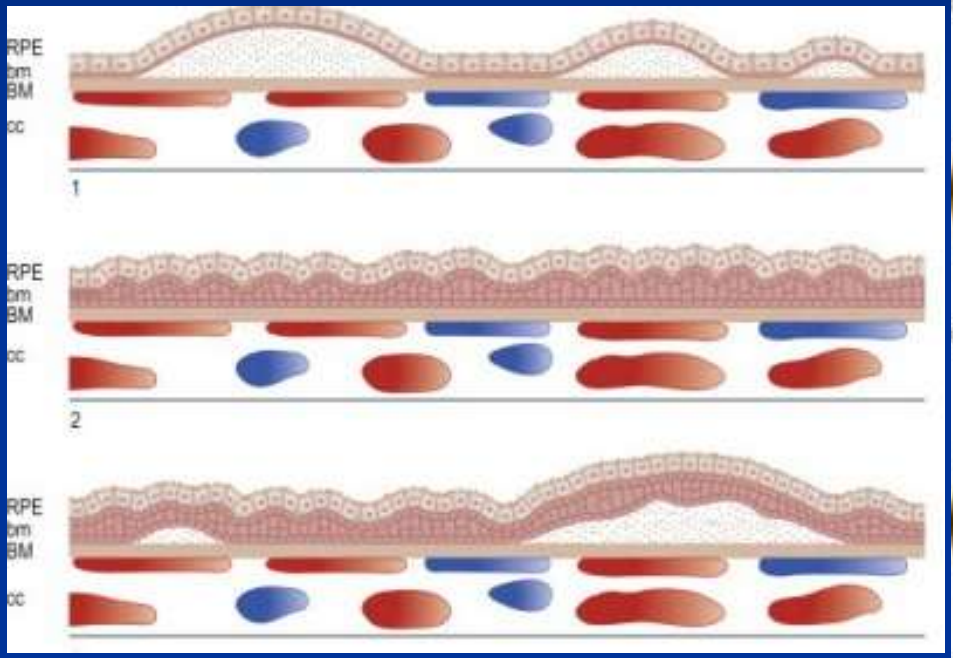
Gass, Atlas of Macular Diseases 2010

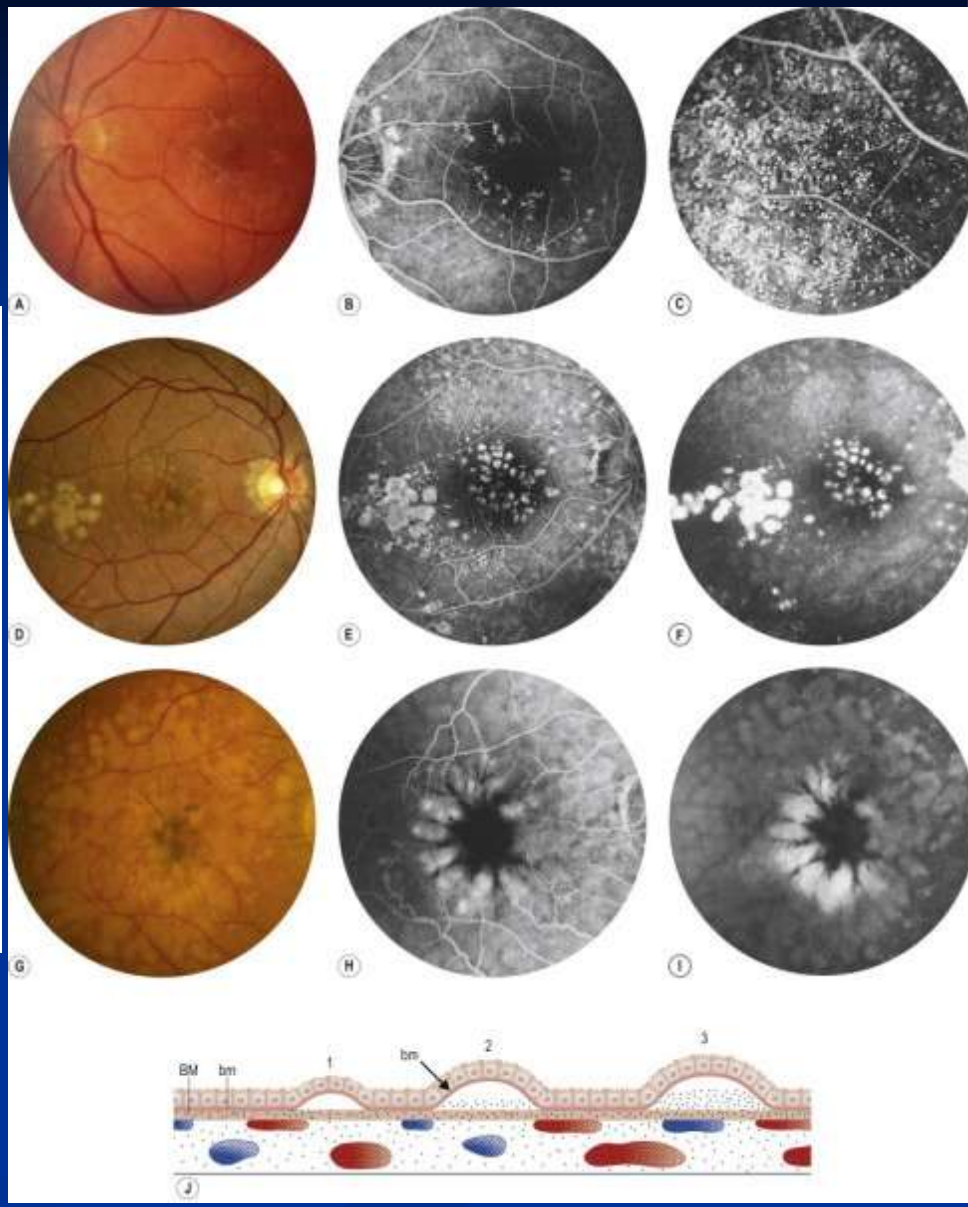


Basal Laminar Drusen

- FA show lesions better than clinically
- Later stage associated with pseudo-vitelliform exudative lesion
- Associated with macular edema without CNV
- SRF can spontaneously resolve

Gass, Atlas of Macular Diseases

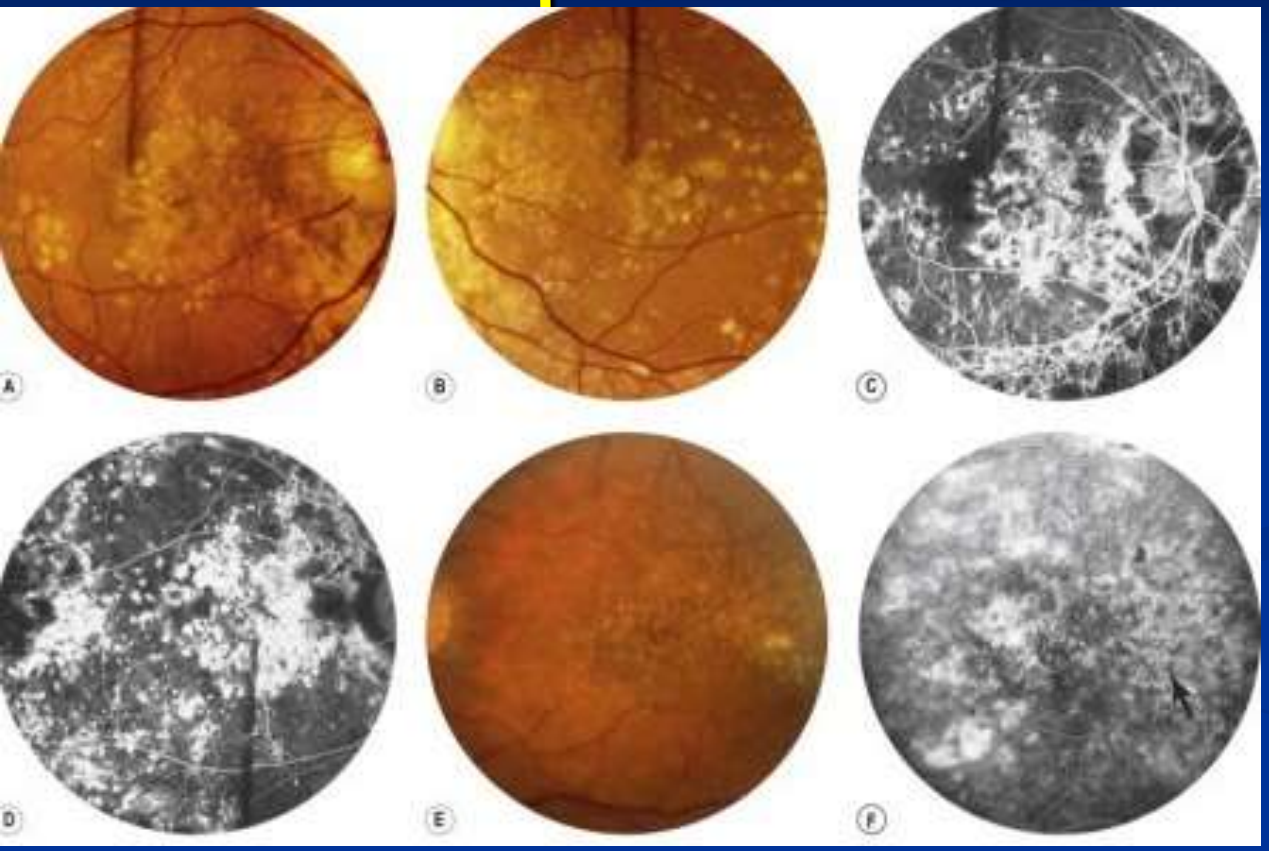




Gass, Atlas of Macular Diseases

Medical Knowledge

Basal Laminar Drusen Associated with Membranoproliferative Glomerulonephritis

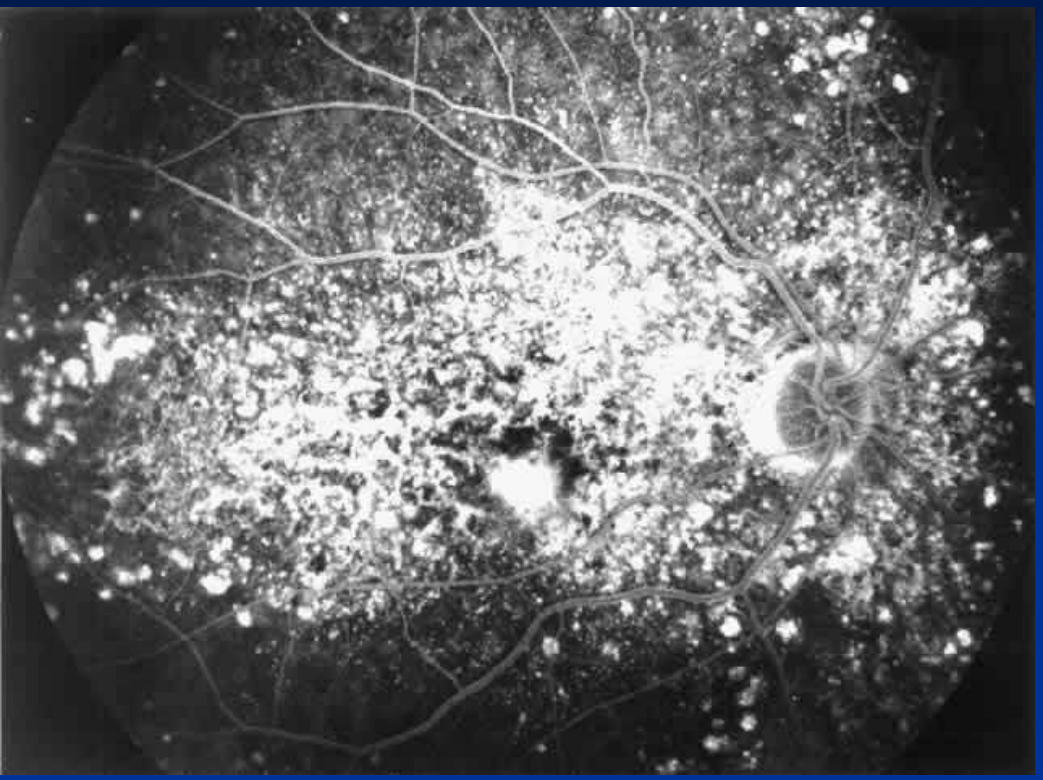


A–D: A 50-year-old man, who had a history of renal transplant at ages 36 and 39 years because of type II MPGN, complained of blurred vision of 2 months' duration. Visual acuity in right eye was 20/200, left eye 20/20. Variable size, calcified. FA: cuticular and calcified drusen

E and F: A 19-year-old man with childhood onset diabetes and nephrotic syndrome associated with type II MPGN. Visual acuity in the right eye was 20/20 and of the left eye was 20/25.

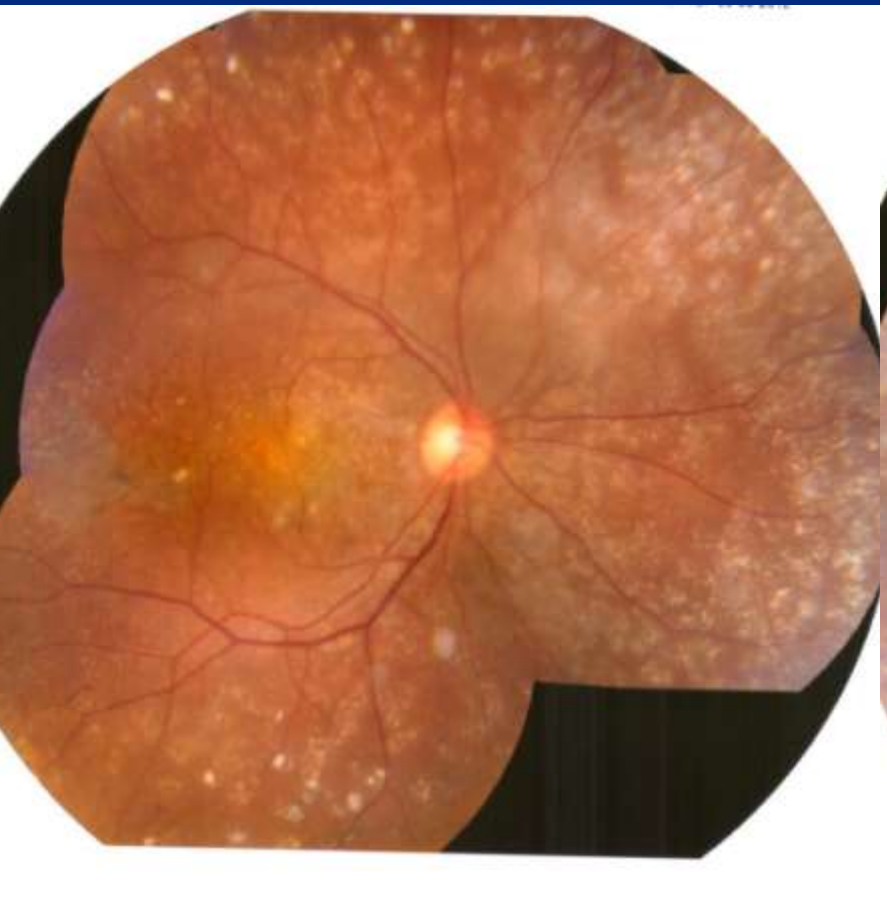
(Agarwal, Gass' Atlas of Macular Diseases, 2012)

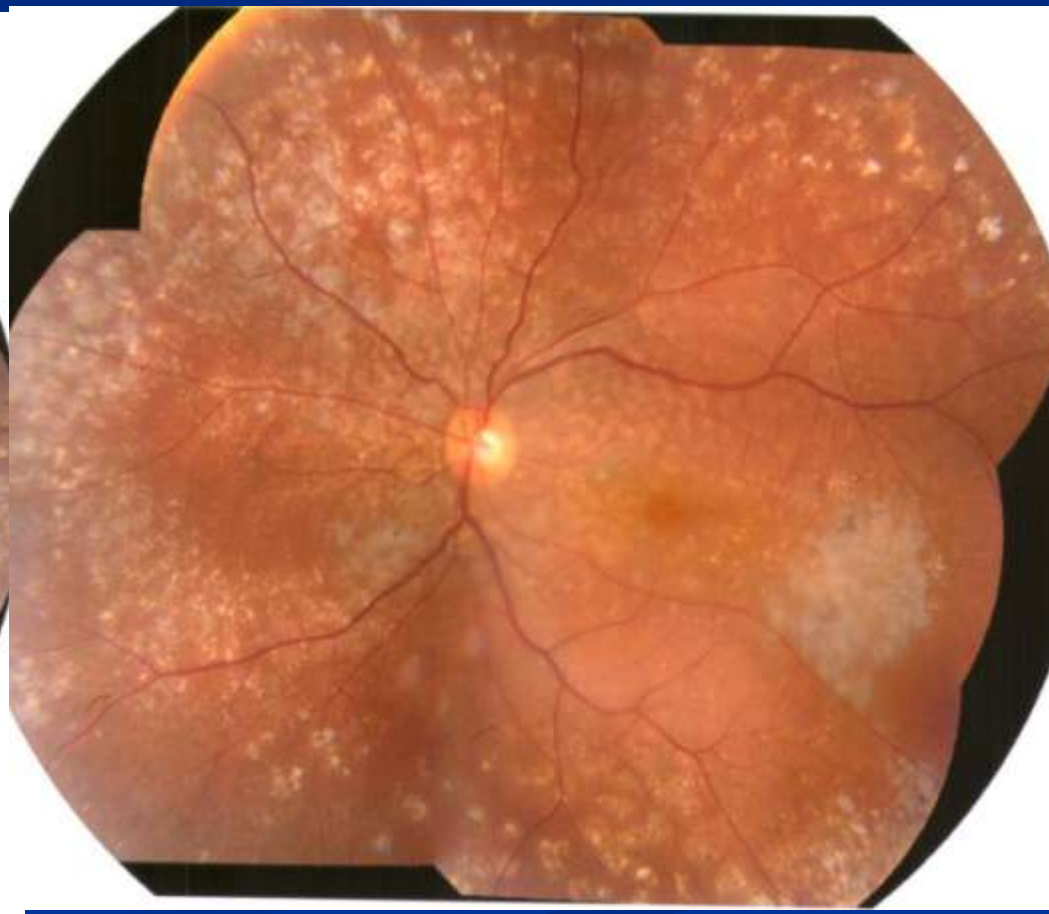
Our Patient



32-year-old patient with renal signs of membranoproliferative glomerulonephritis type II since the age of 9 years. Numerous small and larger drusen-like lesions, atrophic changes, and a small infrafoveolar subretinal neovascular membrane
(Leys A, Graefes Arch Clin Exp Ophthalmol., 1990)

Our patient





Our Patient

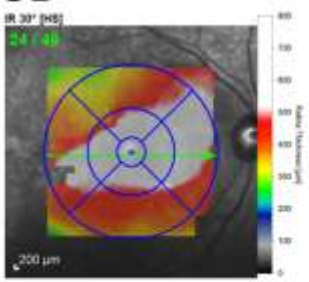
- Unknown Diagnosis: Secondary Acquired Oxalosis vs Basal Laminar Drusen from Type II Membranoproliferative Glomerular nephritis

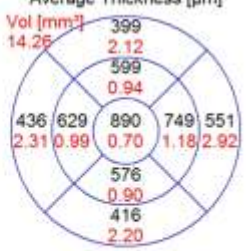
Patient offered intravitreal Avastin. Opted for conservative measures → Given Diamox 250 mg QID PO, f/up in 1 month.

- Urine studies, including 24 hr Oxalate: WNL

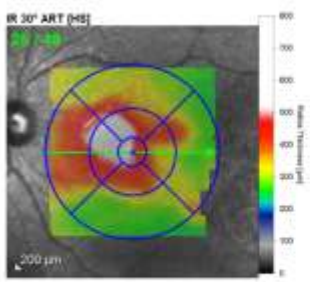
Interpersonal skills/Patient Care/Professionalism

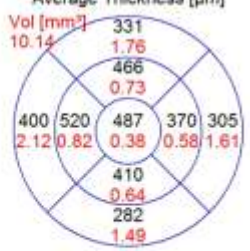
9/5/12
VA
CF OD
20/50 OS



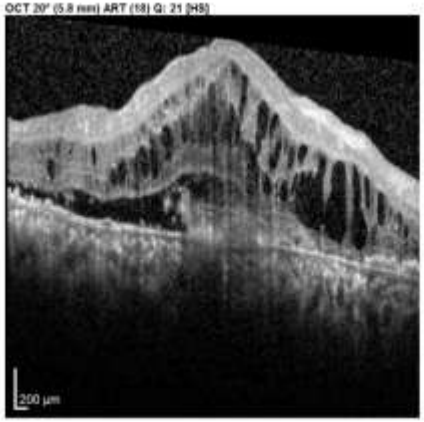


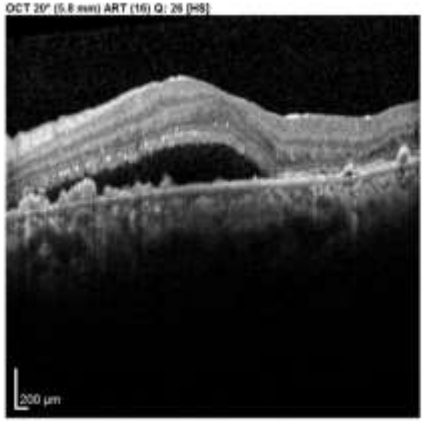
Center: 946 µm
Central Min: 754 µm
Central Max: 972 µm
Circle Diameters: 1, 3, 6 mm ETDRS





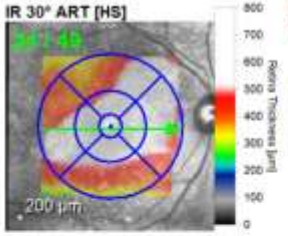
Center: 489 µm
Central Min: 393 µm
Central Max: 578 µm
Circle Diameters: 1, 3, 6 mm ETDRS

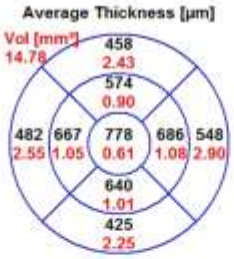




12/19/12
VA:
CF OD
20/50 OS

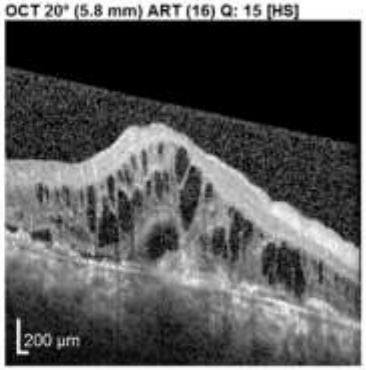
Follow-Up - 12/19/2012

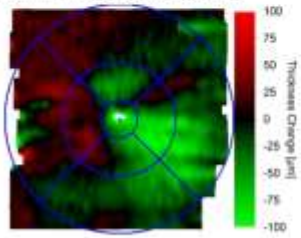


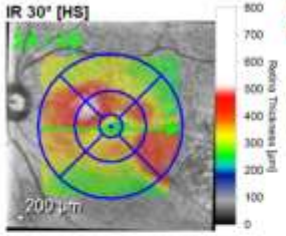


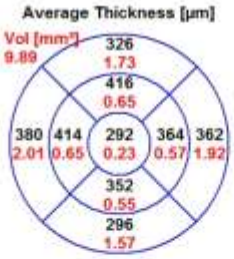
Center: 748 µm
Central Min: 647 µm
Central Max: 884 µm

Circle Diameters:
1, 3, 6 mm ETDRS



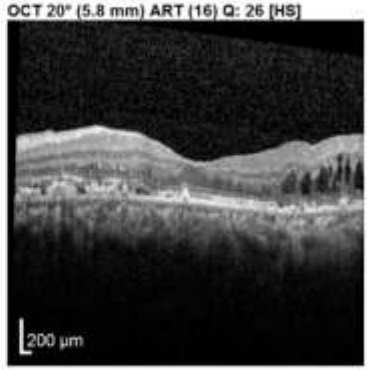


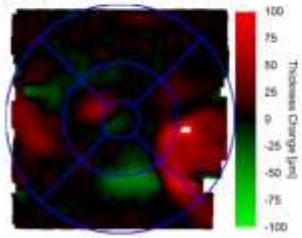




Center: 235 µm
Central Min: 225 µm
Central Max: 413 µm

Circle Diameters:
1, 3, 6 mm ETDRS





Notes:

Our Patient

- Subjective Improvement
- Diagnosis tentative
- Patient will be following up in 1 month…

Works Cited

Agarwal A. Various chapters in: Gass' Atlas of Macular Diseases– 5th Ed.[online] Elsevier Saunders, 2012.

Duvall-Young J, Short CD, Raines MF et al: Fundus changes in mesangiocapillary glomerulonephritis type II: Clinical and fluorescein angiographic findings. Br J Ophthalmol 73:900, 1989.

Genead M. Spectral-domain pptical coherence tomography and fundus autofluorescence characteristics in Patients with fundus albipunctatus and retinitis punctata albescens. Ophthalmic Genetics; Vol. 31(2); 2010: 66-72.

Klein ML, et al. Retinal precursors and the development of geographic atrophy in age-related macular degeneration. Ophthalmology, 115 (6): 2008, 1026–1031.

Leys A, Michielsen B, Leys M et al: Subretinal neovascular membranes associated with chronic membranoproliferative glomerulonephritis type II. Graefes Arch Clin Exp Ophthalmol 228:499, 1990 Ophthalmol 73:900, 1989

Mataftsi A et al. Bietti's crysalline corneoretinal dystrophy: A cross-sectional study.. Retina Volume 24(3); June 2004: 416-426.

Punjabi OS et al. Crystalline retinopathy in primary hyperoxaluria. J AAPOS 2011;15:214-216.

Small KW, et al. Ocular findings in primary hyperoxaluria. Arch Ophthalmol.Vol 108; Jan 1990: 89-93

Yannuzzi, LA . Various chapters in: The Retinal Atlas. [online]. Saunders Elsevier: 2010.

Core Competencies

Patient Care: The patient received compassionate care, based on the appropriate and most effective management techniques that addressed his physical, emotional, and mental health issues

Medical Knowledge: The literature was reviewed, a differential was formed. Diagnostic and therapeutic modalities were discussed using evidence-based medicine and general practice guidelines. The basic and clinical science of the disease was reviewed to better understand this condition

Practice-Based Learning and Improvement: The literature was reviewed. The clinical evidence was assimilated to better treat the patient as well as learn from his clinical course in order to manage patients in the future.

Interpersonal and Communication Skills: We communicated extensively with the patient regarding the process of diagnosing and treating his disease. All of his questions were answered in a compassionate manner. We worked as a team to limit his fears of vision loss.

Professionalism: Our responsibility as a physician to do no harm was adhered to at all times. Necessary tests were suggested and the ethical principles of informed consent were utilized. The patients clinical information remained confidential at all times.

Systems-Based Practice: We showed awareness of the healthcare system, using cost-effective mechanisms of diagnosis and management.

Reflective Practice

This case demonstrated a rare disease process with severe implications for vision loss. After considering a wide differential diagnosis and examining the literature, the appropriate diagnostic modalities were chosen to narrow our differential and formulate a diagnosis. An interdisciplinary approach was taken with the renal team as well as multiple retinal specialists. The patient was appropriately and compassionately managed. He was educated about his disease processes and their natural courses.

Thank You

- Dr. Kenneth Olumba
- Dr. Renelle Pointdujour
- Dr. Yannuzzi, Dr. Freund