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
OD
IR 30° [HS]

Average Thickness [µm]
Vol [mm²] 399
14.27
2.12
599
0.94
436
629
2.31
0.99
890
0.70
749
551
1.18
2.92
576
0.90
416
2.20

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

OS
IR 30° ART [HS]

Average Thickness [µm]
Vol [mm²] 331
10.14
1.76
466
0.73
400
520
2.12
0.82
487
370
0.38
305
0.58
1.61
410
0.64
282
1.49

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

OCT 20° (5.8 mm) ART (17) Q: 20 [HS]

OCT 20° (5.8 mm) ART (16) Q: 26 [HS]

Patient care
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)
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

(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)
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
Peripheral Drusen

Yannuzzi, The Retinal Atlas, 2010
Peripheral reticular Pseudodrusen

Yannuzzi, The Retinal Atlas, 2010
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: hyperfluorescence 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
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.

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
Our Patient

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


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


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 patient's 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.
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

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