Mysterious Maculopathies

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11 March 2017
The Problems in Diagnosis

- Tissue access
- Patient behaviour
- Media clarity
- No obvious pathology
- Lack of specifying history
The Allies in Diagnosis

- Specifying history
- Words & descriptions that localize
- Imaging - OCT, FAF, FA, B-Scan, Photography
- Lab results
- Medical history including medicines
- Clear media
- Patient patient
Obvious maculopathies

- The diagnosis and patient complaint are easily connected- large drusen with subretinal blood, big yellow central cysts in an eye with obvious diabetic retinopathy, a clear subretinal fluid blister in a intense 43 year old man holding on to two cell phones...
And some are not so much...

• 20/200 vision and a benign appearing macula
• Sparkling lights and gray vision in retina that looks OK at first pass
• A “blind” eye without an RAPD, normal maculae, and perfect vision in the other eye
• Severe headaches with poor vision and a few subtle gray spots here and there
28 year old female
• 10 days of headaches
• Poor vision in left eye coinciding with headaches (20/15 OD 20/200 OS)
• No usual analgesic stopped the headaches
• Clear thinking, no other neurologic signs
Left eye
Early, left
FA, mid, right
FA, mid, left
FA, late, right
FA, late, left
APMPPE

- Young healthy men or women (25)
- Rapid loss of vision in usually both eyes
- Inflammation at level of RPE
- 1/3 have flu-like symptoms prior
- 1-4% have CNS involvement: death possible from stroke or vasculitis
- This was an emergency due to the headaches
60 mg prednisone daily
4 days later headaches gone
20/15 OD 20/100 OS
Lesions were becoming pigmented on the edge
2 weeks later, off taper, mild headache returned
20/20 OD, 20 60 OS
1 month later 20/15, 20/20, no headaches
Referral for odd-looking macula

• No visual complaints, 33 years old
• 20/20 OU
• Mixed heritage- mainly north European
• No family history of early visual problems
Malattia Levantinesse

- AKA Doyne’s honeycomb dystrophy
- First described in a family from the Levantine valley in Switzerland
- Often good vision, often subretinal fibrosis with CNVM
- Genetically the two named diseases are the same
- Conical deposits between RPE & Bruch’s
AMD Referral

• 39 years old (?!)
• Long, long list of medical problems, esp. cardiomegaly, diabetes, OSA
• 20/20 vision, not happy about second dilation in a week
• Morbid obesity
PEPSI

- Paget’s disease of the bone
- Ehler’s – Danlos syndrome
- Pseudoxanthoma elasticum
- Sickle cell anemia
- Idiopathic

- Accompanied by peau d’ orange pseudo-texture in temporal macula
CHRPE vs. CHRPE

• Congenital hypertrophy of the retinal pigment epithelium
• Often sent to me as possible melanoma
• Evenly thickened, round, jet-black (in first years) with pale-white encircling border, developing lacunae with time
• No malignant potential, natural history shows depigmentation over time
Atypical CHRPE

- Multiple, bilateral
- Pointed, torpedo-shaped
- Pointing toward optic nerve
- Associated with FAP - familial adenomatous polyposis & Gardner’s syndrome
FAP/Gardner’s

- Soft-tissue tumours in 1\textsuperscript{st} 2 decades (fibromas, lipomas, sebaceous cysts)
- Bony tumours (osteomas) in 2\textsuperscript{nd} decade
- Risk of adenocarcinoma of colon in life is 100%
- Usual age of onset 30
Our patient

• Colon adenocarcinoma at 35, s/p colectomy, ileostomy, Whipple procedure
• Stage 1 adenocarcinoma of lung at 32
• Not married, no children
• Mother with early-onset colon CA
• Sister with lupus
Sun Allergy

- PMLE (polymorphic light eruption): itchy red rash, often hereditary
- Actinic prurigo (hereditary, native Americans)
- Photo-allergic eruption (sun-provoked reaction to antibiotics, fragrances, sunscreen, diuretics, oral contraceptives, naproxen, ibuprofen, et al)
- Solar urticaria (hives) rare, young females
Treatments

• Avoidance
• Thalidomide (!), chloroquine, beta-carotene, corticosteroids
• In 1962 our patient was put on the anti-malarial drug chloroquine (close kin of hydroxychloroquine, AKA Plaquenil) by her family doctor for sun allergy
6 years

• The drug worked, he continued to refill it without question
• At 16, a pharmacist asked her if she was getting regular eye exams...no
• He refused to fill the prescription
• Ophthalmologist found macular changes and subnormal vision uncorrectable with glasses
Preventable Tragedy

• By 17 she was 20/200 OU and has remained so since
23 Year Old Man

• 3 day history of flickering lights and blurred (hazy) vision in the left eye
• Recent viral illness
• Mild photophobia
• A dark spot next to central vision- not complete, more like a gray zone
MEWDS

- Multiple evanescent white dot syndrome
- Young healthy people F>M
- Related to and probably of same causation as idiopathic blind-spot enlargement syndrome
- Our patient’s grey dots and symptoms faded away over a week
76 Year Old Man

- Abrupt decrease in vision 2-3 months ago OD
- Assumed cataract, scheduled visit
- Cataract surgeon unimpressed with cataracts, saw macular elevation, 20/80 OD 20/40 OS
- 1+ NS OU
- Soft drusen OU, mild hyperope
- Asteroid hyalosis
Polypoidal Choroidal Vasculopathy

- Inject OD with bevacizumab
- 1 month later second injection- at that visit SRF gone, irregular PED still present, 20/40
- BUT- 2 weeks after 1st visit, diagnosed with lung CA with brain mets, already had 10 brain radiation treatments
- Still no path diagnosis as to type of cancer
Random Finding

• Employee of local optometry clinic
• All volunteered to be photographed for new widefield imaging system
• 3 (!) employees were told by rep to see retina specialist ASAP
• This patient had minimal medical history other than Barret’s esophagus
Exam

- Patient reported a few wavy lines on Amsler grid testing - had not noticed previously
- VA 20/20 OU
- Did report that bits and pieces of visual field had been missing for a while
- Normal pressure, no cataract, rest of exam other than retina normal
StARgardt’s Disease

• Unusual late diagnosis
• AKA fundus flavimaculatus
• Lipofuscin storage disease- classic silent or dark choroid on FA
• Autosomal recessive, so often family history is non-contributory
• Peripheral flecks common
• Several subtypes
Blurry Vision

- Healthy woman in late 50s
- Noticed blurry vision, more at night than day
- 20/40 OD, 20/25 OS
- Anterior subcapsular cataracts, unimpressive
- Butterfly-type yellow lesions in maculae, symmetrical
Pattern Dystrophies

• Generally good prognosis
• Symmetrical
• Can be seen in 2nd-4rth decade
• Slow-moving
• Rare to have CNVM complications
• Cataract surgery improved her 20/30 OD, 20/25 OS, with manifest refraction to 20/20 OU
More Dystrophy

• This one is not as fun- for everyone
• Long-term patient with stable awful central vision- which she thinks is just fine, especially when driving
• 20/200 OU best corrected
• Clear lenses, OD – 7.5 -2.00; OS -9.00 -2.00
• Doctor-shops for signature on driver’s license
• Drives anyway
Cone Dystrophy

- This is part of a vast spectrum of cone, cone-rod, and rod dystrophies, all part of the broad retinitis pigmentosa spectrum.
- When the cones are primarily affected, colour and central visual acuity are most affected; peripheral vision remains good.
- They tend to become manifest in the 1st-2nd decades then remain stable.
Peculiar Macula

• This 78 year old man complained of decreased vision in his left eye for several months
• He was generally healthy except for gout
• Vision was 2/25 OD and 20/40 OS
• Normal pressures, nerves, pseudophakic, status post YAG capsulotomies
Choroidal Osteoma

• Slow leakage of serous fluid into macula responsible for visual complaints as well as pigmentary changes
• I planned to do PDT but his insurance changed and he was lost to follow-up
Too Much

• Sometimes an eye just can’t take it anymore
• This 62 year old man had severe pigmentary glaucoma with multiple procedures over the years: ALT, SLT, trabeculectomy, shunt, and finally a shunt revision.
• After the last procedure, he noticed decreasing vision OD (20/200). Left eye remained a steady 20/30
Scleritis, CD, RD

• Started on Ibuprofen 800 mg tid
• 3 days later, vision measured the same, he said it seemed worse
• I told him to keep up the anti-inflammatory drug and come back in two days
Better and Worse

• Exudative RD- if anything worse
• Hand-motion vision
• Choroidal detachment resolved
• Sclera still thickened
• Sub-Tenon’s Kenalog administered, 40 mg
1 month later

- Mild RD improvement, vision CF
- Referred to vitreoretinal surgeon for possible surgical intervention
- Pressure fine at 23mm Hg
- 1 month later, vitrectomy
- 6 months after that, retina remained attached, vision 20/150
Blind Eye

• 52 year old, “my left eye is blind”
• Same complaints 4 years ago, then, 20/150 OD and HM OS
• Here for diabetic exam- mild NPDR
• VF (black OS, patchy OD), OCT & FA normal
• MRI read as normal
Blind eye

- 4 years later, multiple no shows in between, arrives for complaint of “my left eye is blind” for the last 2-3 months
- Wearing +1.25 sph OD; plano -/.50 OS (20/250 OD, NLP OS)
- Manifest: +0.50 sph OD, 20/200 OD
- No APD
- Pressures normal, 0.3 healthy nerves, symmetrical, 1+ NS
Blind eye

• OCT, FA normal
• While covering the right eye, I slowly advanced my finger toward her left eye.
• Her comment: “I wouldn’t blink when you do that on the left eye but I would on the right.”
• I was able to reassure her the vision would return in the left eye, and not to forget to come back for her annual diabetic exam
Bilateral Central Blindness

• 64 year old woman with 25 year history of central scotoma. Stopped driving at 40.
• Vision is CF @ 3” OD; 20/200 OS eccentric
• Never any chloroquine or hydroxychloroquine use
• Grandmother (paternal) had similar early central blindness
Testing

- ffERG: no cone-rod dystrophic markers, mild decrease in photopic/scotopic signals
- VF: central scotomata
- FAF: central hypo with perimacular hyper flecks
- OCT: outer retinal atrophy & disorganization with RPE loss
North Carolina Dystrophy

• 545 families from western NC descended from 3 Irish brothers documented over 7 generations
• Has been documented in a Chicago African-American family over 4 generations as well as families from Belize and Korea
• Visual loss usually apparent in teens and invariable by 4rth decade
Optometric Referral

• 72 year old man saw optometrist for blurry vision in left eye

• She noted macular edema & blood, referred

• Vision at visit was 20/25 OD, 20/40 OS

• He thought his vision declined for 1-2 months

• Exam revealed soft drusen, intraretinal blood and edema
Types of Neovascularization

• Type 1: occult, the growth occurs under the retinal pigment epithelium
• Type 2: classic, the growth occurs upward into the sub-sensory retinal space
• Type 3: Retinal angiomatous proliferation (RAP): growth begins in the outer plexiform layer of the retina downward to anastomotic connections with the choroid
Treatment

• All are promoted by VEGF and therefore susceptible to anti-VEGF drugs
• Spectral-domain OCT has allowed Type 3 lesions to be defined in unprecedented detail
• RAP was first described in 1990 and thought to be a rare lesion
• Now it is believed Type 3 lesions may account for 1/3 of exudative AMD
Enough

• I chose these cases out of over 300 in my notebook of noteworthy cases

• They just keep coming