

On behalf of Vision Expo, we sincerely thank you for being with us this year.

Vision Expo Has Gone Green!

We have eliminated all paper session evaluation forms. Please be sure to complete your electronic session evaluations online when you login to request your CE Letter for each course you attended! Your feedback is important to us as our Conference Advisory Board considers content and speakers for future meetings to provide you with the best education possible.



1



A MULTIMODAL IMAGING GRAND ROUNDS

Carolyn Majcher, OD, FAAO, FORS
Oklahoma College of Optometry

2

Contact:

- majcher@nsuok.edu
- 918-444-4155

Disclosures:

- Paid consultant/speaker for:
 - Carl Zeiss Meditec
 - Regeneron Pharmaceuticals
 - Optomed
- Paid advisory board member for Apellis Pharmaceuticals, Iveric Bio, Ocuteira, Notal Vision, LENZ Therapeutics
- Non-financial support (writing assistance) from Roche
- All relevant relationships have been mitigated

3

OVERVIEW

Technology

- Widefield (WF)/UltraWF (UWF) color photography
- Fundus autofluorescence (FAF)
- OCT imaging
 - Conventional structural
 - OCT angiography (OCTA)
 - Enhanced depth imaging (EDI)
- Ultrasound
 - Ocular and carotid
- Neuroimaging (CT/MRI)

4

WIDEFIELD vs ULTRA-WIDEFIELD



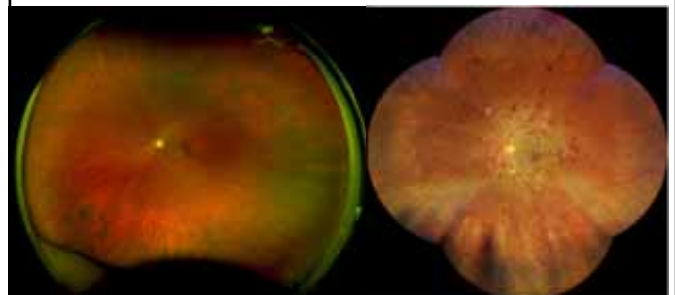
WF= Up to the vortex vein ampullae, >50°



UWF= Includes at least 4 vortex vein ampullae, ~200° and 80% retinal surface

5

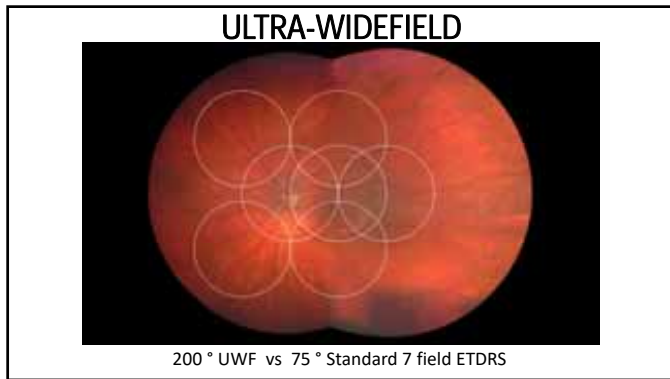
ULTRA-WIDEFIELD



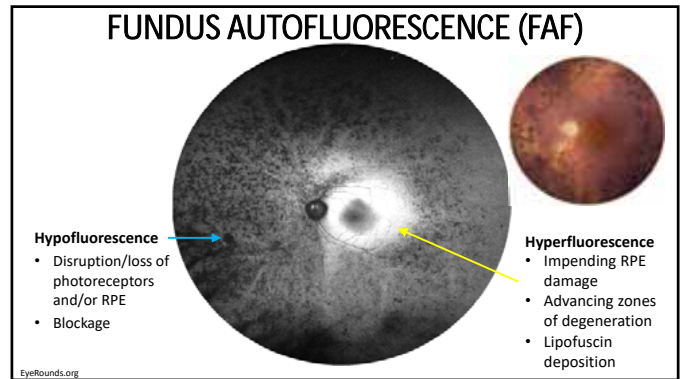
Single Capture

Montage

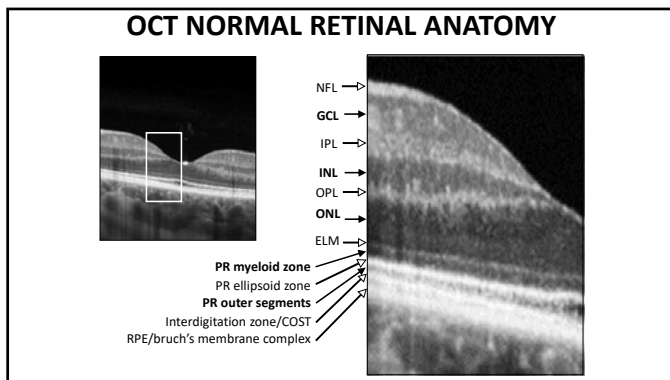
6



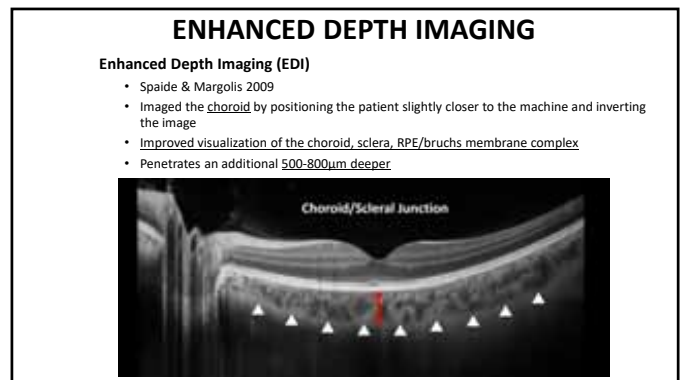
7



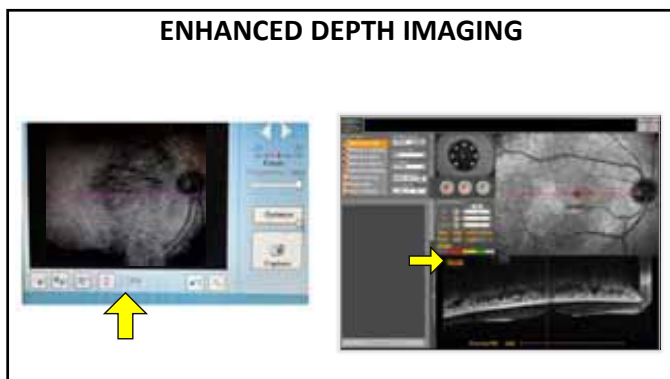
8



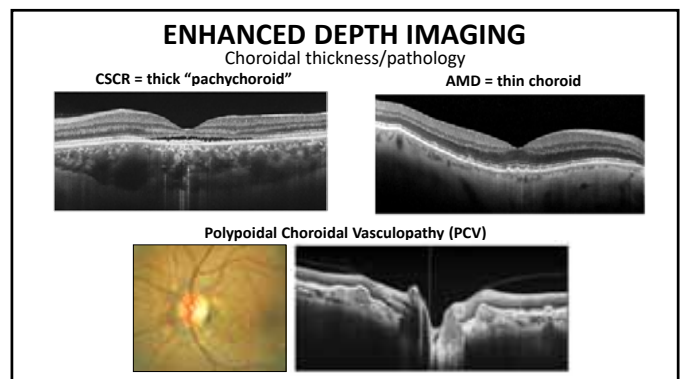
9



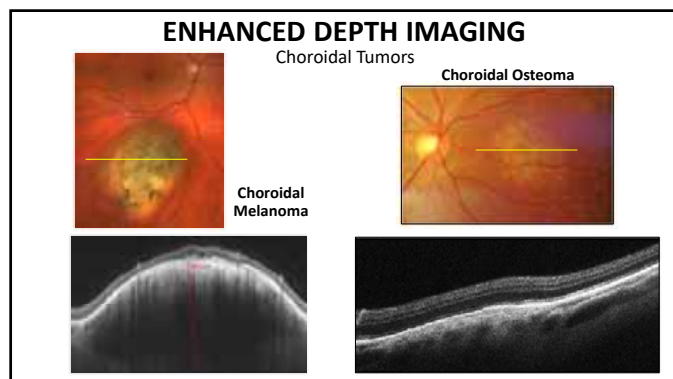
10



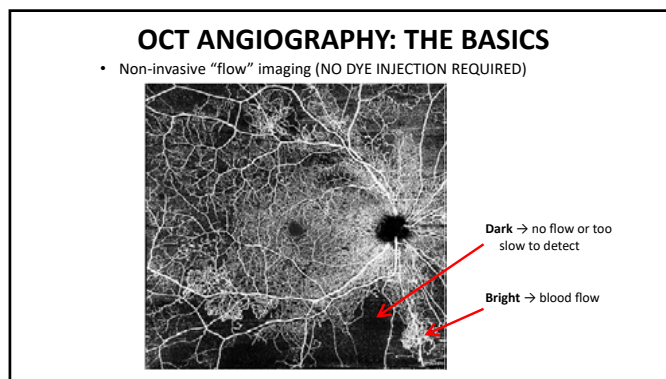
11



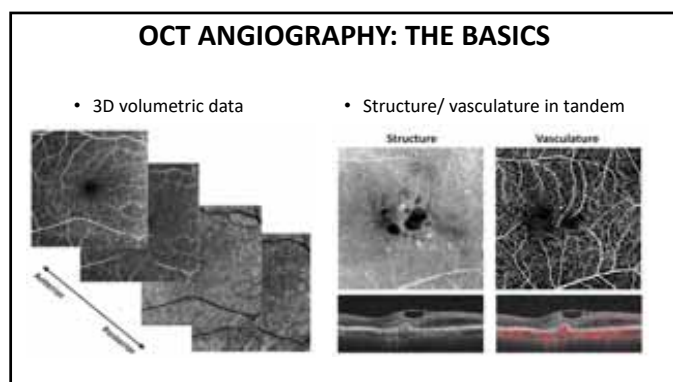
12



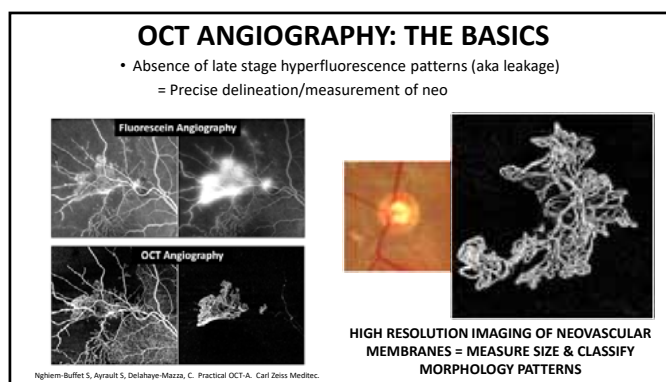
13



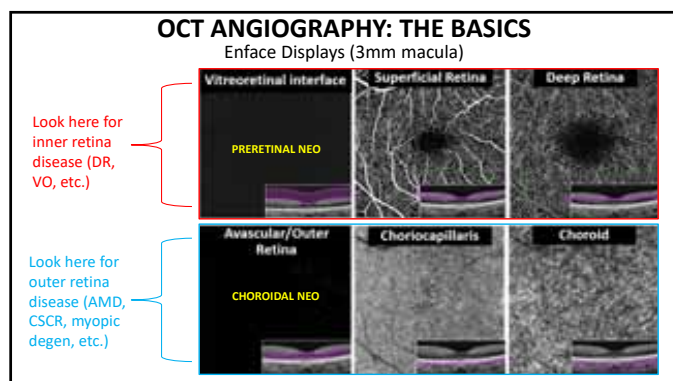
14



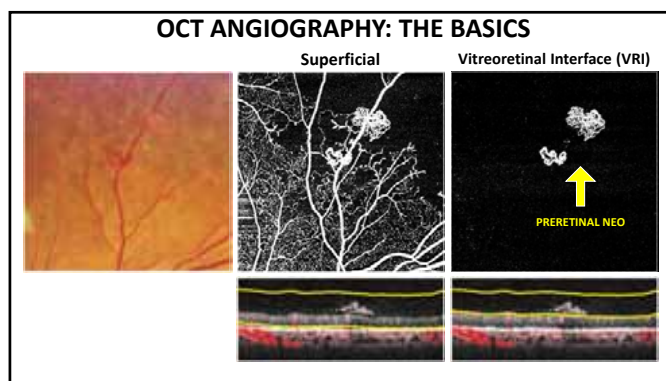
15



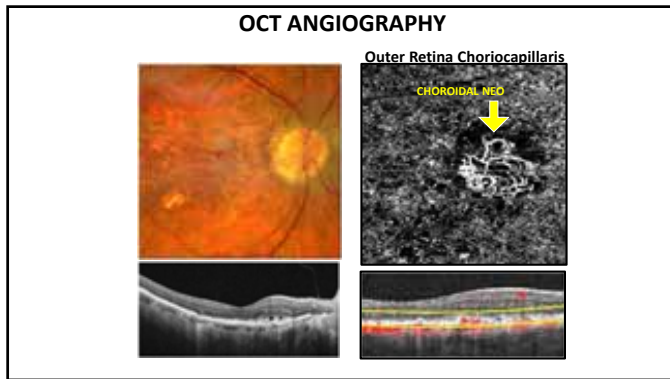
16



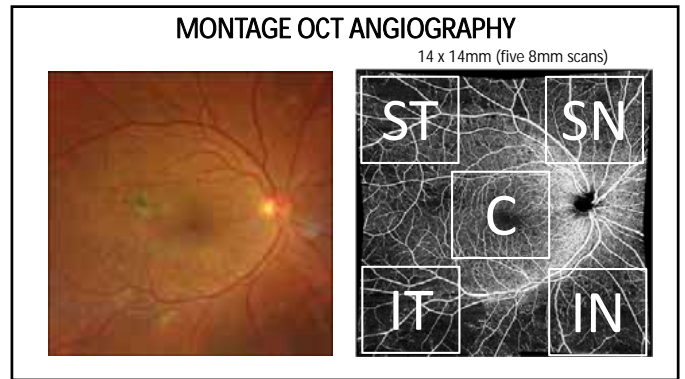
17



18



19



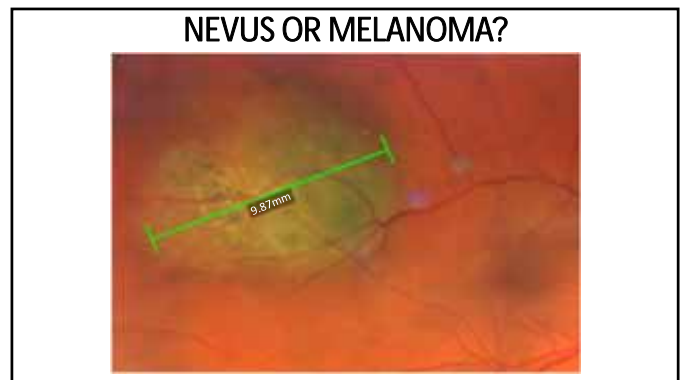
20

NEVUS OR MELANOMA? YOU DECIDE!

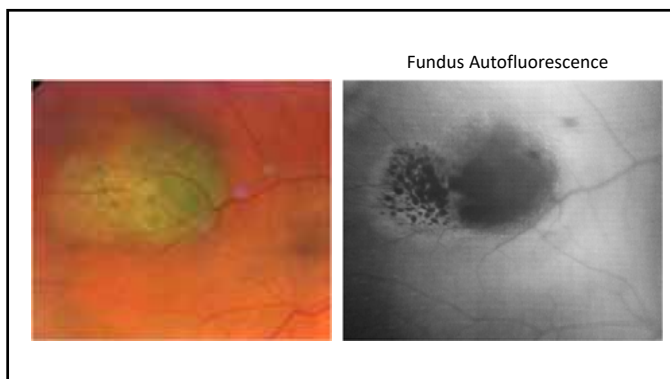
72yo American Indian female

- CC: Doctor directed visit 6mo cataract FU
- Oc Hx:
 - Cataracts OU
 - LEE & DFE 6 months ago
 - Choroidal nevus OD (description from DFE 6mo ago: "Large choroidal nevus superior temporal 3DD flat, distinct borders," took photos but not linked to chart)
- BCVA
 - OD 20/40 PHNI
 - OS 20/30

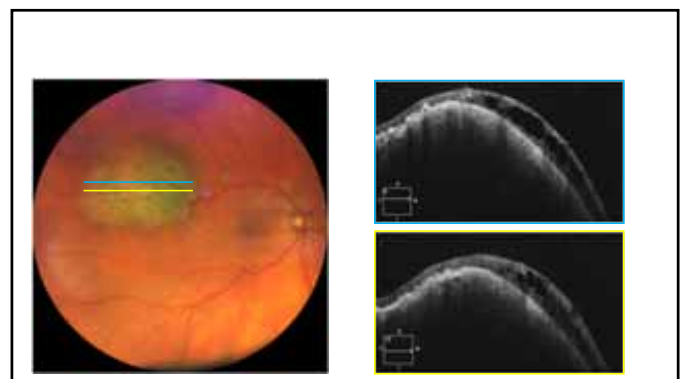
21



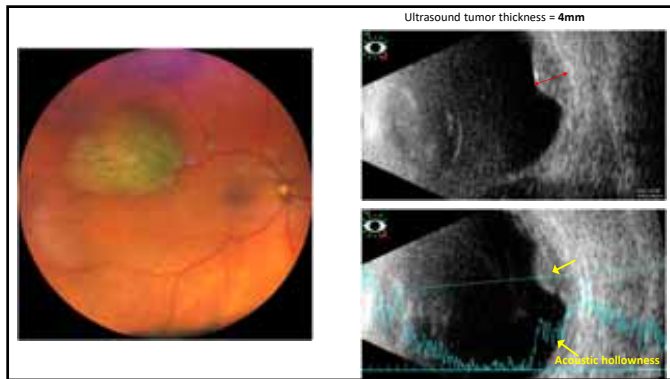
22



23



24



25

NEVUS OR MELANOMA?

Differentiating small choroidal melanoma from choroidal nevus 2019 UPDATE ★

To Find Small Ocular Melanoma Doing IMaging (TFSOM-DIM)
 T- Thickness (>2mm US = ~ 890µm OCT)
 F- Fluid, SRF
 S- Symptomatic VL (VA ≤20/50)
 O- Orange pigment (FAF)
 M- Melanoma acoustic hollowiness
 DIM- DiaMeter >5mm

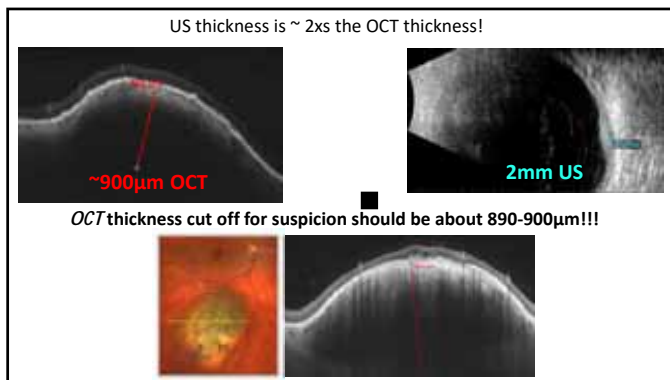
Risk for growth within the next 5 years:

- 0 risk factors = 1.1%
- 1 factor = 11%
- 2 factors = 22%
- 3 factors = 34%
- 4 factors = 51%

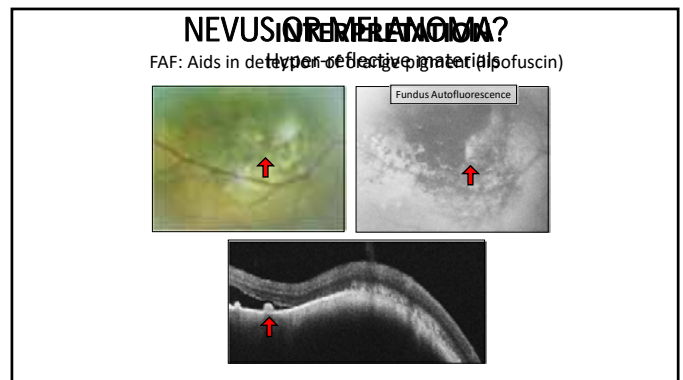
Shields CL. Small choroidal melanoma: detection with multimodal imaging and management with plaque radiotherapy or Au-011 nanoparticle therapy. Curr Opin Ophthalmol. 2019

| Factor | Factor | Factor | Factor | Factor | Factor |
|----------------|--------|----------------|----------------|----------------------|---------------|
| Thickness >2mm | Fluid | Symptomatic VL | Orange pigment | Acoustic hollowiness | Diameter >5mm |
| 1 | 1 | 1 | 1 | 1 | 1 |
| 2 | 2 | 2 | 2 | 2 | 2 |
| 3 | 3 | 3 | 3 | 3 | 3 |
| 4 | 4 | 4 | 4 | 4 | 4 |
| 5 | 5 | 5 | 5 | 5 | 5 |
| 6 | 6 | 6 | 6 | 6 | 6 |
| 7 | 7 | 7 | 7 | 7 | 7 |
| 8 | 8 | 8 | 8 | 8 | 8 |
| 9 | 9 | 9 | 9 | 9 | 9 |
| 10 | 10 | 10 | 10 | 10 | 10 |

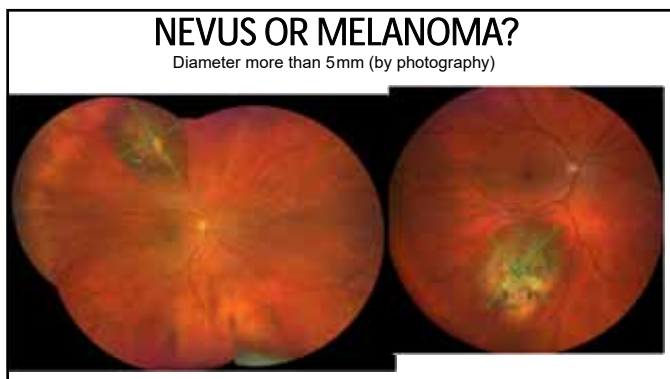
26



27



28



29

NEVUS OR MELANOMA?

Decision Dx-Uveal Melanoma: Gene Expression Profiling

Assessment

- Choroidal melanoma

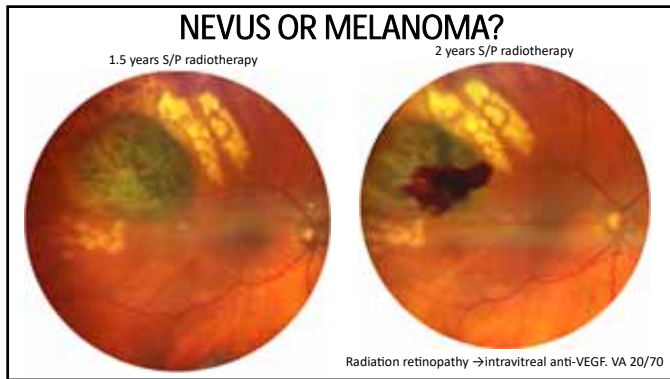
Plan

- Ocular oncologist treated with **plaque radiotherapy** and took sample for **gene expression profile testing**
 - Class 1A
- Followed by adjunctive laser thermotherapy and intravitreal bevacizumab to prevent radiation retinopathy
- Liver ultrasound, liver function tests with LDH, and chest x-ray all WNL

Decision Dx-UM test results are reported as follows:

- Class 1A - very low risk (2%) of metastasis within 5 years?
- Class 1B - moderate risk (21%) of metastasis within 5 years?
- Class 2 - high risk (72%) of metastasis within 5 years?

30



31

Take Home Message

Differentiating Choroidal Nevus from Small Melanoma

- Fundus autofluorescence (FAF) aids in detection of subclinical orange pigment (lipofuscin) = ↑ malignancy risk
- Photography diameter >5mm = ↑ malignancy risk
- Ultrasound acoustic hollowness = ↑ malignancy risk
- OCT aids in the detection of subtle SRF and overlying retinal abnormalities = ↑ malignancy risk
- EDI OCT aids in measuring tumor thickness (OCT measurements approx. half of ultrasound measures)

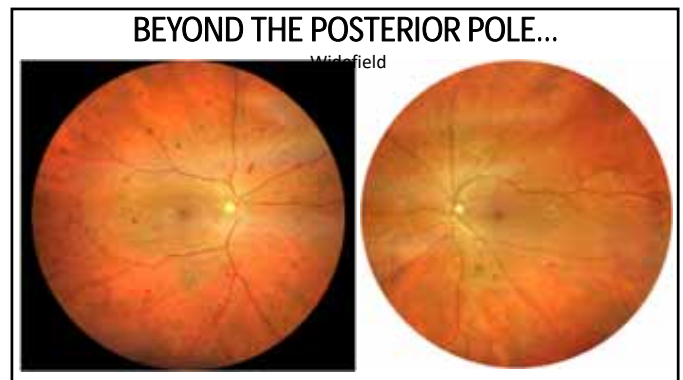
32

BEYOND THE POSTERIOR POLE...

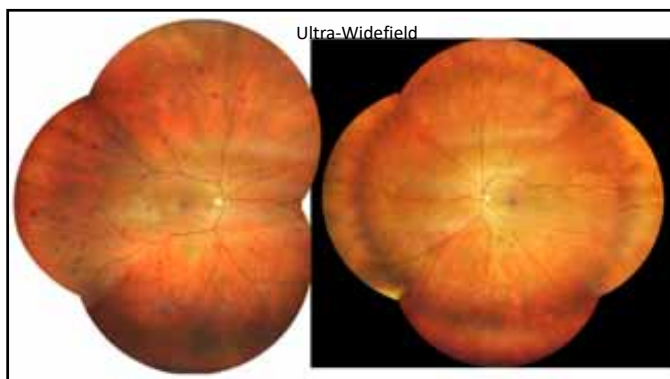
70yo American Indian male – Routine exam, notes blur and ↓ vision OD since cat surg 2 years ago

- Med Hx:
 - Type 2 DM x 24 yrs (last A1c 9.1%)
 - Paroxysmal atrial fibrillation, HTN, previously diagnosed with hypercholesterolemia but PCP recently Dcd atorvastatin due to improved labsw
- Meds: Metformin, ozempic, insulin, furosemide, lisinopril, metoprolol, ASA 81mg
- BCVAs @dist:
 - OD 20/20, OS 20/20
- Entrance testing: all WNLs
- SLE: Clear PCIOLs OU
- IOPs: OD 13/ OS 14 mmHg
- BP: 155/90

33



34



35

BEYOND THE POSTERIOR POLE...

Assessment

- Type 2 DM with:
 - OD Severe NPDR without DME
 - OS Moderate NPDR without DME
- Hypertensive retinopathy OU

Why is the retinopathy so asymmetric??

WHEN PRESUMED DR IS ASYMMETRIC THINK ABOUT OCULAR ISCHEMIC SYNDROME (OIS) AND CRVO!!!

36

BEYOND THE POSTERIOR POLE...

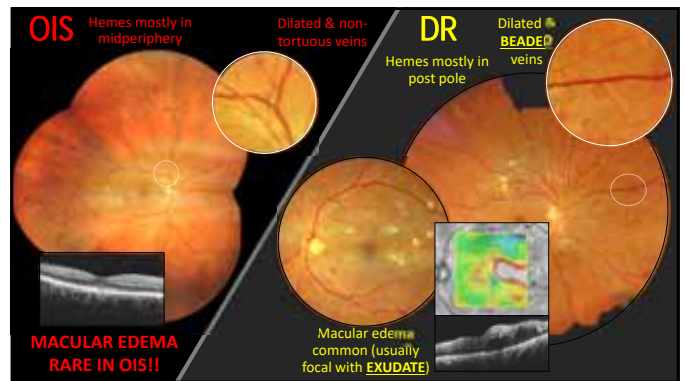
| | Ocular Ischemic Syndrome (OIS) | Diabetic Retinopathy | CRVO |
|-----------------|--|---|--|
| Symmetry | asymmetric | symmetric | asymmetric |
| Heme Shape | blot | blot | flame > blot |
| Heme Location | Midperiphery/periphery > post pole | Usually more concentrated within the post pole | Usually more concentrated within the post pole |
| Retinal Vessels | Veins dilated and non-tortuous, arteriolar attenuation | Veins dilated & beaded | Veins dilated and tortuous |
| ONH | Normal to pale | Normal | Edematous |
| Mac Edema | Rare | Common, usually focal (decentered) with exudate | Common (central & diffuse CME) |
| Onset | Insidious | Insidious | Acute |

Why is the retinopathy so asymmetric??

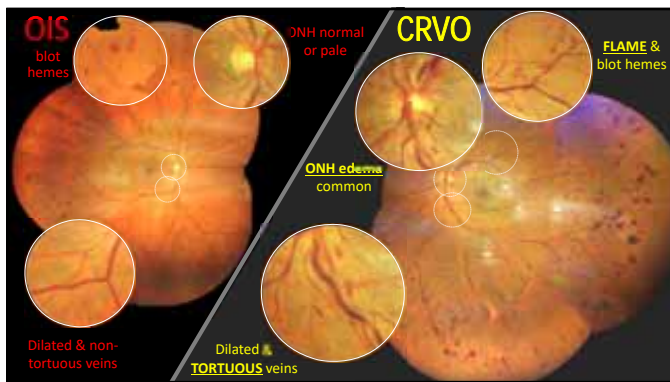


WHEN PRESUMED DR IS ASYMMETRIC THINK ABOUT OCULAR ISCHEMIC SYNDROME (OIS) AND CRVO!!!

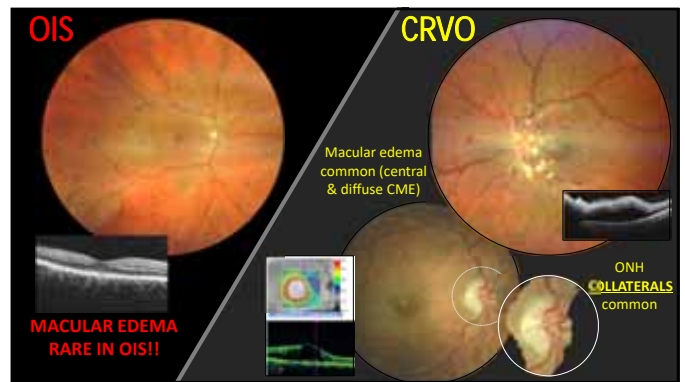
37



38



39

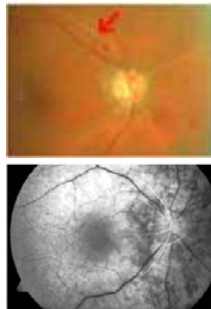


40

BEYOND THE POSTERIOR POLE.....

Other Findings suggestive of OIS

- Arteriolar attenuation
- Spontaneous arterial pulse present or can be easily induced
- ↓ IOP initially
- AC rxn flare > cells
- IVFA delayed choroidal filling, prolonged arm-to-retina time



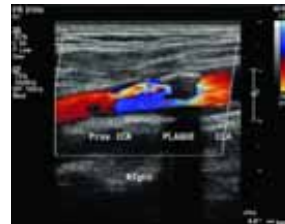
Hsieh J. Ophthalmic artery steal. Clinical and refractive optom 2017

41

BEYOND THE POSTERIOR POLE.....

Carotid Doppler US

- 70% stenosis of the right ICA
- 50-69% stenosis of the left ICA



CTA Head & Neck

- High grade 80% stenosis of the right proximal ICA 14mm distal to its origin
- 50% stenosis of the left proximal ICA



PT UNDERGOES RT CAROTID ENDARTERECTOMY!

42

BEYOND THE POSTERIOR POLE.....

OCULAR ISCHEMIC SYNDROME

What is it?

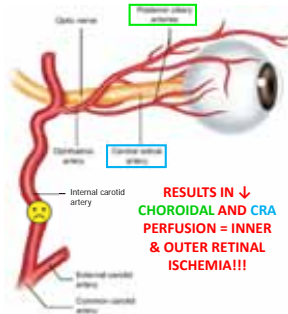
- Ocular symptoms/signs 2° to chronic, severe ocular hypoperfusion due to stenosis of the common carotid, ICA, or ophthalmic artery

Why does it happen?

- Usually secondary to atherosclerosis but can also be caused by GCA, carotid artery dissection, or other inflammation
- Typically $\geq 90\%$ ipsilateral stenosis is necessary to cause OIS (\downarrow CRA perfusion by 50%)

Who gets it?

- Usually >55 yo (mean 65yo)
- Twice as likely to affect males



43

BEYOND THE POSTERIOR POLE.....

OCULAR ISCHEMIC SYNDROME

Symptoms

- Progressive, insidious VL over weeks to months
 - 35% 20/20 to 20/40, 35% CF to LP
- Amaurosis fugax (5%)
- Light-induced amaurosis fugax
- Ocular angina
- Other transient neurologic symptoms (TIA)
- May be asymptomatic

Vision threatening complications

- Ant neo (66%) & NVG
- Post seg neo (37%)
- CRAO or ophthalmic artery occlusion



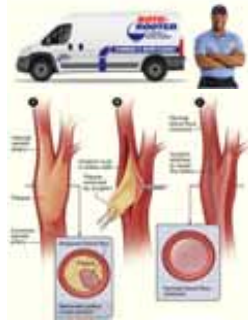
44

BEYOND THE POSTERIOR POLE.....

OCULAR ISCHEMIC SYNDROME

Systemic Management

- Consider carotid endarterectomy when:
 - Asymptomatic with 70-99% stenosis
 - Symptomatic (hx of ipsilat CVA or TIA) with 50-99% stenosis
- Medical management
 - Anti-platelet (usually ASA 81mg)
 - Statin therapy (target LDL <100 to 70mg/dL)
- Risk factor modification
 - BP, glucose, cholesterol & weight control
 - Exercise/diet
 - D/C smoking
- Thorough cardiovascular evaluation



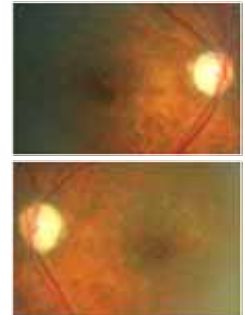
45

HYDROXYCHLOROQUINE RETINAL TOXICITY OR AMD?

66yo American Indian female

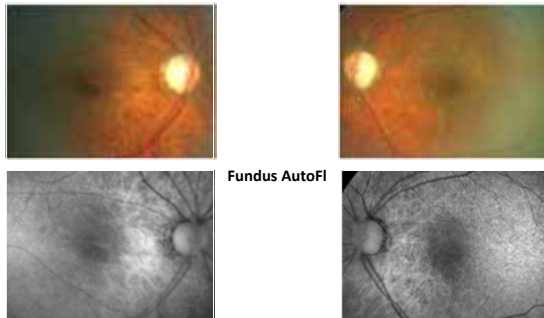
- Taking Plaquenil 200mg BID x 20+ years
- Weight: 157lbs (max daily dose = 356mg)
- + ANA, possible SLE
- Stage 3 CKD
- Ex- heavy smoker of 45yrs
- History of nonexudative AMD OU
- BCVAs OD 20/25⁺², OS 20/25

| | |
|---------|---------------------------|
| Stage 1 | 1-2 mild and/or moderate |
| Stage 2 | 3-4 mild and/or moderate |
| Stage 3 | 5-6 mild and/or moderate |
| Stage 4 | 7-8 mild and/or moderate |
| Stage 5 | 9-10 mild and/or moderate |



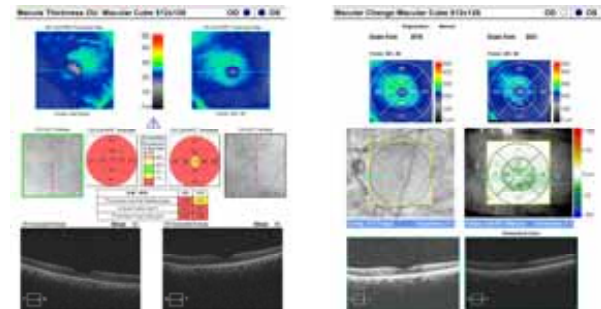
46

HYDROXYCHLOROQUINE RETINAL TOXICITY OR AMD?

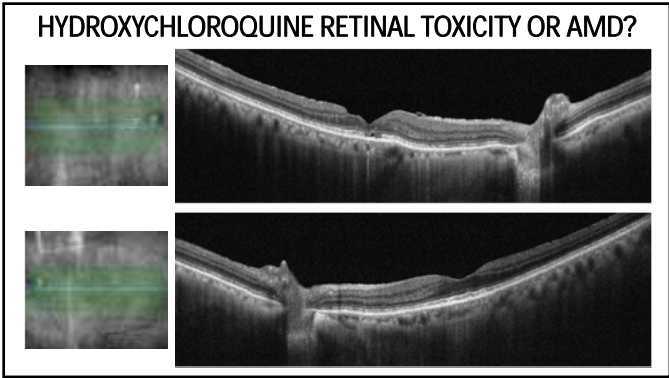


47

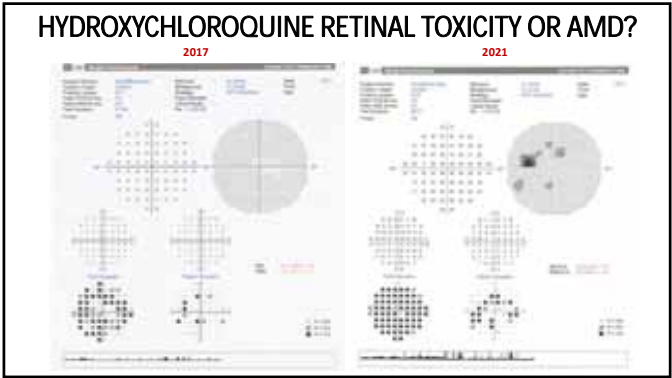
HYDROXYCHLOROQUINE RETINAL TOXICITY OR AMD?



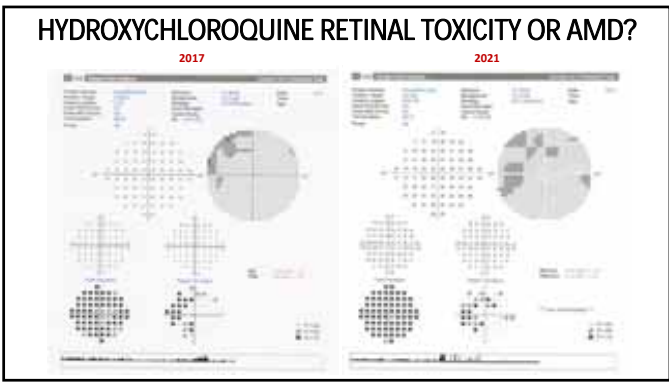
48



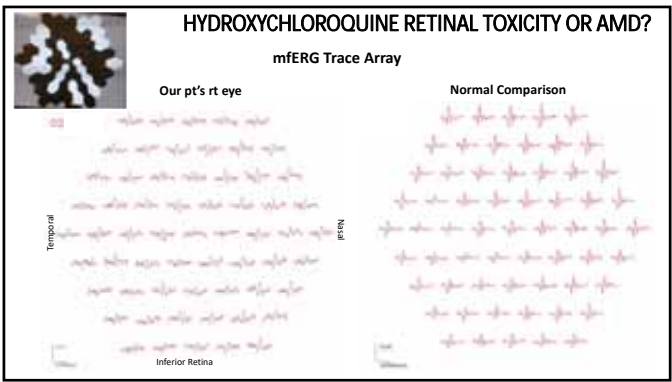
49



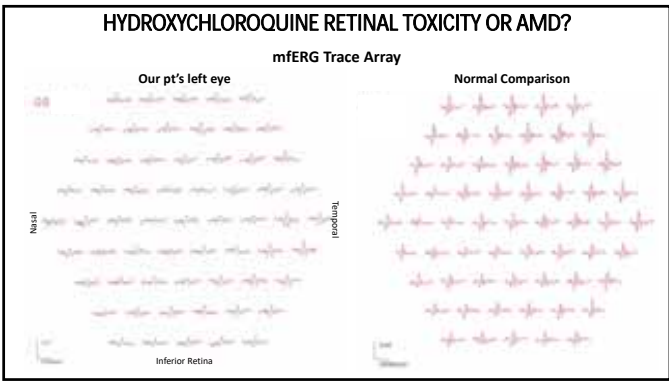
50



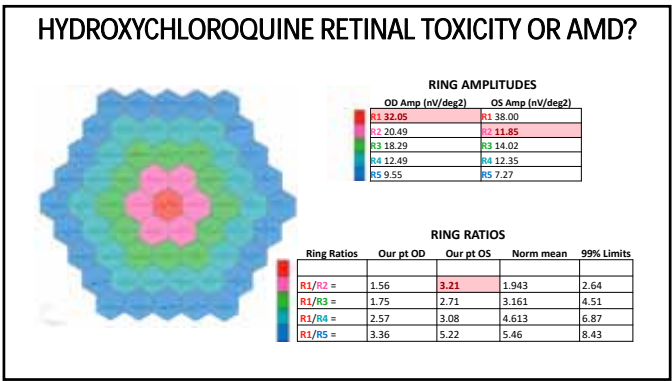
51



52



53



54

Hydroxychloroquine Retinal Toxicity

AAO 2016 Recommendations on Screening for Chloroquine and Hydroxychloroquine Retinopathy

Table 1. High-Risk Factors for Toxic Retinopathy

| | |
|-------------------|---|
| High-dose (16.5%) | >12 mg/kg total weight |
| Duration of use | >5 yrs, screening for other risk factors |
| Concomitant drugs | Hydroxychloroquine, chloroquine, tamoxifen, etc. |
| Baseline disease | Pre-existing retinal disease |
| Baseline disease | High-dose screening and monitoring is not recommended |

16.5% = chloroquine 16.5% or hydroxychloroquine 16.5%

Table 2. Clinical Examination Techniques

Recommended Screening Tests


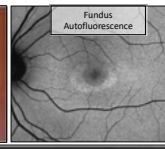
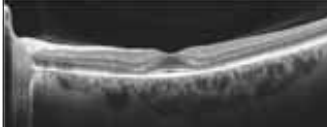
- Annual visual field (perimetry) to detect 30-2 arcuate, 30-2 arcuate
- 30-2 arcuate test (if available)
- 30-2 arcuate test (if available)
- 30-2 arcuate test (if available)

Screening schedule: baseline fundus exam, then annual screening starting at 5 yrs

ANNUAL SCREENING WITH SD-OCT AND VF AFTER 5YRS IS SOC!!!

55

Hydroxychloroquine Retinal Toxicity


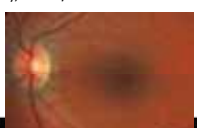


Fundus Autofluorescence

Screening schedule: baseline fundus exam, then annual screening starting at 5 yrs

56

Hydroxychloroquine Retinal Toxicity

35yo with SLE on HCQ x 16 years (200mg bid), VA 20/20 OD & OS

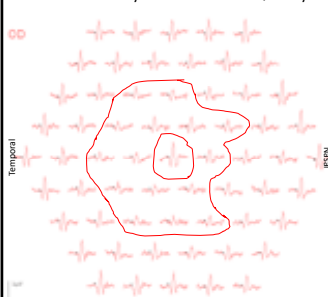






Very early OCT Changes with normal HVF!!

57

Hydroxychloroquine Retinal Toxicity

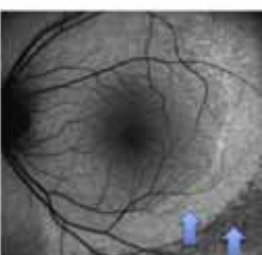
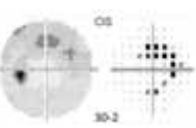
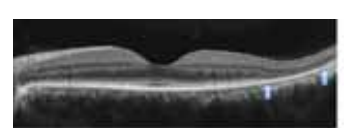
35yo with SLE on HCQ x 16 years (200mg bid), VA 20/20 OD & OS

58

Hydroxychloroquine Retinal Toxicity

Toxicity often occurs outside the macula and near the arcades in Asians

PERFORM 30-2 VF AND OCT NEAR THE ARCADES IN ASIANS!!!

AAO. Recommendations on Screening for Chloroquine and Hydroxychloroquine Retinopathy 2016.

59

PENTOSAN POLYSULFATE TOXIC MACULOPATHY

- Maculopathy associated with chronic pentosan polysulfate (Elmiron) therapy
- A common treatment for interstitial cystitis
 - A chronic regional pain syndrome of the bladder and pelvis
 - Predominately affects females manifesting with urinary urgency and dyspareunia

Table 1: Consolidated Clinical Observations

| | |
|-------------------------------------|--|
| Median Age: | 60 years (range: 37 to 79 years) |
| Median Duration of PPS Intake: | 14.5 years (range: 3 to 22 years) |
| Common Presenting Symptoms: | Blurred vision while reading (48.6 percent) Prolonged dark adaptation (48.6 percent) Metamorphopsia (11.4 percent) |
| Median Duration of Visual Symptoms: | 4 years (range: 1 to 9 years) |
| Median Visual Acuity: | OD: 20/25 OS Range: 20/20 to 20/30 OS Range: 20/15 to 20/40 |

Data documented in a series of 26 confirmed cases of PPS-associated maculopathy.¹¹

<https://www.reviewofophthalmology.com/article/clinical-pearls-for-a-new-condition>

60

PENTOSAN POLYSULFATE TOXIC MACULOPATHY



<https://www.reviewofophthalmology.com/article/clinical-pearls-for-a-new-condition>

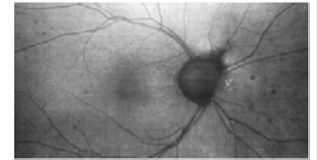
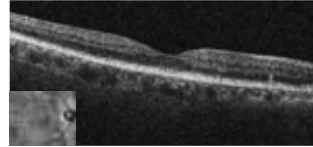
61

PENTOSAN POLYSULFATE TOXIC MACULOPATHY

Case in Point!

62yo American Indian female

- Took pentosan polysulfate for ~ 10 yrs for interstitial cystitis
- BCVAs 20/20 OD and OS

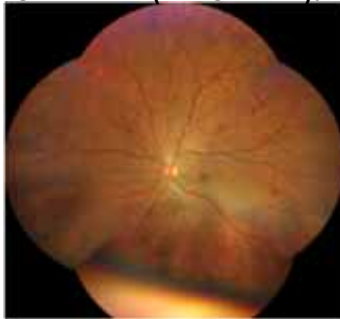


62

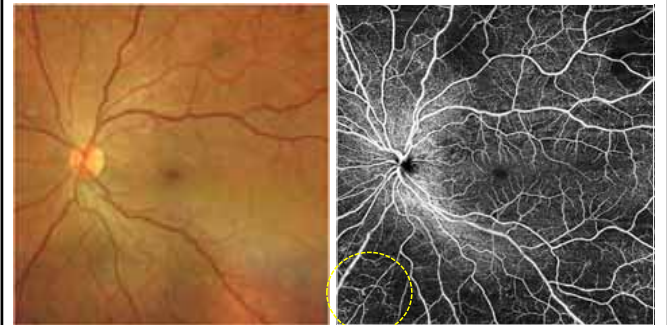
PROLIFERATIVE OR NONPROLIFERATIVE (NVE OR IRMA)?

43yo American Indian male

- CC: Stable vision OU.
- LEE: 1 yr ago diagnosed with moderately-severe NPDR
- Type 2 DM x 18 years (HbA1C 8.5%)
- VA (cc @ dist)
 - OS: 20/20
- BP: 142/85

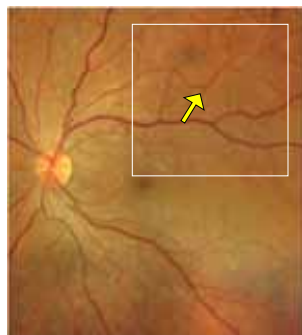
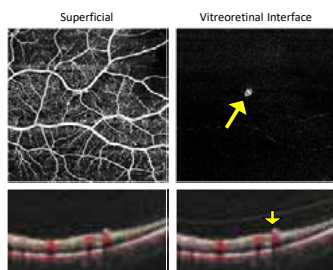


63



64

8mm OCTA Superior Temporal Arcade Tiny NVE!

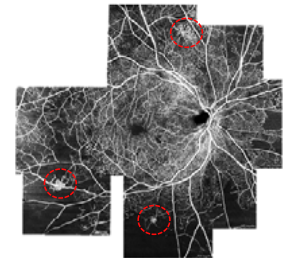
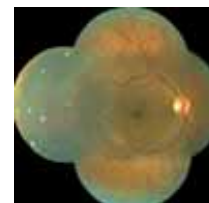


65

PROLIFERATIVE OR NONPROLIFERATIVE?

You QS et al. Detection of Clinically Unsuspected Retinal Neovascularization with Wide-field OCTA. 2019

- Performed wide-field OCTA on 27 eyes with NPDR via DFE & color fundus photography
- Of the 7 eyes originally graded as severe NPDR, wide field OCTA detected neovascularization in 4 eyes (57%)
- 2 of these eyes would have been missed with 6x6mm scan alone



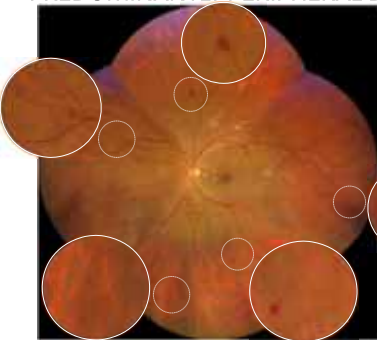
66

PREDOMINANTLY PERIPHERAL DIABETIC RETINOPATHY

S, et al. UWF Peripheral Lesions Predict Progression. Ophthalmology 2015.

- Followed 200 DR eyes for ~ 4 yrs
- Eyes with predominantly peripheral DR defined as majority of DR lesions outside the 75° ETDRS standard 7 fields
- Compared to eyes without, eyes with predominantly peripheral DR had a 3.2-fold ↑ risk of 22-step DR progression (11% vs. 34%), and a 4.7-fold ↑ risk for progression to PDR (6% vs. 25%).

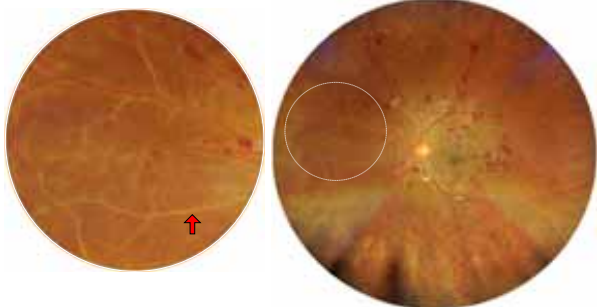
EYES WITH PREDOMINANTLY PERIPHERAL DR HAVE A GREATER RISK FOR DR PROGRESSION AND DEVELOPMENT OF PDR!!



67

PREDOMINANTLY PERIPHERAL DIABETIC RETINOPATHY

Peripheral vascular sheathing = massive retinal ischemia/NP

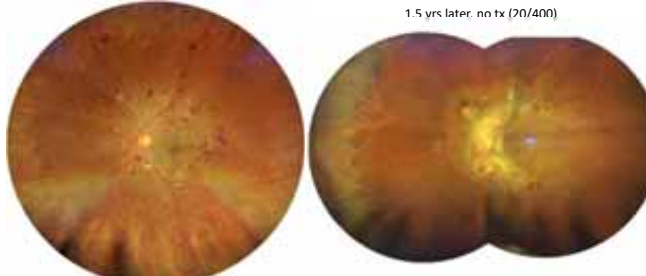


68

PREDOMINANTLY PERIPHERAL DIABETIC RETINOPATHY

Peripheral vascular sheathing = massive retinal ischemia/NP

Baseline (20/50) 1.5 yrs later, no tx (20/400)



69

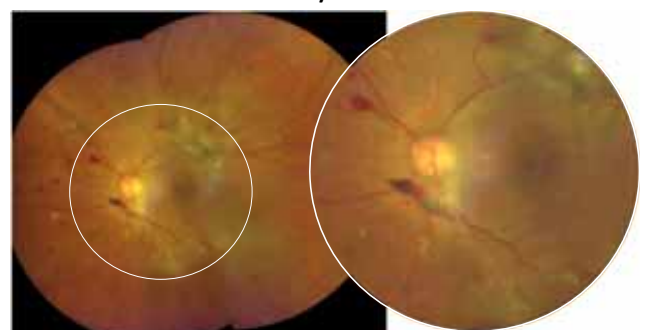
TRACTION RD OCT/B-SCAN CORRELATES

63yo American Indian male

- CC: Central blur OS, prior severe NPDR OU per LEE 1.5yrs ago. Went to retina consult but no tx done.
- Type 2 DM x 20 years (HbA1C 7.7%), CHF
- VA (cc @ dist)
 - OD: 20/25-2
 - OS: 20/250 PHNI (20/40 1.5 yrs ago)

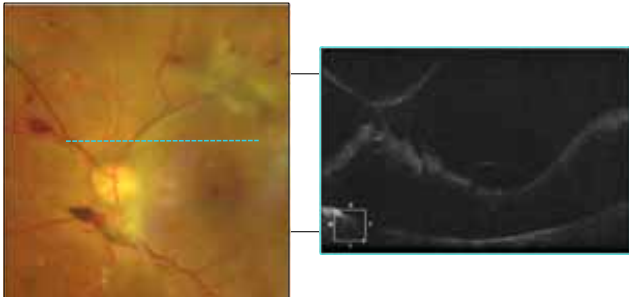
70

TRACTION RD OCT/B-SCAN CORRELATES

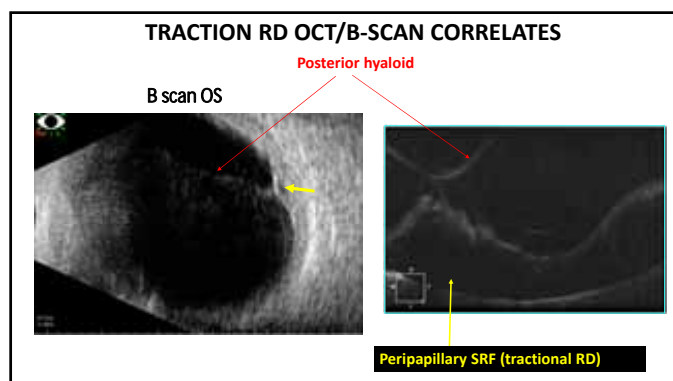


71

TRACTION RD OCT/B-SCAN CORRELATES



72



73

HOPE ON THE HORIZON

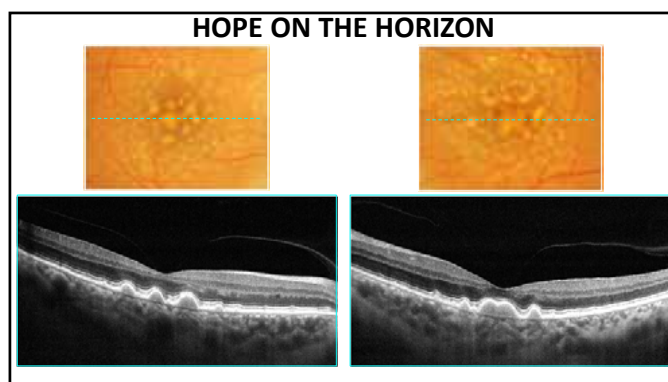
60yo American Indian female

- No vision complaints, no changes via home amsler screening
- Dry AMD x 2 yrs, taking AREDS 2
- Med Hx: preDM, dyslipidemia, former smoker
- LEE approx. 15 months ago
- BCVA OD: 20/25⁻¹, OS: 20/30⁺¹

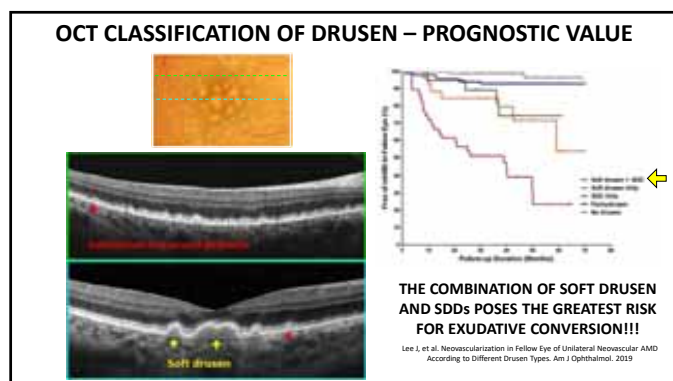
74



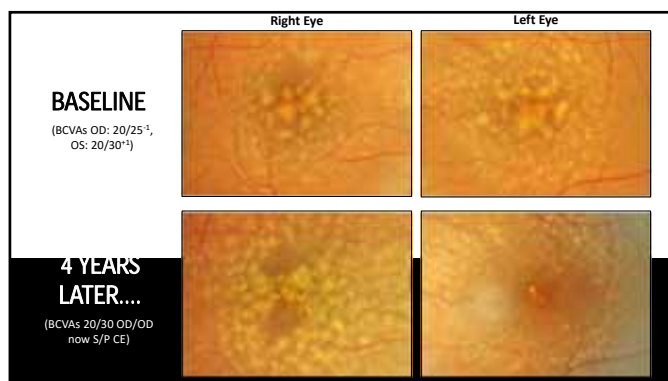
75



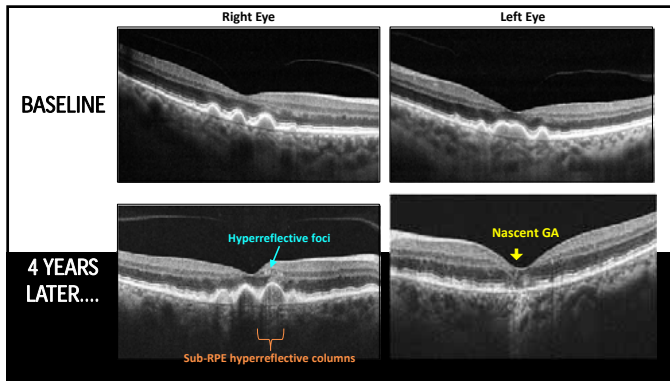
76



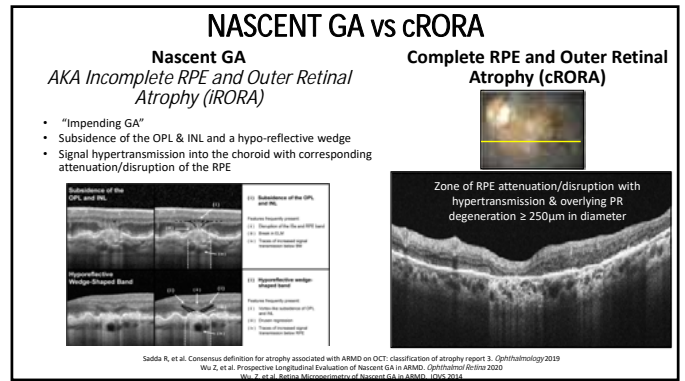
77



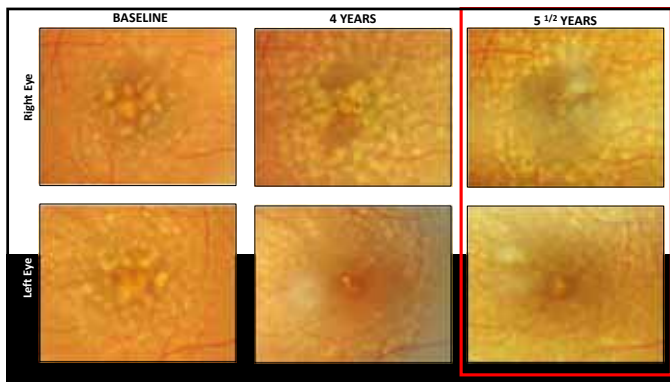
78



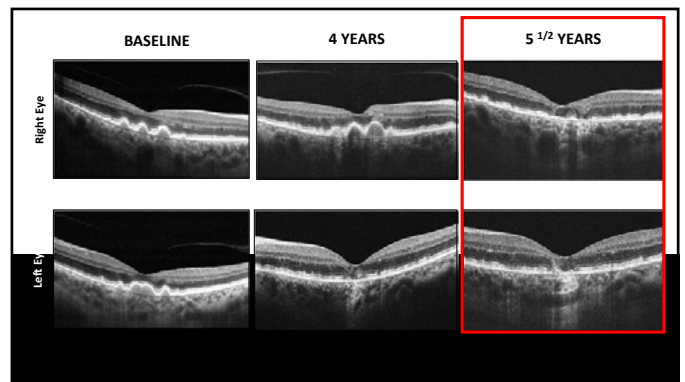
79



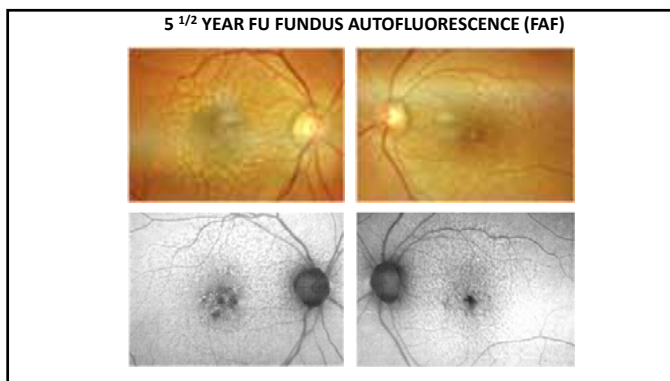
80



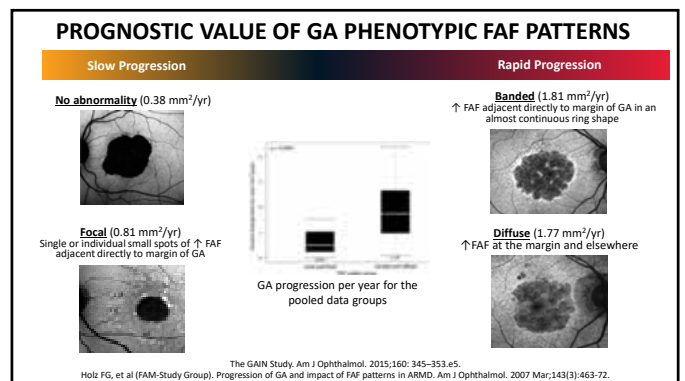
81



82



83



84

HOPE ON THE HORIZON Emerging Treatments for Geographic Atrophy (GA)

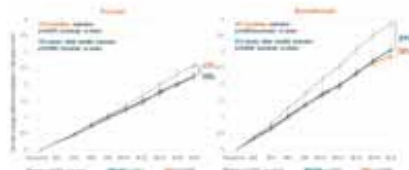
- GA progression is irreversible
- In the AREDS study, the median time from any GA diagnosis to foveal involvement was 2.5 yrs
- Compliment inhibitors
 - Pegcetacoplan (APL-2) – submitted to FDA for approval
 - Avacincaptad Pegol
 - Danicopan

AREDS Research Group. Change in area of GA in the AREDS. AREDS report number 26. Arch Ophthalmol 2009

85

EMERGING TREATMENTS FOR GEOGRAPHIC ATROPY (GA)

- Pegcetacoplan (APL-2): 18 month combined results from phase 3 studies DERBY & OAKS
 - Inclusion:
 - BCVA 20/320 or better
 - Total GA area between 2.5 - 17.5 mm² (1 - 7 disc areas) via FAF
 - Any pattern of FAF hyperautoFL in the junctional zone of GA
 - Greater effectiveness in extrafoveal GA



<https://investor.apellis.com/news-releases/news-release-details/apellis-announces-detailed-18-month-results-phase-3-derby-and-oaks>

86

Take Home Message

- Recognize OCT biomarkers for conversion from intermediate nonexudative AMD to advanced AMD
- Begin looking for patients with GA & nascent GA that may benefit from future therapies
- Use OCTA to screen for vascularization in large, presumed drusenoid PEDs
- Use FAF to detect GA and look for high risk phenotype patterns suggestive of progression



87

DON'T WAKE THE SLEEPING DRAGON

68yo male

CC: Routine exam, no visual complaints

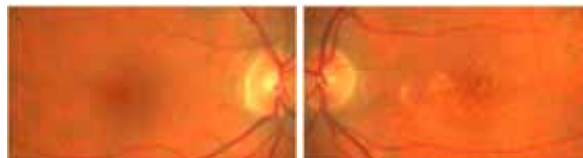
Oc Hx:

Dry AMD x 5 years OU, taking AREDS 2
Cataract NS 1+ OU

Med Hx:

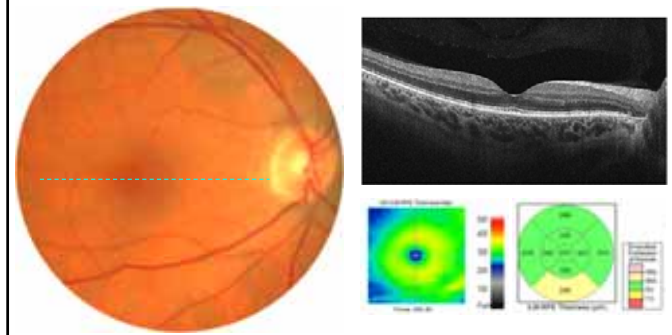
HTN, Type 2 DM
Never smoker

Vision: BCVAs @dist
OD 20/20
OS 20/40+1



88

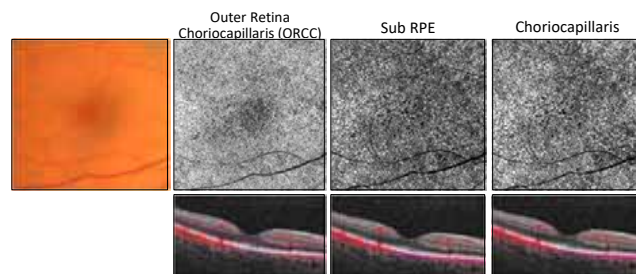
DON'T WAKE THE SLEEPING DRAGON



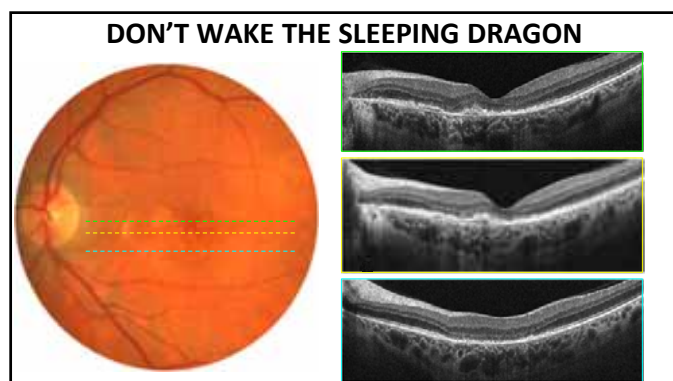
89

DON'T WAKE THE SLEEPING DRAGON

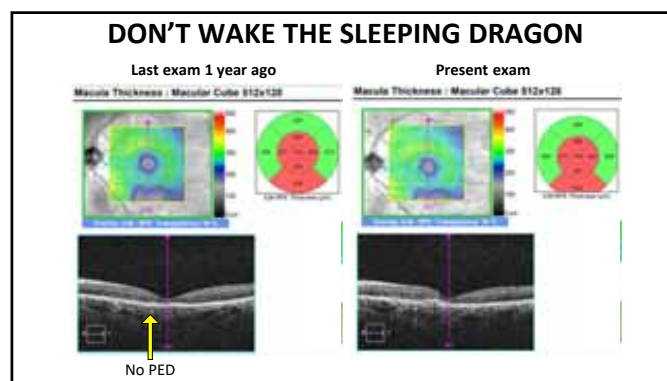
OCT Angiography 6mm Macula OD



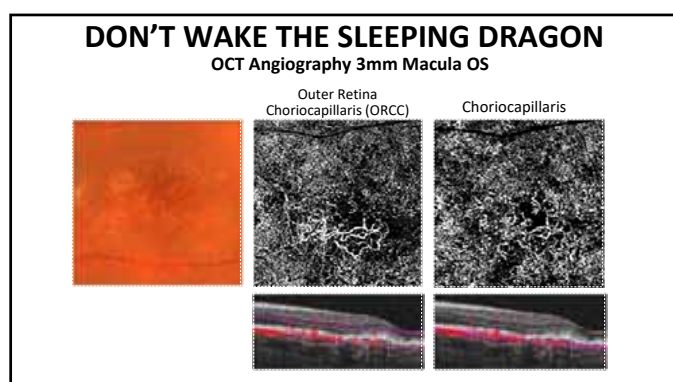
90



91



92



93

DON'T WAKE THE SLEEPING DRAGON

| Assessment |
|---|
| <p>OD Early stage non-exudative AMD</p> <p>OS Non-exudative AMD with probably quiescent CNV</p> |
| Management |
| <p>FU 3 months</p> <p>Cont. amsler & AREDS 2</p> |

94

DON'T WAKE THE SLEEPING DRAGON

No shows 3 month follow up appt, returns 6 months later

CC: Still no vision complaints, reports stable vision, no changes noted on amsler

Vision: BCVAs @dist

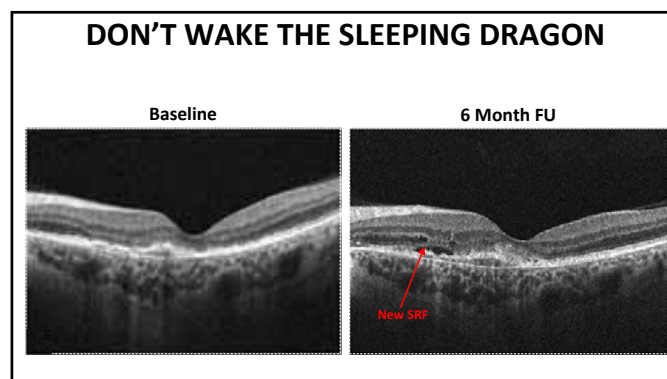
OD 20/20⁻¹

OS 20/40⁻¹

No change in fundus or OCT appearance OD

No appreciable change in fundus appearance OS via ophthalmoscopy.....

95



96

DON'T WAKE THE SLEEPING DRAGON

OCT Angiography 3mm Outer Retina Choriocapillaris OS

Baseline

6 Month FU (6mm)

97

DON'T WAKE THE SLEEPING DRAGON

Assessment

OD Early stage non-exudative AMD
OS **EXUDATIVE** AMD

Management

Refer to retina for intravitreal anti-VEGF

98

NONEXUDATIVE CNV

- Well-defined neovascular complex via OCTA
- No signs of exudation via ophthalmoscopy such as exudate or blood
- No fluid via structural OCT
- No leakage with IVFA

Present in approx. 10% of high risk AMD eyes (intermediate AMD, exudative fellow eye)

Carnevali A, et al. OCTA: A Useful Tool for Diagnosis of Treatment-Naïve Quiescent CNV. Am J Ophth. 2016.

Or C, et al. Incidence of Vascularized Drusen in Non-Exudative ARMD using SD-OCTA. ARVO 2018.

99

NONEXUDATIVE CNV

Prognosis

Rate of future exudation, eyes with nonexudative CNV vs eyes without nonexudative CNV
Bailey S ARVO 2017. 60% vs 4% (5 months)
De Oliveira Dias J Ophthal 2018. 21% vs 4% (12 months)
15x greater risk of exudation after detection of nonexudative CNV

EYES WITH NONEXUDATIVE CNV ARE AT HIGH RISK FOR EXUDATIVE CONVERSION!

Bailey S et al. Early detection of CNV with OCTA. ARVO 2017.

De Oliveira Dias JR, et al. Natural History of Subclinical Neovascularization in Nonexudative ARMD Using SS-OCTA. Ophthalmol 2018.

100

Exudative & Neovascular AMD Features

Hemorrhage, blood, fluid, exudate, PED, OCTA membranes

EARLY DETECTION AND PROMPT TREATMENT OF NEO IS CRITICAL TO MAXIMIZE VISUAL OUTCOMES!!!

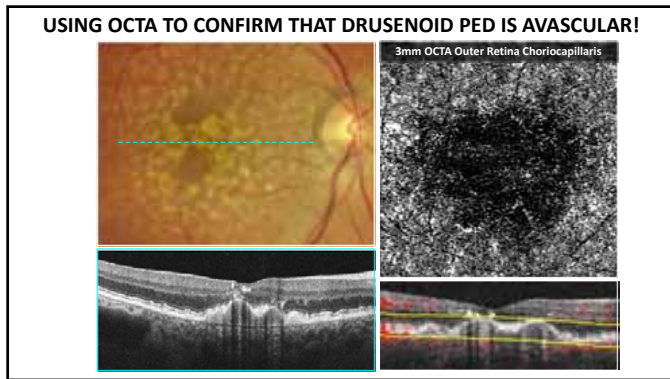
101

Subtypes of RPE Detachments (PEDs)

fibrovascular, hemorrhagic, serous, drusenoid

102

17



103

DON'T WAKE THE SLEEPING DRAGON
Take Home Message

Non-exudative CNV

Non-exudative CNV is a well-formed CNV membrane in an untreated eye that has no OCT (fluid) or ophthalmoscopic signs of leakage (hemorrhage, exudative, fluid) and does not leak with IVFA.

OCTA is the only method of detecting and monitoring growth of non-exudative CNV membranes.

Non-exudative CNV in AMD carries a substantial risk for conversion from nonexudative to exudative AMD.

104

THE CURIOUS CASE OF THE SWOLLEN DISCS

6yo white female

- CC: First eye exam. No ocular or systemic complaints per patient and mother.
- Oc Hx: unremarkable
- Medical Hx: unremarkable
- Meds: none
- Final Refraction
 - OD: +1.00 -1.25x176: 20/20-3
 - OS: +1.50 -1.75x002: 20/20-2
- Entrance testing and SLE of ant seg: Unremarkable OU
- IOPs 21/20mmHg
- BP: 98/66

105

THE CURIOUS CASE OF THE SWOLLEN DISCS

(-) SVP

106

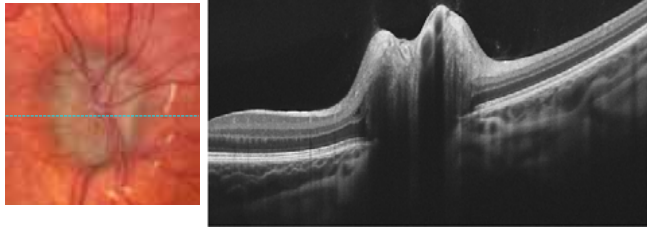
THE CURIOUS CASE OF THE SWOLLEN DISCS
 Fundus Autofluorescence

107

THE CURIOUS CASE OF THE SWOLLEN DISCS

108

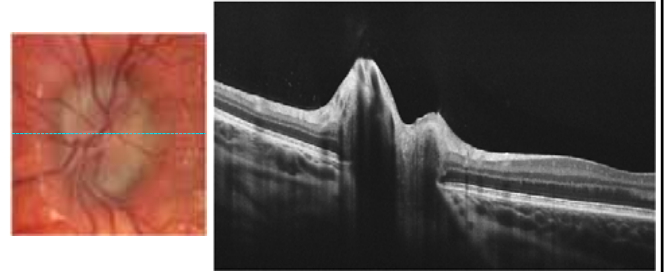
THE CURIOUS CASE OF THE SWOLLEN DISCS



OCT: Subtle intraretinal peripapillary cystic fluid and probable anterior angling of Bruchs temporal

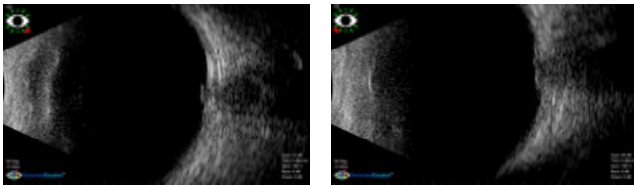
109

THE CURIOUS CASE OF THE SWOLLEN DISCS



110

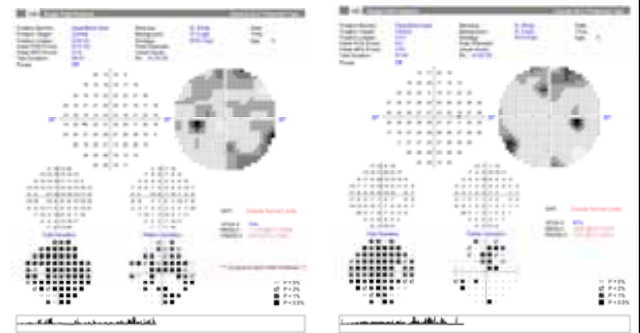
THE CURIOUS CASE OF THE SWOLLEN DISCS B-scan OD/OS



No disc drusen, crescent sign visible OD, increased fluid in subarachnoid space OU

111

And yes.....I put a 6-year-old through a VF



112

THE CURIOUS CASE OF THE SWOLLEN DISCS MRI/MRV brain and orbits with and without contrast

MRI Head

- Mild concavity to the superior margin of the pituitary gland (AKA: partially empty sella)...otherwise no intracranial space-occupying lesion and no evidence of acute hydrocephalus.

MRI Orbits

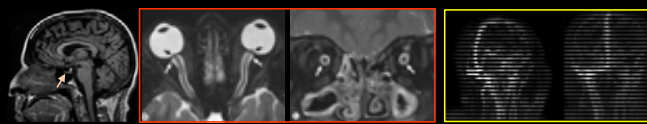
- Bilateral optic disc bulge and **increased nerve sheath CSF** (distended).

MRI Head

- The **left transverse sinus appears to be significantly hypoplastic**. Rt transverse sinus is dominant. No evidence of dural venous sinus thrombosis.

IMPRESSIONS

- Findings suggestive of raised intracranial pressure.



113

THE CURIOUS CASE OF THE SWOLLEN DISCS Lumbar Puncture

Opening Pressure

- Elevated at **288 mm of H2O**

ASSESSMENT

- Pediatric IIH

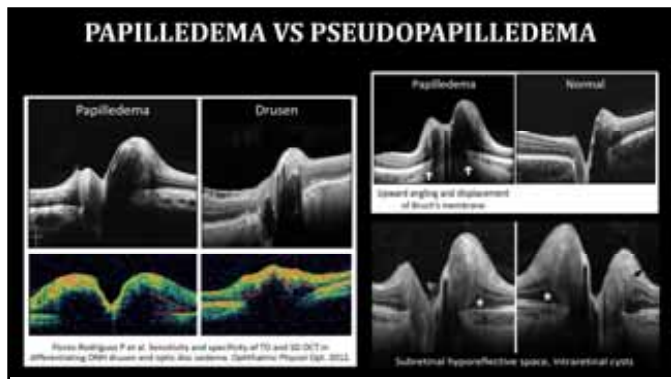
CSF Analysis/cytology

- Glucose, lymphocytes, monocytes, protein and macrophages **all WNL**. No atypical or malignant cells identified. VDRL non-reactive and cryptococcus antigen negative.

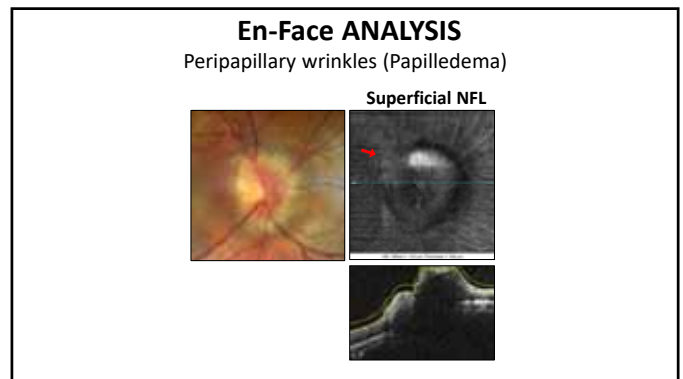
PLAN

- Acetazolamide 9.4ml (235mg total) by mouth every 12 hours

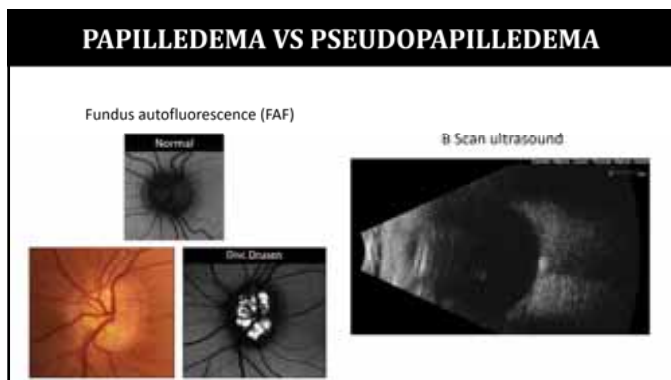
114



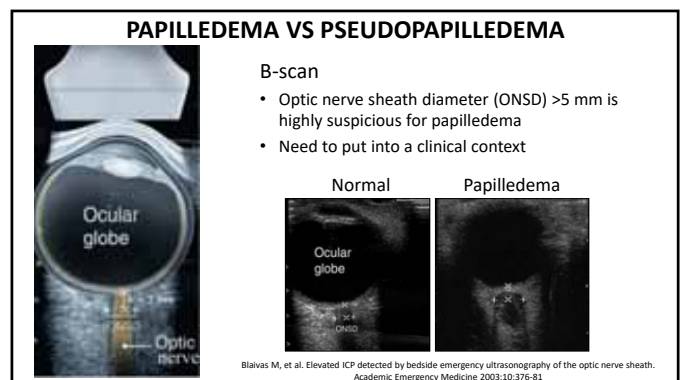
115



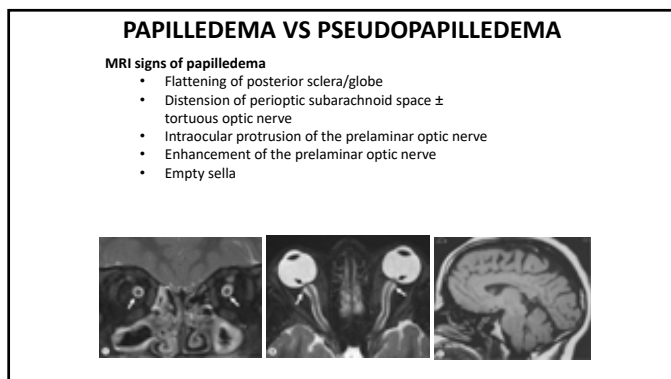
116



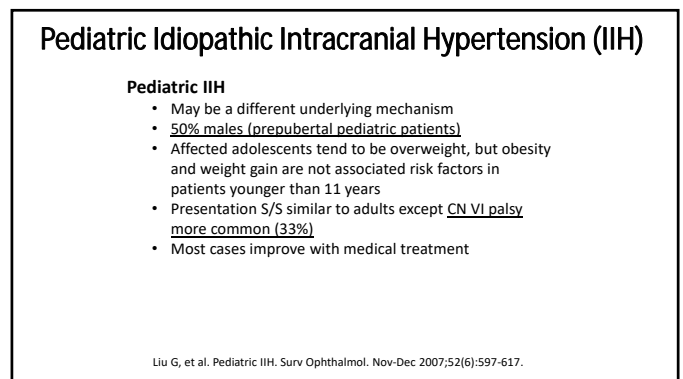
117



118



119



120

Take Home Message

Differentiating Papilledema from Pseudopapilledema

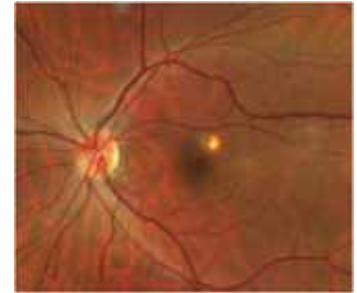
- Symptoms/signs
- FAF: rule out superficial disc drusen
- B-scan: rule out buried/deep drusen, look for ↑ subarachnoid fluid and thickening of the ONSD
- OCT: Peripapillary slope/contour of swelling, fluid, contour Bruch's membrane
- MRI: Rule out hydrocephalus, features of papilledema include flattening of globe, distension of ON subarachnoid space, empty sella

121

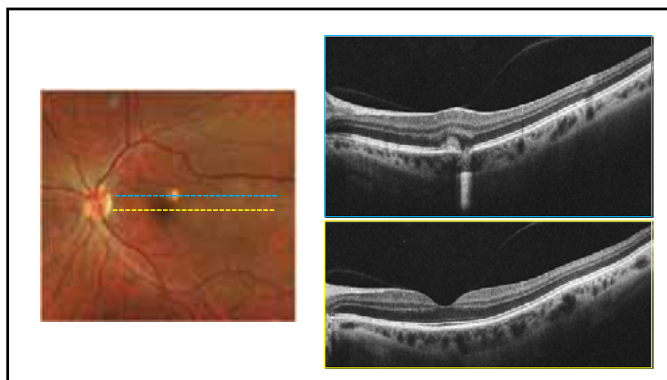
ACUTE ONSET METAMORPHOPSIA

38yo American Indian female

- CC: new onset floaters and slightly distorted vision OS x 3 days
- Oc Hx: LASIK OU 2010, pt reports there were "old scars" seen in the back of the eyes when she had LASIK done
- Med Hx: WNL
- BCVA :
 - OD: 20/20
 - OS: 20/25+2



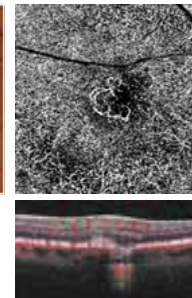
122



123

OD OCT Angiography 3x3mm Macula

Outer Retina Choriocapillaris (ORCC)



Why does a 38yo have macular CNV?



Myopia?
Trauma (choroidal rupture)?
POHS?
Angioid streaks?
Idiopathic?

124

PRESUMED OCULAR HISTOPLASMOSIS SYNDROME (POHS)



Clinical POHS Triad:

1. Histo spots: multiple atrophic "punched out" chorioretinal scars within the midperiphery/posterior pole
2. Peripapillary atrophy
3. Maculopathy: CNV & subretinal neo possible

NO VITRITIS!!!

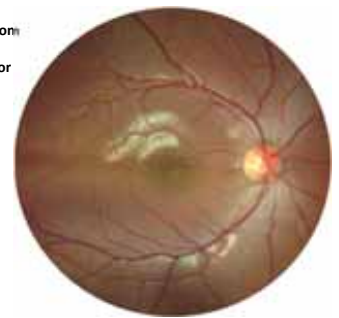
125

WHY THE REDUCED VA?

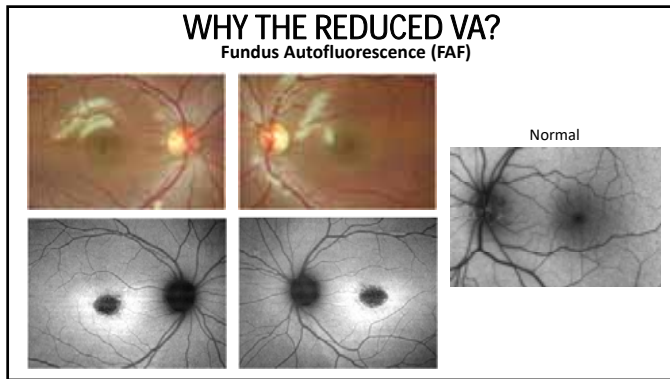
15yo American Indian female- Complains worsening blur at D and N with new glasses from 6 months ago

- POH: VT for accommodative spasm, prior dx of Streff syndrome
 - Hx of ↓ BCVAs:
 - 2016 - 20/25 OD/OS
 - 2019 - OD 20/70, OS 20/50
 - 2020 - 20/70 OD/OS
 - 2022
 - OD -1.50-0.50x180 20/150 PHNI
 - OS -1.75-0.50x180 20/150 PHNI

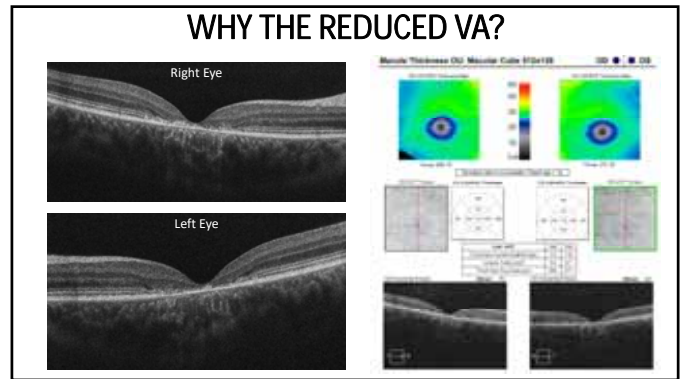
- MH: Unremarkable
- Oc fam Hx: Unremarkable
- Entrance testing: Normal
- External exam: Normal OU
- Tonometry: 14/14 mmHg



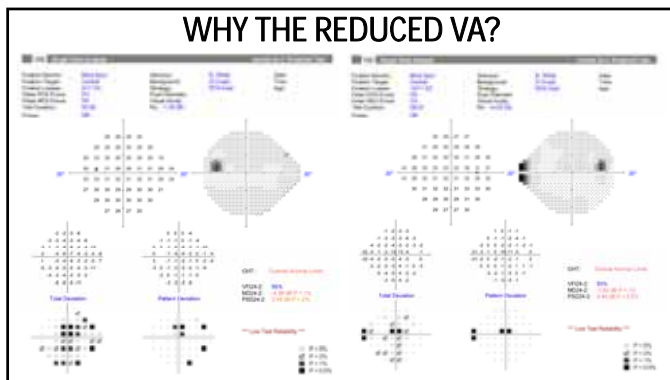
126



127



128



129

| WHY THE REDUCED VA? |
|---|
| Assessment |
| <ul style="list-style-type: none"> Probable inherited retinal disease (IRD) <ul style="list-style-type: none"> suspect Stargardt disease |
| Management |
| <ul style="list-style-type: none"> Get genetic testing- results showed RDH12 (Retinal Dehydrogenase 12) mutation consistent with autosomal recessive Leber Congenital Amaurosis (LCA) Refer to LV |

130

GENETIC TESTING FOR IRD

Why perform testing?

- Can confirm or change diagnosis
- More accurate prognosis
- Confirms inheritance pattern, risk for other family members
- Potential qualification for clinical trial or gene therapy

131

NO COST GENETIC TESTING, COUNSELING AND ACCESS TO CLINICAL TRIALS

OPEN ACCESS, NO COST GENETIC TESTING!!!

- 2 programs available
 - ID YOUR IRD via Invitae (Sponsored by Spark Therapeutics) - 325 gene panel
 - My Retina Tracker via Blueprint genetics (Sponsored by Foundation Fighting Blindness) - 322 gene panel
- Need clinical diagnosis or symptoms of IRD
- Identifies copy number variants (insertions, deletions) and non coding variants (intronic mutations)
- OD orders genetic test online, collects saliva sample and mails to lab
- Results in ~1 month

132

NO COST GENETIC TESTING, COUNSELING AND ACCESS TO CLINICAL TRIALS

Step 2: Genetic counseling

- Telephone based
- 60-75 minute sessions
- Following the session pts and ODs are given formal summary report and a detailed pedigree

Step 3: My Retina Tracker® Registry

- Way to connect pts with IRDs to researchers recruiting for clinical trials
- Patient can upload genetic test results
- Patient controlled, secure and HIPAA compliant
- Only de-identified data is shared with researchers
- ~ 16,000 registrants
- www.MyRetinaTracker.org

133

LEBER CONGENITAL AMAUROSIS (LCA)

What is it?

- Family of congenital retinal dystrophies
- Most common cause of inherited blindness in childhood (esp <1yo)
- Represents ~ 5% of all IRDs
- Estimated birth prevalence is 2 to 3 per 100,000 births

What causes it?

- 17 phenotypes (LCA1 to LCA17) with 25 genotypes have been identified that account for 70-80% of cases
 - CEP290 (15%), GUCY2D (12%), and CRB1 (10%) and RPE65 (8%) mutations most frequent
- Usually autosomal recessive
- Disrupts the phototransduction/visual cycle

What is the clinical presentation?

- Central foveal atrophy, "bone-spicule" like pigmentation, subretinal flecks, abnormal ERG
- Vision loss is usually severe ($\leq 20/400$), present at birth, and is generally stable or very slowly progressive
- May be associated with keratoconus, cataract, hyperopia, neurodevelopmental delay/intellectual disability rare

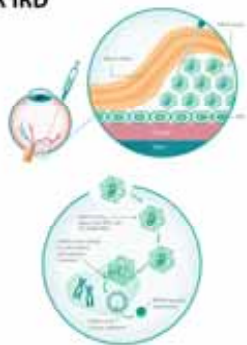
134

GENE THERAPY FOR IRD

Luxturna® (voretigene neparvovec-rzyl)

- First FDA-approved gene therapy for an eye IRD, Dec 2017
- Administered via one-time subretinal injection w/ PPV
- Must have mutations in both copies of the RPE65 gene to be eligible
 - Clinical presentation is leber's congenital amaurosis or RP
- Can locate retinal specialist who will perform treatment here: <https://luxturna.com/specialist-locator/>

Phase 3 gene therapy clinical trials underway currently for choroideremia and X-linked RP

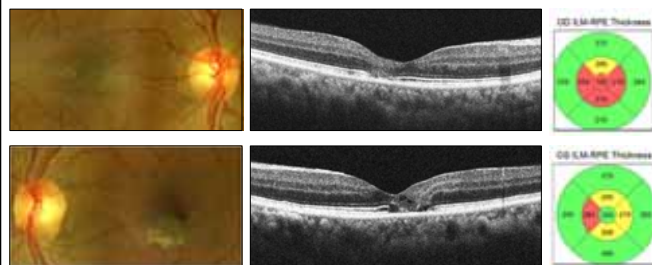


135

Macular Holes or Macular Mystery?

55yo American Indian Female

Referred from outside OD for vision loss OU and poss macular holes
BCVAs: OD 20/20⁻¹, OS 20/20



136

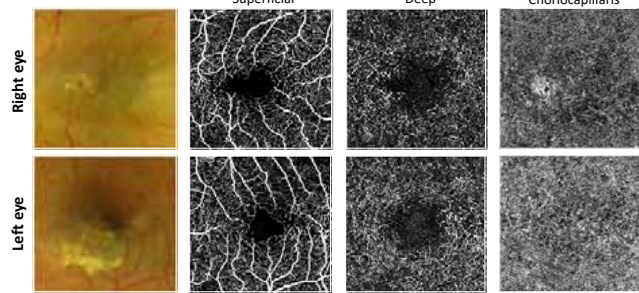
Macular Mystery

OCT Angiography (3mm macula)

Superficial

Deep

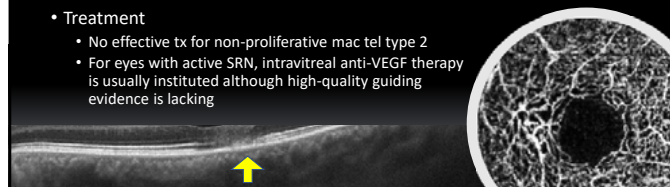
Outer Retina
Choriocapillaris



137

MACULAR TELANGIECTASIA TYPE 2

- Bilateral peripapillary telangiectatic capillaries
- **OCT: Bilateral temporal peripapillary outer retinal atrophy, ILM draping in later stage disease**
- Onset 5-6th decade, M=F
- Pathogenesis involves environmental and genetic factors
 - Autosomal dominant transmission has been described
- Treatment
 - No effective tx for non-proliferative mac tel type 2
 - For eyes with active SRN, intravitreal anti-VEGF therapy is usually instituted although high-quality guiding evidence is lacking



138

Proliferative Mac Tel Type 2

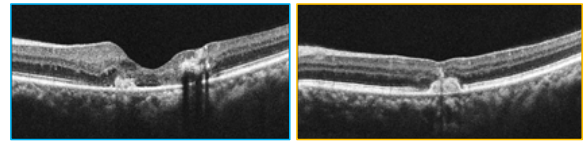
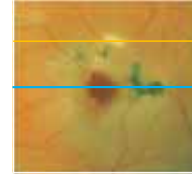
57yo Hispanic female – Referred from primary care for possible macular dystrophy/macular scarring OU

- CC: gradually progressive vision loss OU x several years
- Oc Hx: No known illness or fam Hx. No prior surg or trauma.
- Med Hx: Unremarkable
- BCVAs @dist:
 - OD 20/100 PHNI
 - OS 20/200 PHNI
- Entrance testing: WNL
- SLE: 1-2+ NS cat OU



139

Proliferative Mac Tel Type 2



140

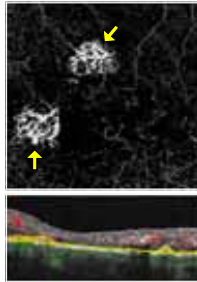
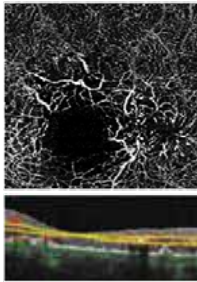
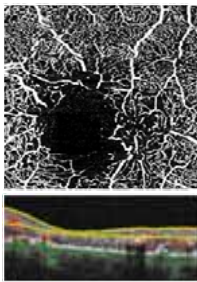
Proliferative Mac Tel Type 2

OCTA 3mm Macula OS

Superficial

Deep

Outer Retina (Custom)



141

The "Take Home Message"

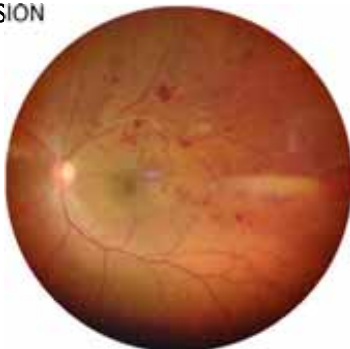
- OCTA allows for earlier detection of perfoveal telangiectasia in early, subclinical mac tel type 2
- OCTA aids in the detection of subretinal neovascularization in mac tel type 2
- Mac tel type 2 eyes with active subretinal neovascularization should be referred for consideration of treatment
 - Better prognosis with earlier treatment and small size neo

142

RETINAL VEIN OCCLUSION

40yo American Indian female

- CC: Routine exam, no complains
- Medical Hx:
- HTN (BP in-office 133/83)
- VA sc @dist
- OS 20/20
- Pupils: Equally round, (-) APD
- CVF: OD restriction inf nasal
- Ant seg: WNLs
- IOP: 15/16



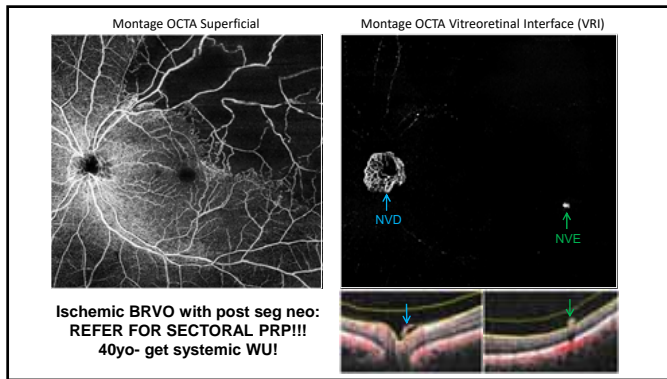
143

RETINAL VEIN OCCLUSION

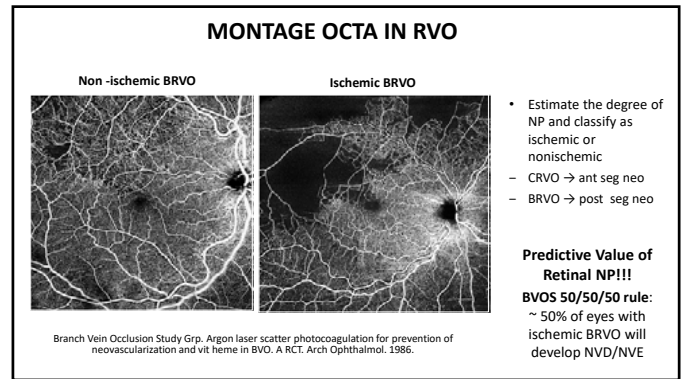
Montage OCTA superficial



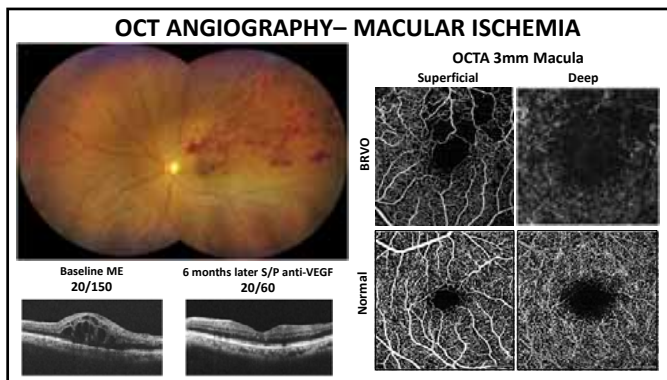
144



145



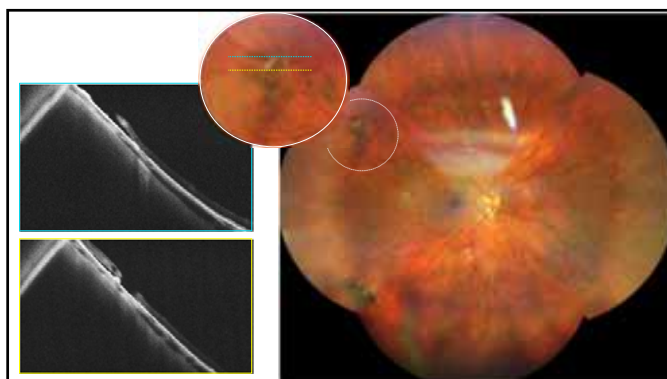
146



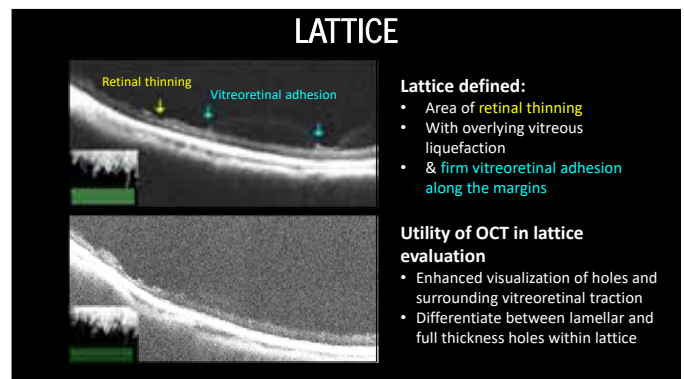
147



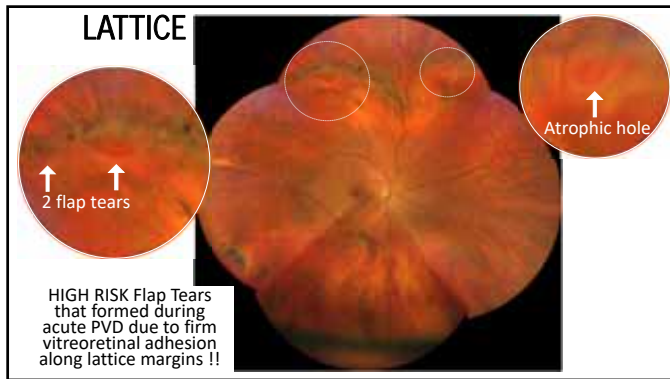
148



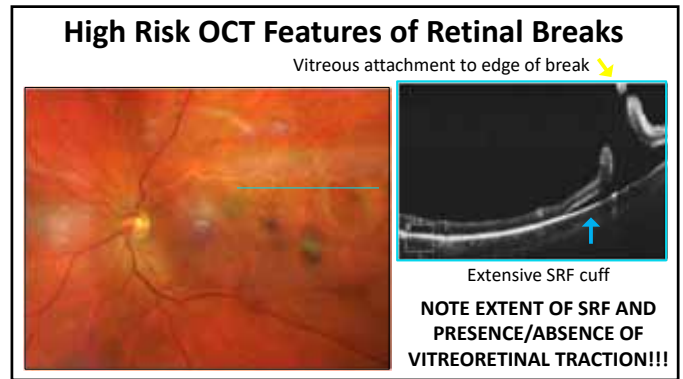
149



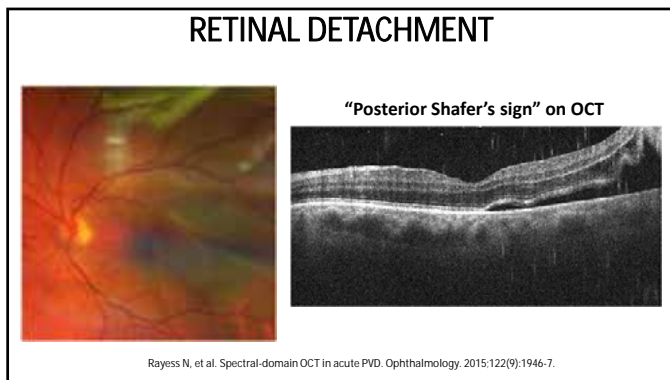
150



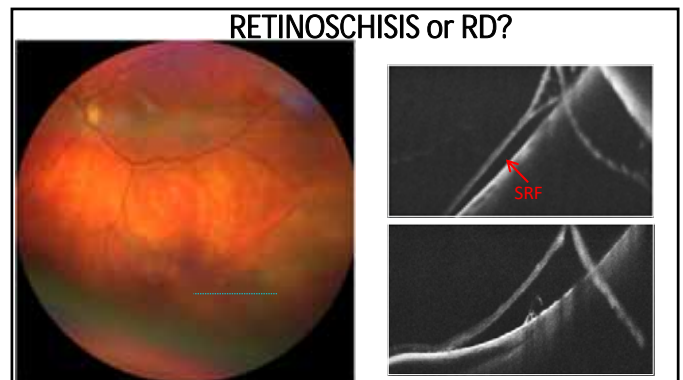
151



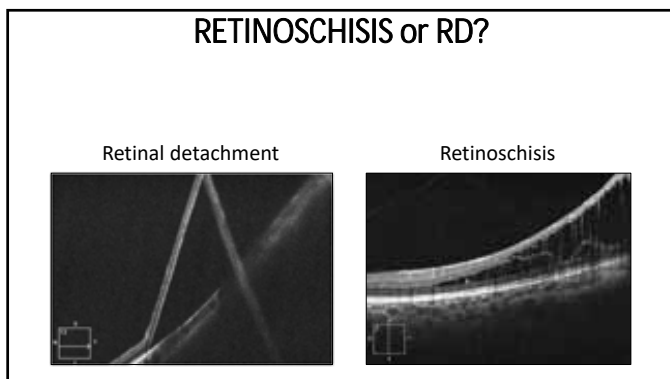
152



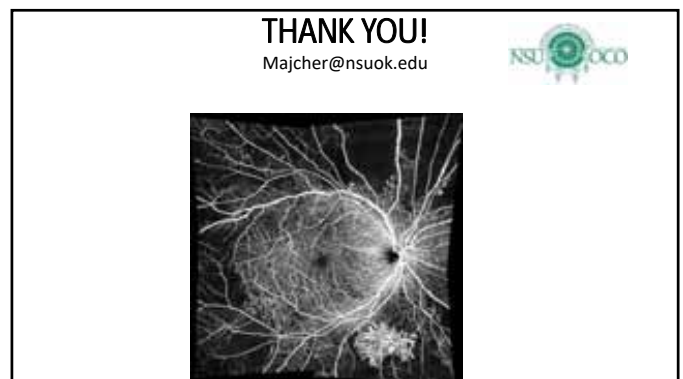
153



154



155



156