ESCS
Expanding the clinical spectrum

Retina Society 2020

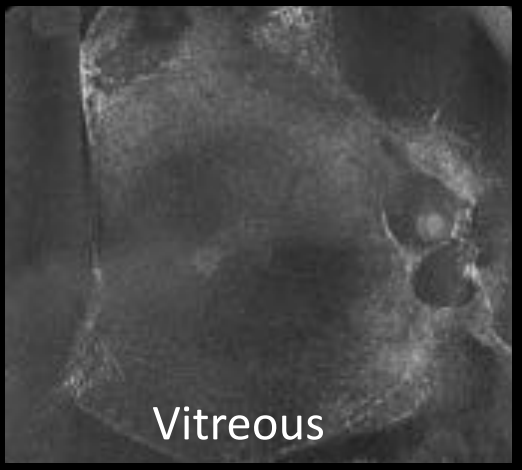
Lawrence A. Yannuzzi

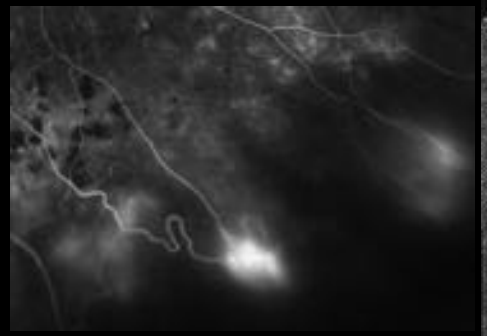
THE MACULA FOUNDATION
EXPANDING THE FIELD OF VISION

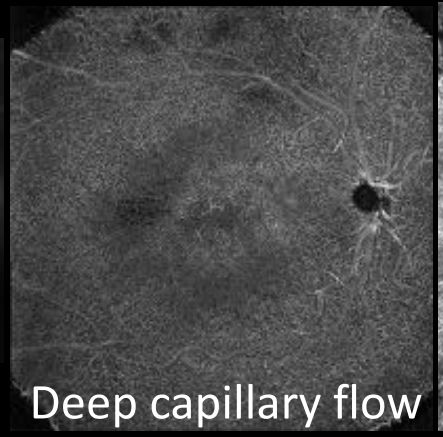
VRMNY | Vitreous Retina Macula Consultants of New York

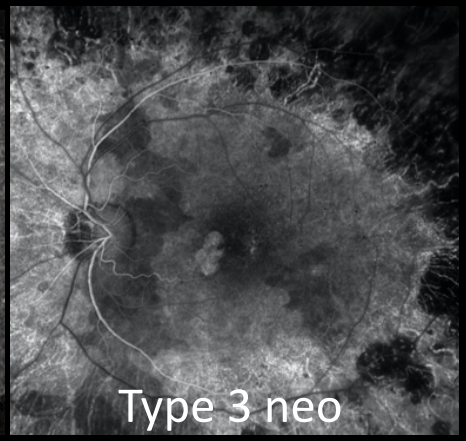
Financial Disclosures: None

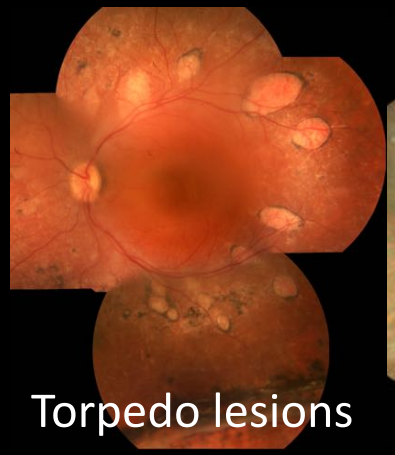
ESCS or NR2E3 is Favre-Goldmann Summary


Vitreous

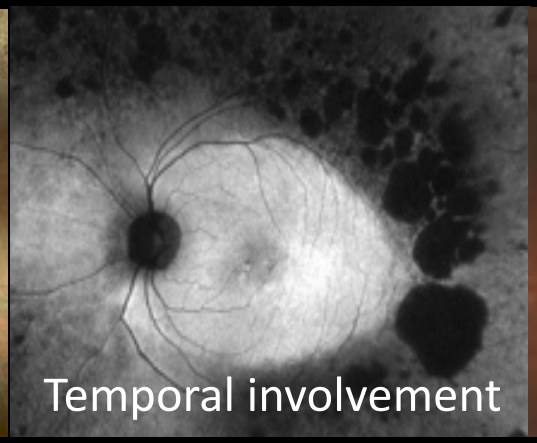

Retinal neo


Deep capillary flow


Type 3 neo


Torpedo lesions


Nummular lesions


Temporal involvement


Fibrosis

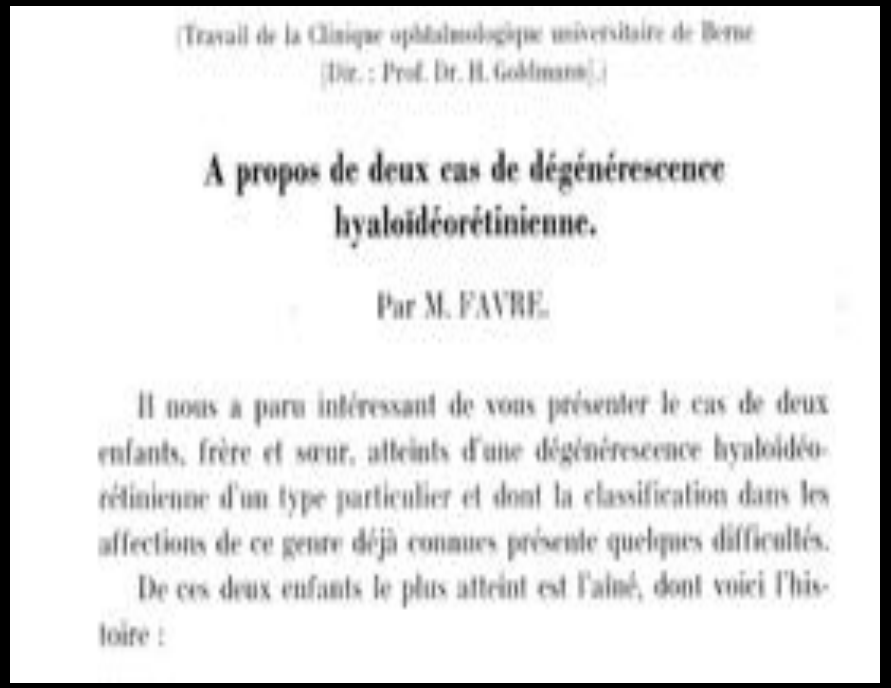
Goldmann-Favre Syndrome

Hans Goldmann
(Mentor)

Maurice Favre
(Fellow)

Favre Syndrome

Maurice Favre



(Travail de la Clinique ophtalmologique universitaire de Berne
[Dir. : Prof. Dr. H. Goldmann].)

A propos de deux cas de dégénérescence hyaloïdéorétinienne.

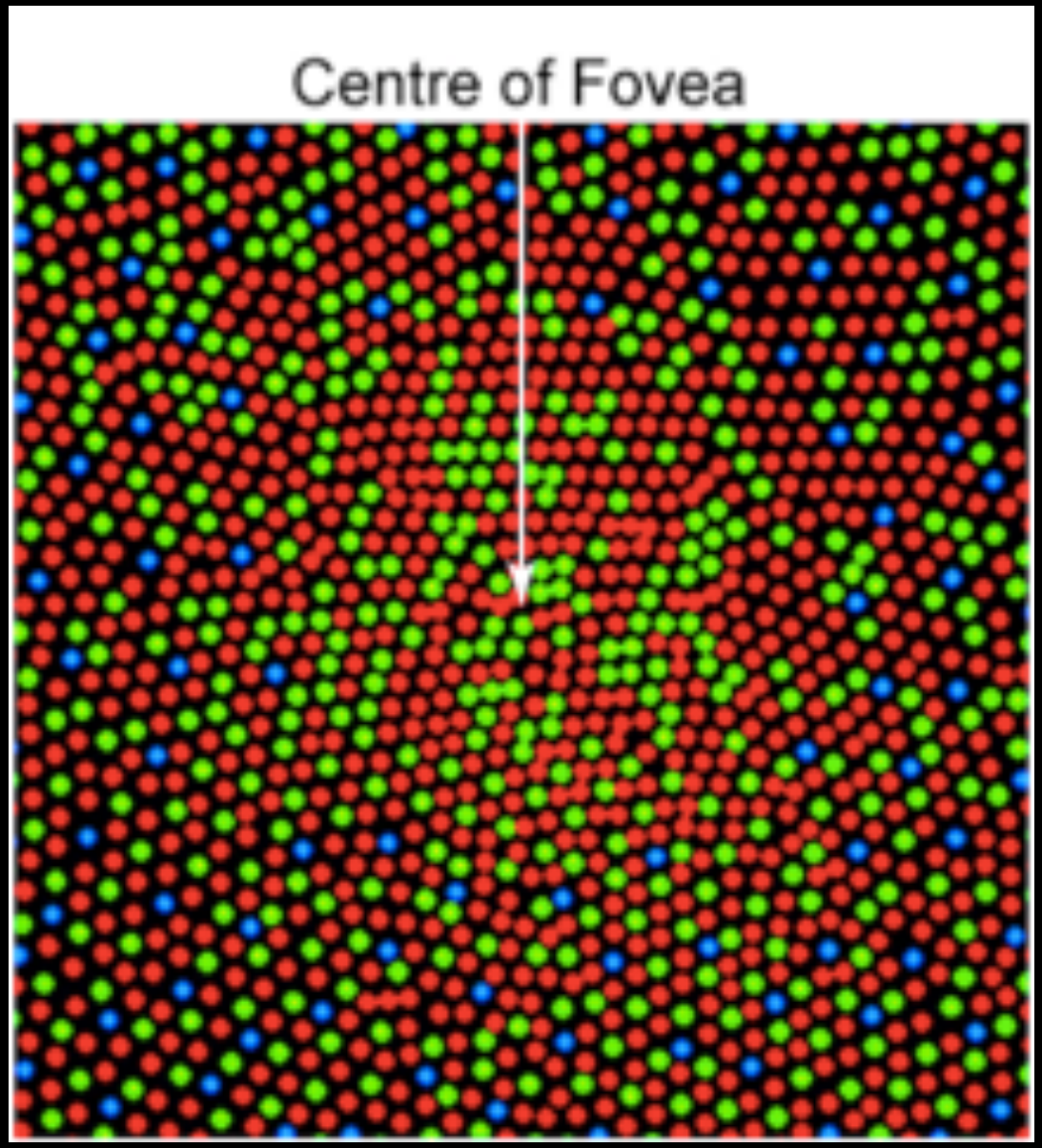
Par M. FAVRE.

Il nous a paru intéressant de vous présenter le cas de deux enfants, frère et sœur, atteints d'une dégénérescence hyaloïdéorétinienne d'un type particulier et dont la classification dans les affections de ce genre déjà connues présente quelques difficultés.

De ces deux enfants le plus atteint est l'aîné, dont voici l'histoire :

1958

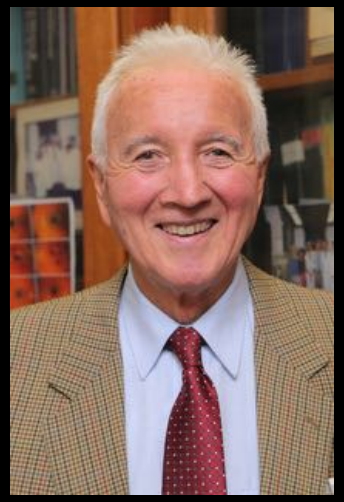
Enhanced S Cone Syndrome



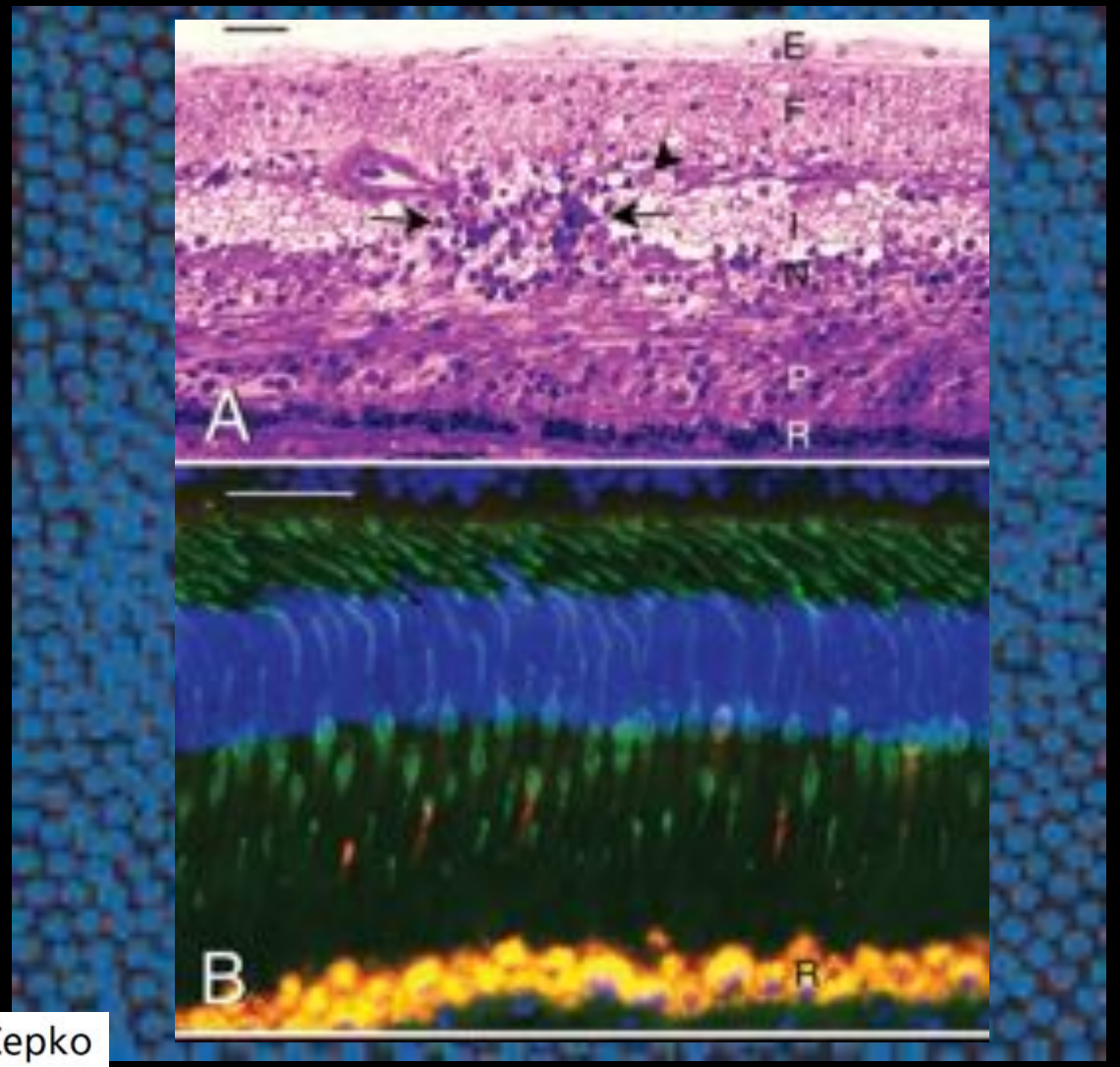
Normal

Enhanced S Cone Syndrome (92% blue cones)

Peter Gouras



Analysis of Blue Cone Sensitivity - ARVO



Connie Cepko

ESCS

Samuel Jacobson

SWS (Blue) Cone Hypersensitivity in a Newly Identified Retinal Degeneration

Samuel G. Jacobson, Michael F. Marmor,* Colin M. Kemp, and Robert W. Knighton

1990

The nuclear receptor *NR2E3* plays a role in human retinal photoreceptor differentiation and degeneration

Ann H. Milam*†‡, Linda Rose*, Artur V. Cideciyan*, Mark R. Barakat*, Wai-Xing Tang*, Nisha Gupta*, Tomas S. Aleman*, Alan F. Wright§, Edwin M. Stone¶, Val C. Sheffield‖, and Samuel G. Jacobson*

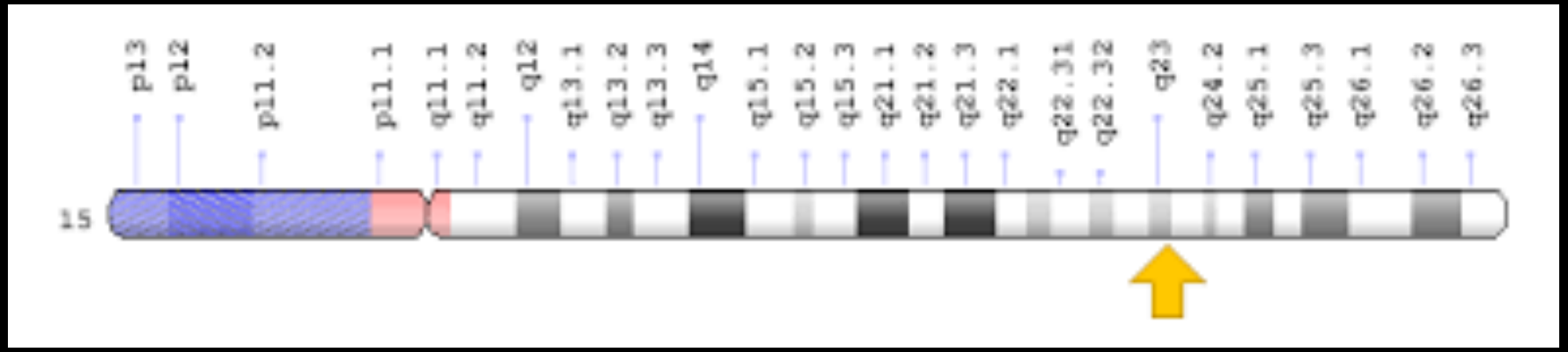
2001

ESCS = NR2E3

Retina

Cone Vision Changes in the Enhanced S-Cone Syndrome Caused by *NR2E3* Gene Mutations

Alexandra V. Garafalo,[1] Giacomo Calzetti,[1] Artur V. Cideciyan,[1] Alejandro J. Roman,[1] Supna Saxena,[1] Alexander Sumaroka,[1] Windy Choi,[1] Alan F. Wright,[2] and Samuel G. Jacobson[1]



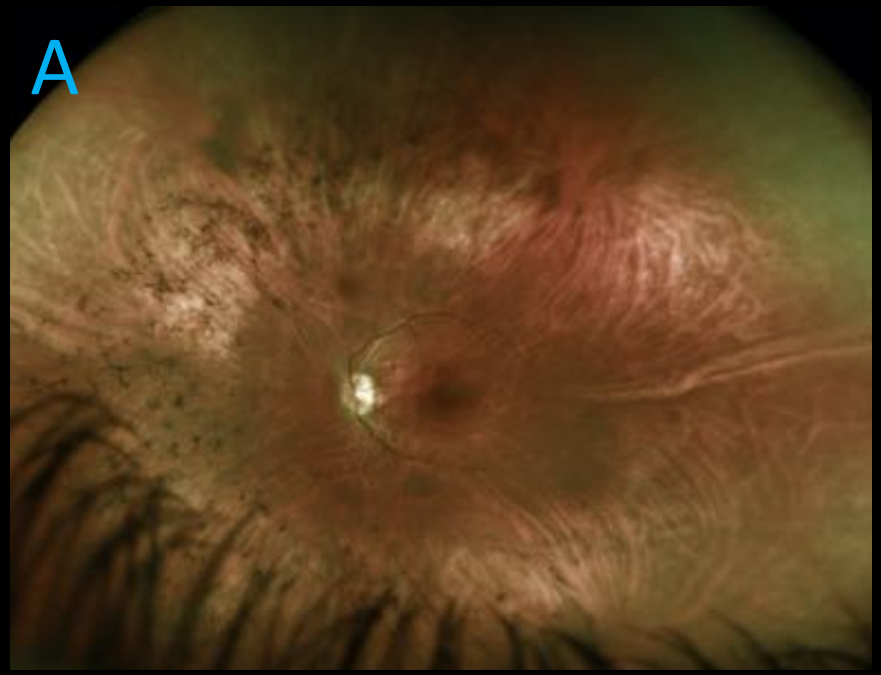
37 mutations

Mostly compound heterozygous

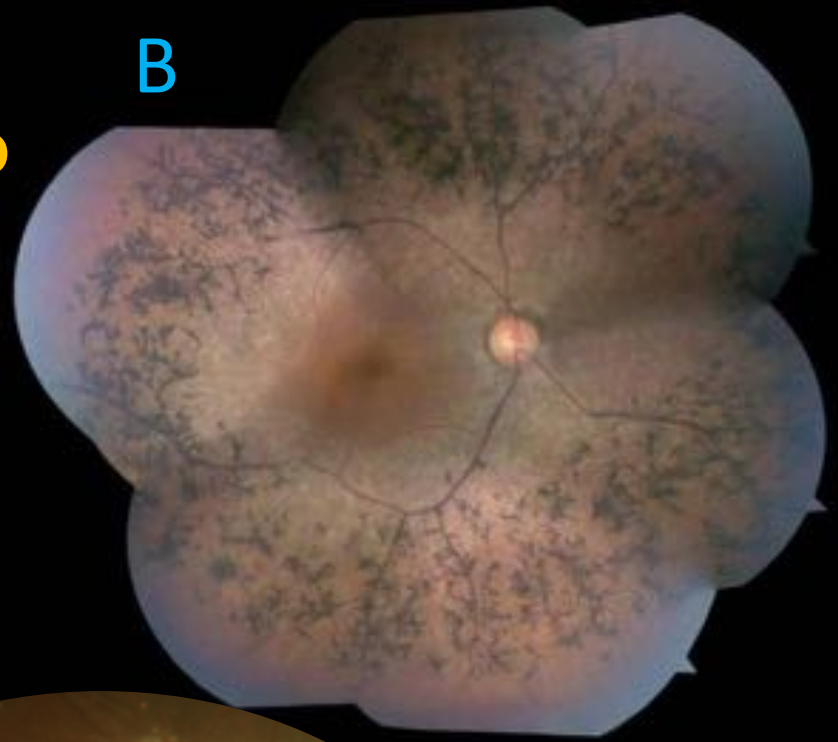
Fewer than 2% no detectable allele

ESCS
Which one fits?

A

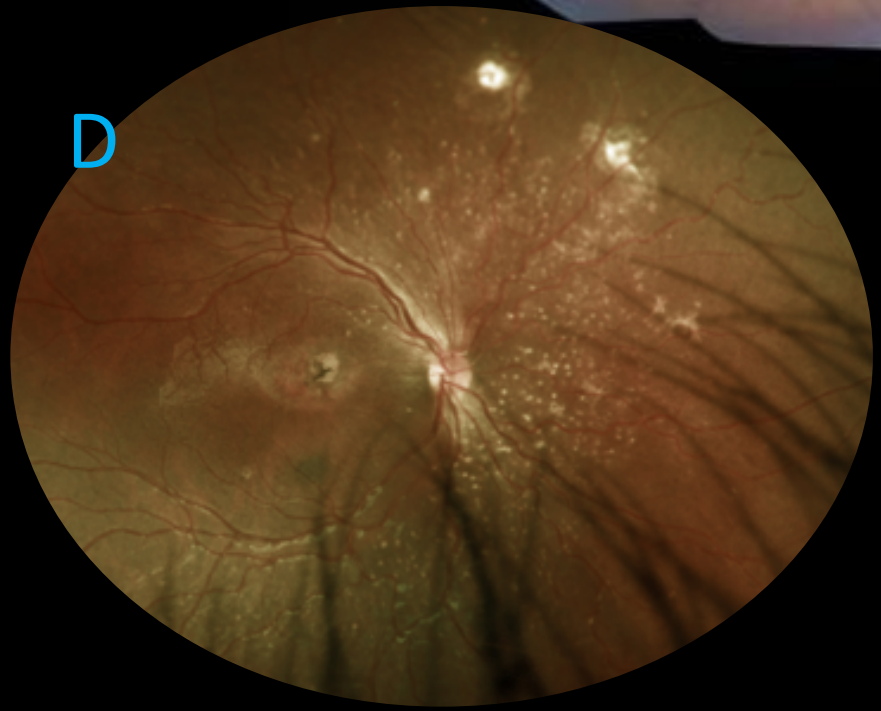


B



C

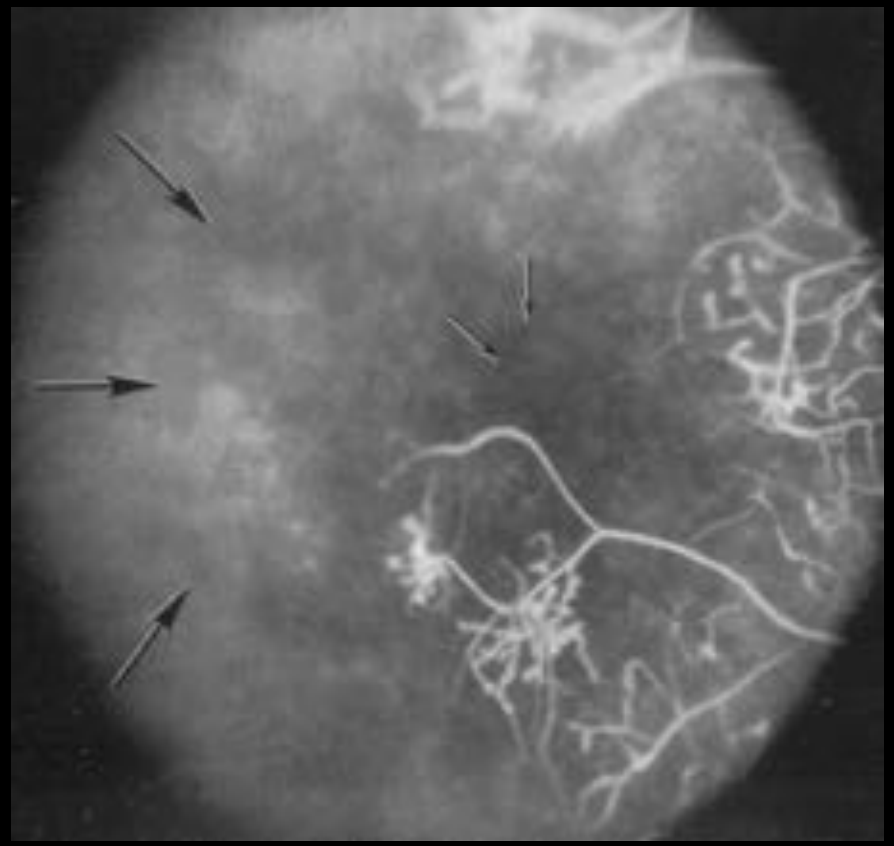
D

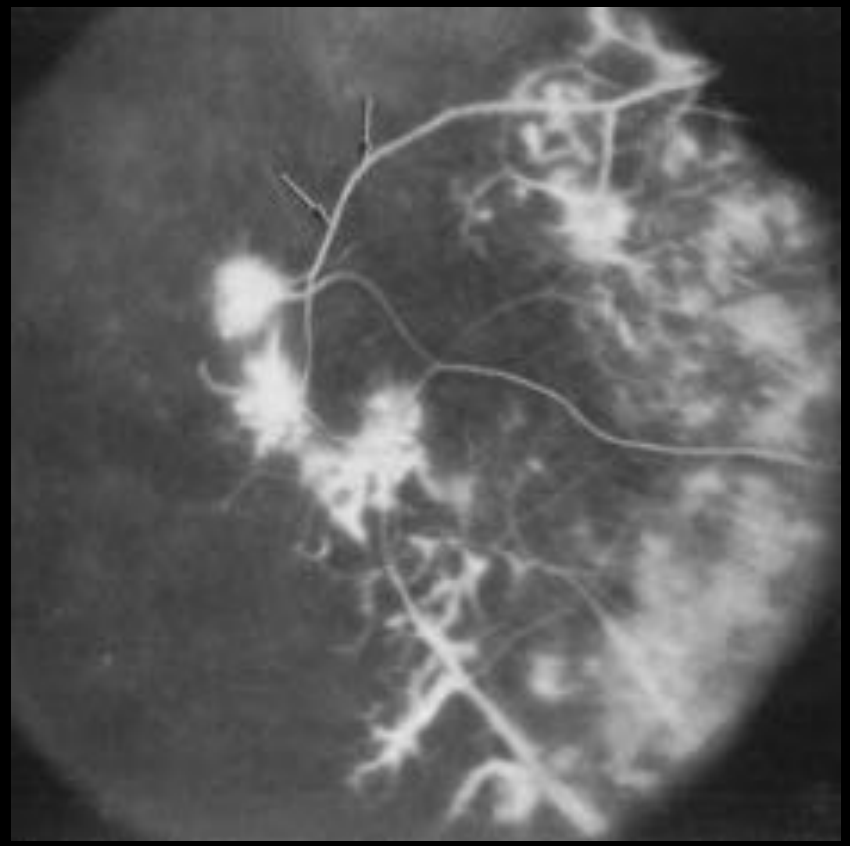


Diagnostic features of the Favre-Goldmann syndrome

GERALD A. FISHMAN, LEE M. JAMPOL, AND MORTON F. GOLDBERG
From the Department of Ophthalmology, University of Illinois Eye and Ear Infirmary, Chicago, Illinois

1976





Early nightblindness

Posterior subcap cataract

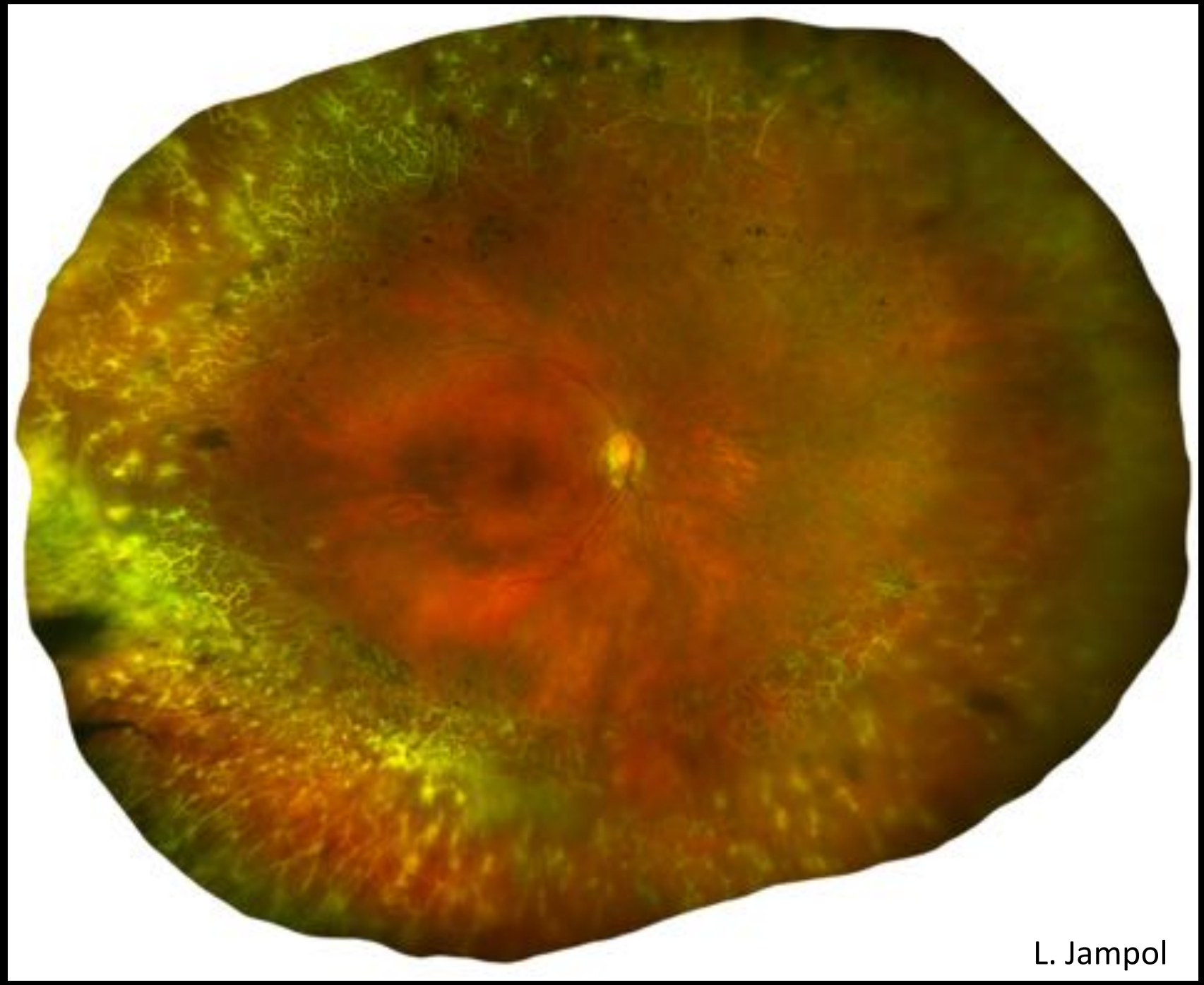
Vitreous degeneration

Pigmentary degeneration

ERG changes

Retinal non-perfusion

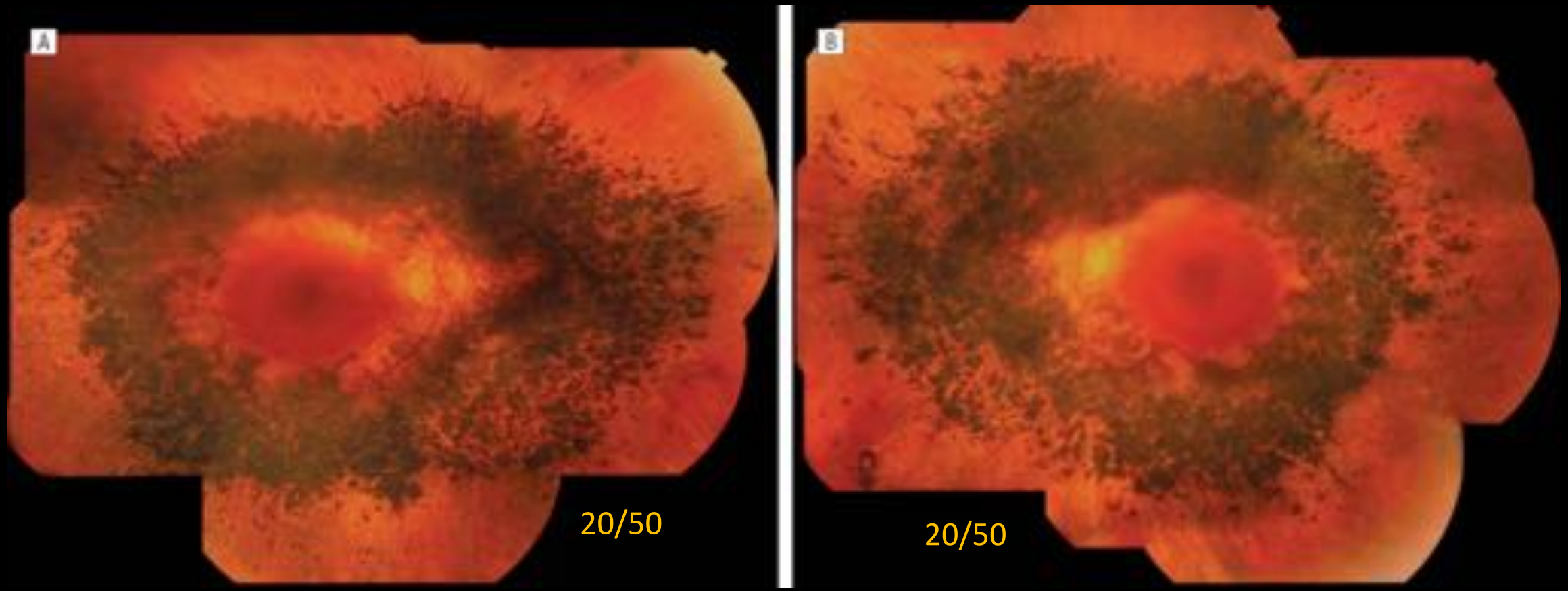
Diffuse retinal leakage


L. Jampol

Phenotypic Features of Patients With *NR2E3* Mutations

2009

Sophia I. Pachydaki, MD; Carolyn C. Klaver, MD; Irene A. Barbazetto, MD; Monique S. Roy, MD; Peter Gouras, MD; Rando Allikmets, PhD; Lawrence A. Yannuzzi, MD



66yo Female

Original Investigation | CLINICAL SCIENCES

Expanded Clinical Spectrum of Enhanced S-Cone Syndrome

Suzanne Yzer, MD, PhD; Irene Barbazetto, MD; Rando Allikmets, PhD; Mary J. van Schooneveld, MD, PhD; Arthur Bergen, PhD; Stephen H. Tsang, MD, PhD; Samuel G. Jacobson, MD, PhD; Lawrence A. Yannuzzi, MD

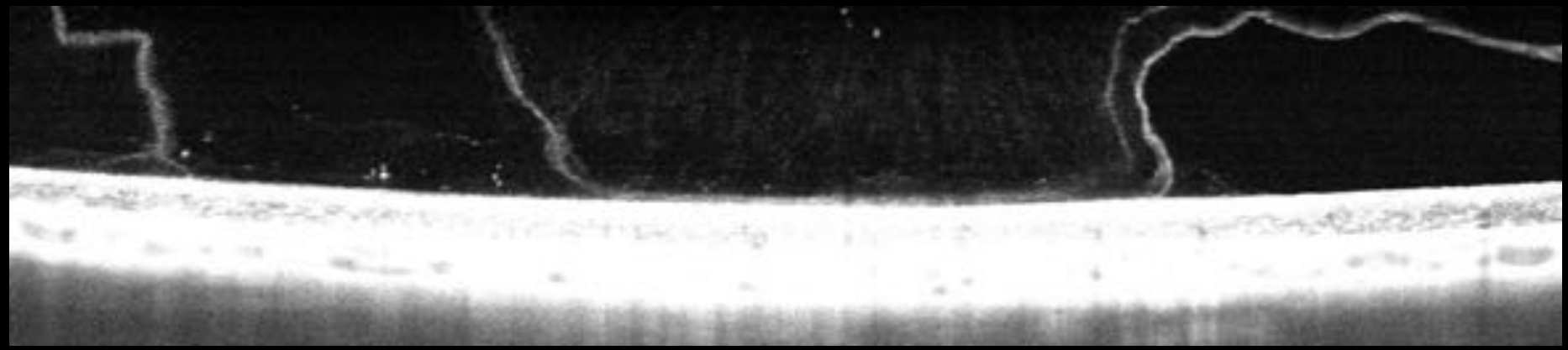
Torpedo-like lesions

- Nyctalopia at early age
- PSC
- CME
- Peripheral retinoschisis
- Peripheral retinal ischemia

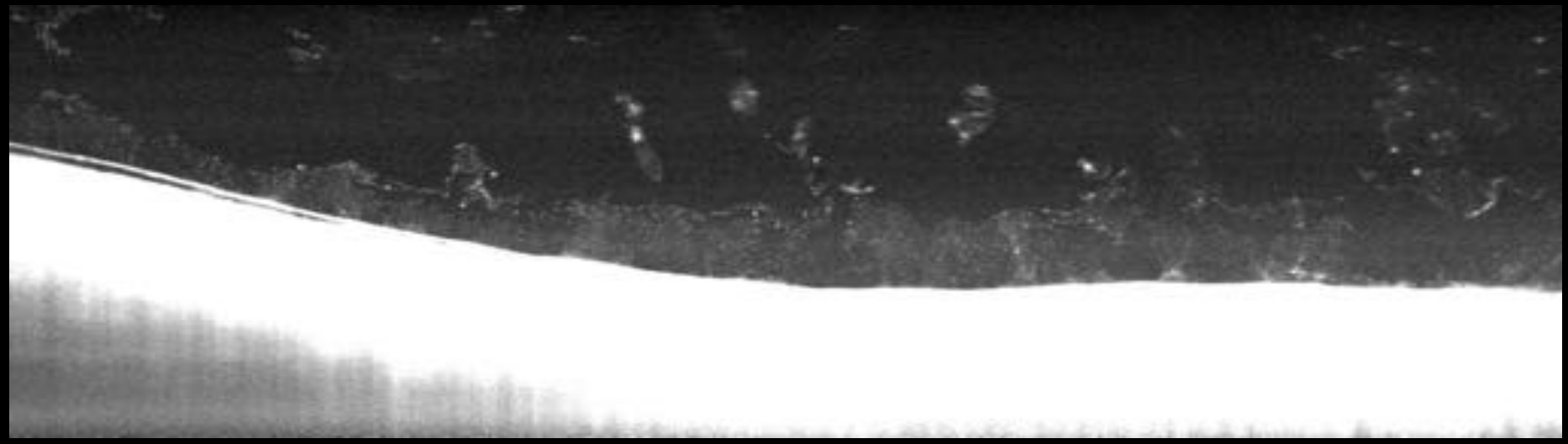
Current Series

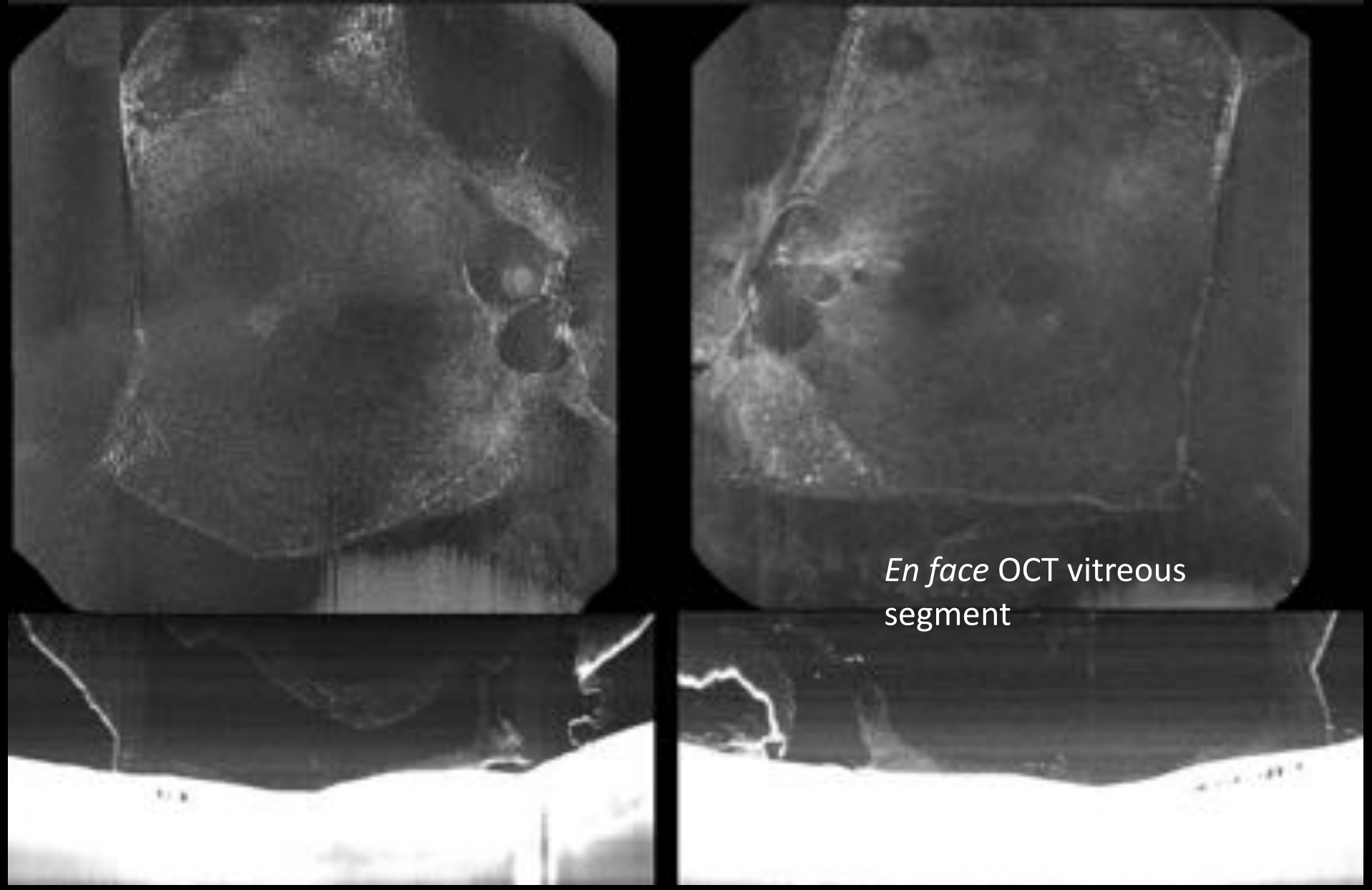
Vitreous In ESCS

Thick posterior hyaloid



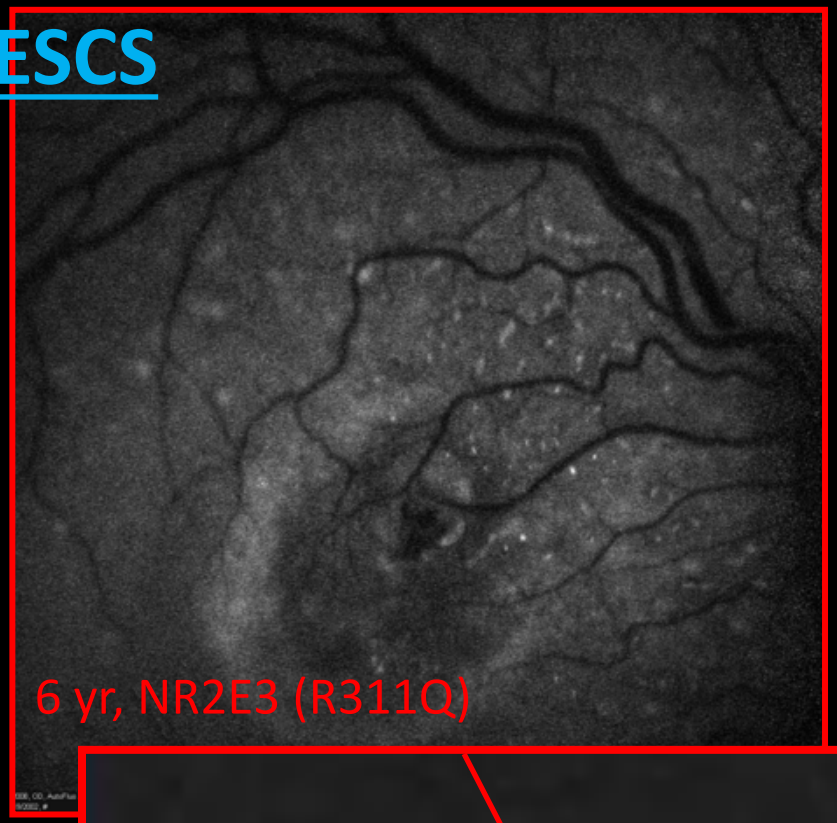
Fibrillar changes

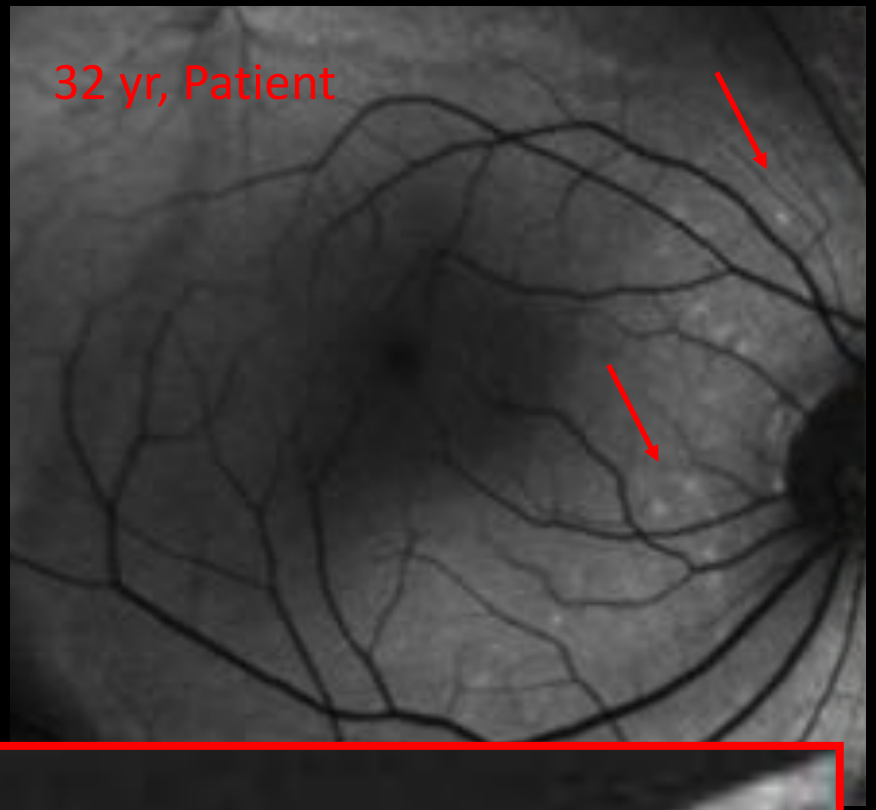




SQUARE or polygonal vitreomacular adhesion + thick posterior hyaloid

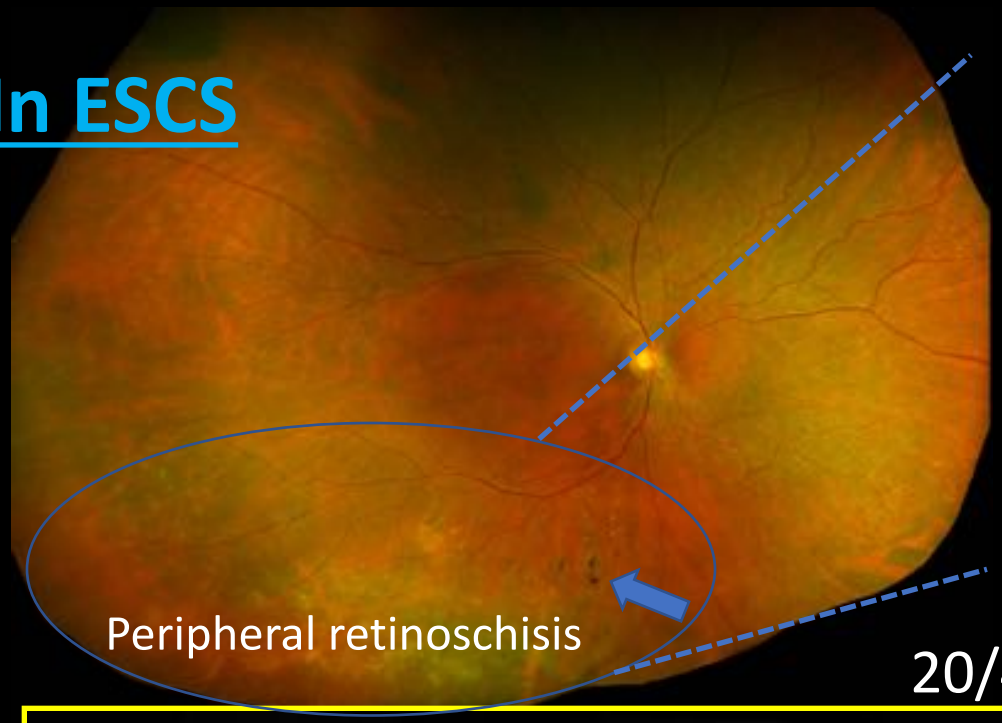
Retina In ESCS





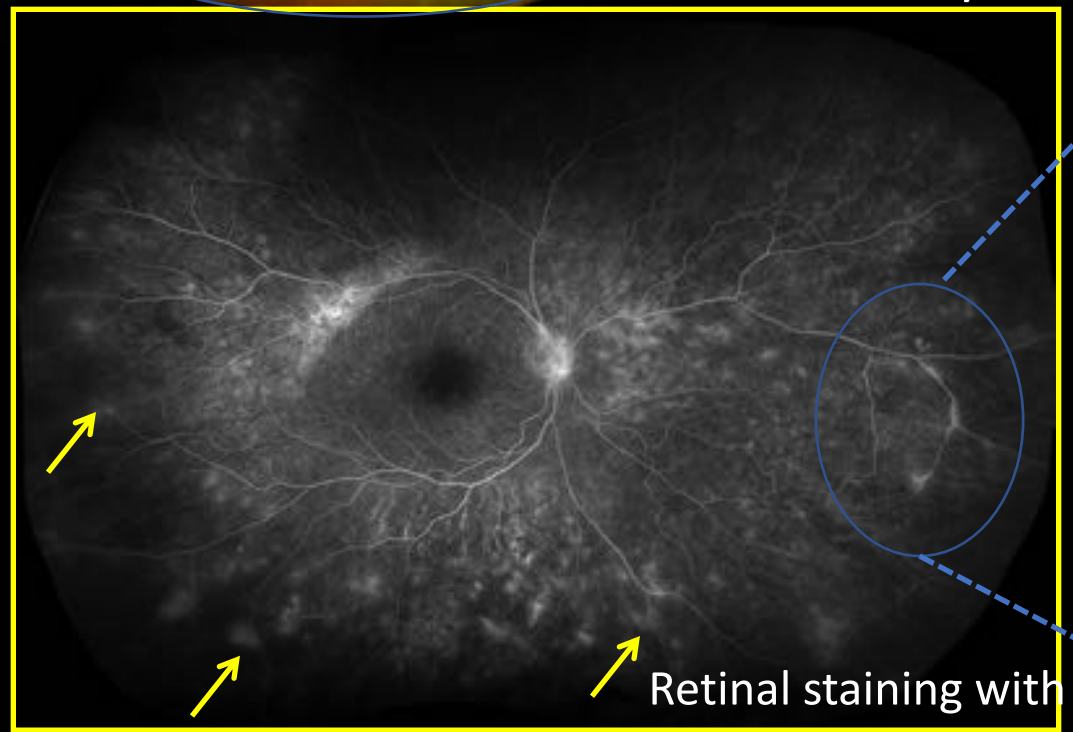
S. Tsang

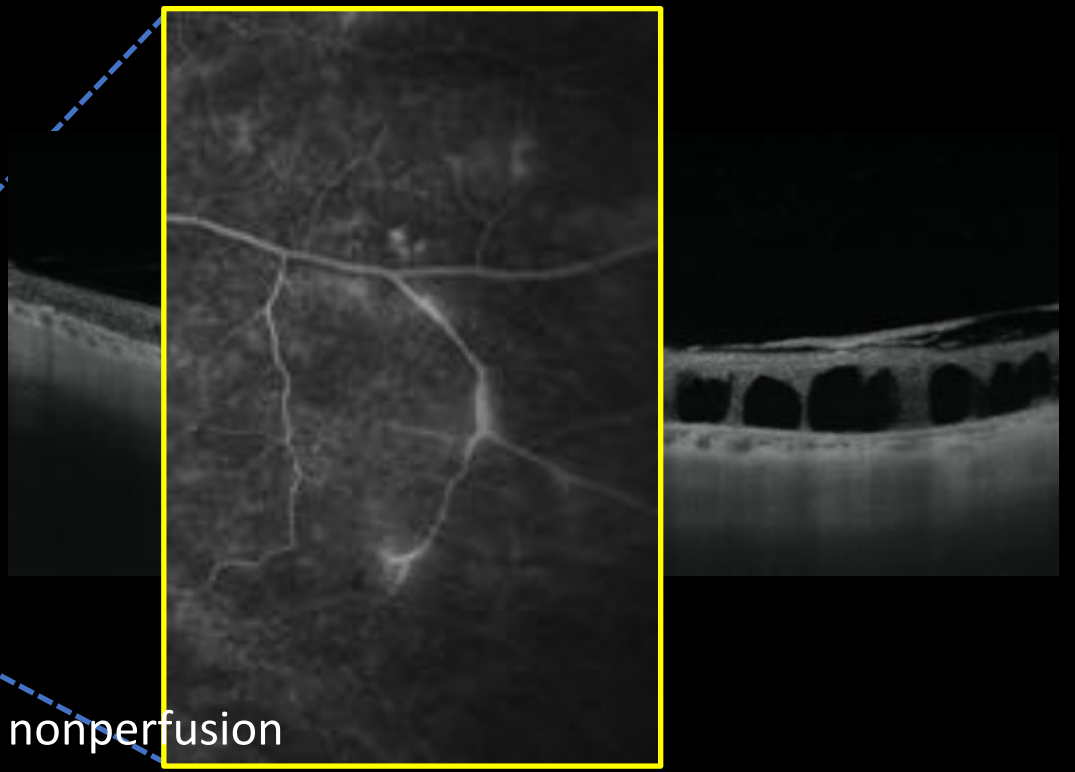
Retina In ESCS



Opaque dendritic vessels

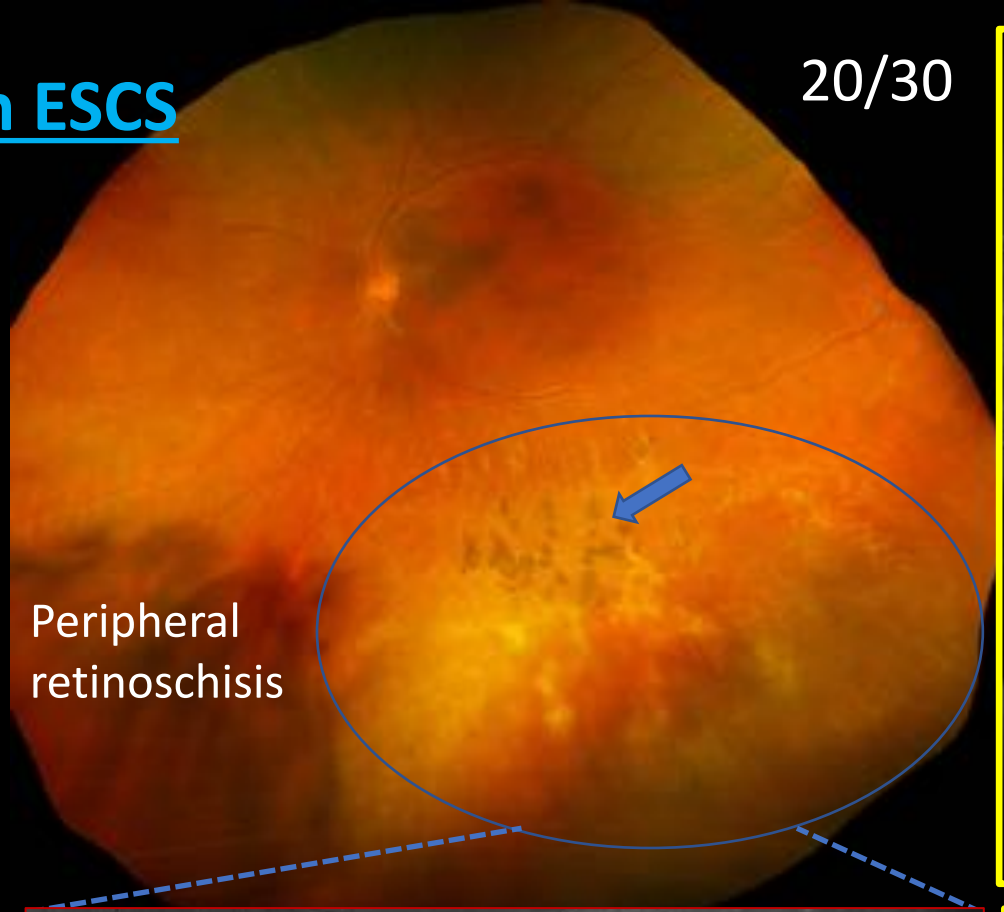
20/40

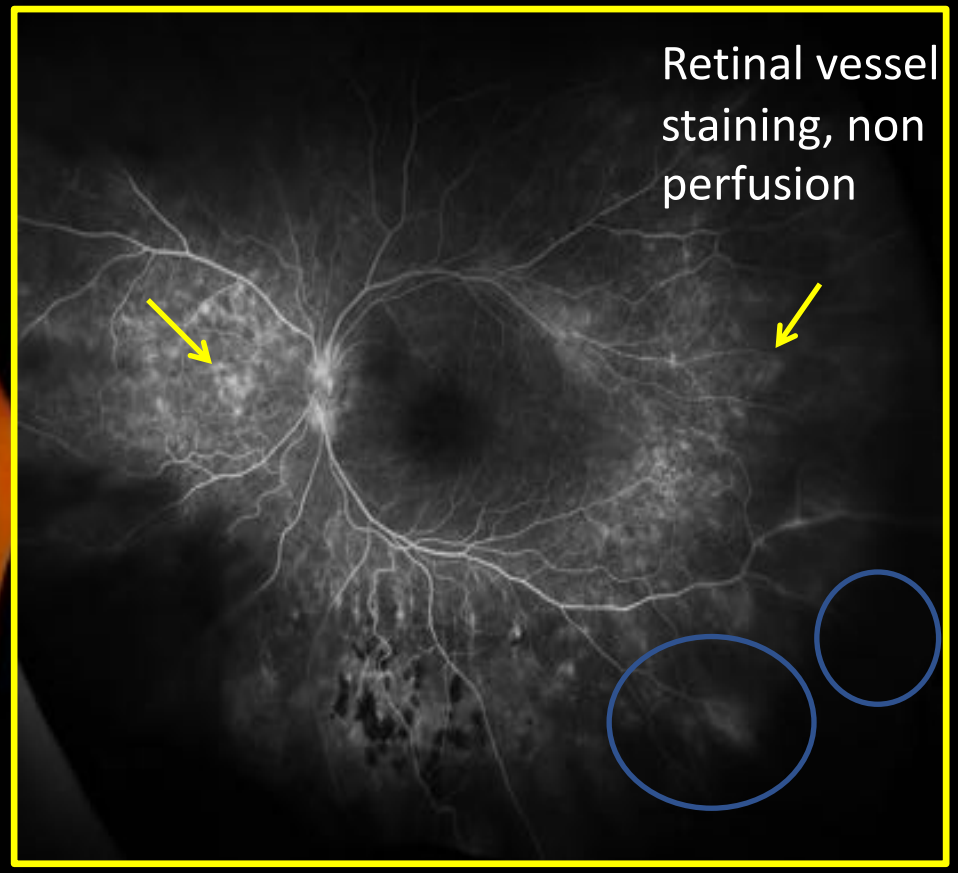




Retina In ESCS

20/30

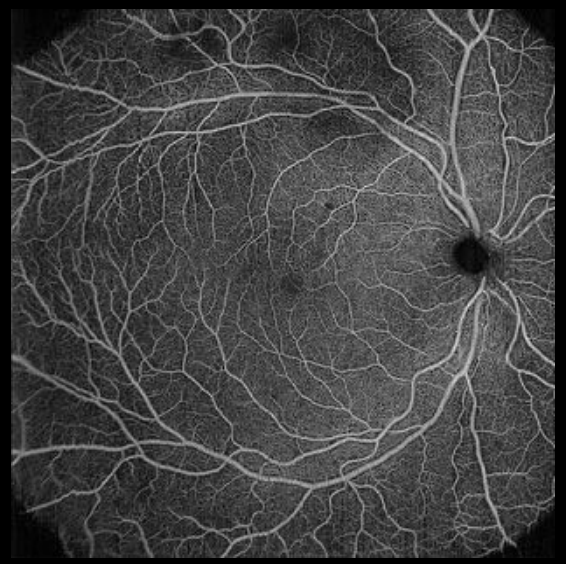




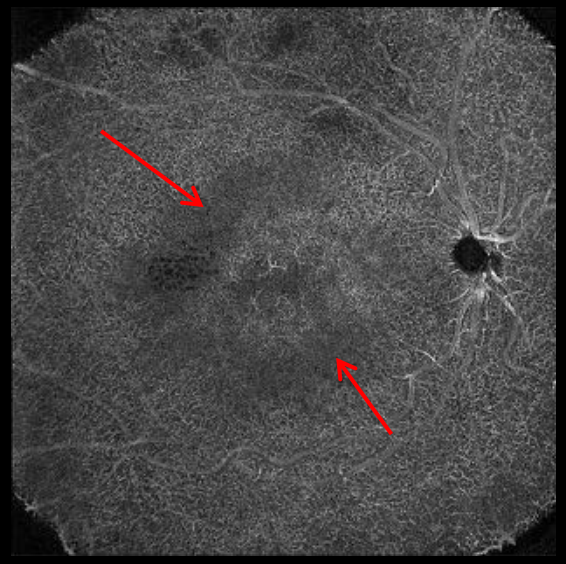
Retina In ESCS

OCT-A

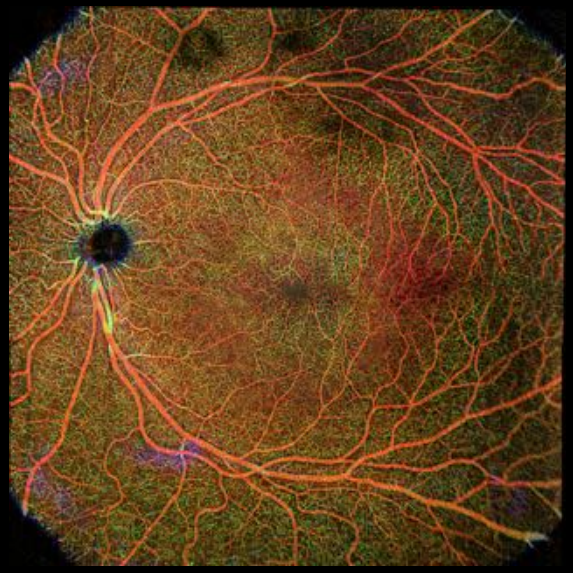
Full retina

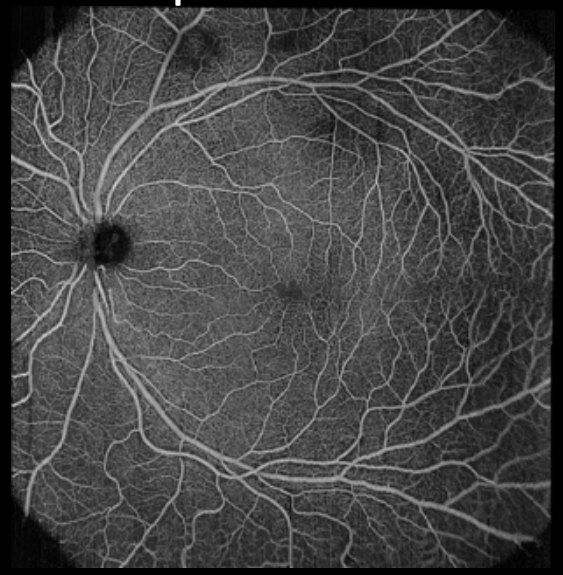


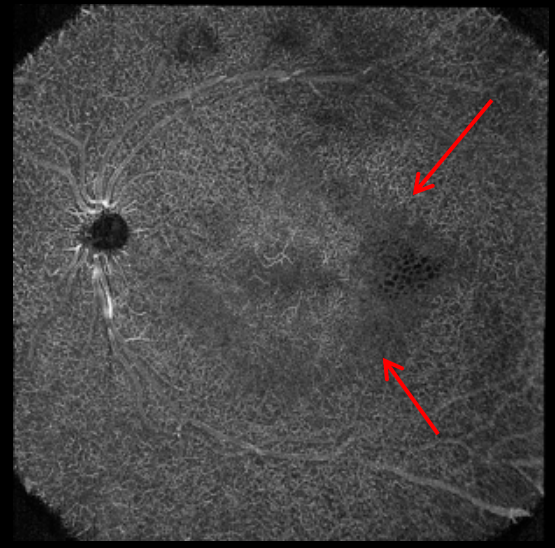
Superficial



Deep

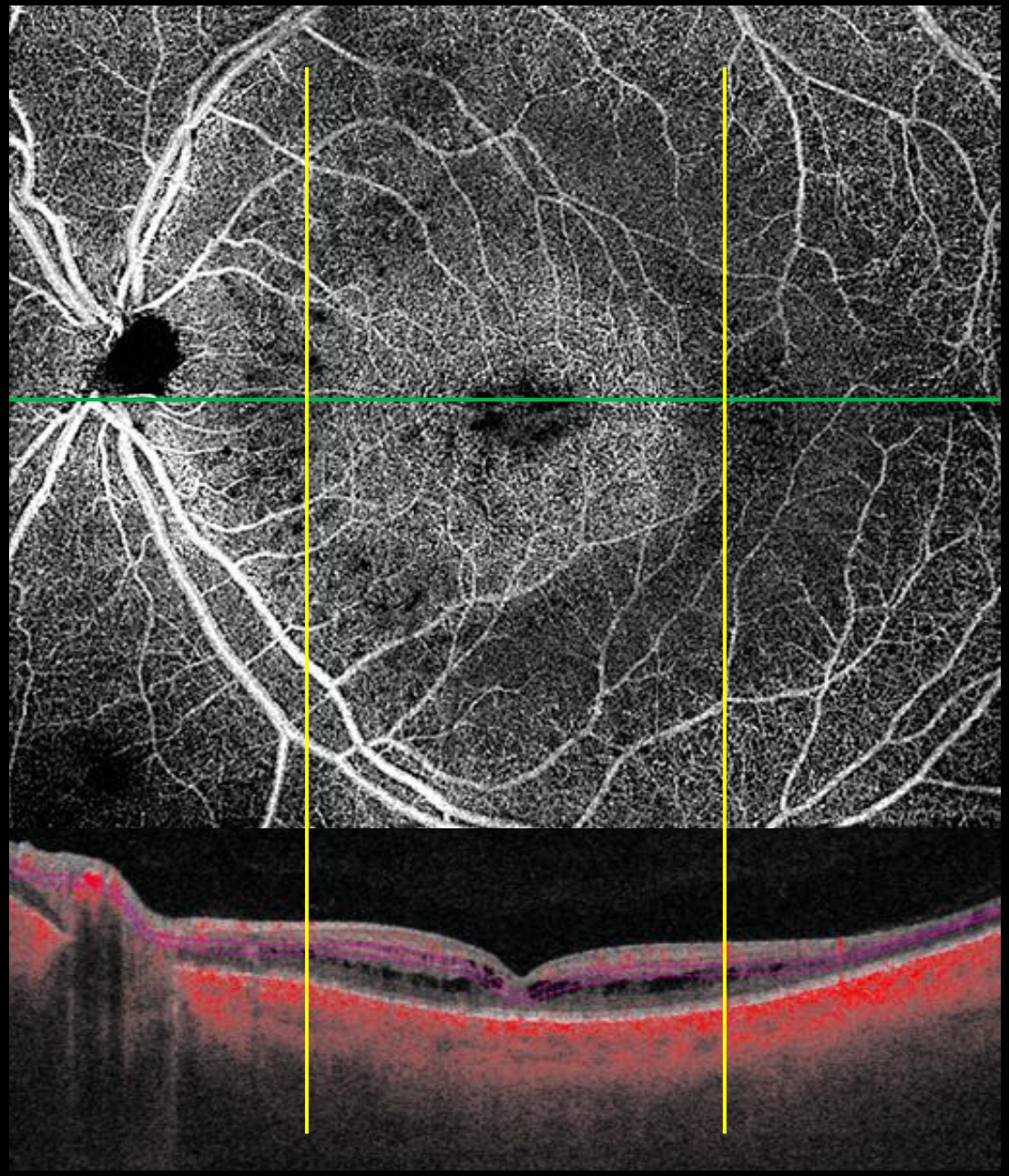




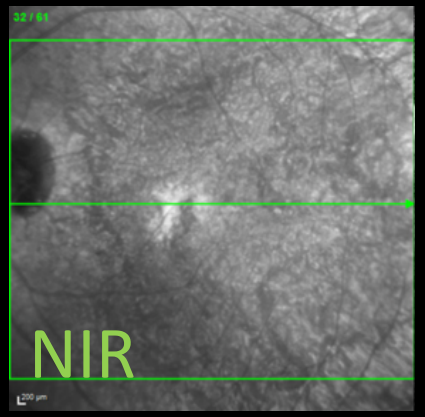


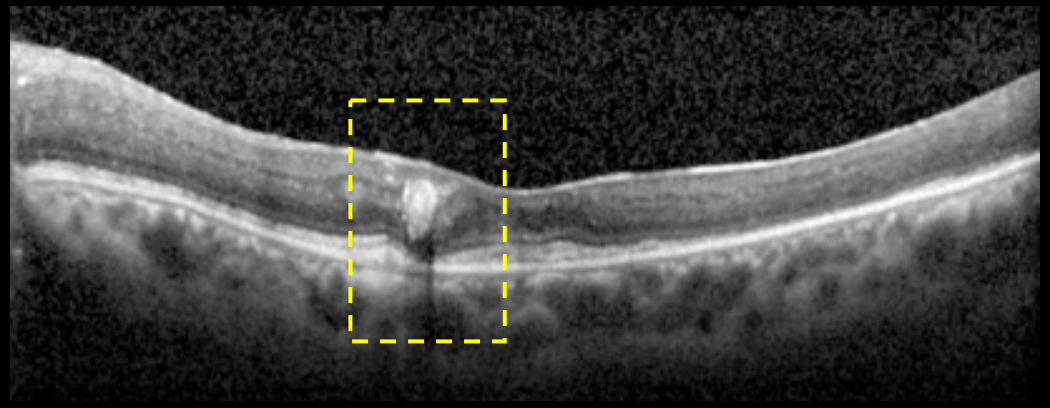
REDUCED SIGNAL OR NON-PERFUSION IN DEEP CAPILLARY PLEXUS

Retina In ESCS



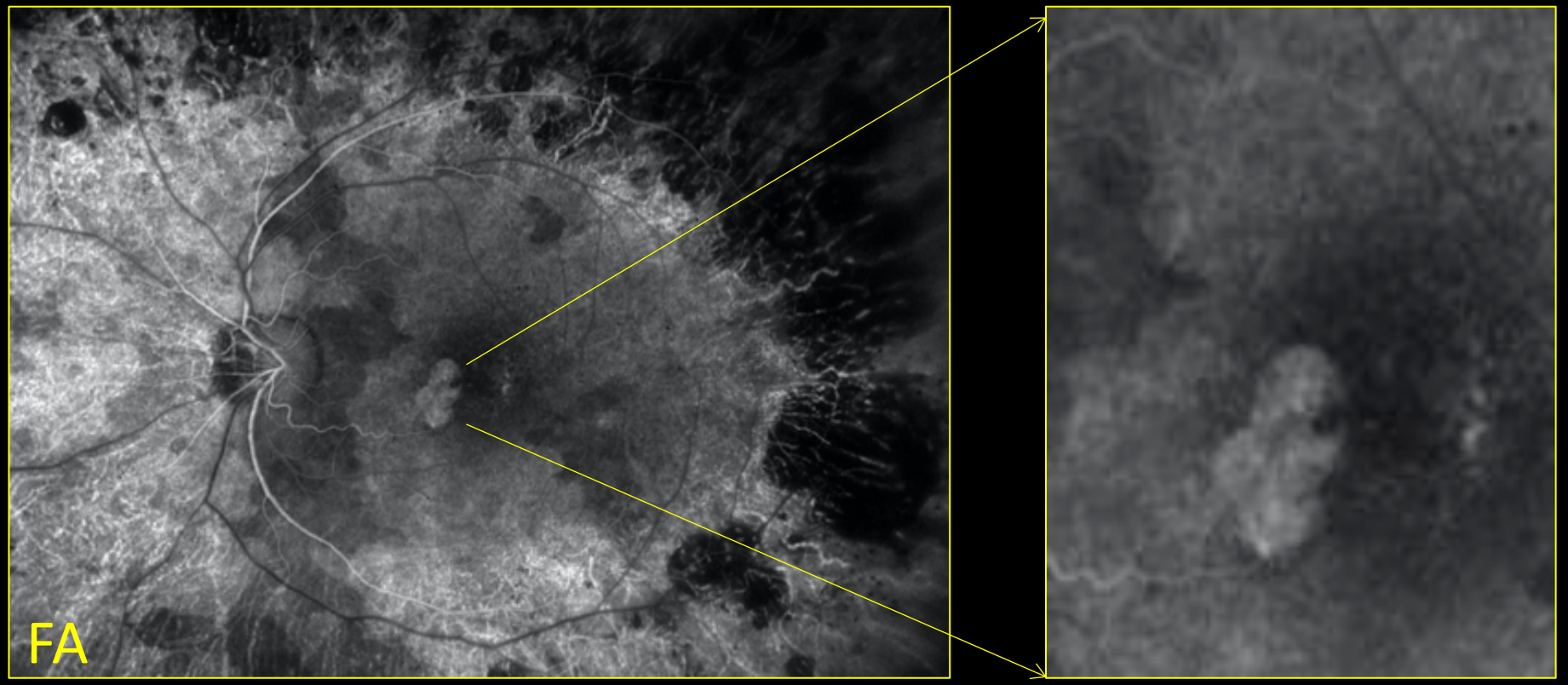
REDUCED DEEP CAPILLARY PLEXUS FLOW DOES NOT CORRESPOND TO RETINOCHISIS





Neovascular changes

Angiomatous lesion
TYPE 3 NEO

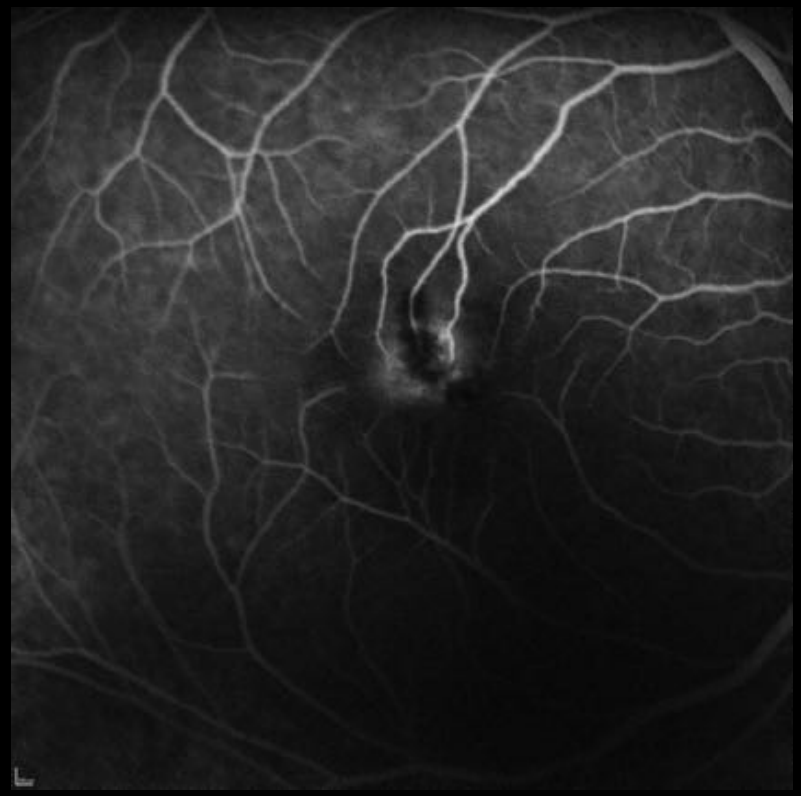


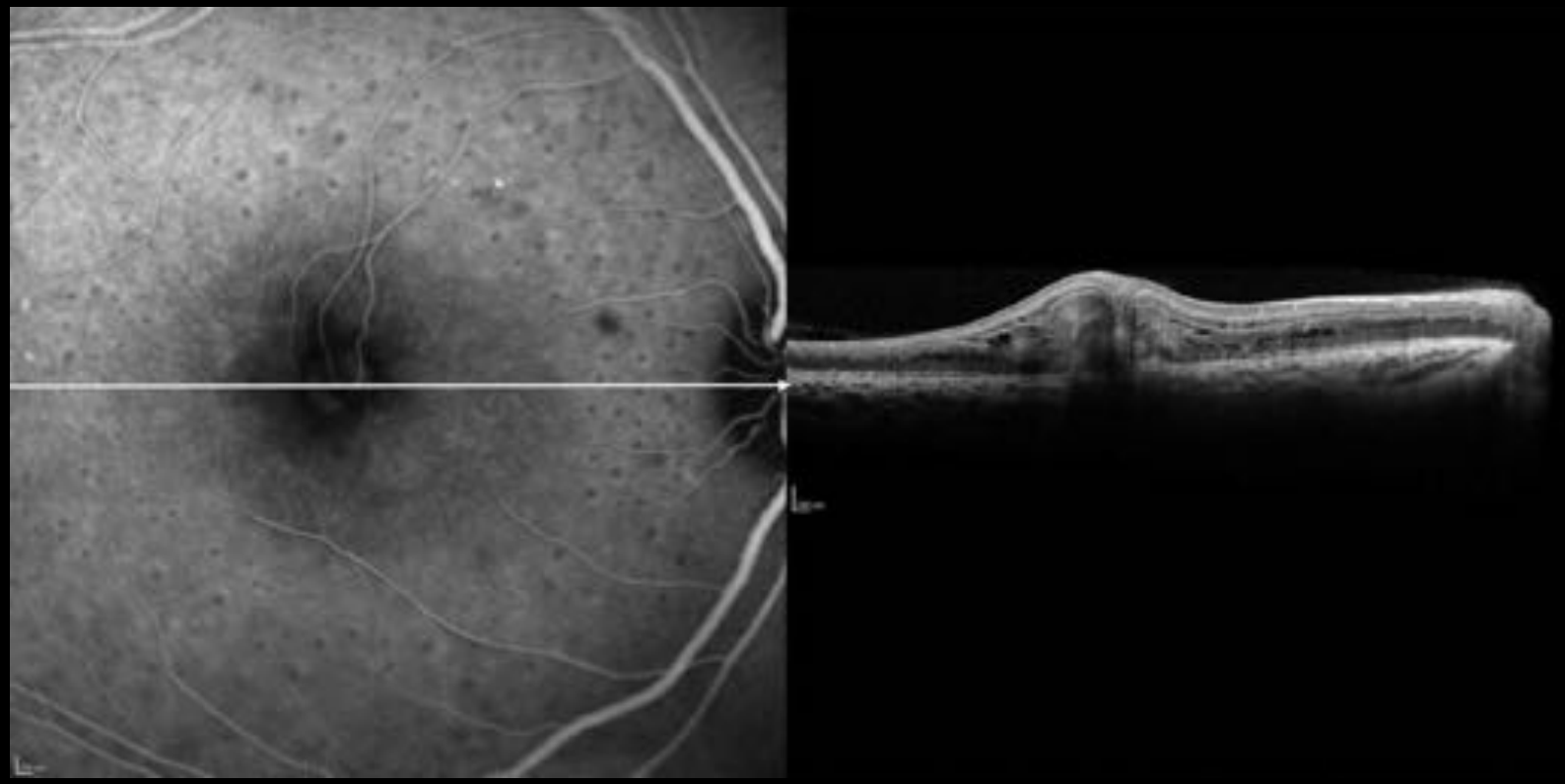
RETINOCHOROIDAL ANASTOMOSIS ASSOCIATED WITH ENHANCED S-CONE SYNDROME

Jennyfer Zerbib, MD,*† Rocio Blanco Garavito, MD,* Sylvie Gerber, PhD,‡
Hassiba Oubraham, MD,* Anne Sikorav, MD,* Isabelle Audo, MD,§
Josseline Kaplan, MD,‡ Jean-Michel Rozet, PhD,‡ Eric H. Souied, MD, PhD*

2019

10 year-old boy

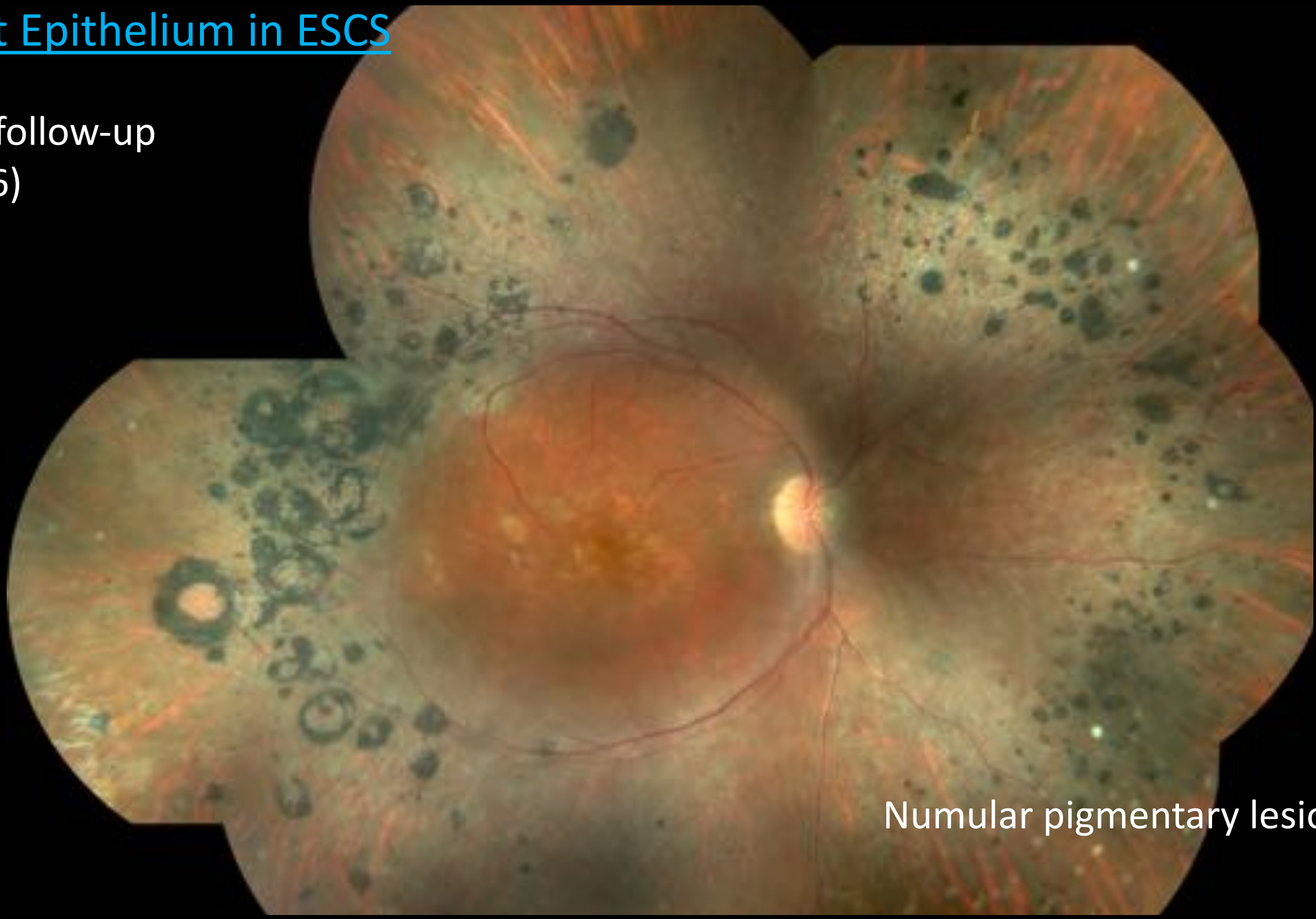




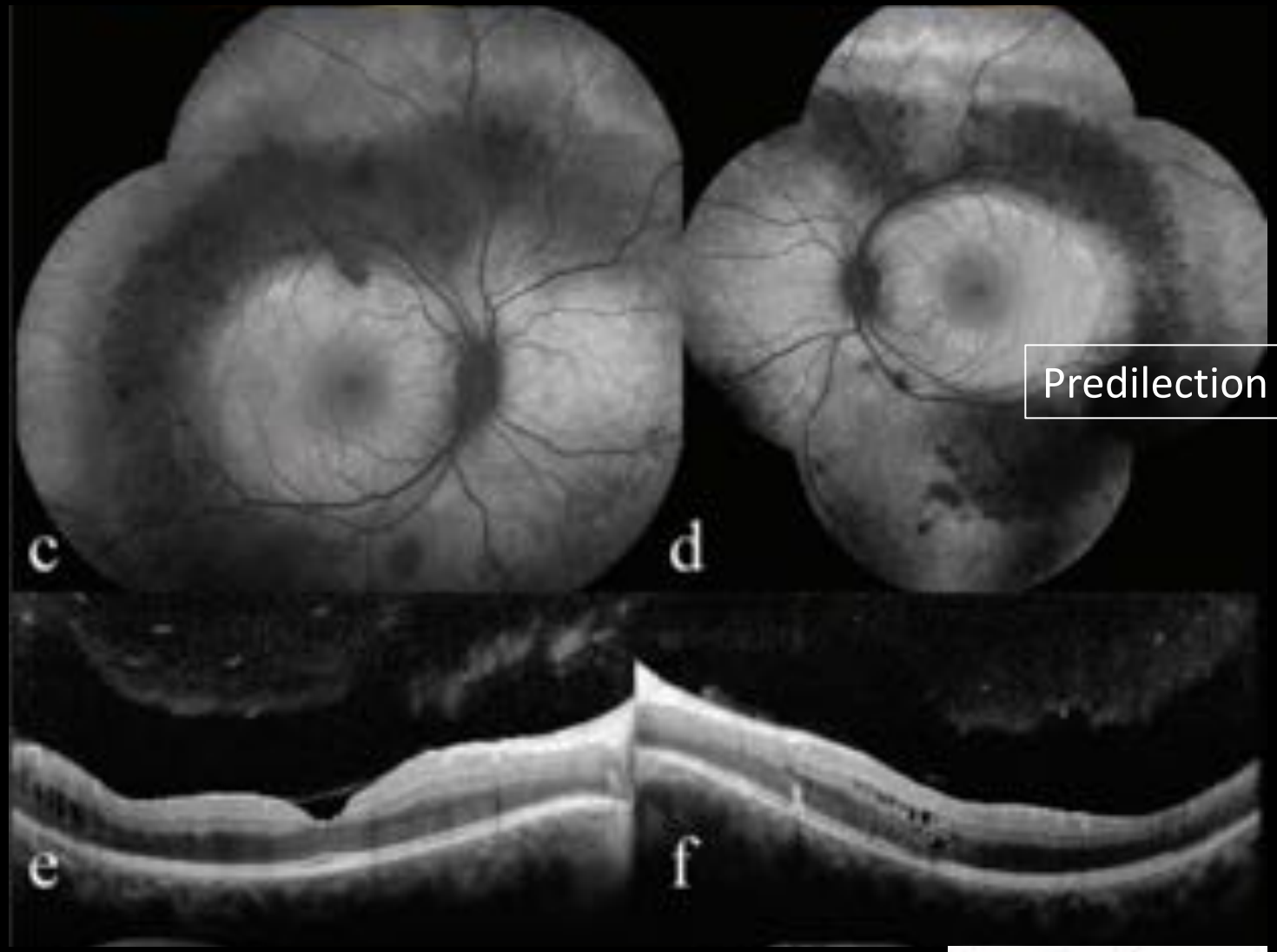
Pigment Epithelium in ESCS

51 year follow-up
(Aged 66)

20/40



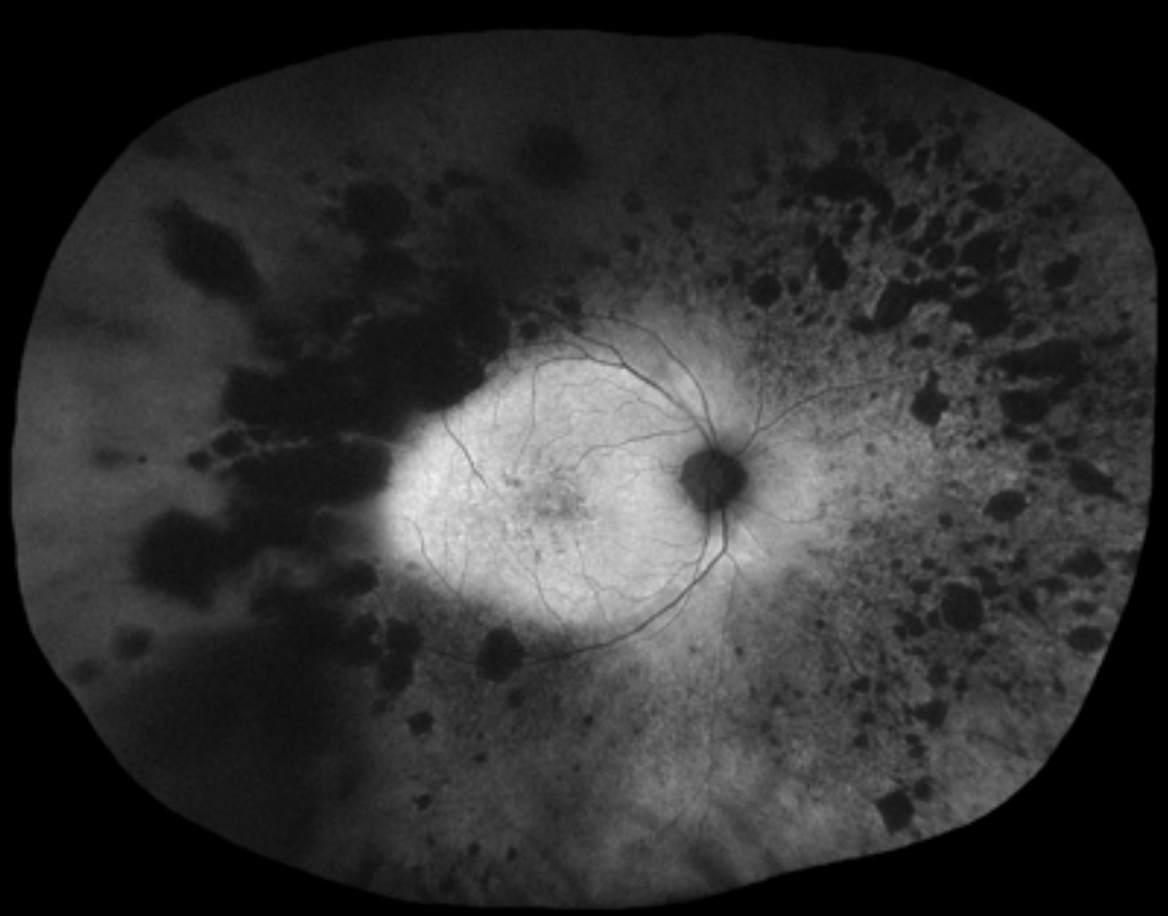
Numular pigmentary lesions



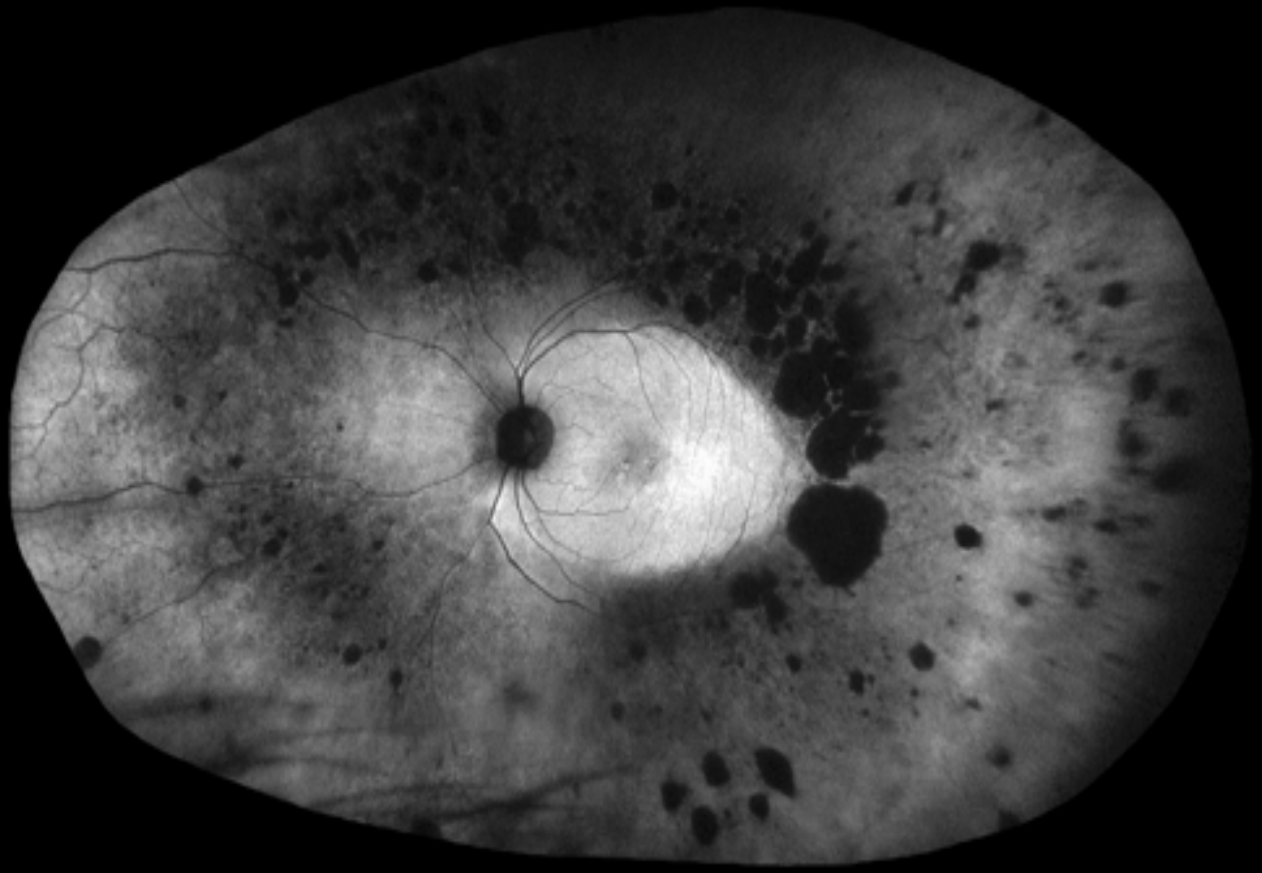
Predilection for superior and temporal

Serdar Özateş

Pigment Epithelium in ESCS



FAF

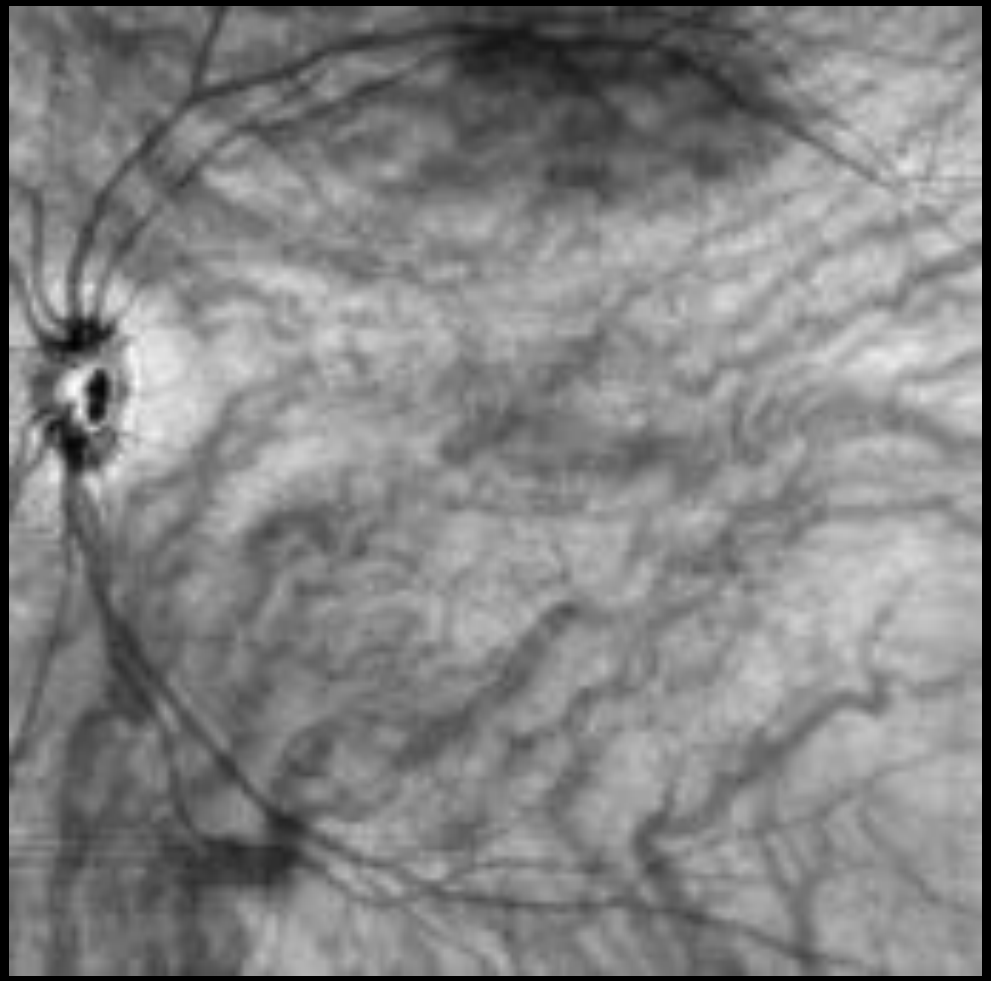


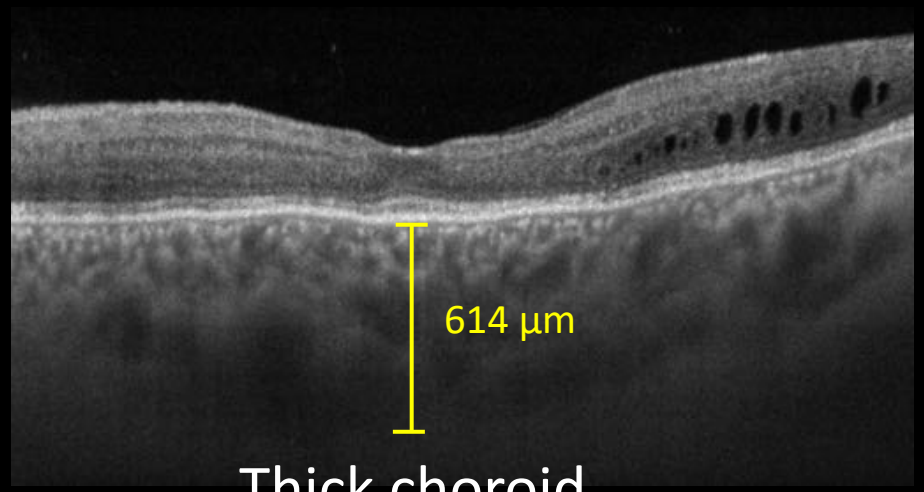
FAF

Hypo FAF numular cincumferential lesions

Yellow-white spots

Choroidal architecture in ESCS



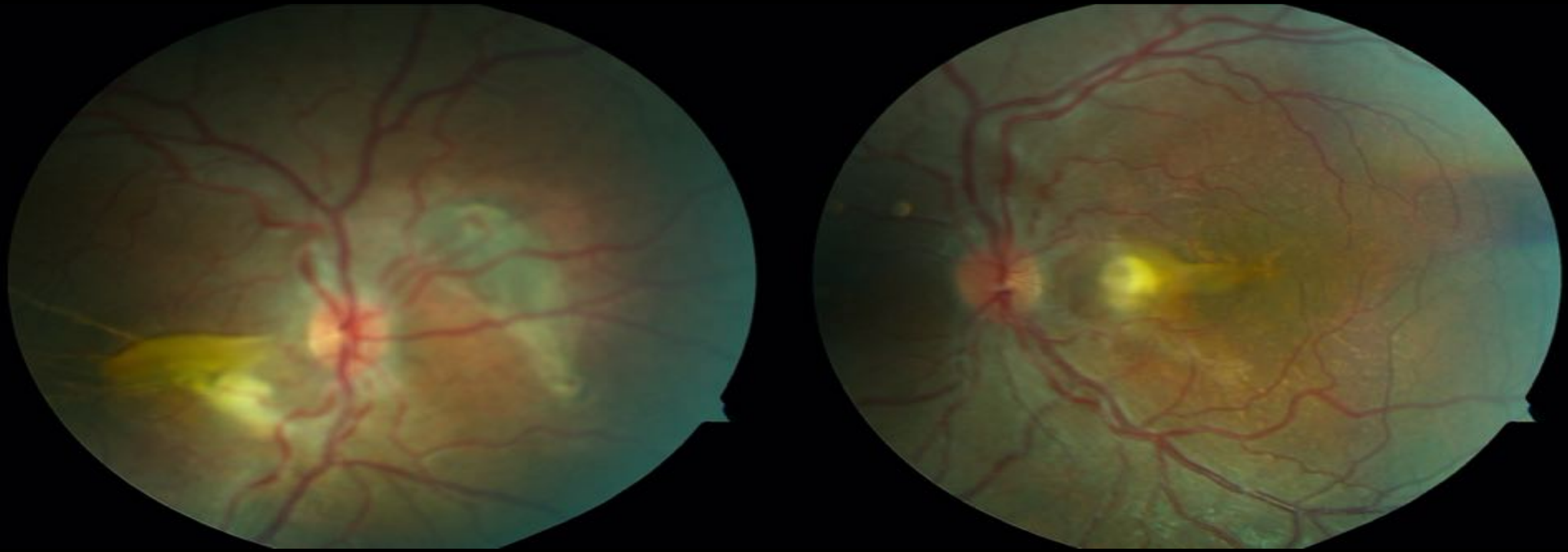


Thick choroid

Hyperopia

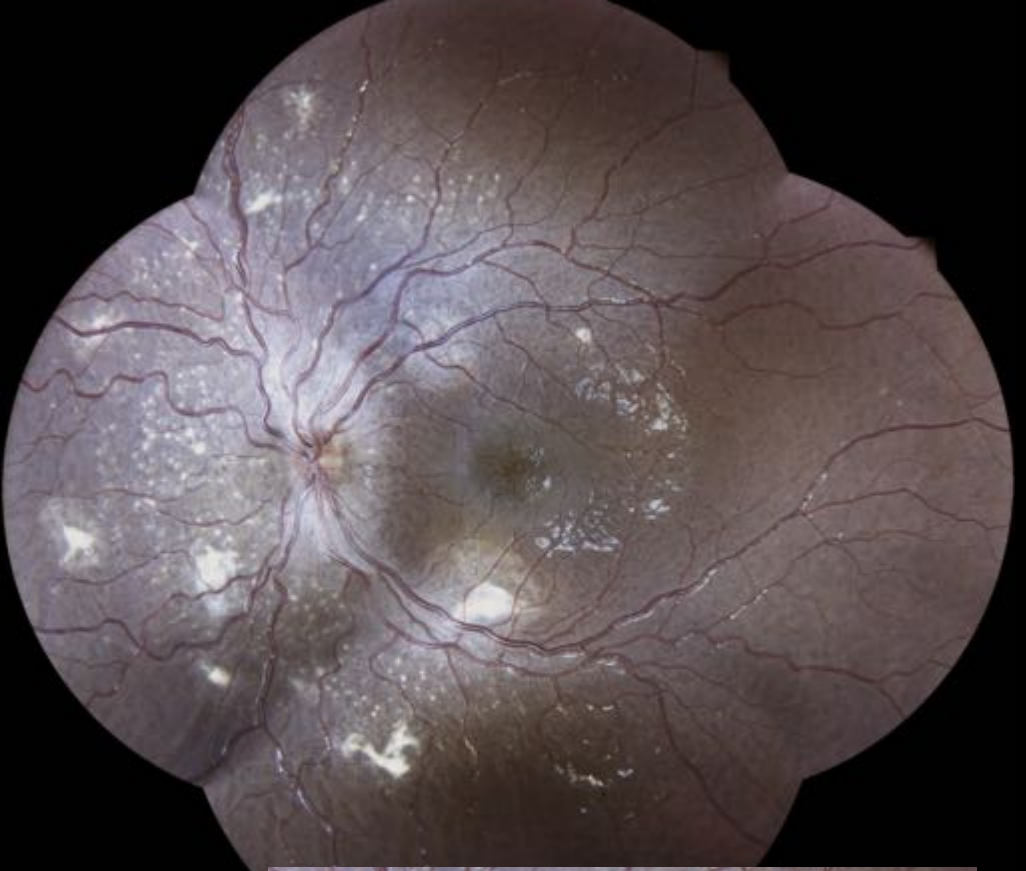
Nyctalopia + CNV = ESCS

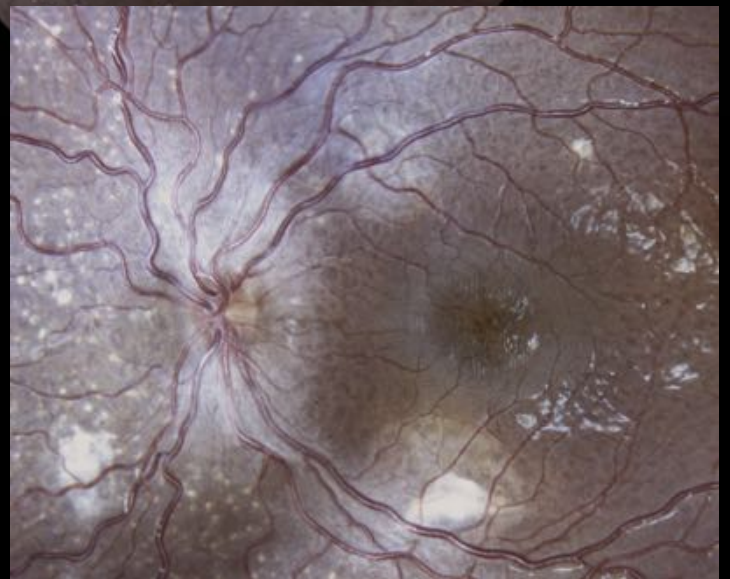
4 year-old boy



Choroidal neovascularization

S. Tsang

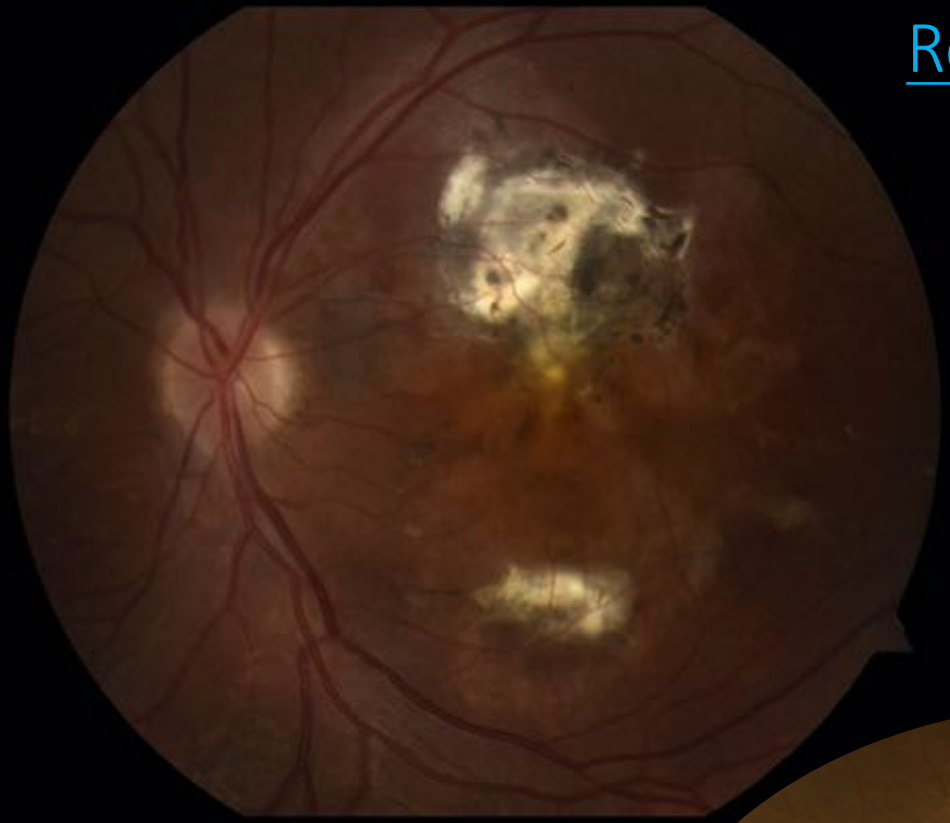


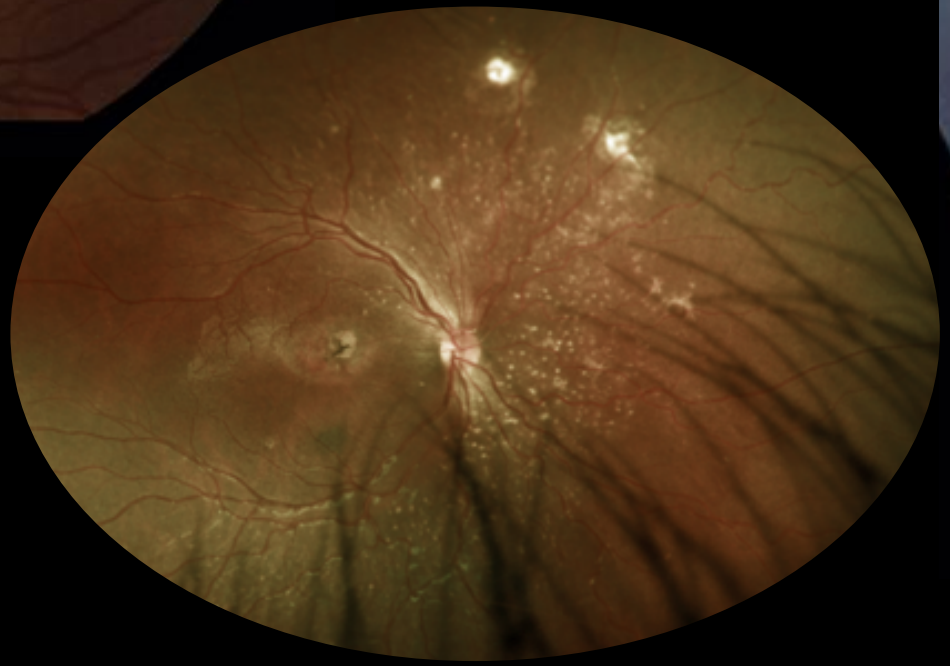


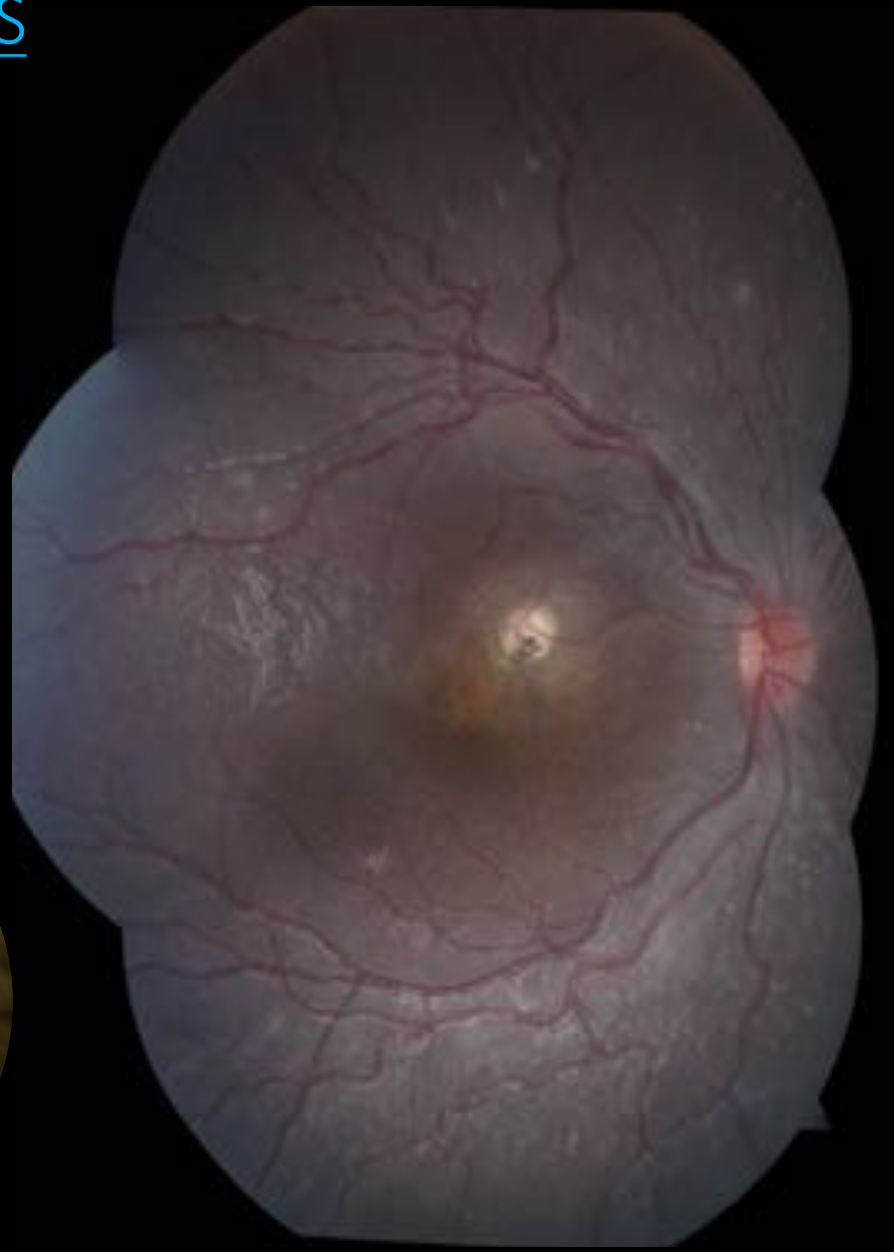
Neovascular changes

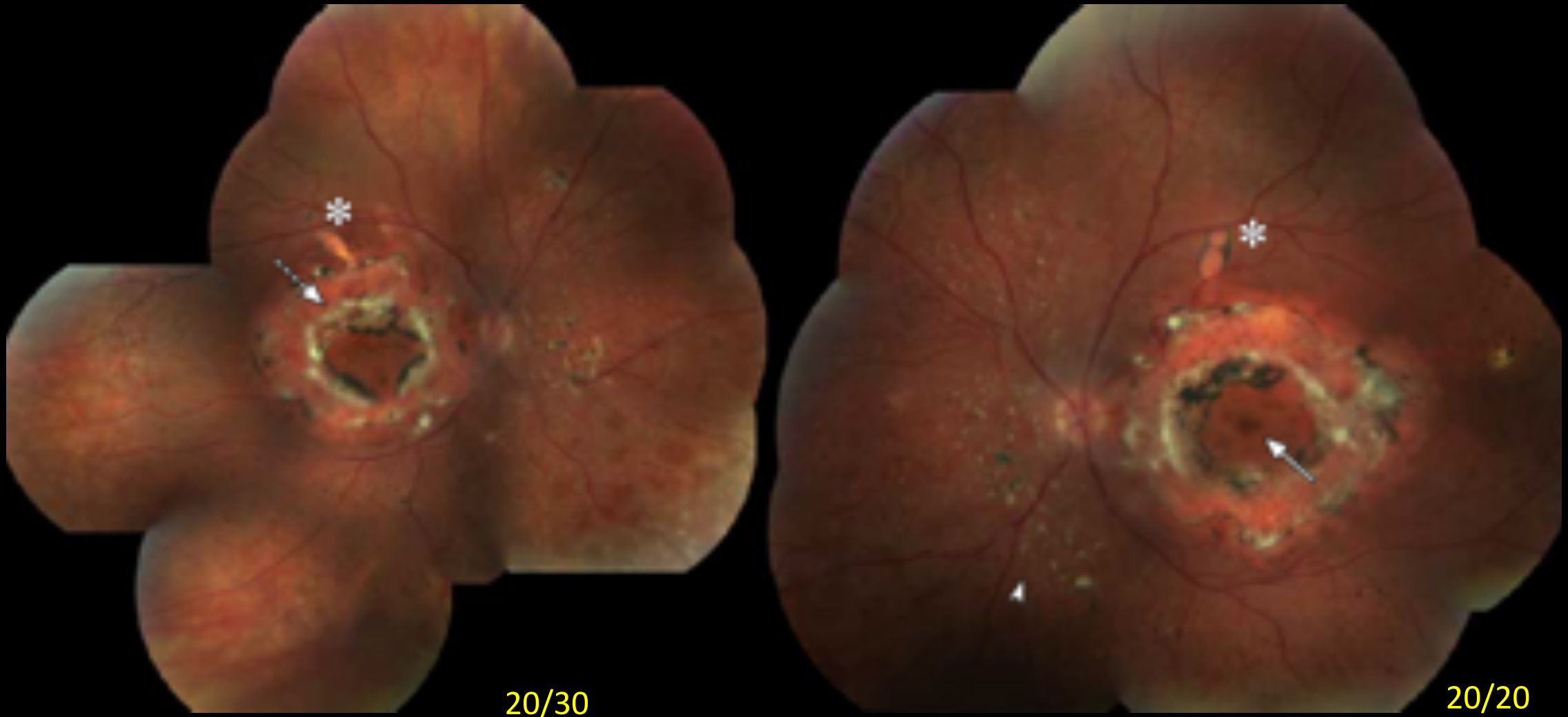
Fibrotic tissue + type 1 CNV

Retinal fibrosis in ESCS









20/30

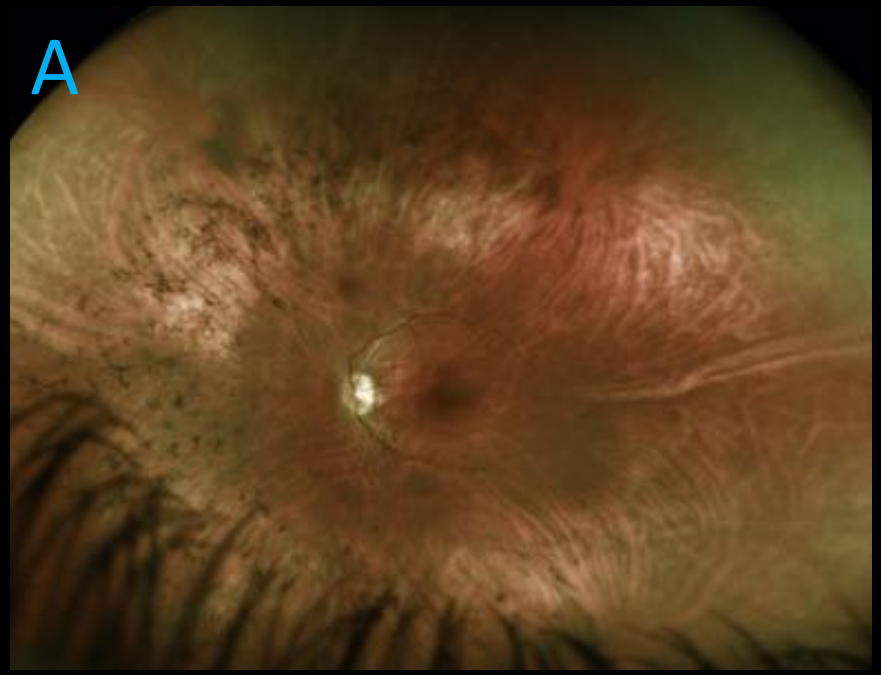
20/20

Helicoid fibrosis

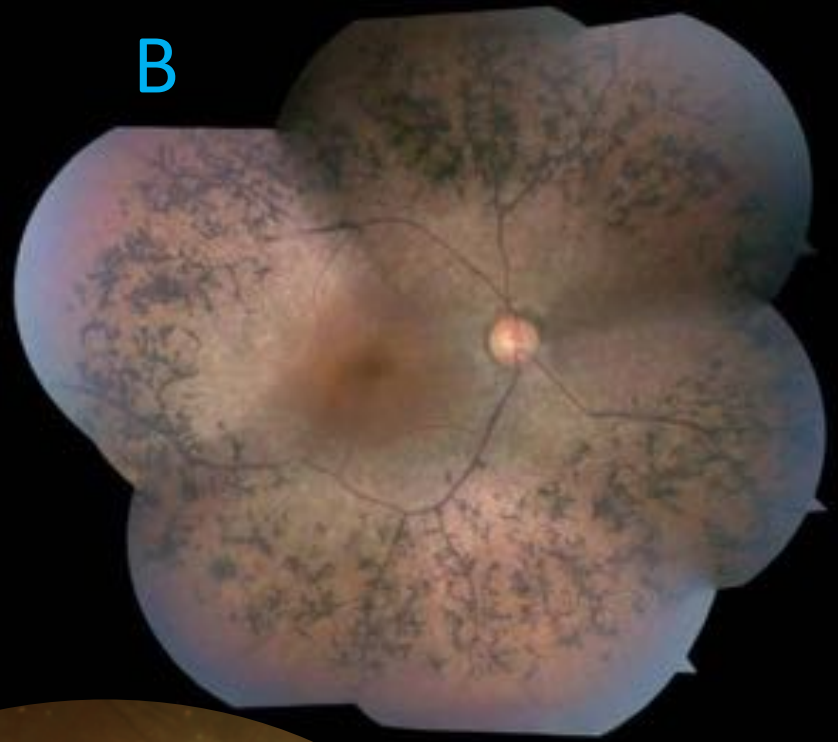
S. Yzer

ESCS = NR2E3
Which one?

A

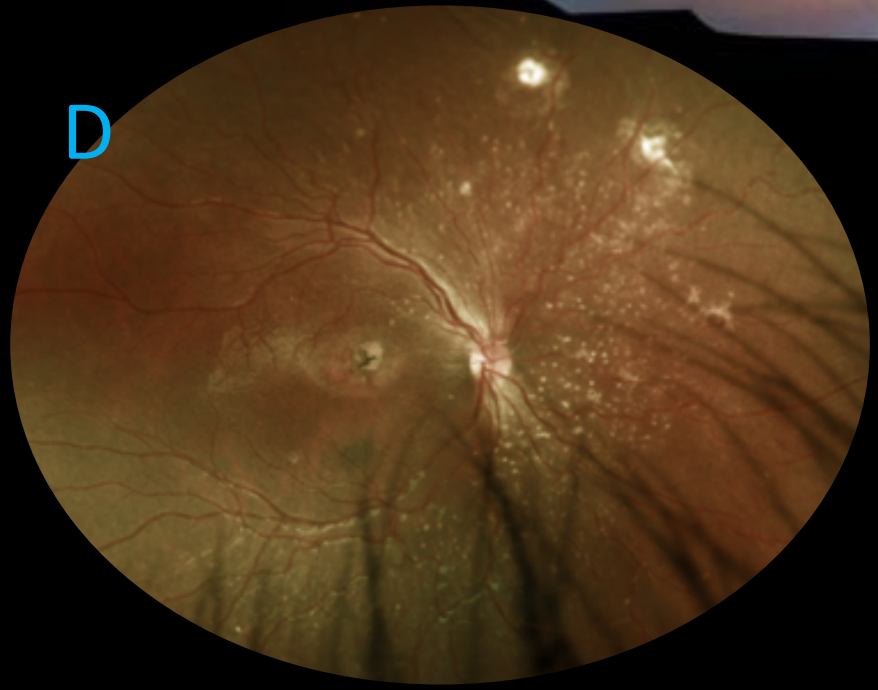


B



C

D

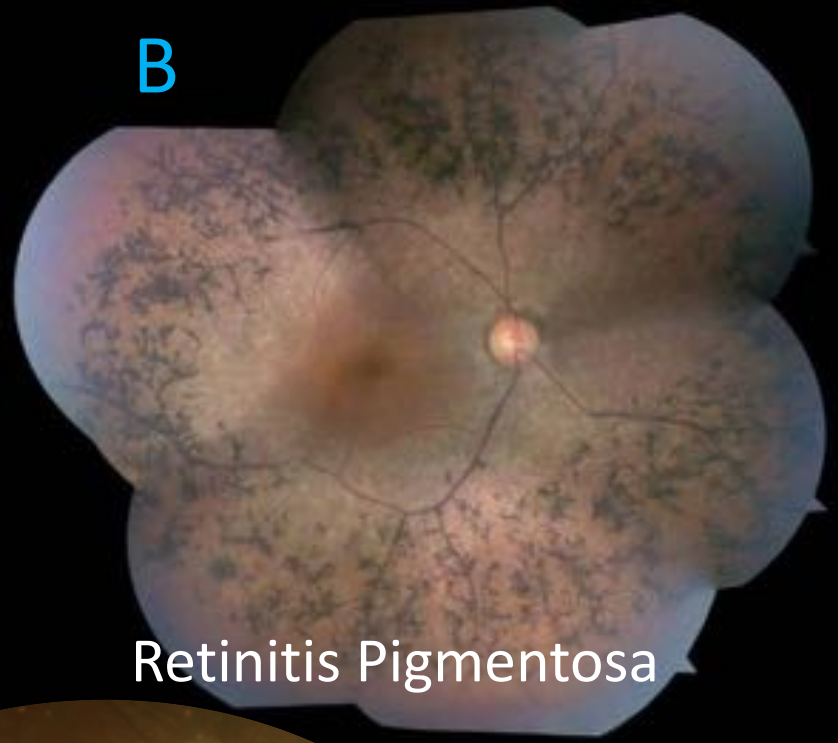


ESCS
Which one?

A

MAK1-associated RP

B

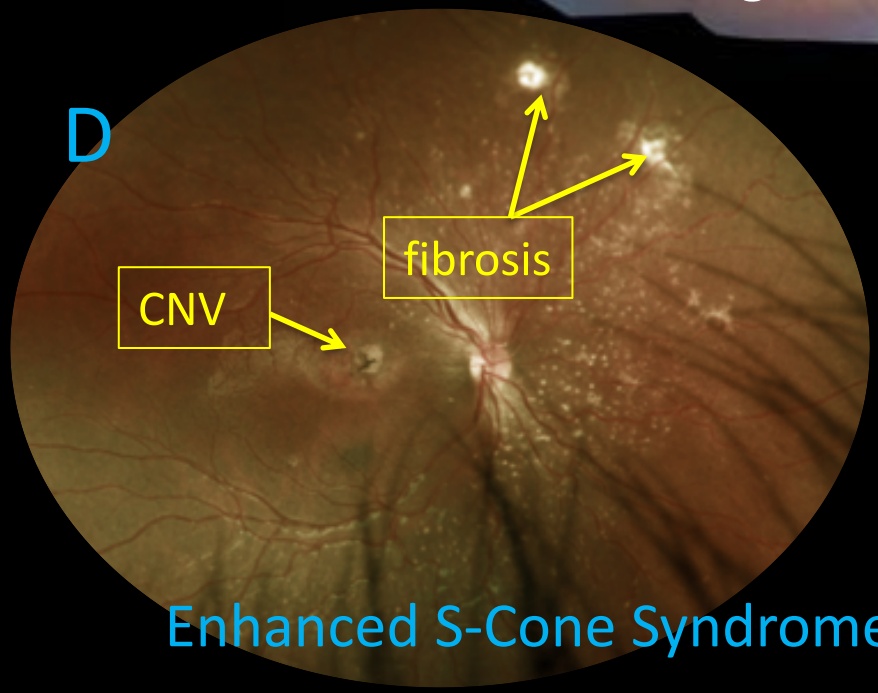


Retinitis Pigmentosa

C

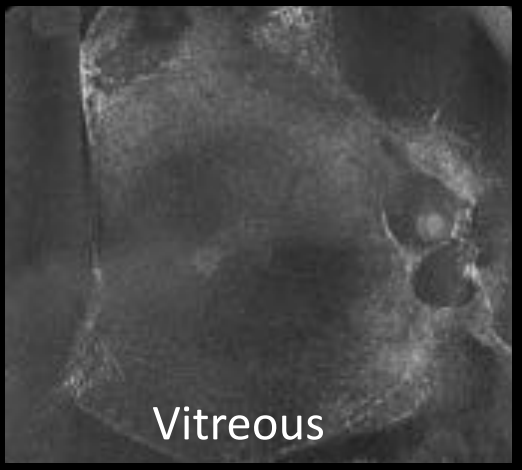
X Linked Retinoschisis

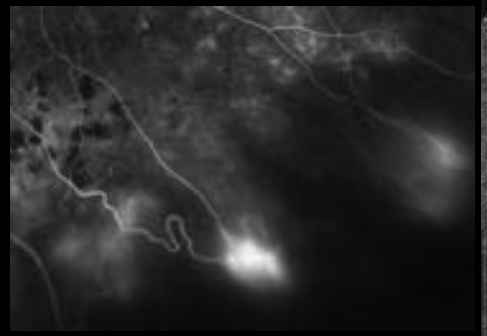
D

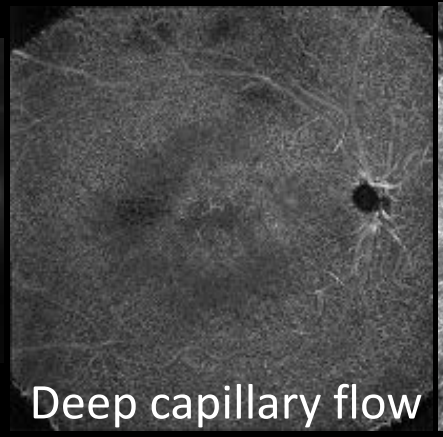


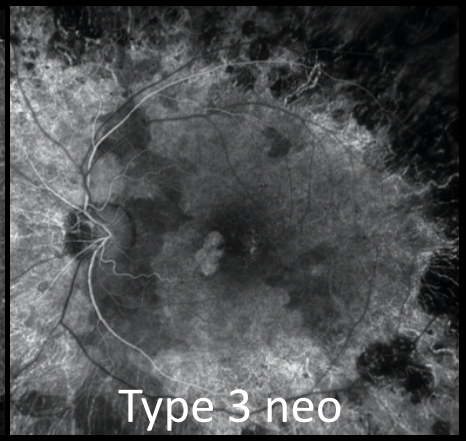
Enhanced S-Cone Syndrome

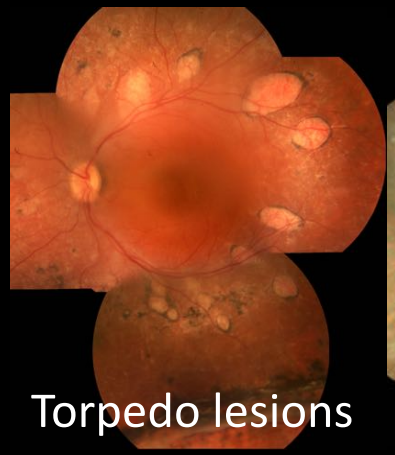
ESCS or NR2E3 is Favre-Goldmann Summary


Vitreous

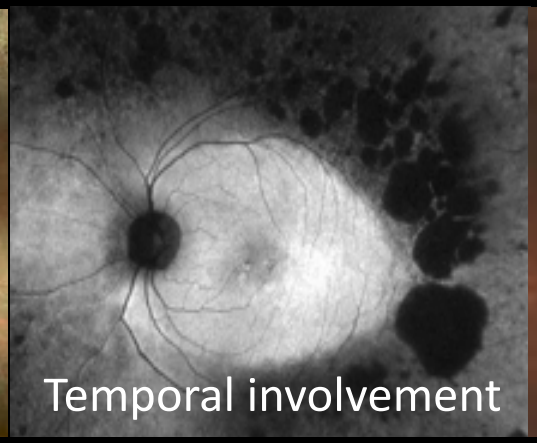

Retinal neo


Deep capillary flow


Type 3 neo


Torpedo lesions


Nummular lesions


Temporal involvement


Fibrosis

The Enhanced S-Cone Syndrome Project

Gerardo Ledesma-Gil

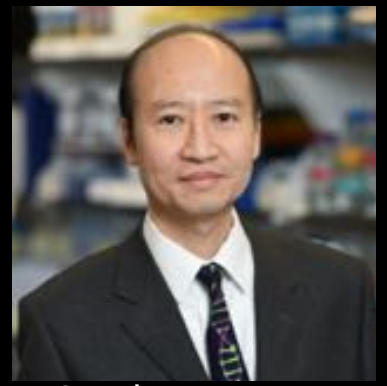
Juliet Essilfie

Mark Breazzano

Pedro Fernandez

Bailey Freund

Richard Spaide



Stephen Tsang