

ESCS

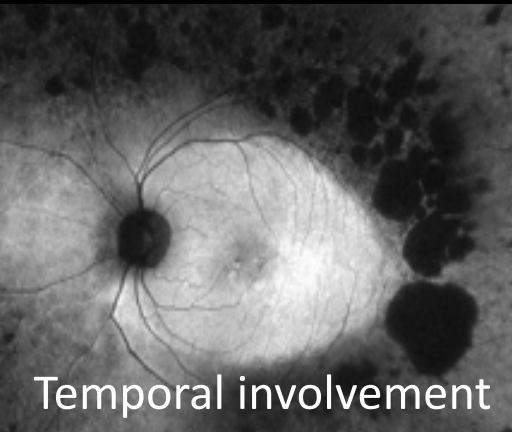
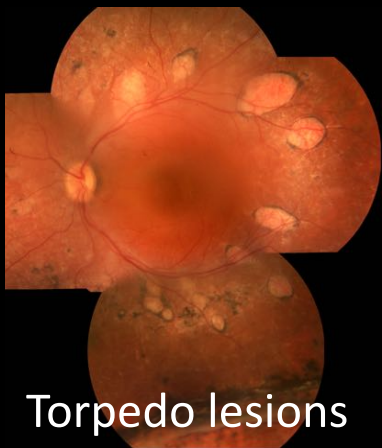
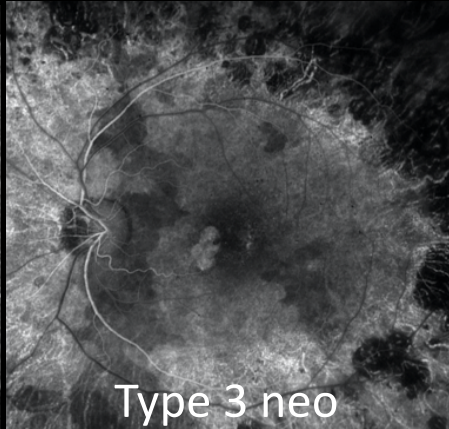
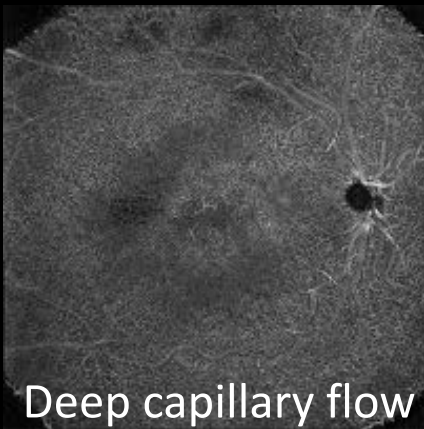
Expanding the clinical spectrum

Retina Society 2020

Lawrence A. Yannuzzi

Financial Disclosures: None

ESCS or NR2E3 is Favre-Goldmann
Summary



Golden-Collar-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 hyaloïdéo-rétinienne.

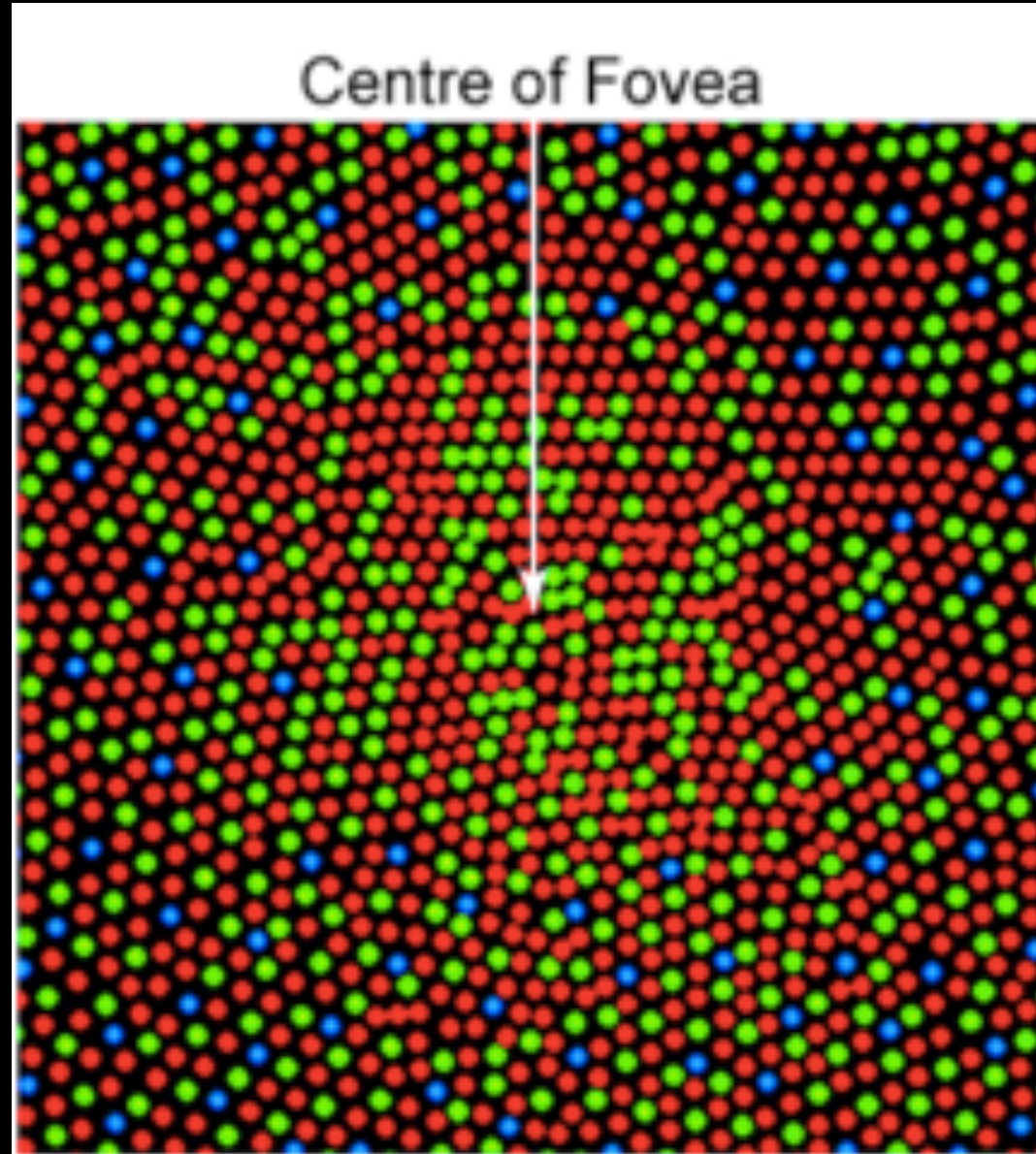
Par M. FAVRE.

Il nous a paru intéressant de vous présenter le cas de deux enfants, frère et sœur, atteints d'une dégénérescence hyaloïdéo-rétinienne 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 :

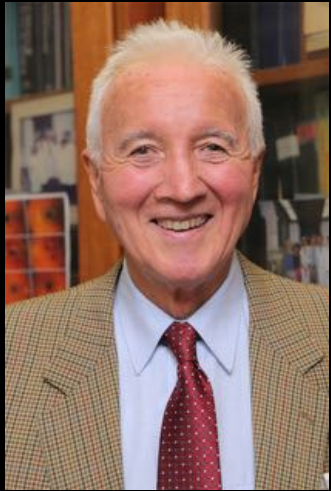
1958

Enhanced S Cone Syndrome



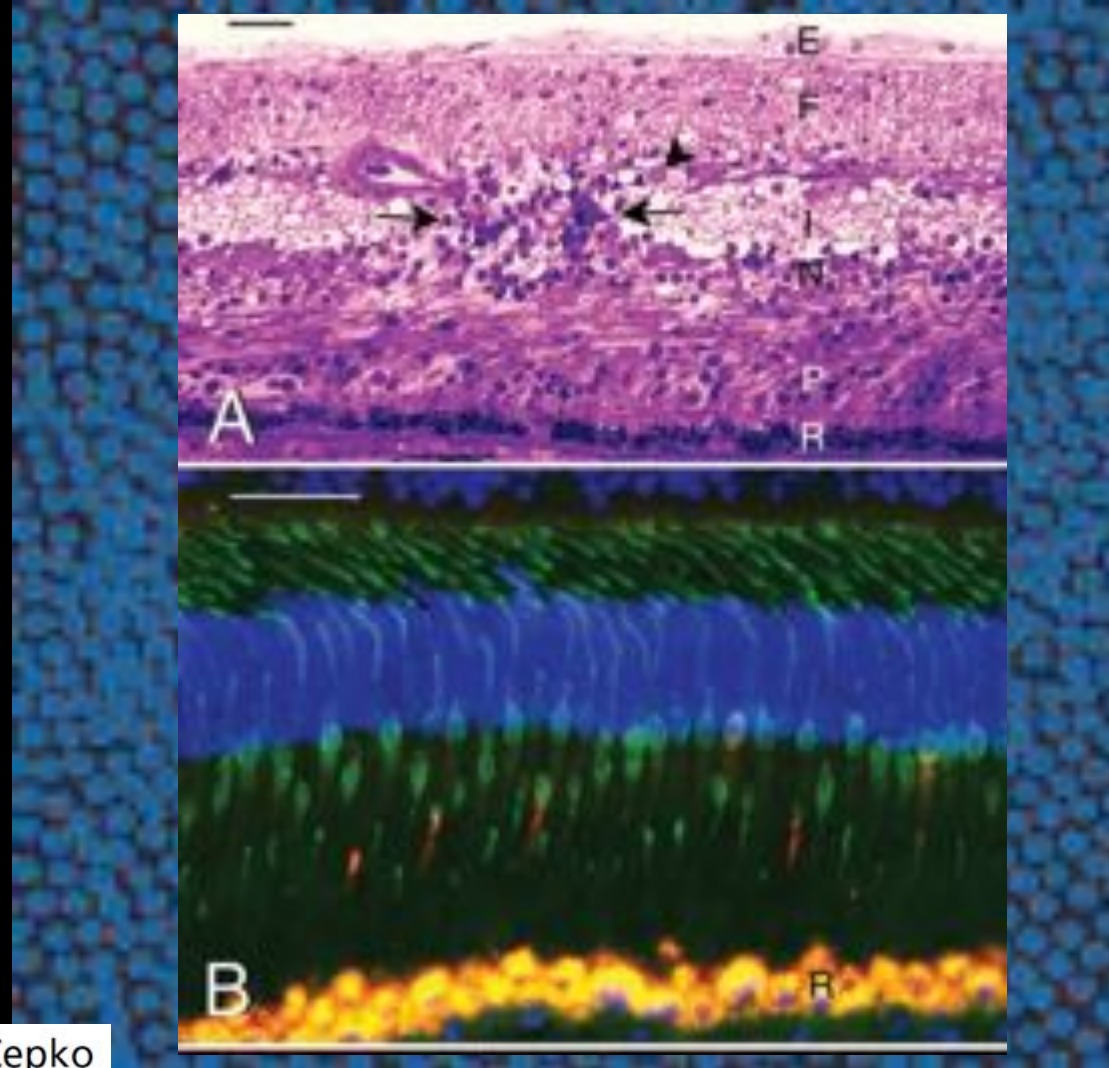
Enhanced S Cone Syndrome (92% blue cones)

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*^{1,2}, Linda Rose*, Artur V. Cideciyan*, Mark R. Barakat*, Wai-Xing Tang*, Nisha Gupta*, Tomas S. Aleman*, Alan F. Wright³, Edwin M. Stone⁴, Val C. Sheffield¹, and Samuel G. Jacobson*

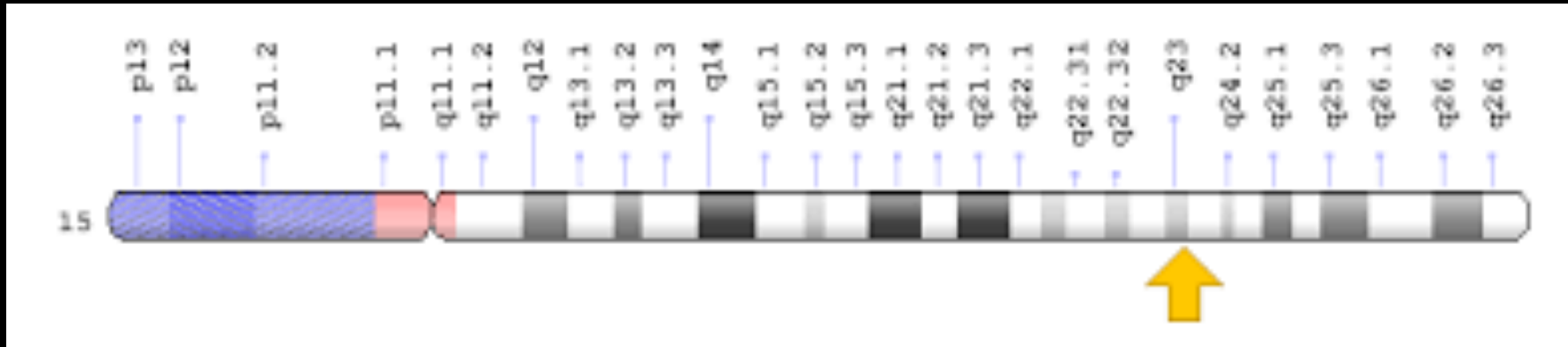
2001

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 E. Wright,² and Samuel G. Jacobson¹

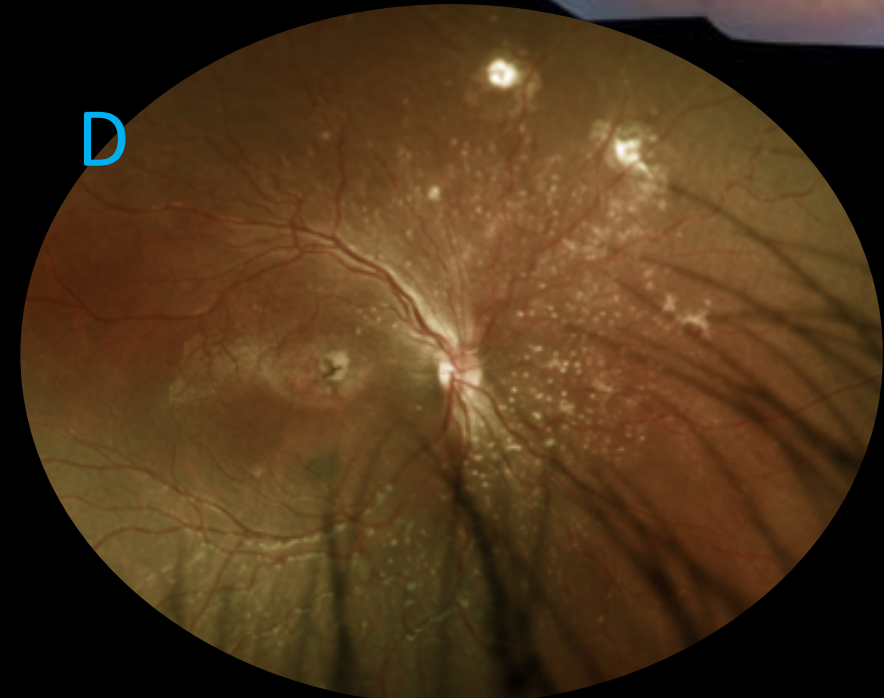
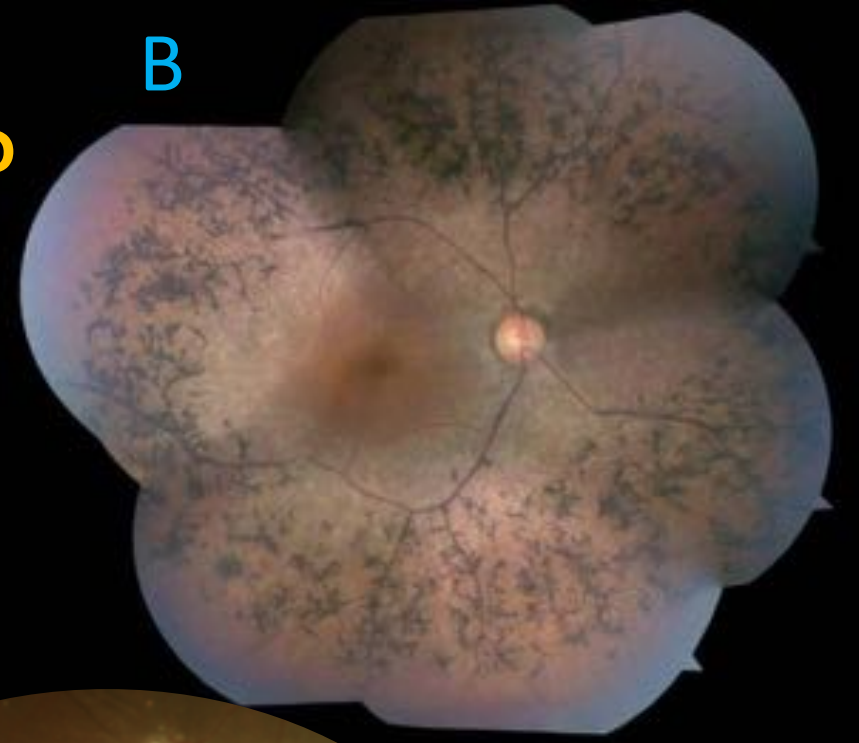
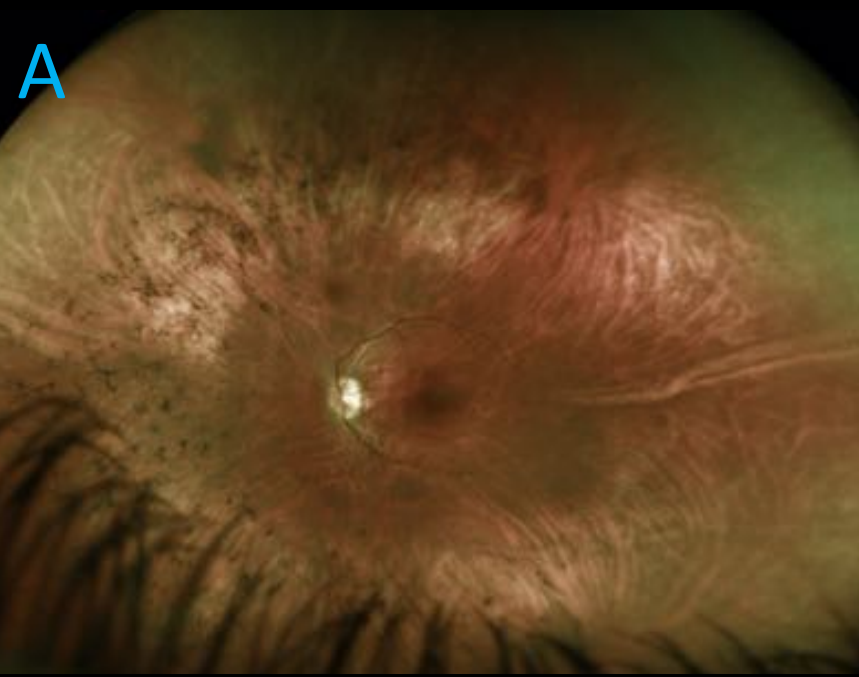


37 mutations

Mostly compound heterozygous
Fewer than 2% no detectable allele

ESCS

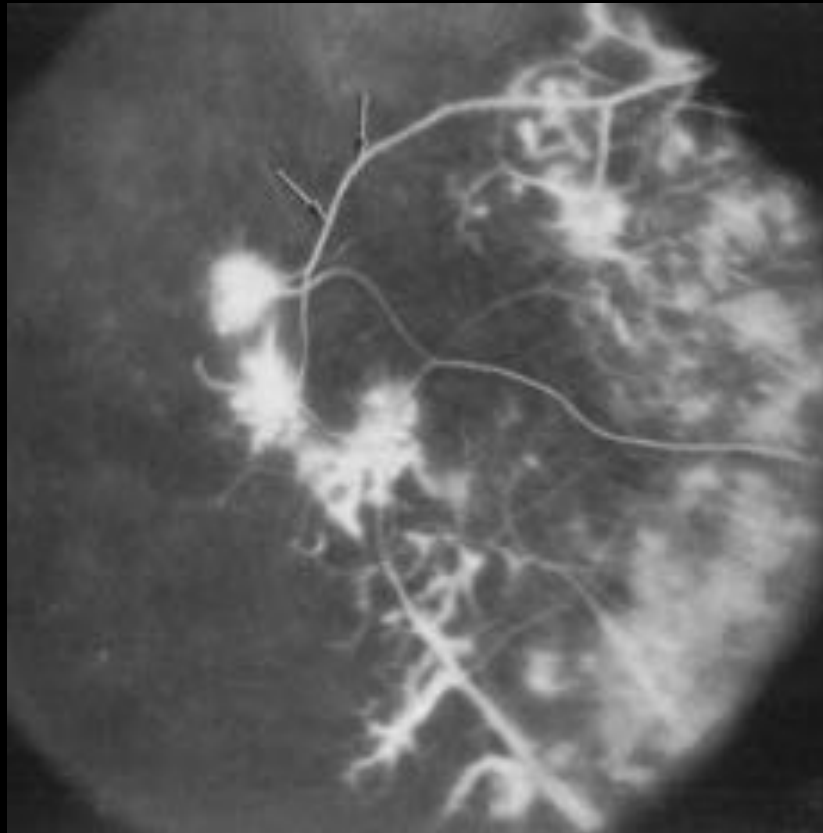
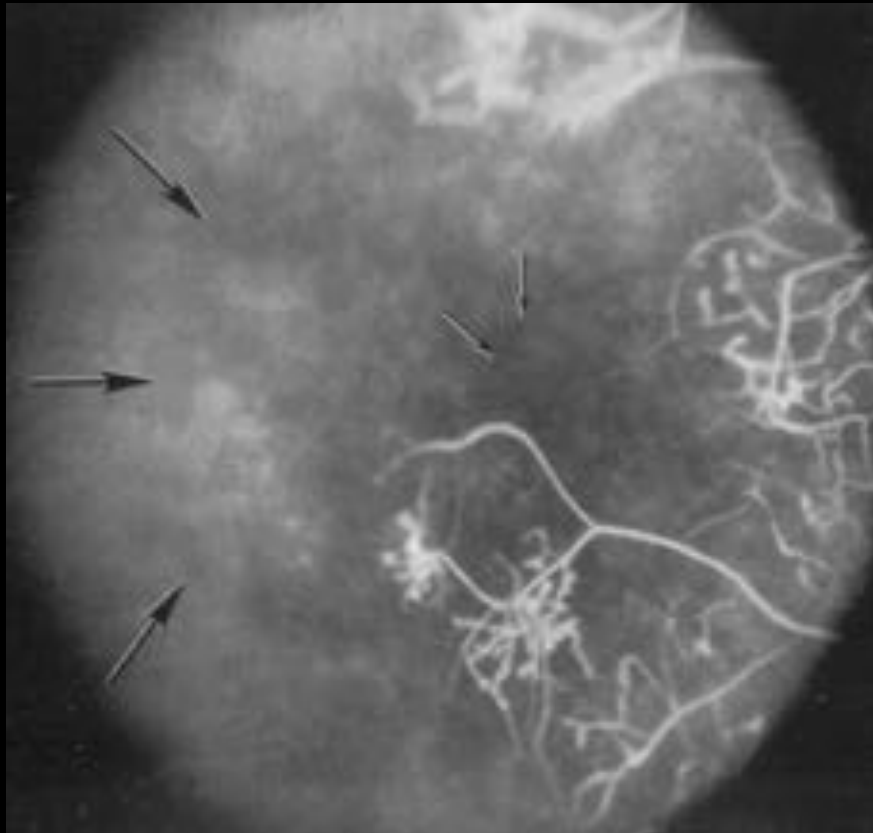
Which one fits?



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

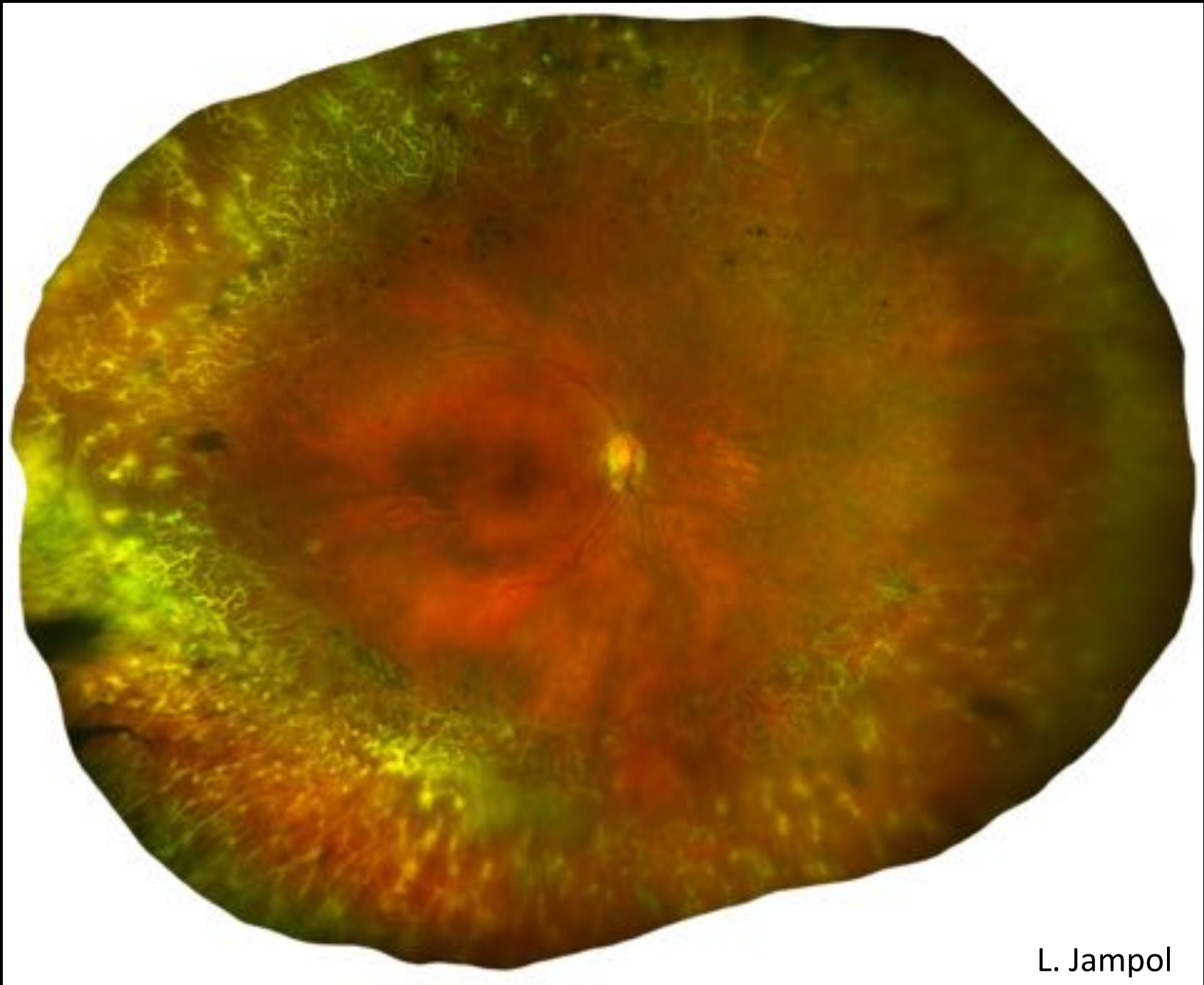
Vitreous degeneration

Pigmentary degeneration

ERG changes

Retinal non-perfusion

Diffuse retinal leakage

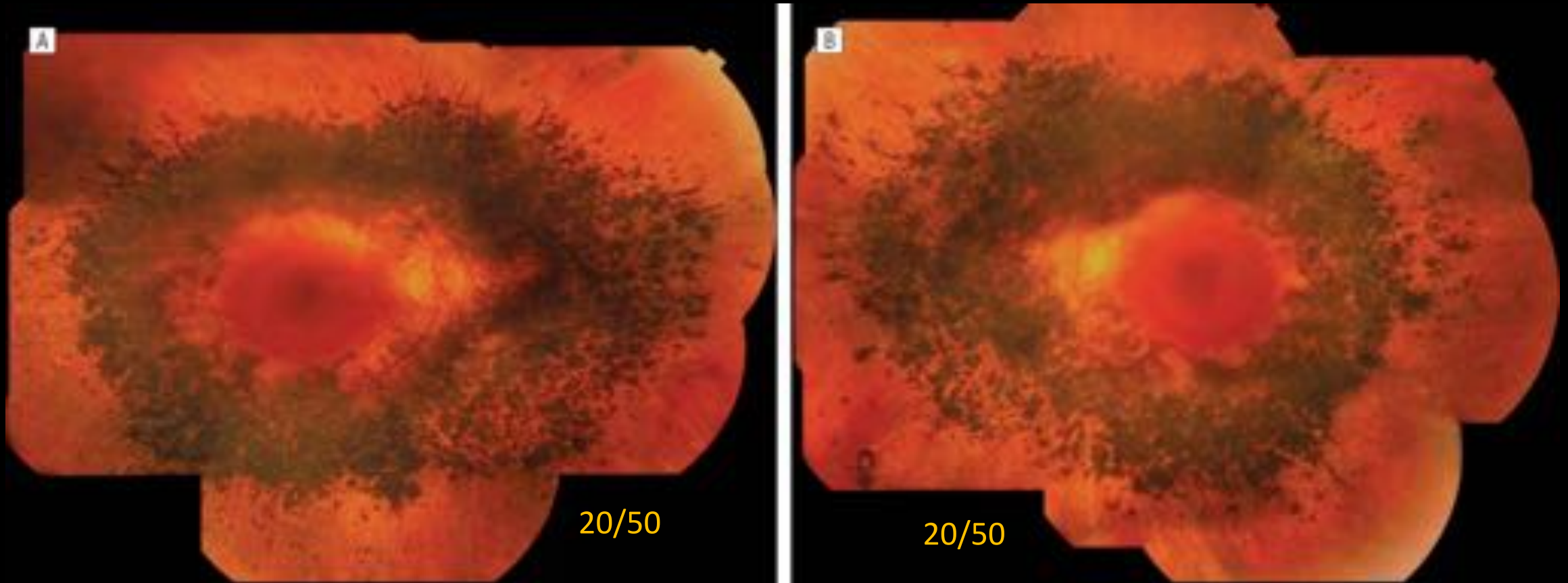


L. Jampol

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

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

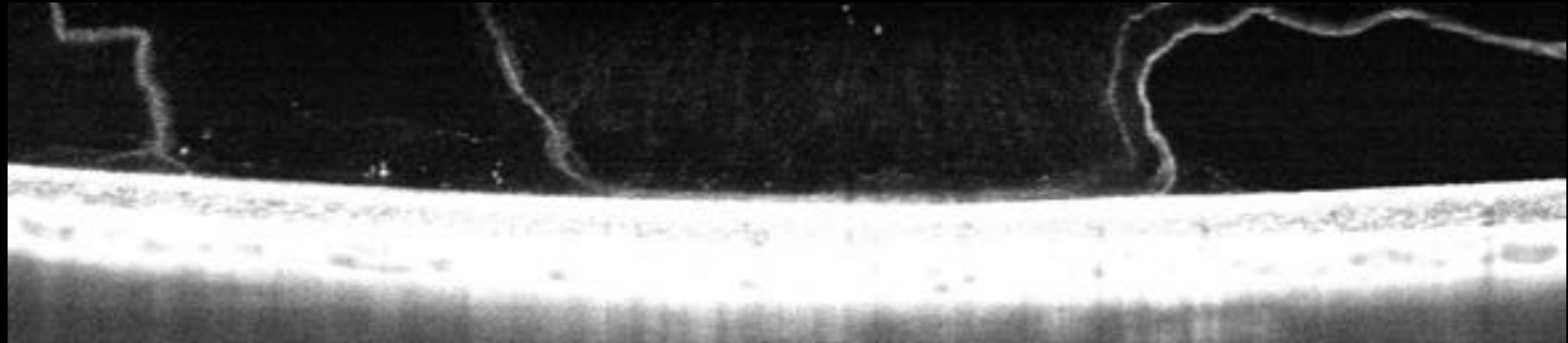


- Nyctalopia at early age
- PSC
- CME
- Peripheral retinoschisis
- Peripheral retinal ischemia

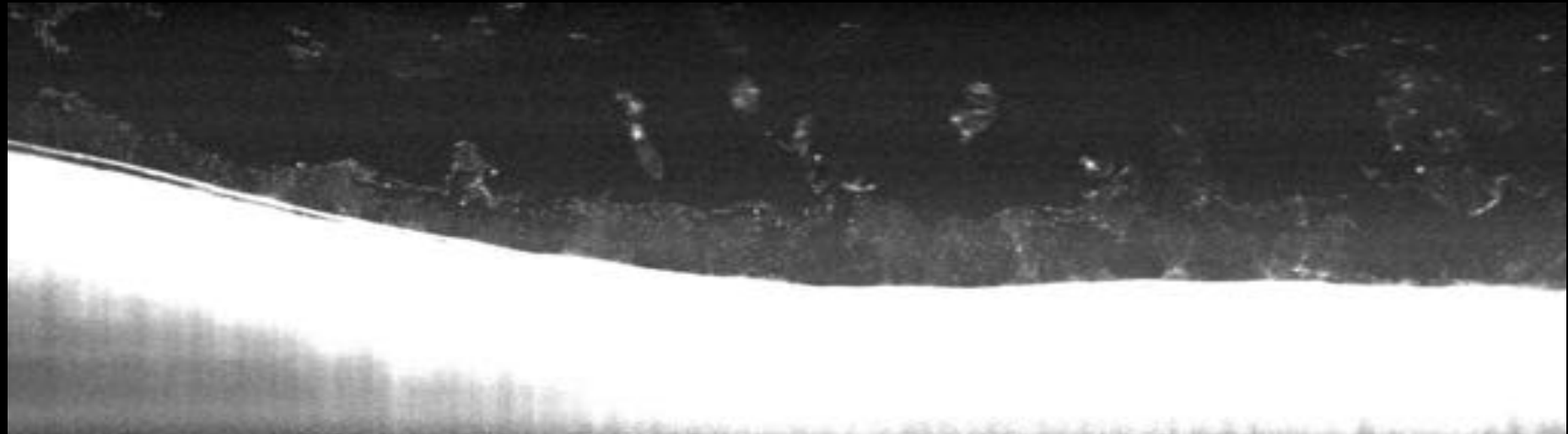
Current Series

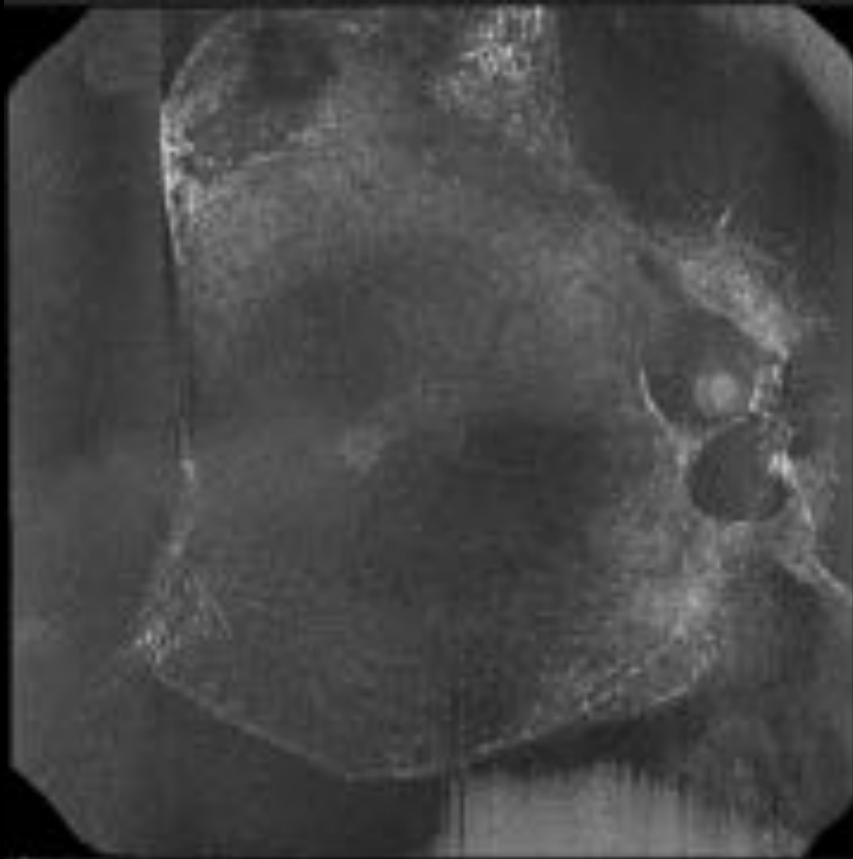
Vitreous In ESCS

Thick posterior hyaloid



Fibrillar changes



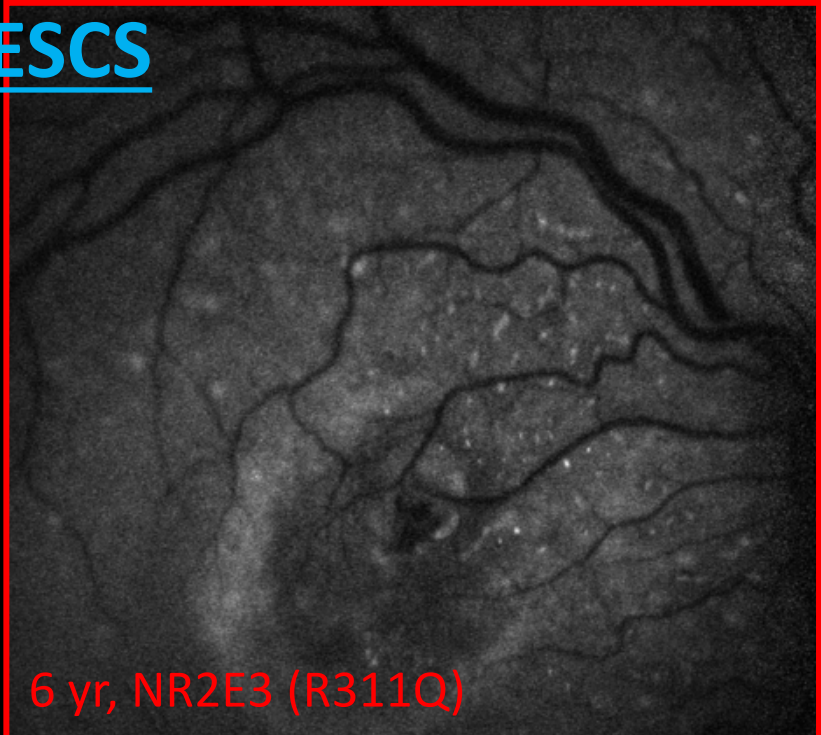


En face OCT vitreous segment

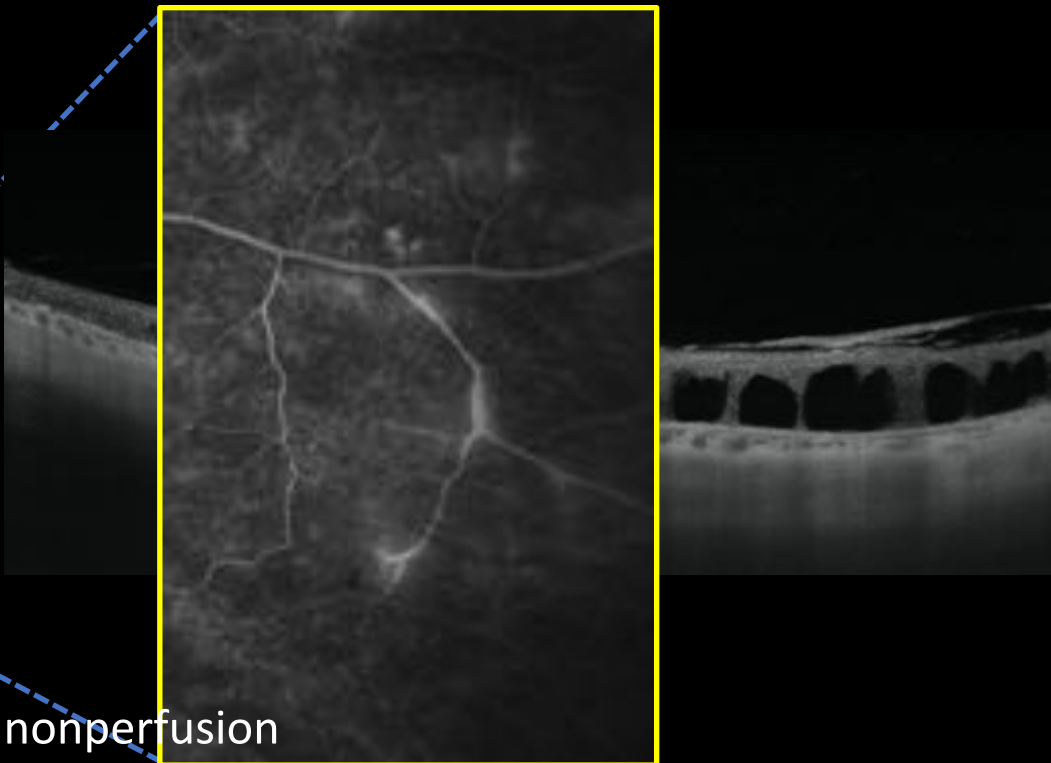
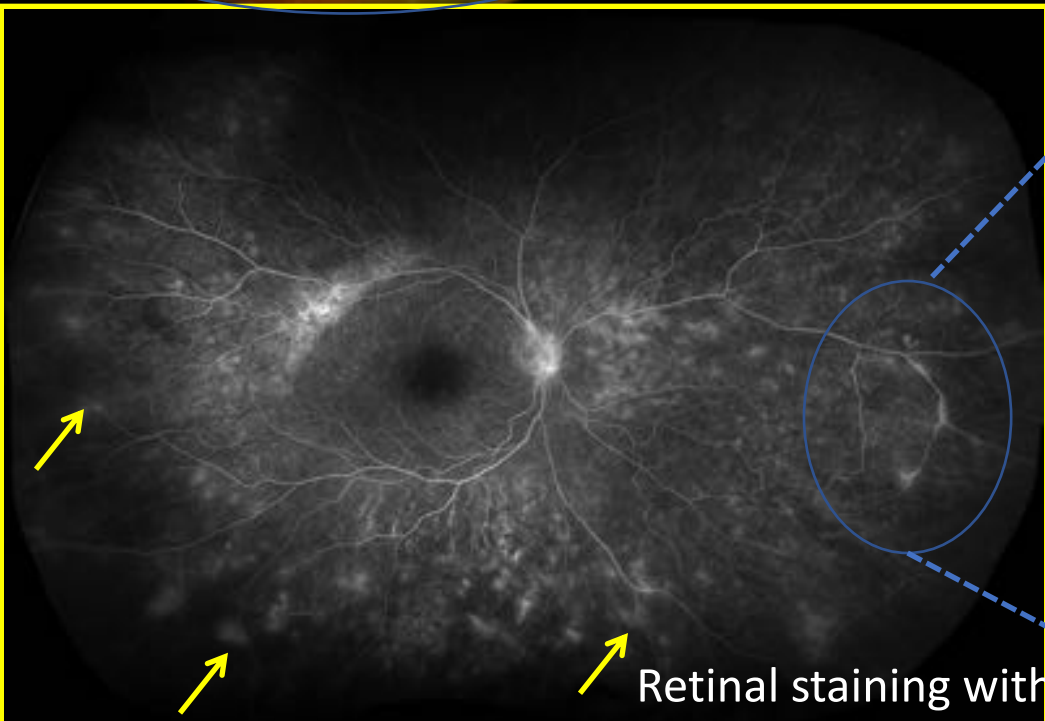
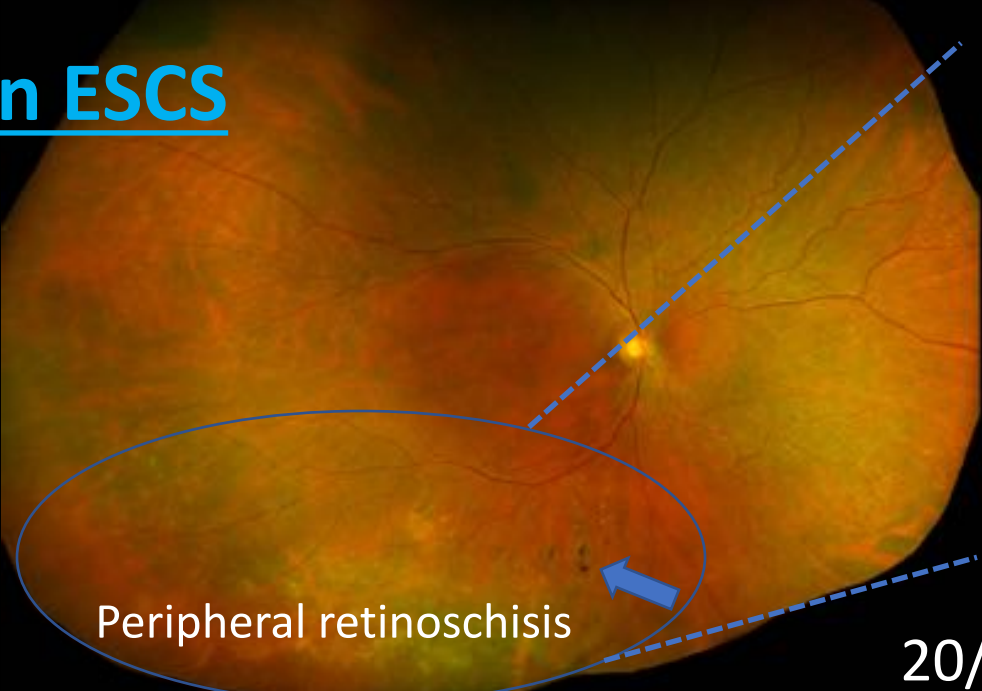


SQUARE or polygonal vitreomacular adhesion + thick posterior hyaloid

Retina In ESCS

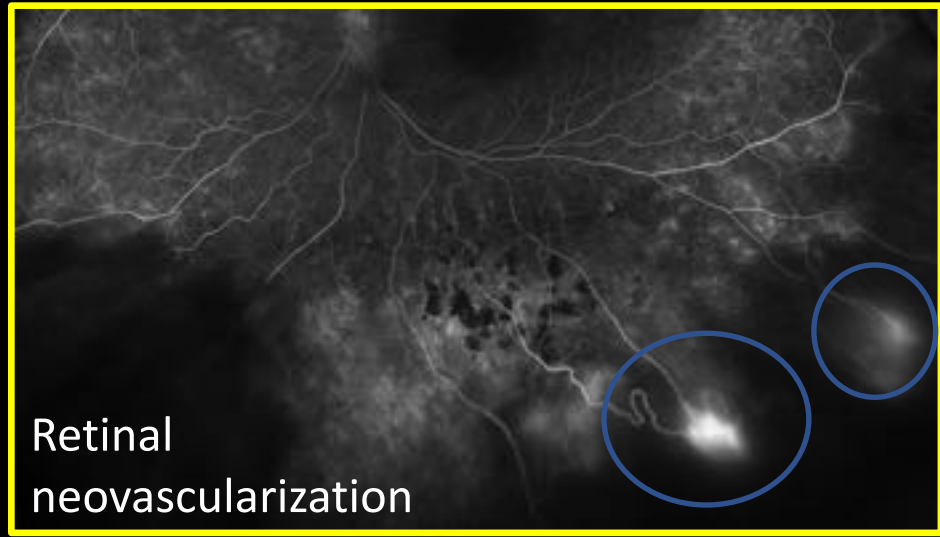
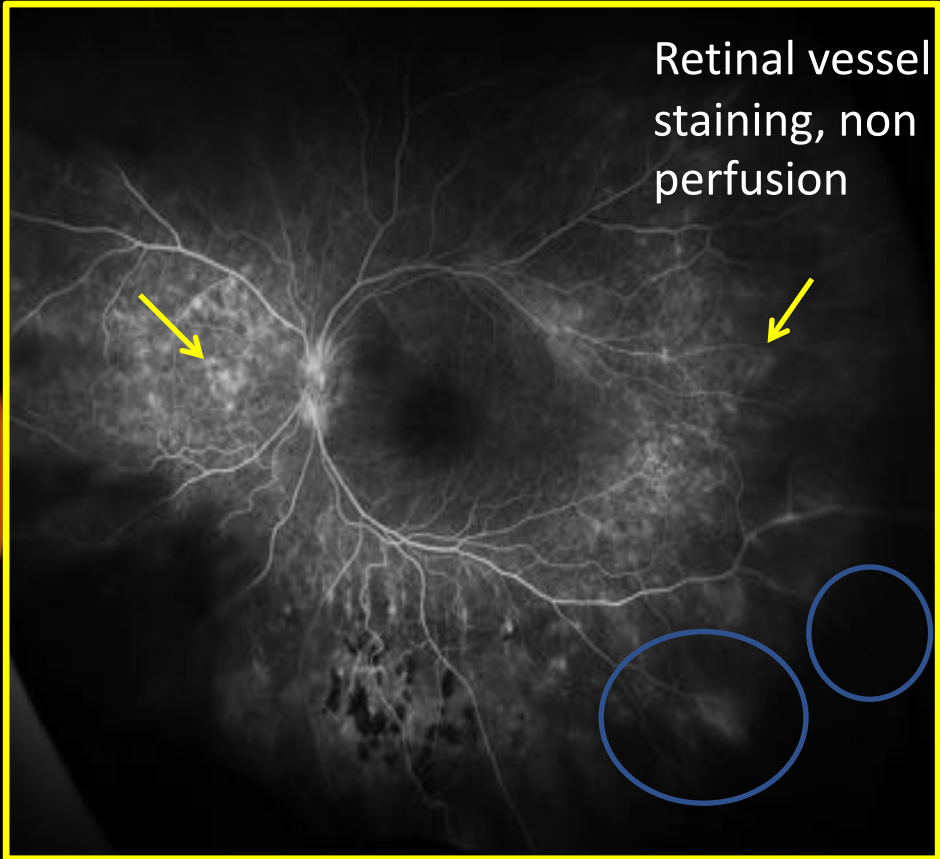
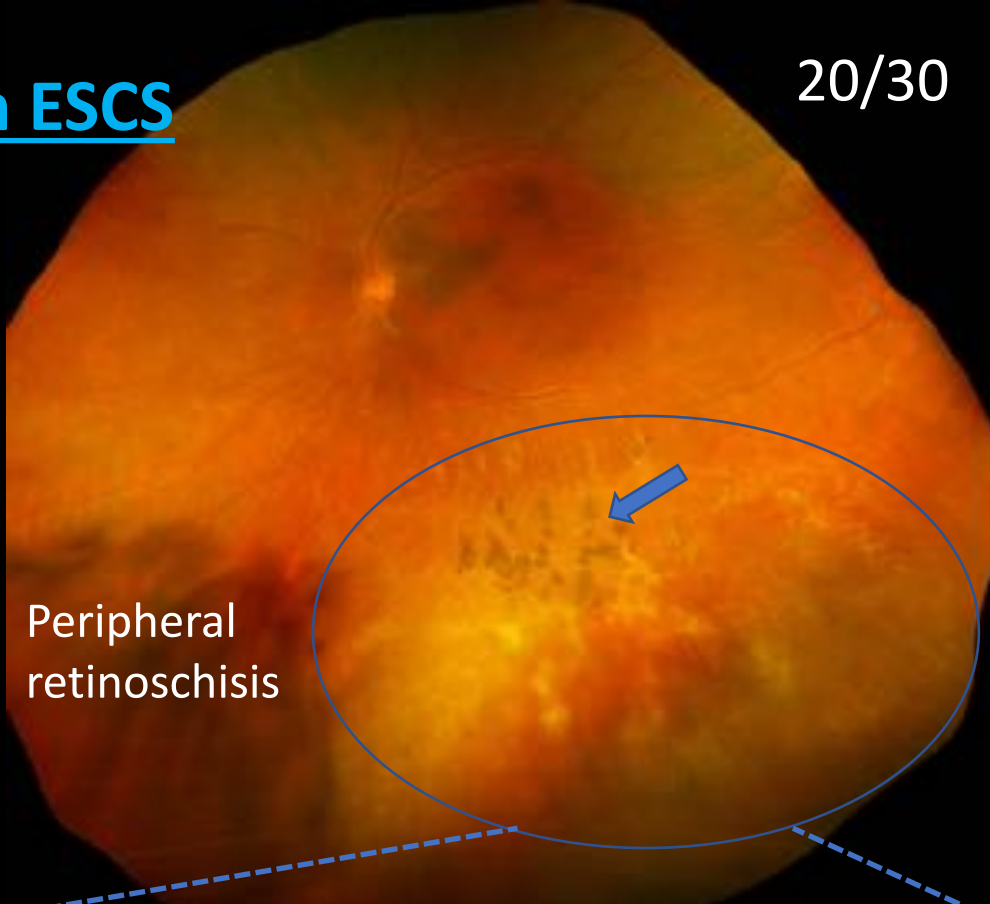


Retina In ESCS



Retina In ESCS

20/30



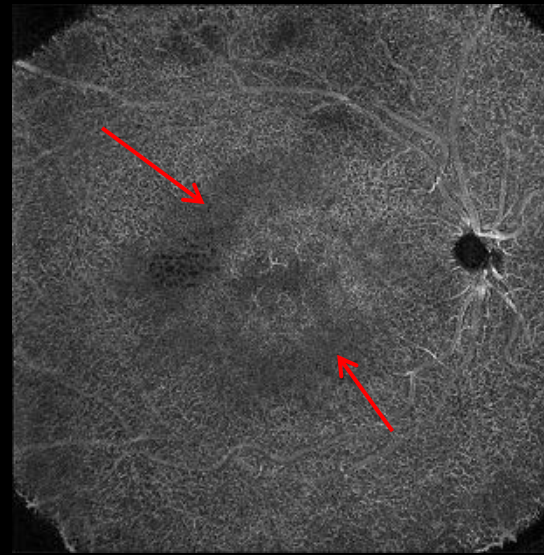
Retina In ESCS



Full retina

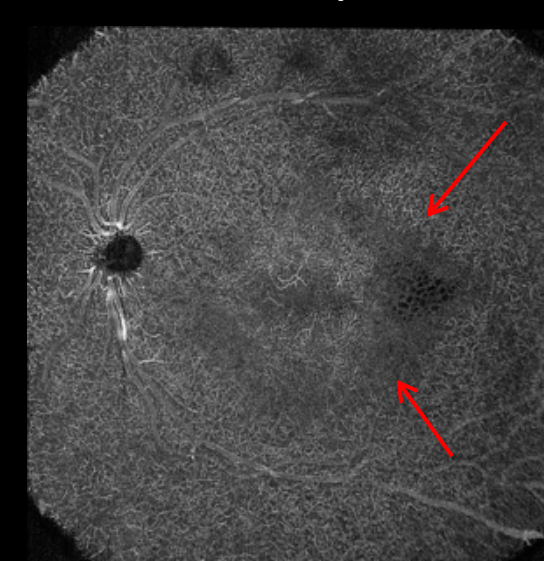
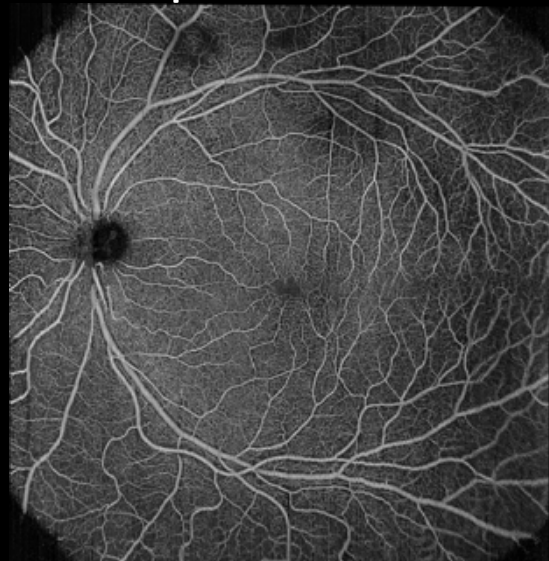
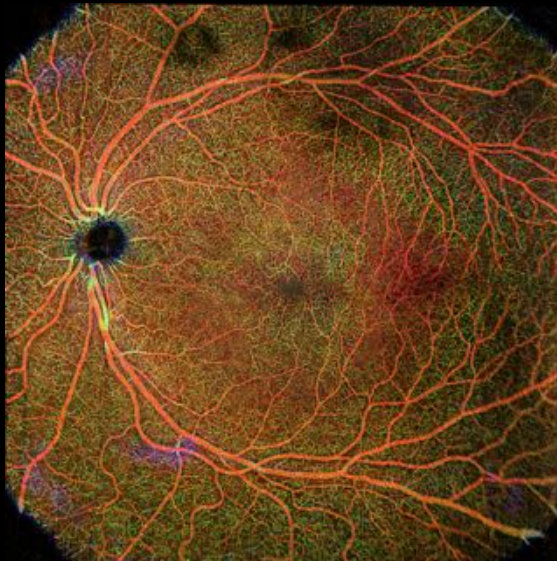


Superficial



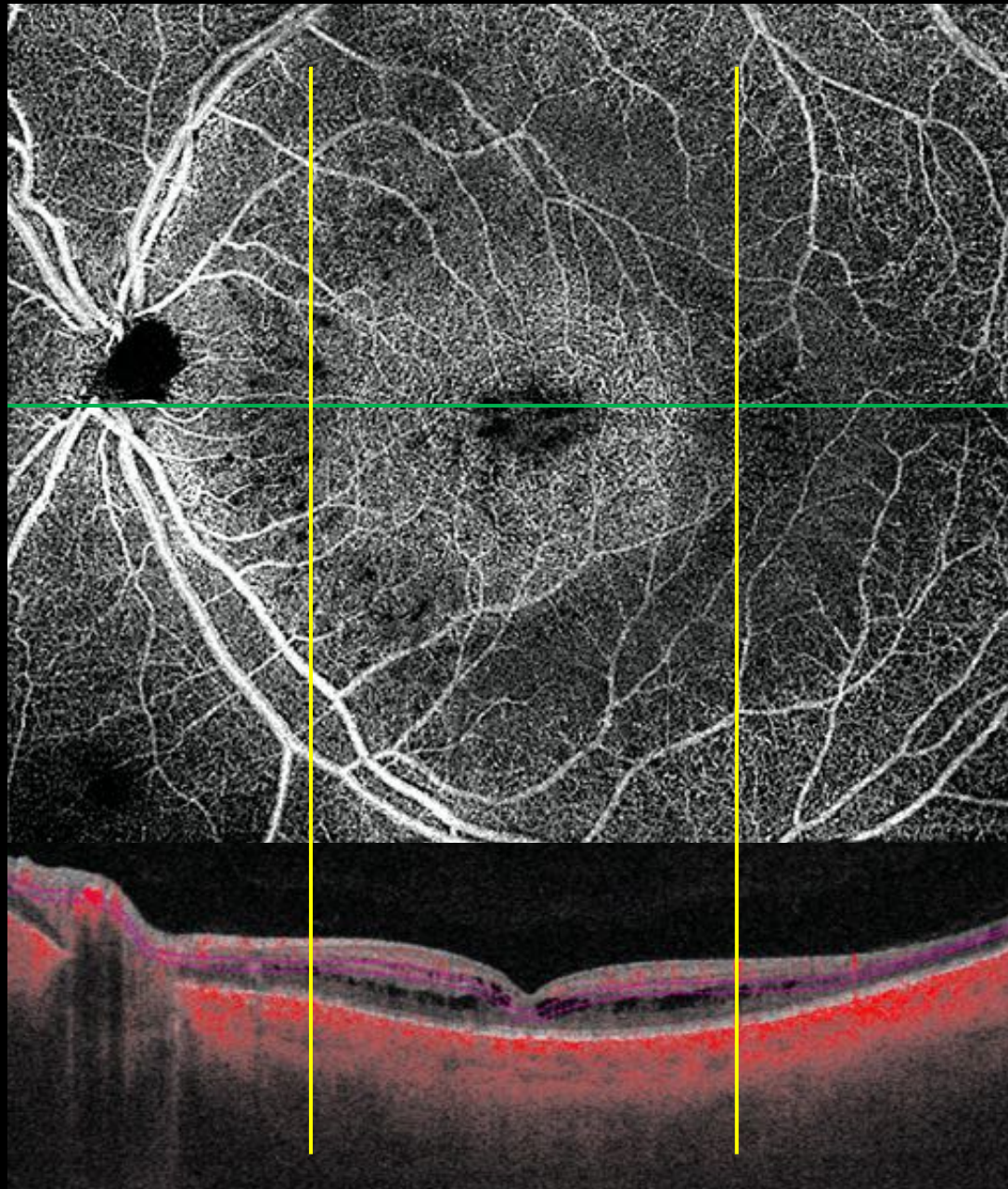
Deep

OCT-A

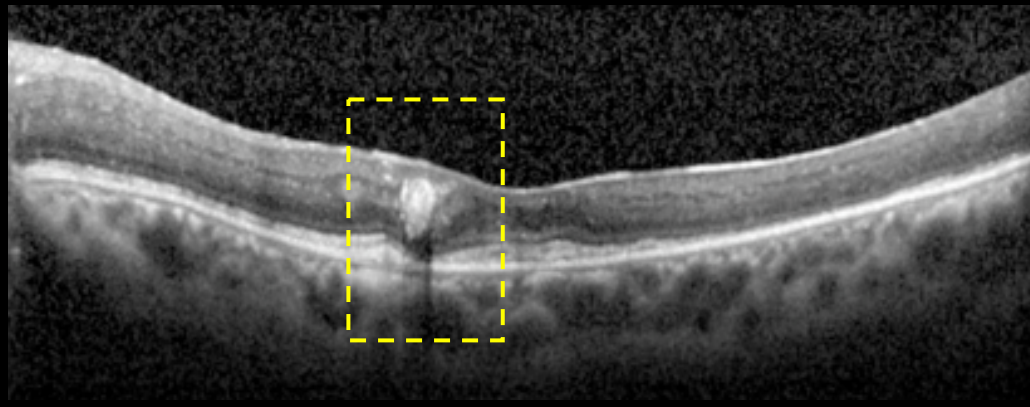
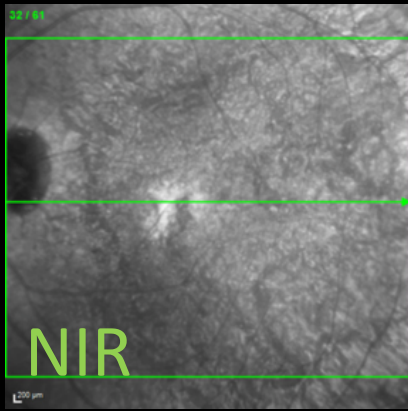


REDUCED SIGNAL OR NON-PERFUSION IN DEEP CAPILLARY PLEXUS

Retina In ESCS

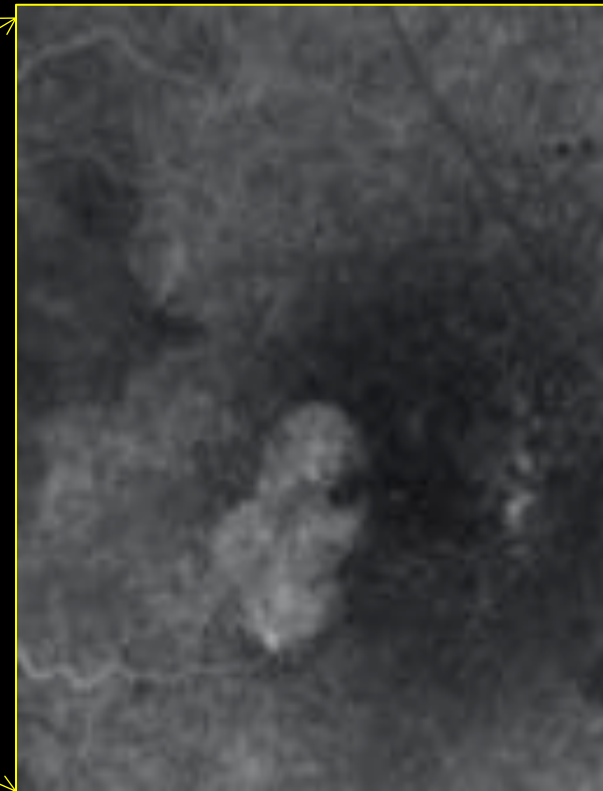
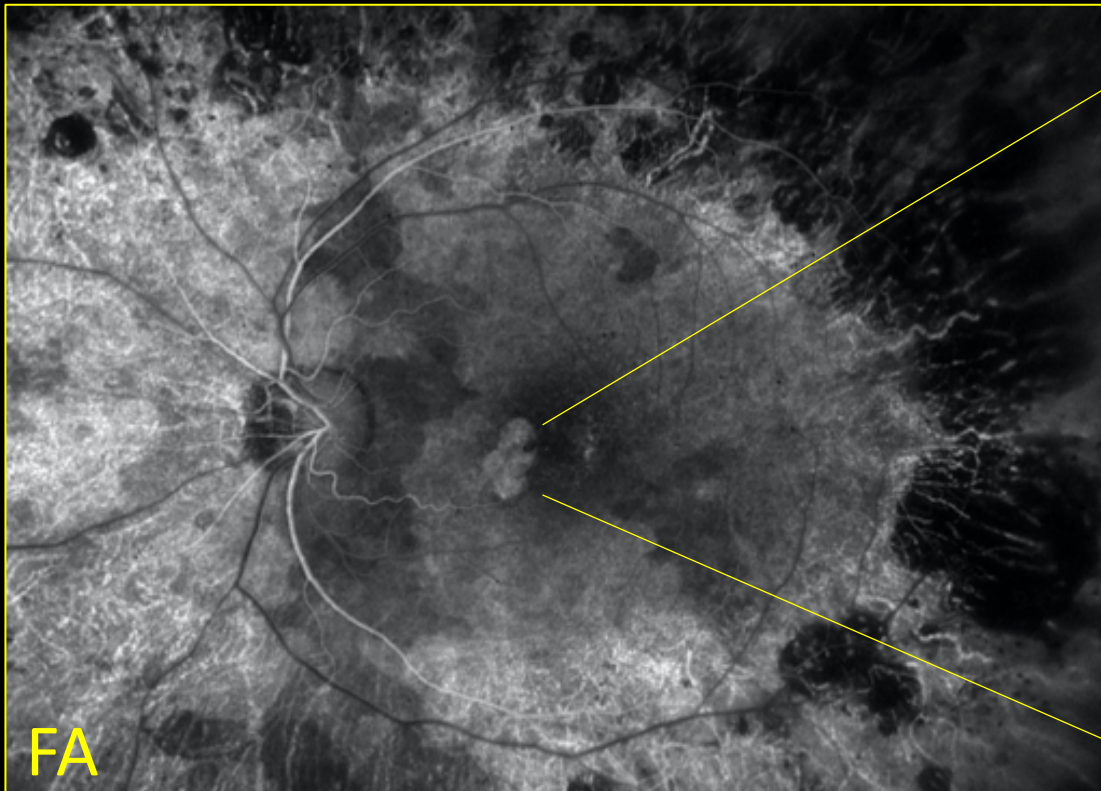


REDUCED DEEP CAPILLARY PLEXUS FLOW DOES NOT CORRESPOND TO RETINOCHISIS



Neovascular
changes

Angiomatous lesion
TYPE 3 NEO

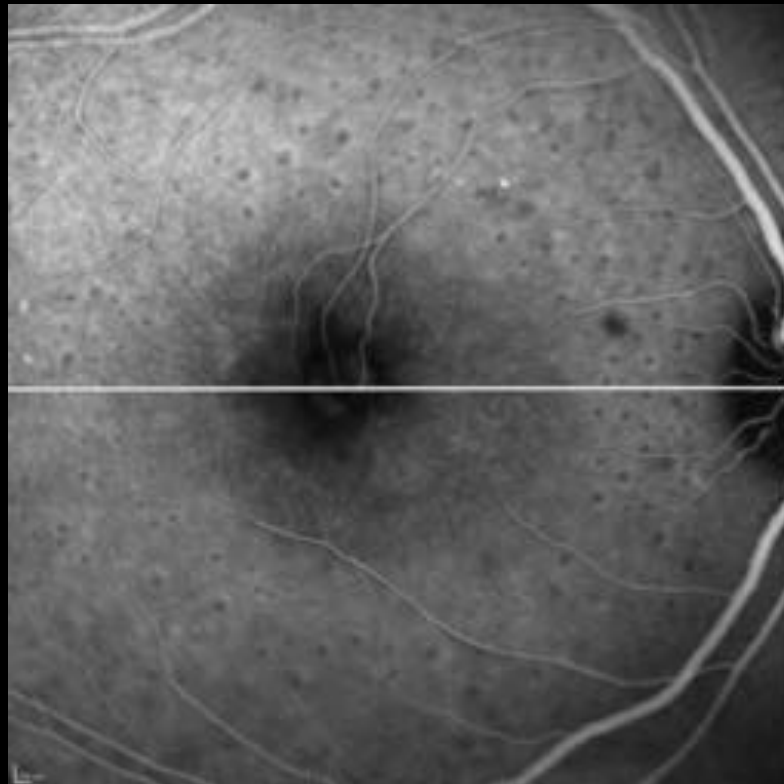


RETINOCHOROIDAL ANASTOMOSIS ASSOCIATED WITH ENHANCED S-CONE SYNDROME

10 year-old boy

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*

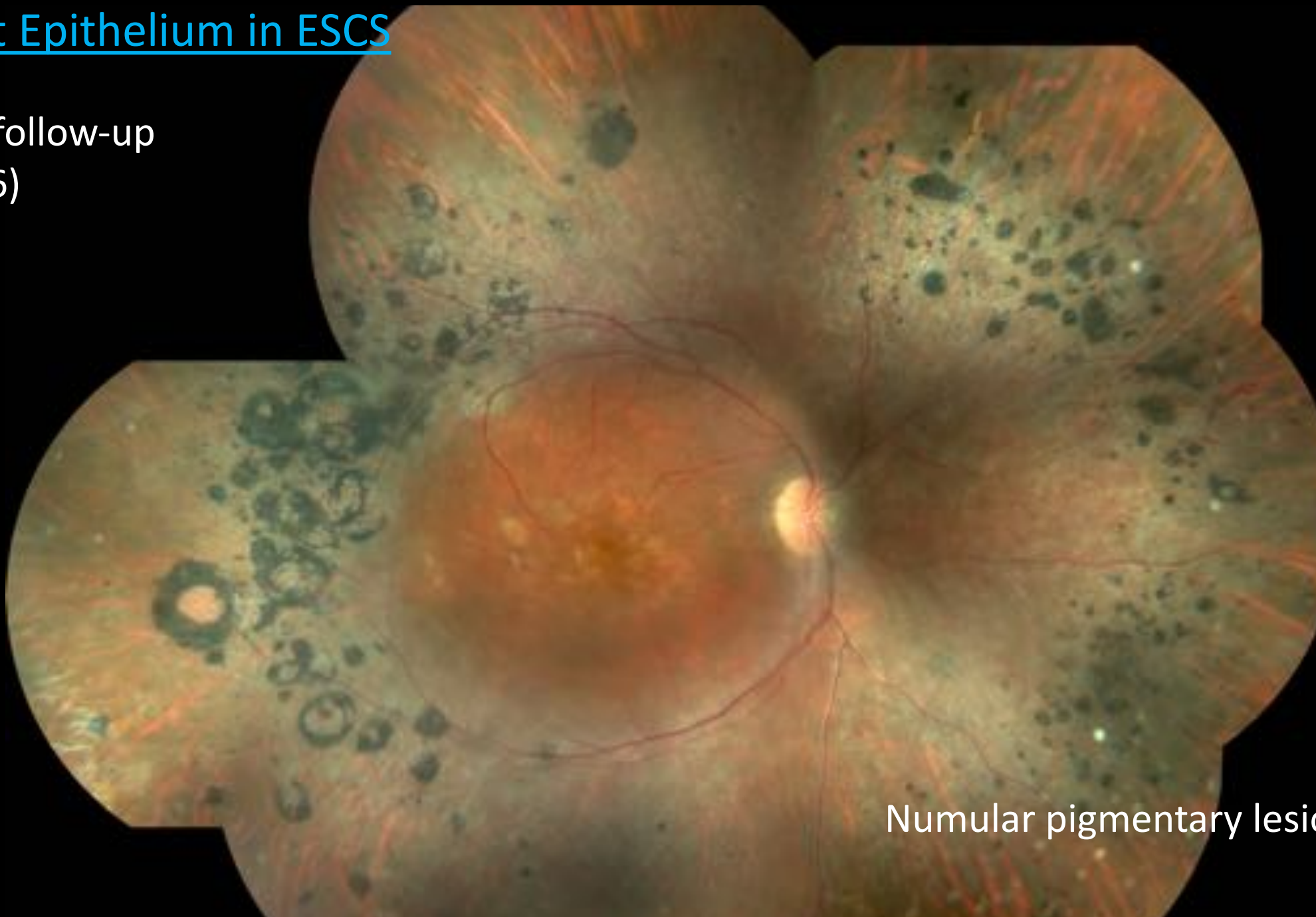
2019



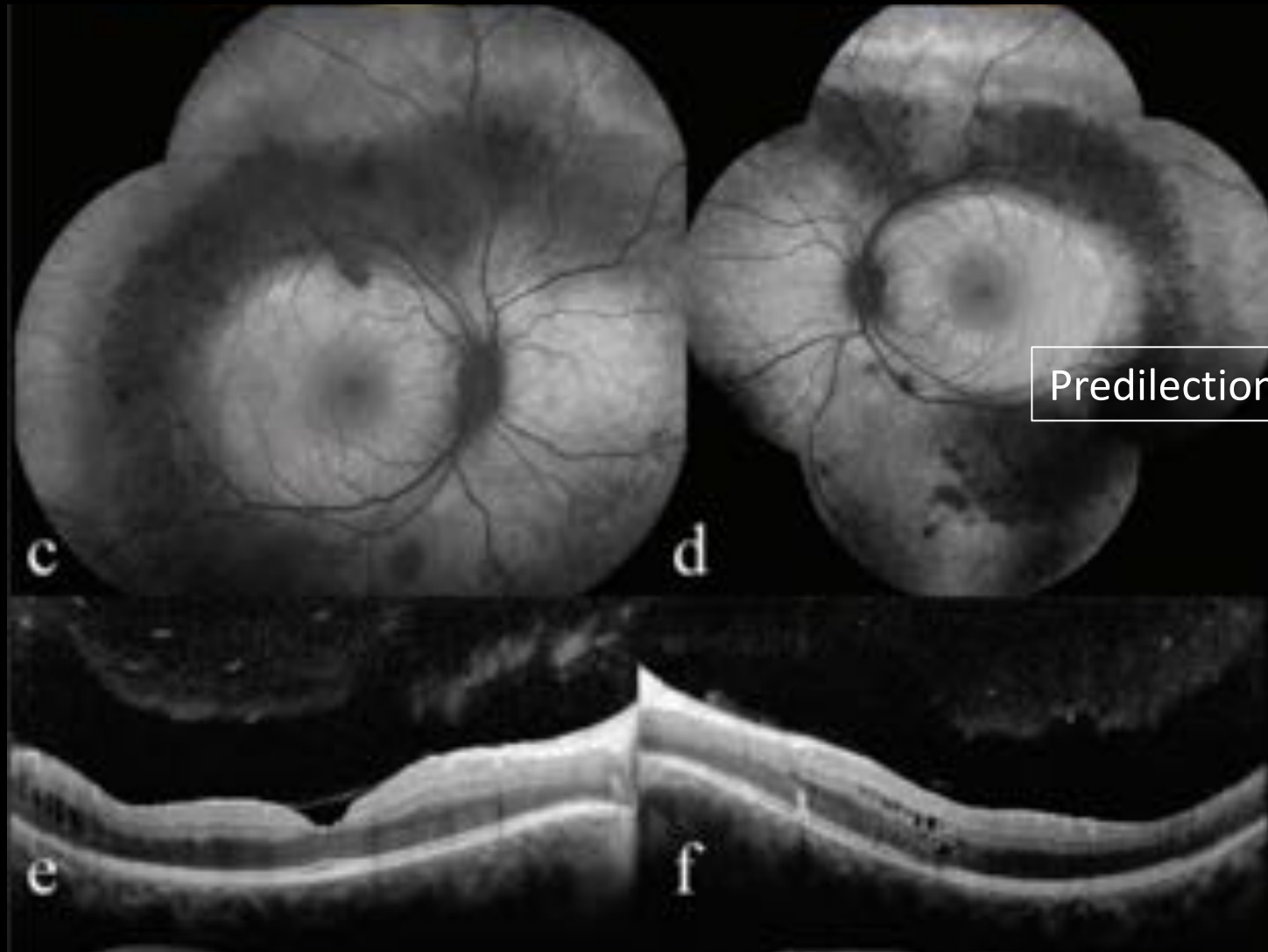
Pigment Epithelium in ESCS

51 year follow-up
(Aged 66)

20/40



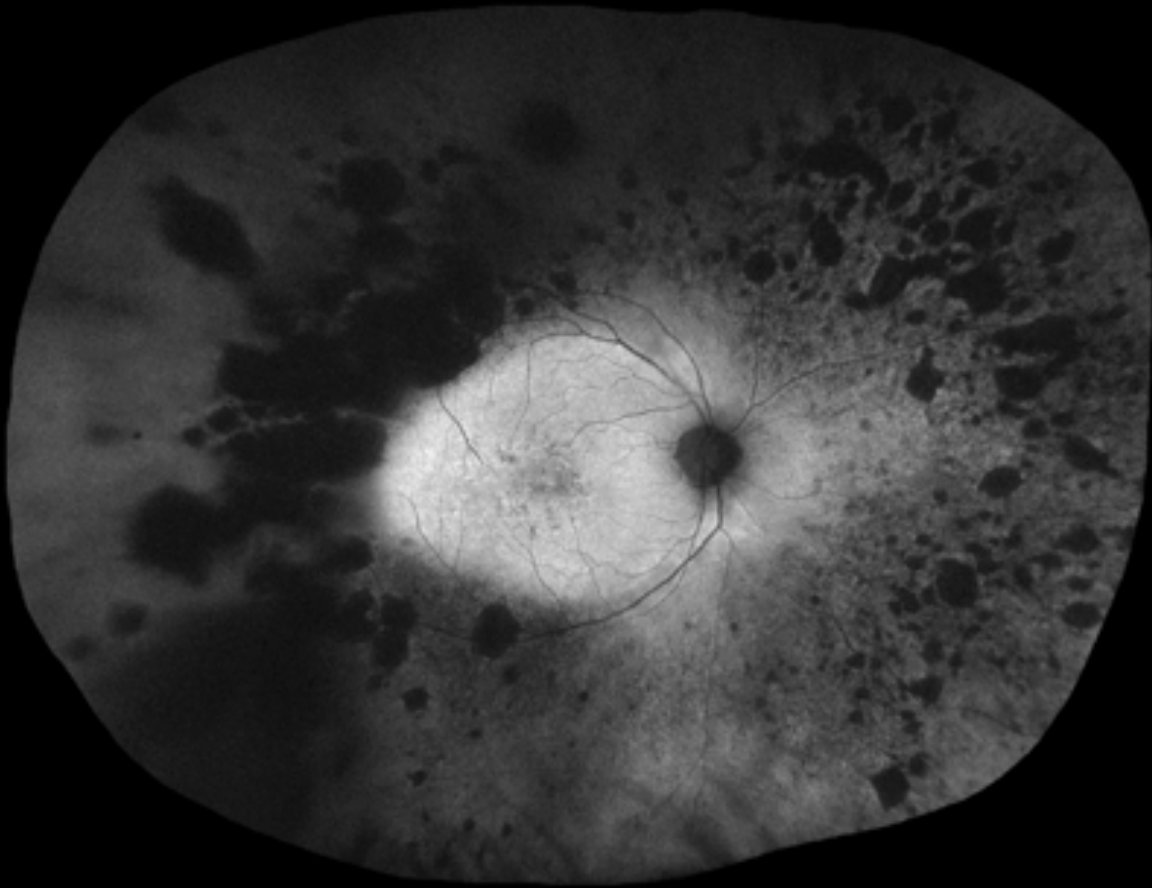
Nummular pigmentary lesions



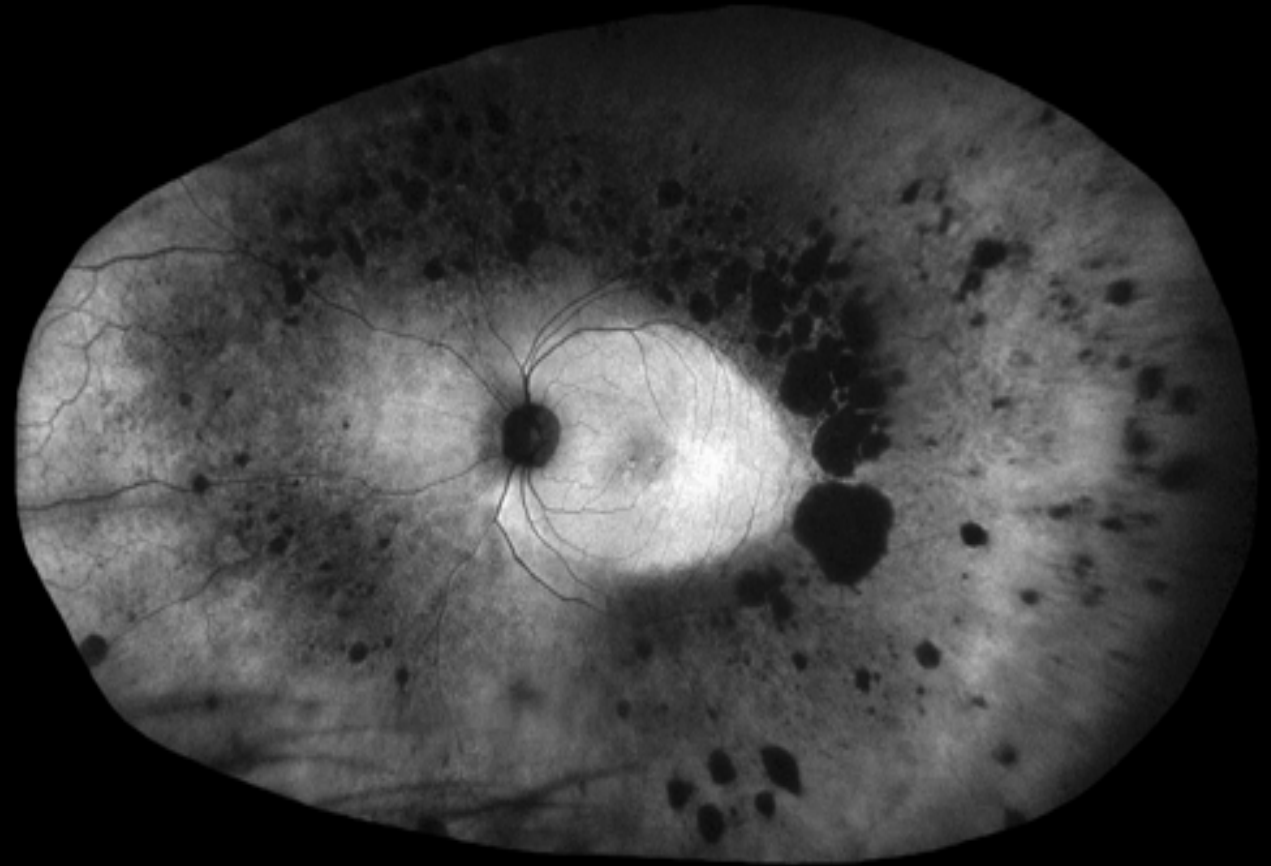
Predilection for superior and temporal

Serdar Özateş

Pigment Epithelium in ESCS



FAF

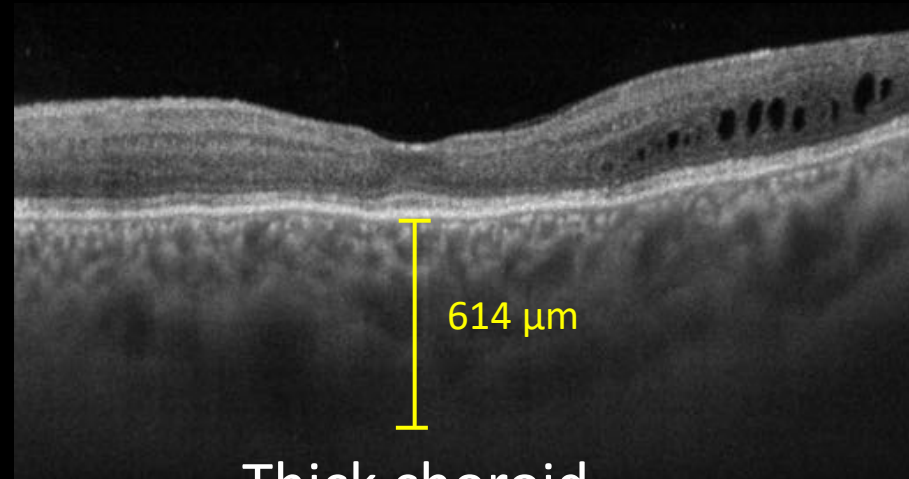
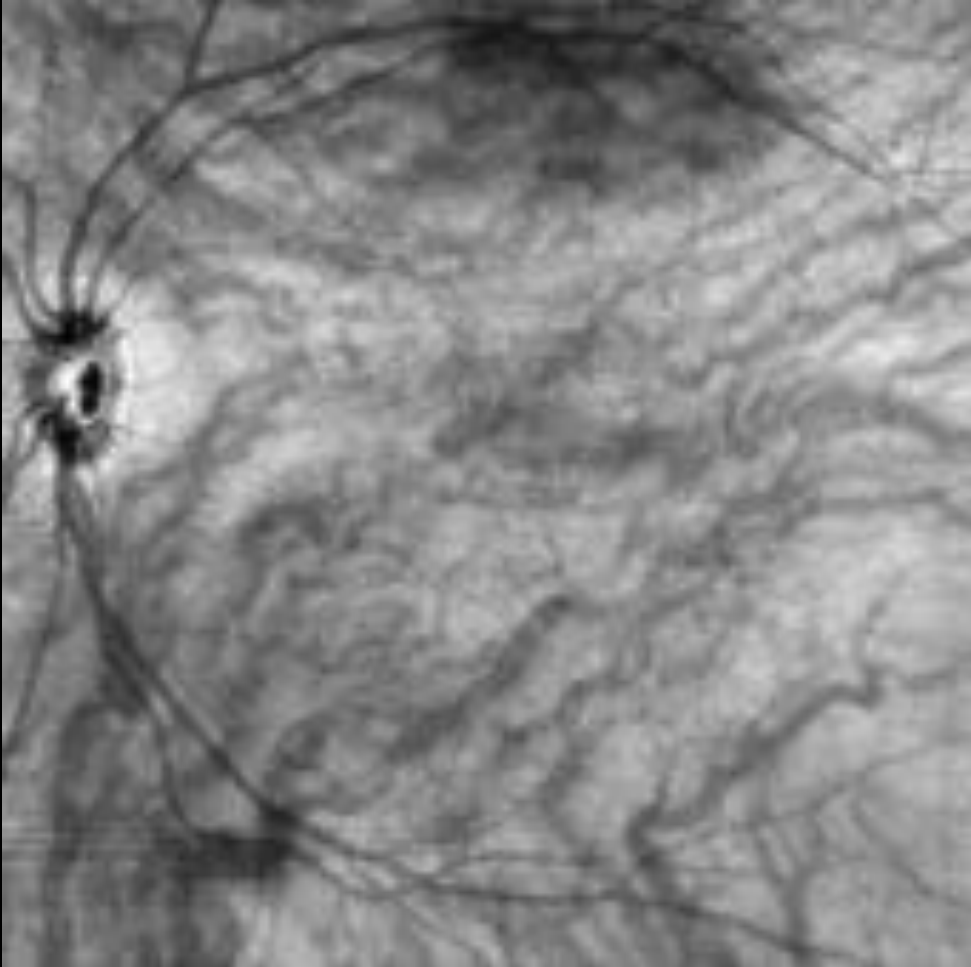


FAF

Hypo FAF numular
circumferential lesions

Yellow-white spots

Choroidal architecture in ESCS

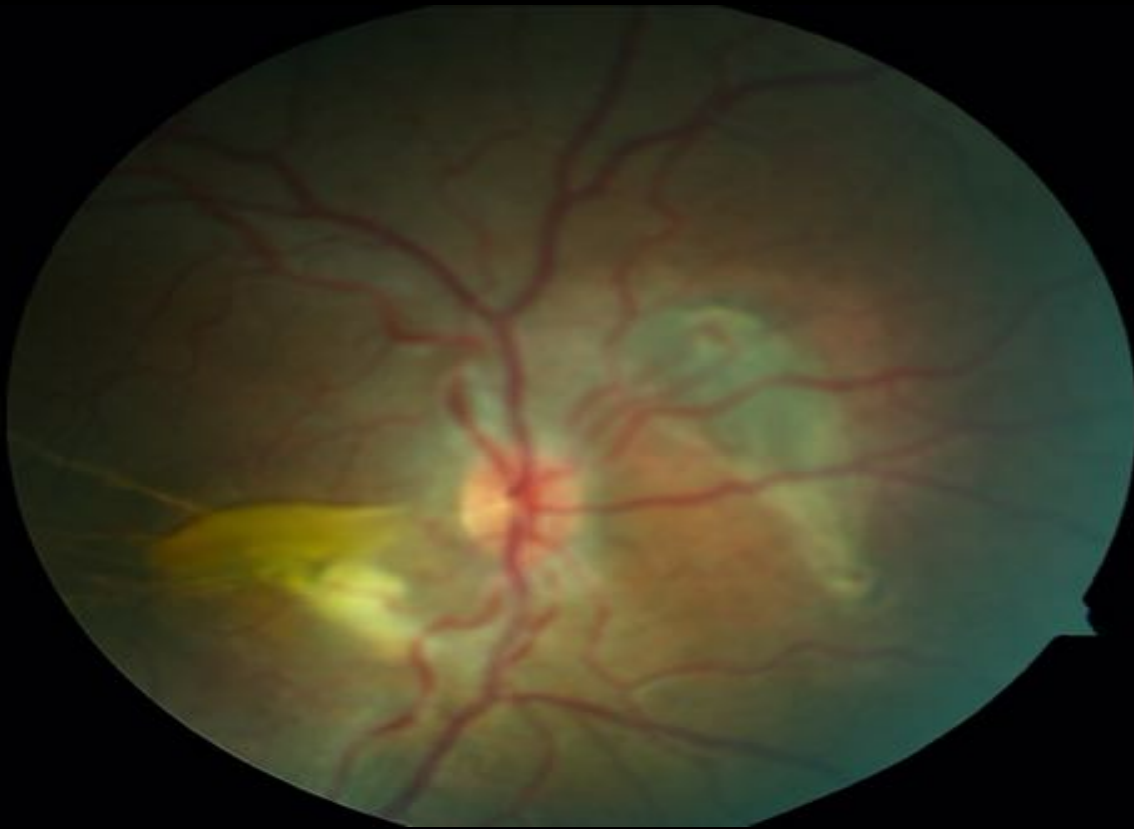


Thick choroid

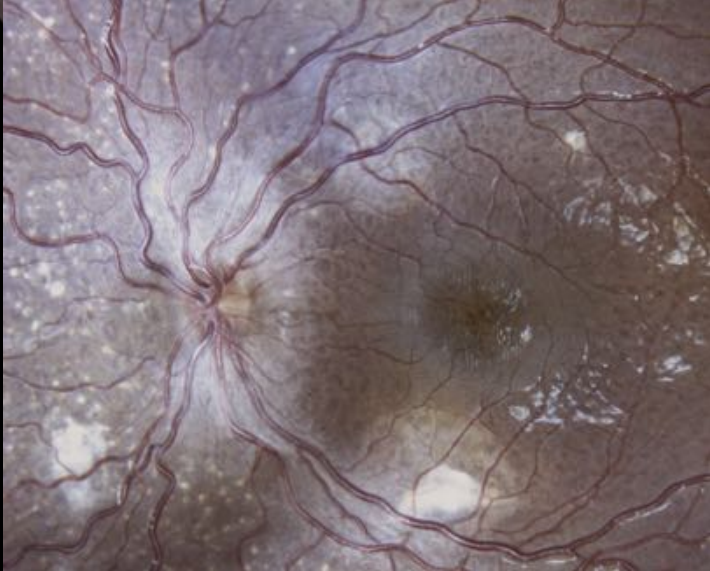
Hyperopia

Nyctalopia + CNV = ESCS

4 year-old boy

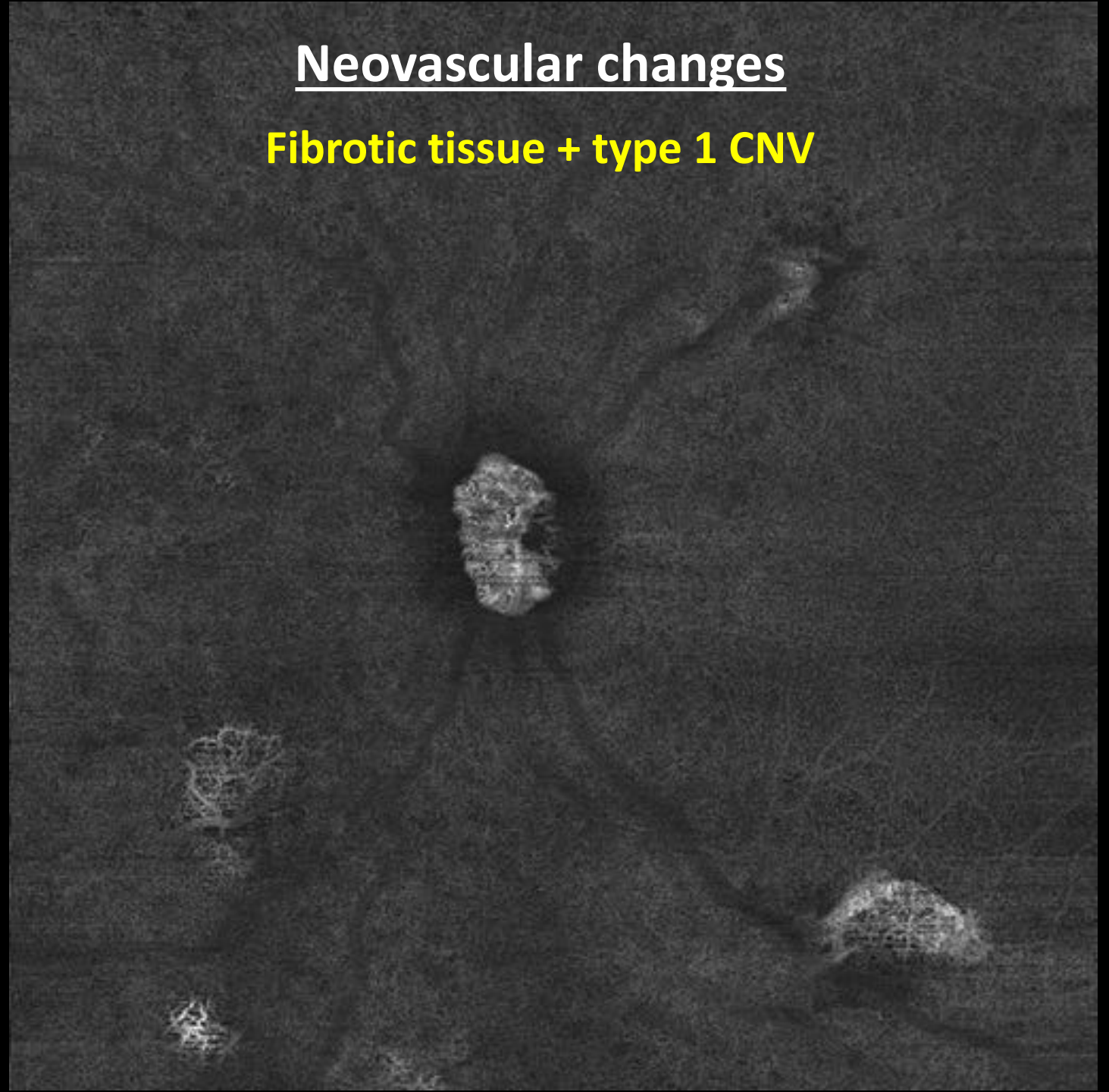


Choroidal neovascularization

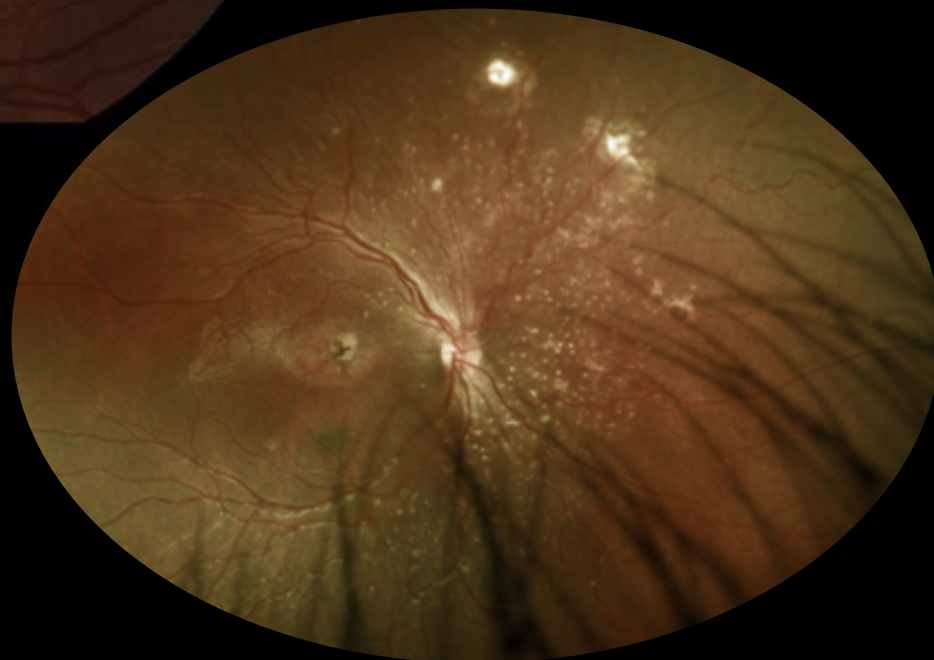
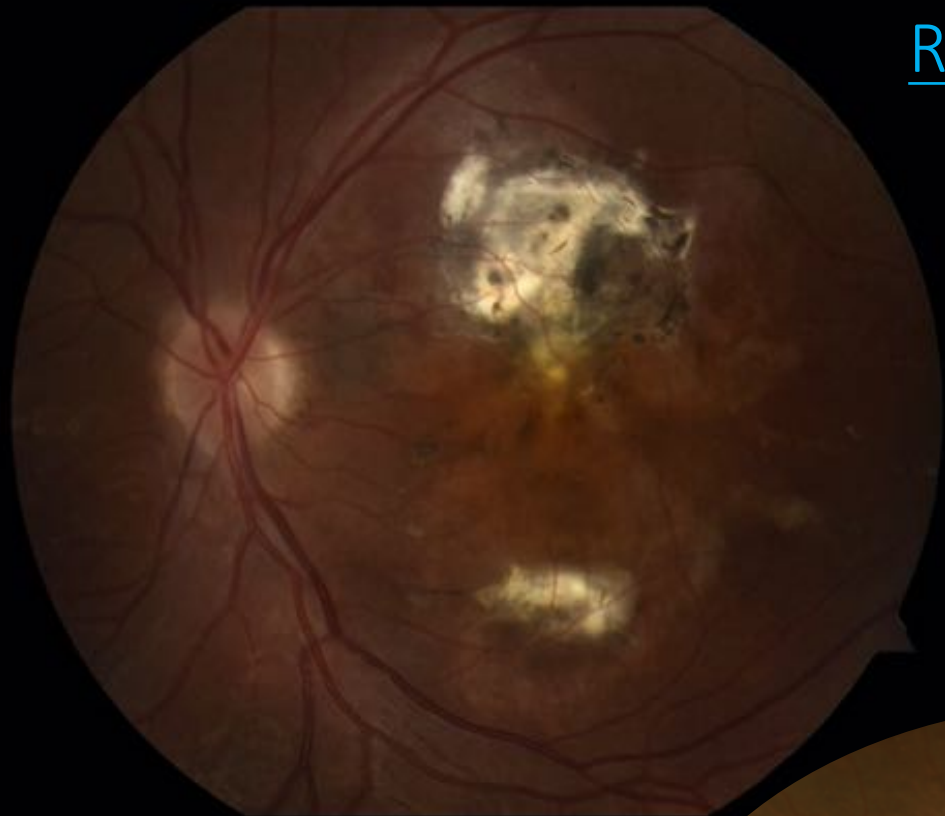


Neovascular changes

Fibrotic tissue + type 1 CNV



Retinal fibrosis in ESCS



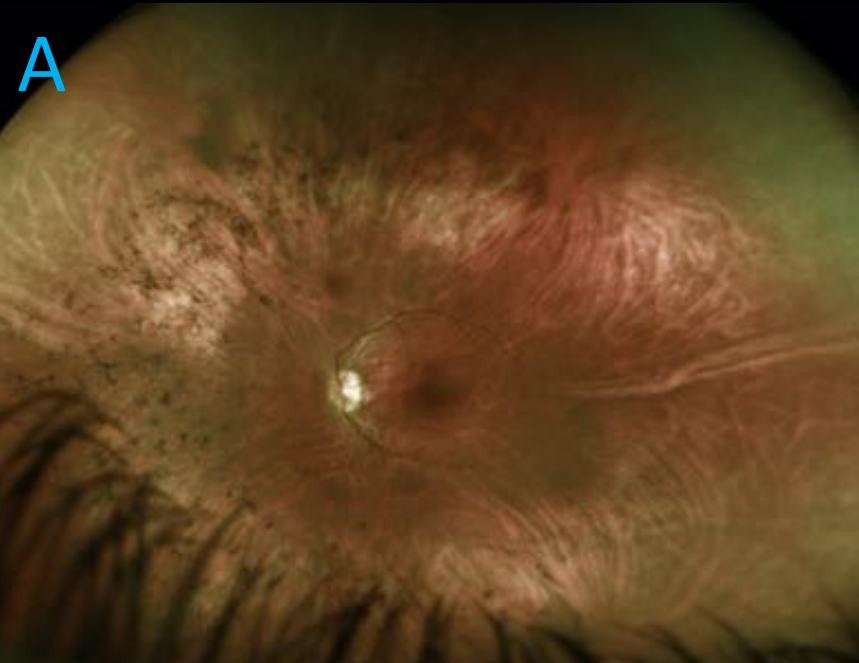


Helicoid fibrosis



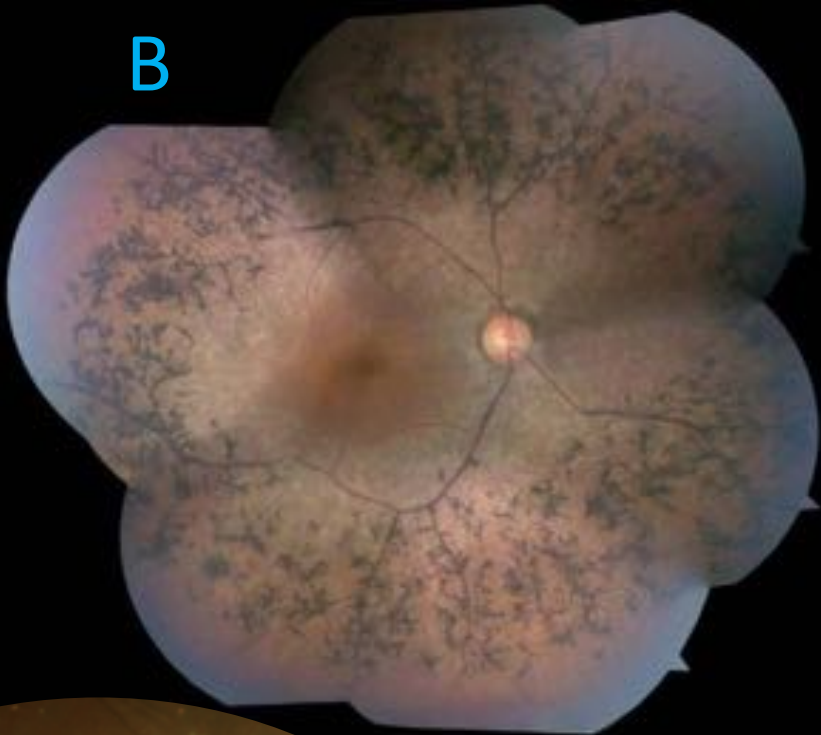
S. Yzer

A



ESCS = NR2E3
Which one?

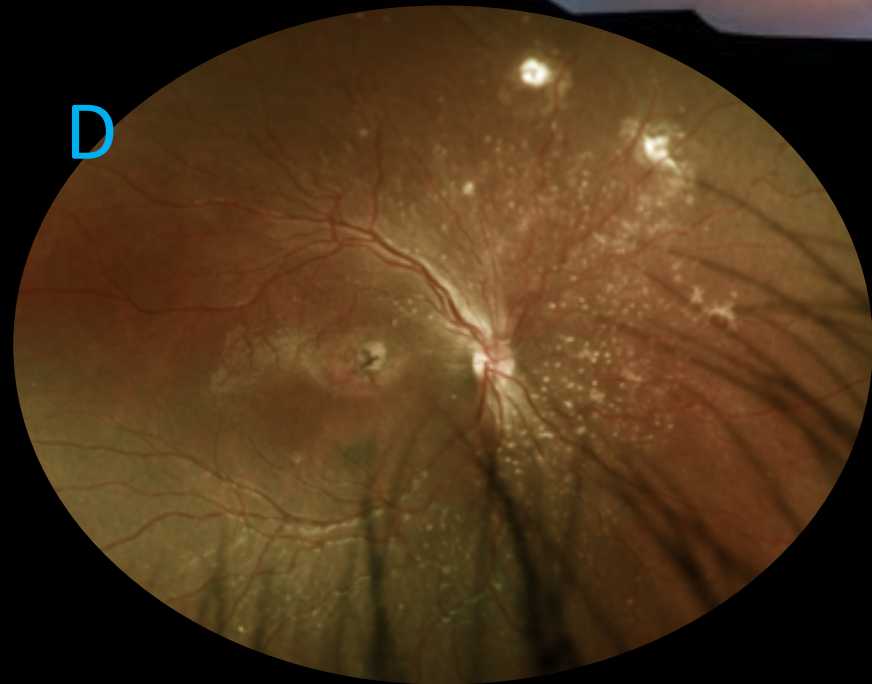
B



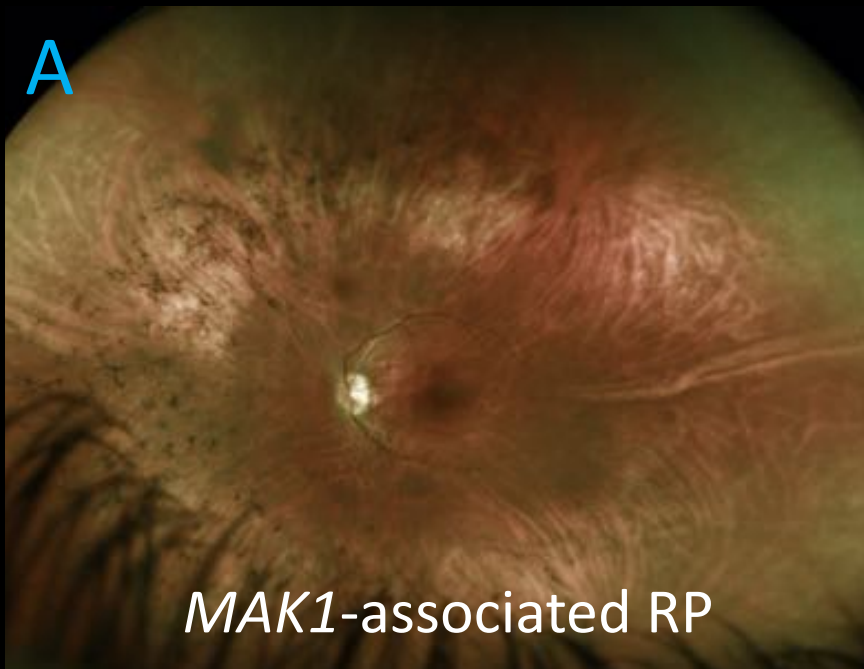
C



D



A

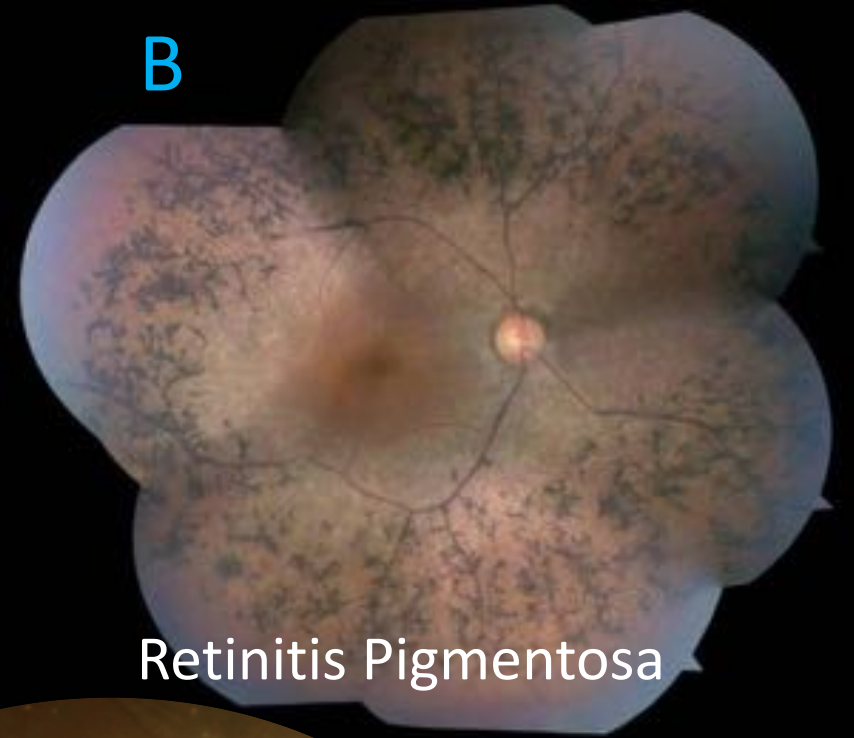


MAK1-associated RP

ESCS

Which one?

B



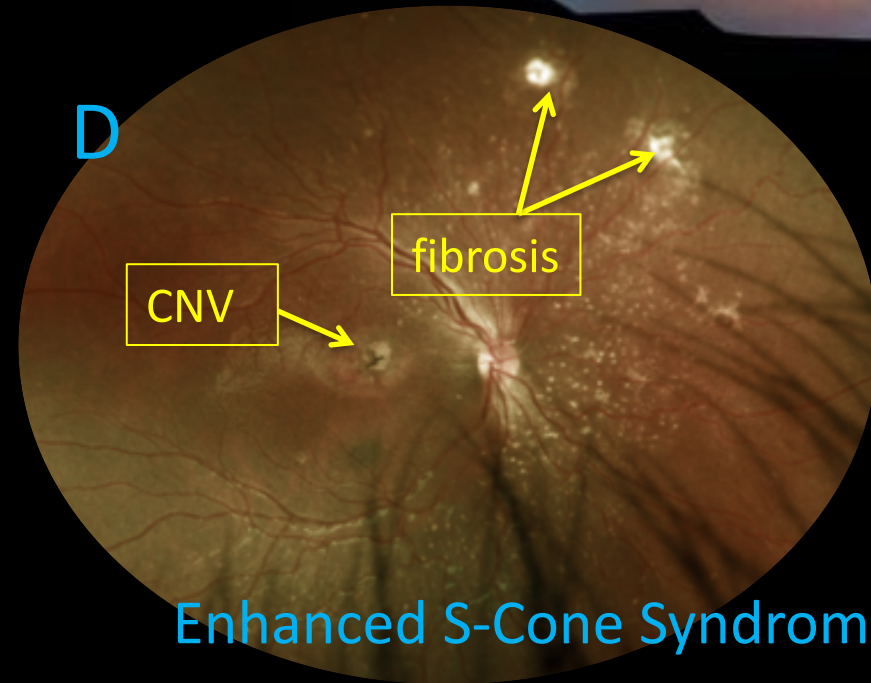
Retinitis Pigmentosa

C



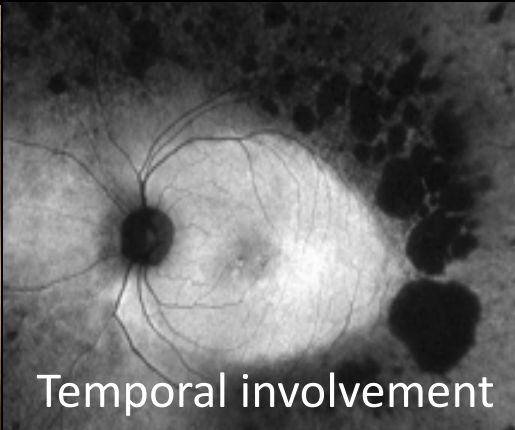
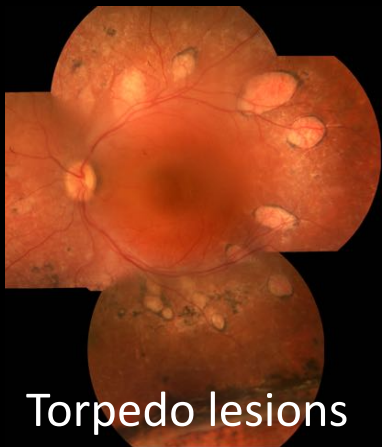
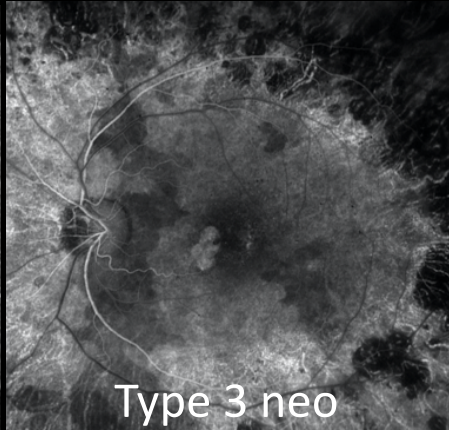
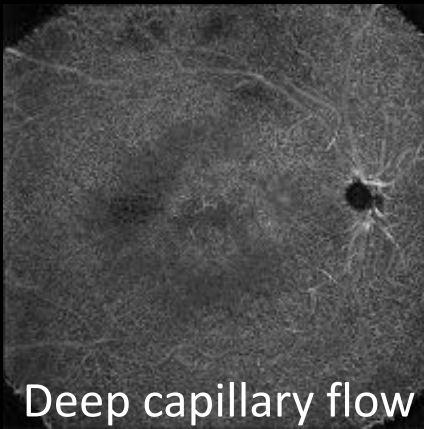
X Linked Retinoschisis

D



Enhanced S-Cone Syndrome

ESCS or NR2E3 is Favre-Goldmann
Summary



The Enhanced S-Cone Syndrome Project



Gerardo Ledesma-Gil



Juliet Essilfie



Mark Breazzano



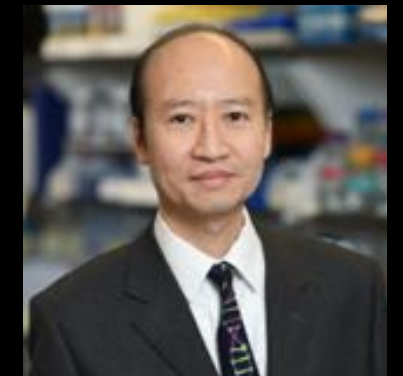
Pedro Fernandez



Bailey Freund



Richard Spaide



Stephen Tsang