ESCS
Expanding the clinical spectrum
Retina Society 2020

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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)
Maurice Favre

Favre Syndrome

1958
Enhanced S Cone Syndrome

Centre of Fovea

Normal
Enhanced S Cone Syndrome (92% blue cones)

Analysis of Blue Cone Sensitivity - ARVO

Peter Gouras

Samuel Jacobson

ESCS

SW5 (Blue) Cone Hypersensitivity in a Newly Identified Retinal Degeneration

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

Connie Cepko
ESCS = NR2E3

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

Early nightblindness
Posterior subcap cataract
Vitreous degeneration
Pigmentary degeneration
ERG changes
Retinal non-perfusion
Diffuse retinal leakage
Phenotypic Features of Patients With NR2E3 Mutations

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
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

*En face* OCT vitreous segment
Peripheral retinoschisis

Opaque dendritic vessels

20/40

Retinal staining with nonperfusion

Red-free image
Peripheral retinoschisis

Opaque dendritic vessels

Retinal vessel staining, non perfusion

Retinal neovascularization
Retina In ESCS

OCT-A

Full retina  Superficial  Deep

REDUCED SIGNAL OR NON-PERFUSION IN DEEP CAPILLARY PLEXUS
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 Oubrahim, MD,† Anne Sikorav, MD,† Isabelle Audo, MD,§
Josseline Kaplan, MD,‡ Jean-Michel Rozet, PhD,‡ Eric H. Souied, MD, PhD*
Pigment Epithelium in ESCS

51 year follow-up
(Aged 66)

20/40

Numular pigmentary lesions
Predilection for superior and temporal
Pigment Epithelium in ESCS

Hyper FAF radial spikes
Hypo FAF segmental pattern
Hypo FAF numular circumferential lesions
Yellow-white spots

FAF
Choroidal architecture in ESCS

Thick choroid

Hyperopia

614 µm
4 year-old boy

Nyctalopia + CNV = ESCS

Choroidal neovascularization

S. Tsang
Neovascular changes

Fibrotic tissue + type 1 CNV
Retinal fibrosis in ESCS
Helicoid fibrosis

S. Yzer
ESCS = NR2E3
Which one?
ESCS
Which one?

A. MAK1-associated RP

B. Retinitis Pigmentosa

C. X Linked Retinoschisis

D. Enhanced S-Cone Syndrome

CNV
fibrosis
ESCS or NR2E3 is Favre-Goldmann

Summary