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)
Favre Syndrome

Maurice Favre

1958
Enhanced S Cone Syndrome

Centre of Fovea

Normal
Enhanced S Cone Syndrome
(92% blue cones)

Analysis of Blue Cone Sensitivity
- ARVO

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

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

Peter Gouras

Samuel Jacobson

Connie Cepko
*ESCS* = *NR2E3*

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

Alexandra V. Garafalo, Giacomo Calzetti, Artur V. Gideciyan, Alejandro J. Roman, Supra Saxena, Alexander Sumaroka, Windy Choi, Alan E. Wright, and Samuel G. Jacobson

37 mutations
Mostly compound heterozygous
Fewer than 2% no detectable allele
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
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
Torpedo-like lesions

- Nyctalopia at early age
- PSC
- CME
- Peripheral retinoschisis
- Peripheral retinal ischemia
Vitreous In ESCS

Thick posterior hyaloid

Fibrillar changes
SQUARE or polygonal vitreomacular adhesion + thick posterior hyaloid
Retina In ESCS

6 yr, NR2E3 (R311Q)

32 yr. Patient

Yellow-white spots

S. Tsang
Retina In ESCS

- Peripheral retinoschisis
- Opaque dendritic vessels
- Retinal staining with nonperfusion
Peripheral retinoschisis

Retinal vessel staining, non perfusion

Opaque dendritic vessels

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 Oubraham, MD, Anne Sikorav, MD, Isabelle Audo, MD
Josseline Kaplan, MD, Jean-Michel Rozet, PhD, Eric H. Souied, MD, PhD

10 year-old boy
Pigment Epithelium in ESCS

51 year follow-up
(Aged 66)

20/40

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

FAF

Hypo FAF numular circumferential lesions

Yellow-white spots
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

20/30

20/20
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

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Vitreous Retinal neo Deep capillary flow Type 3 neo

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Torpedo lesions Nummular lesions Temporal involvement Fibrosis
The Enhanced S-Cone Syndrome Project

Gerardo Ledesma-Gil

Juliet Essilfie

Mark Breazzano

Bailey Freund

Richard Spaide

Stephen Tsang

Pedro Fernandez