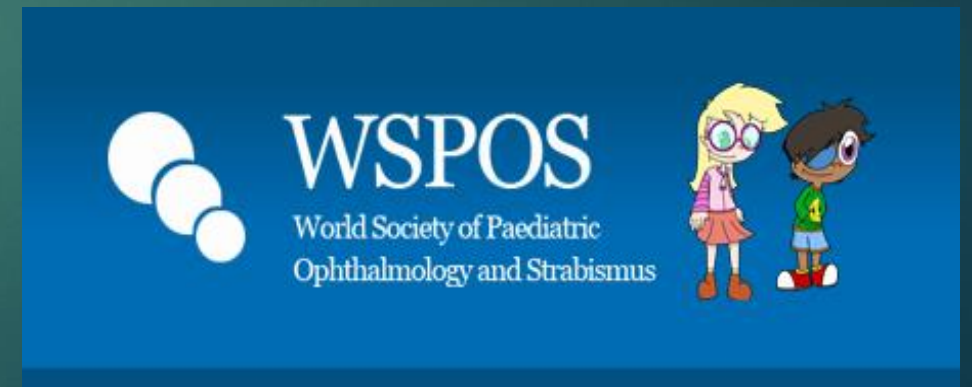


Case 11

Global Case Report Quiz

THALES A. C. DE GUIMARÃES, MD
ALEX V. LEVIN, MD, MHSc, FRCSC

WILLS EYE HOSPITAL, THOMAS JEFFERSON UNIVERSITY - PHILADELPHIA, USA

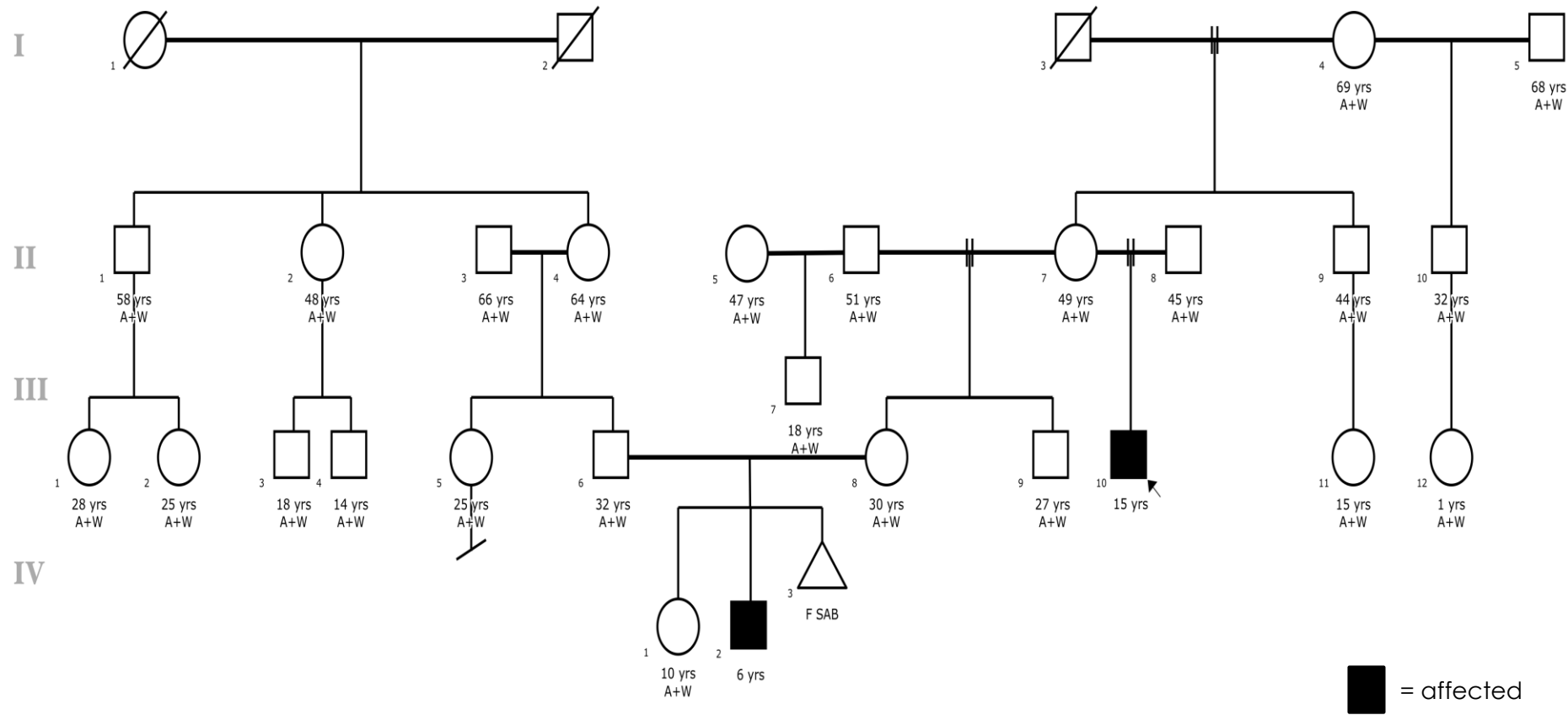


Case Report

- ▶ 15 year old male referred with subnormal best corrected visual acuity. No other symptoms.
- ▶ Review of systems: Unremarkable.
- ▶ Past ocular history: Started feeling vision was getting worse at 8 years old, when he was diagnosed with inferotemporal retinoschisis. Has been seeing his local ophthalmologist every 6 months since then and is reportedly stable.
- ▶ Family history: His maternal nephew has similar findings.

Family pedigree

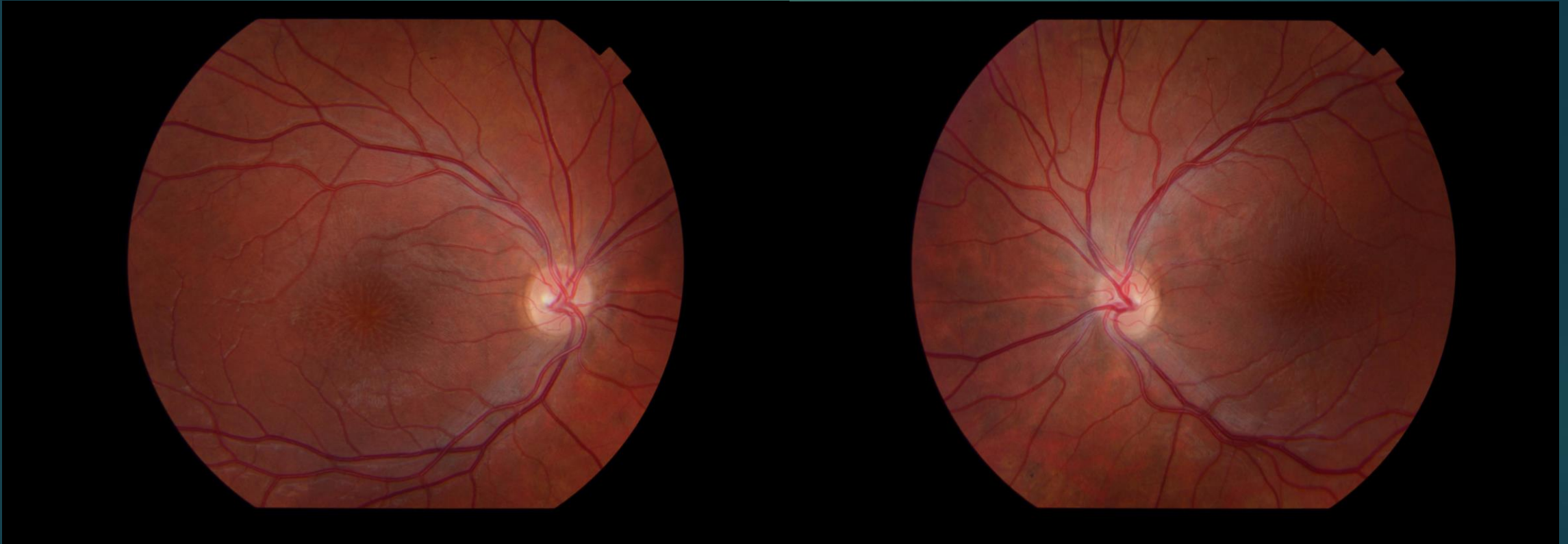
Pedigree



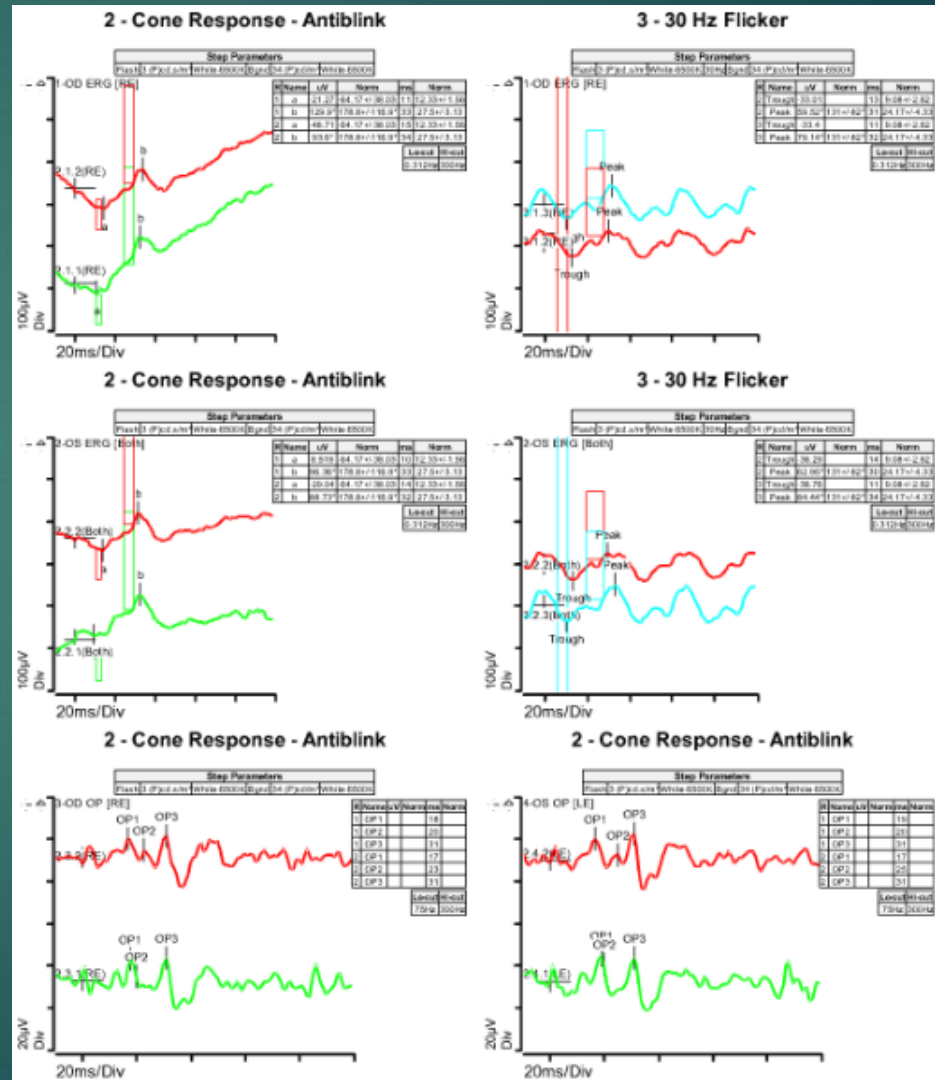
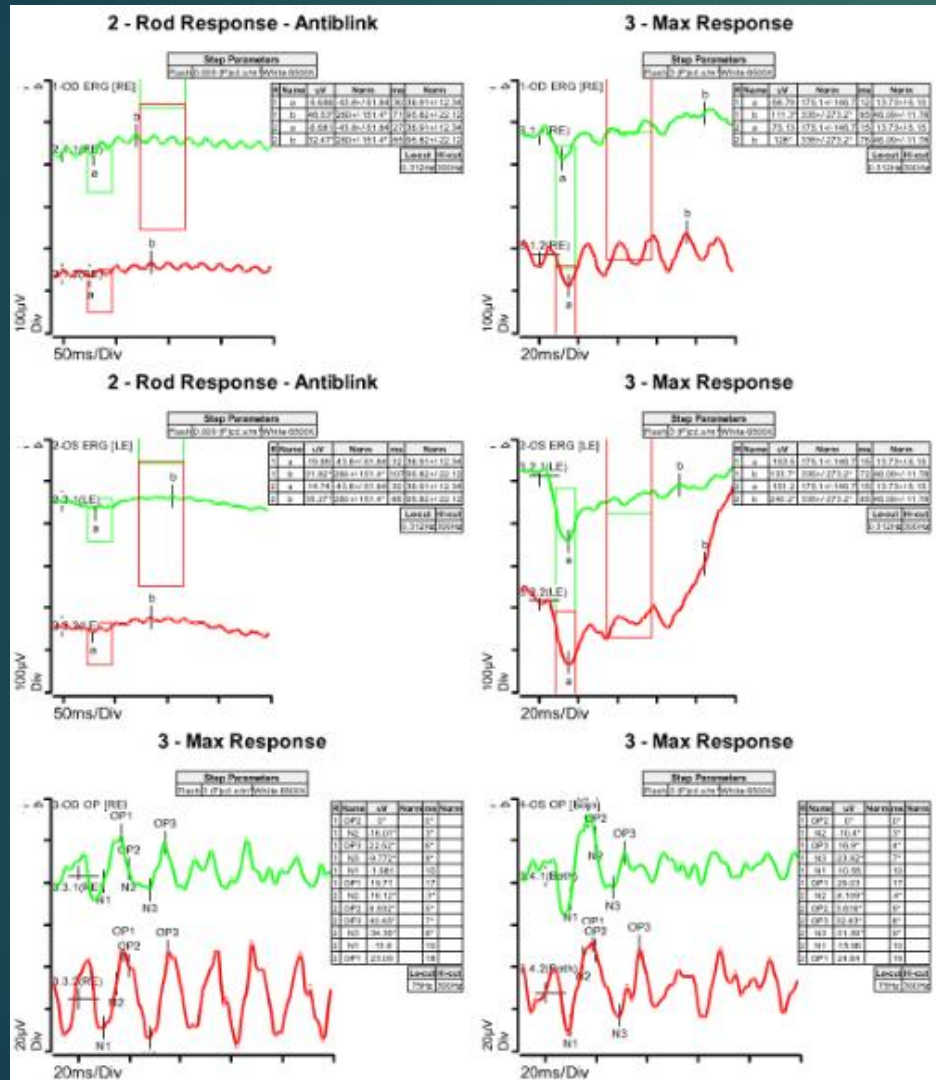
Physical exam

- ▶ Slit lamp examination normal. Pupils normal with no APD, eye movements full with no strabismus or nystagmus. Visual fields full to confrontation. Normal intraocular pressure.
- ▶ Best corrected acuity: 20/40 OD and 20/50 OS.
- ▶ Cycloplegic refraction:
 - 0.25 +1.00 axis 84 OD
 - 0.50 +1.50 axis 104 OS

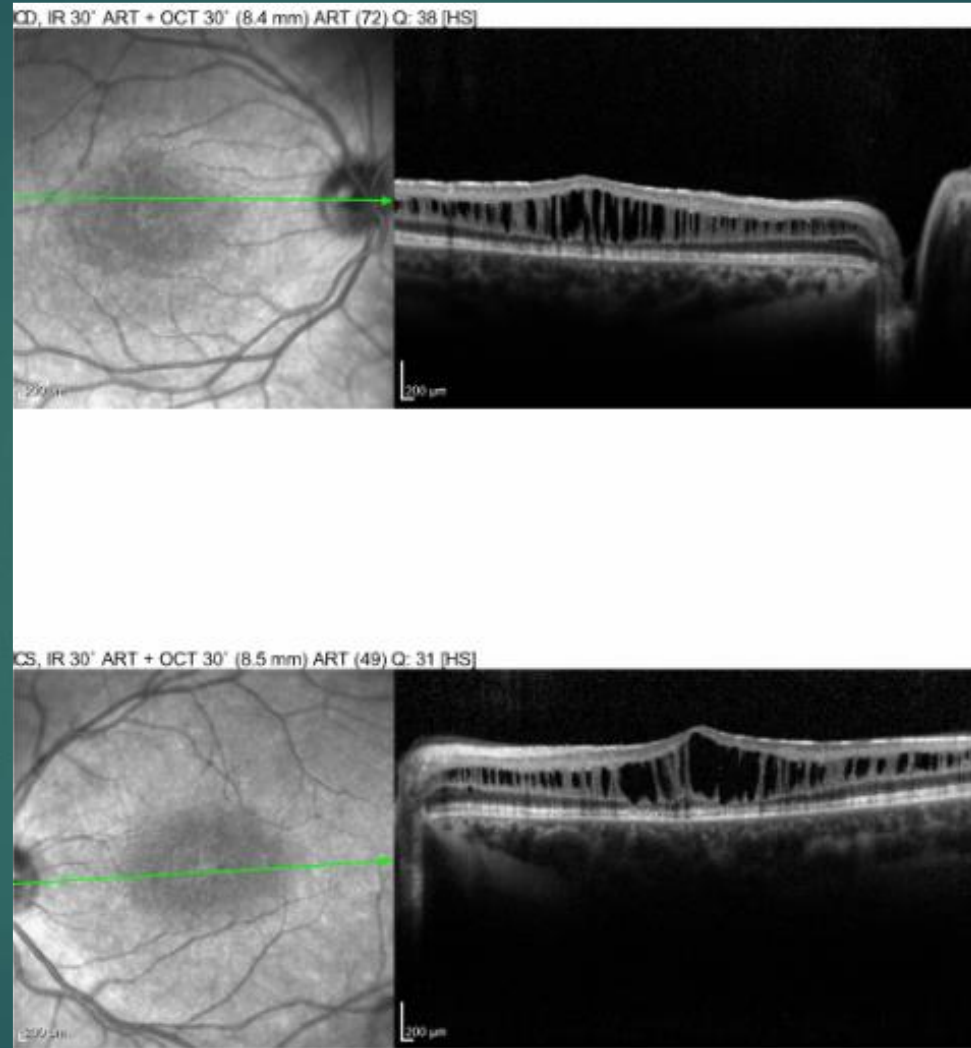
Fundus examination: Radial spoke-wheel pattern of inner retina, infero-temporal far periphery schisis with intact vessels on surface (not shown below). Normal optic nerves. No pigmentary changes.



ERG: Abnormal full-field ERG with signs of both rod and cone dysfunction. There is an electronegative B-wave.



Spectral domain OCT



Diagnosis

- ▶ Based on ocular examination, diagnostic testing and family history, the diagnosis of Juvenile X-linked Retinoschisis (JXLR) was made.

What should be done next?

▶ Which is the gene expected to be mutated in a patient with JXLR?

- a) *PRPH2/RDS*
- b) *ABCA4*
- c) *RS1*
- d) *CRB1*
- e) *USH2A*

▶ Is this patient at risk for retinal detachment?

- () Yes
- () No

▶ Which treatment would you consider starting for this patient?

- a) Intravitreal bevacizumab injection
- b) Intravitreal triamcinolone
- c) Oral corticosteroids
- d) Carbonic anhydrase inhibitors
- e) Non steroidal anti-inflammatory drops

Could our experts answer these questions?

- ▶ What would be your work-up for intraretinal cystoid spaces?
- ▶ What is the role of carbonic anhydrase inhibitors in JXLR?
- ▶ What would you tell the family about the long-term prognosis?
- ▶ Is there any ongoing research that could potentially help patients with JXLR?

Thank you!

