

High Myopia associated retinal detachments in children

Focus on genetics



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ROYAL MAXIM PALACE KEMPINSKI, CAIRO

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Differential diagnosis of paediatric RRD

- **Trauma**
 - **GRT, Dialysis, Contusion breaks**

- **Genetic: Connective Tissue**
 - **Marfans: Fibrillin**
 - **Sticklers: COL2A1(75%), COL11A1(15%)**
 - **Wagners: VCAN**
 - **Knoblochs: COL18A1**

- **Genetic: RPE: 1993: UK Manchester**
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Is the retina pathognomonic?

■ Trauma

- ~~GRT, Dialysis, Contusion breaks~~

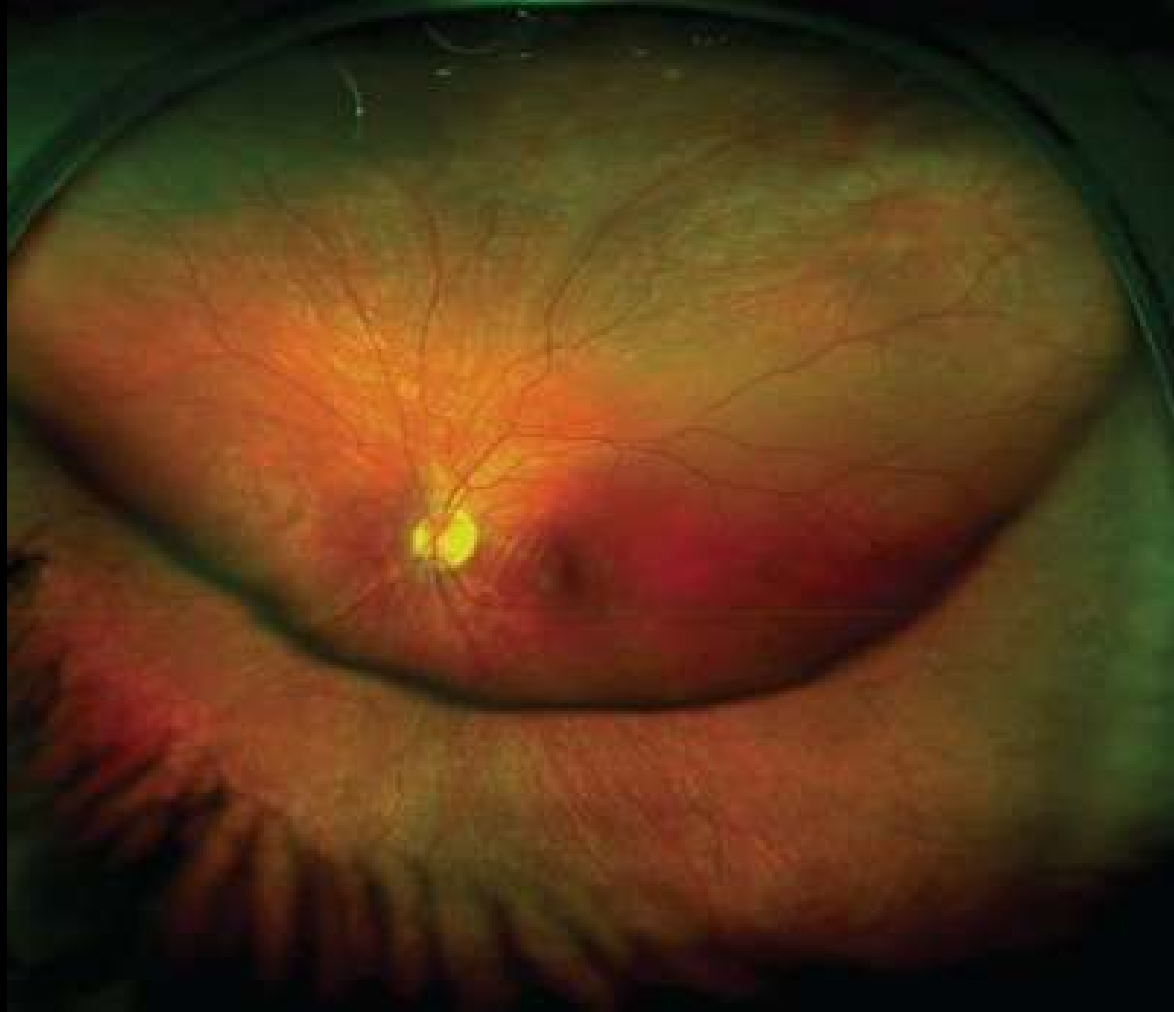
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Genetic diagnosis of RRD: fellow eye ?



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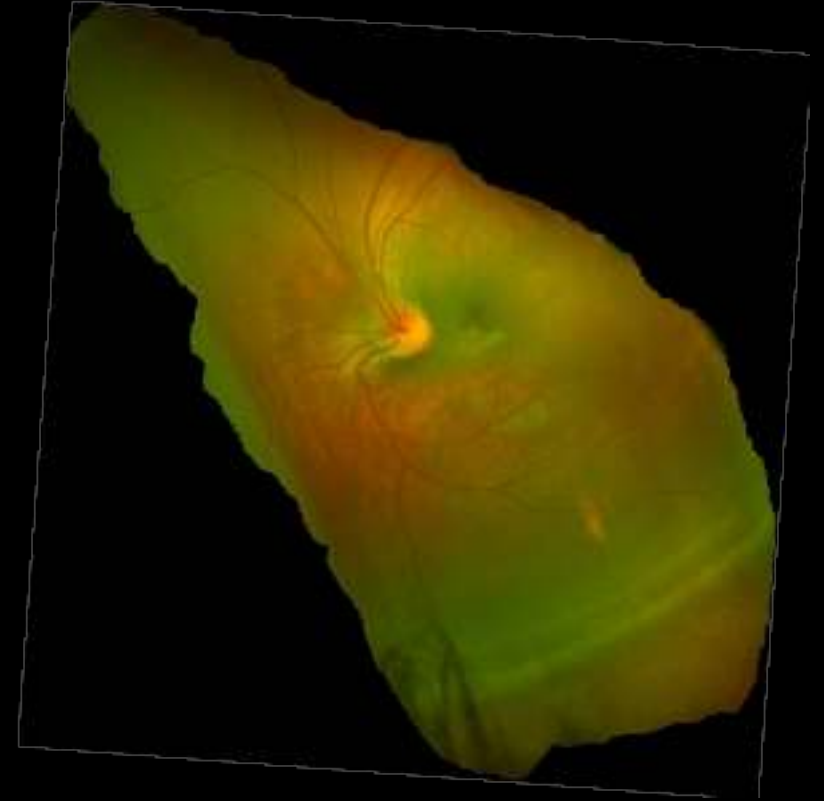
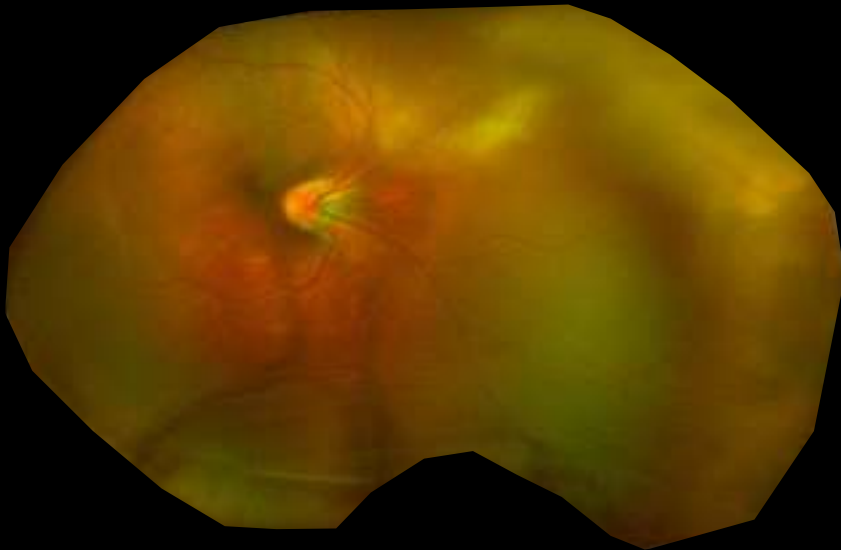
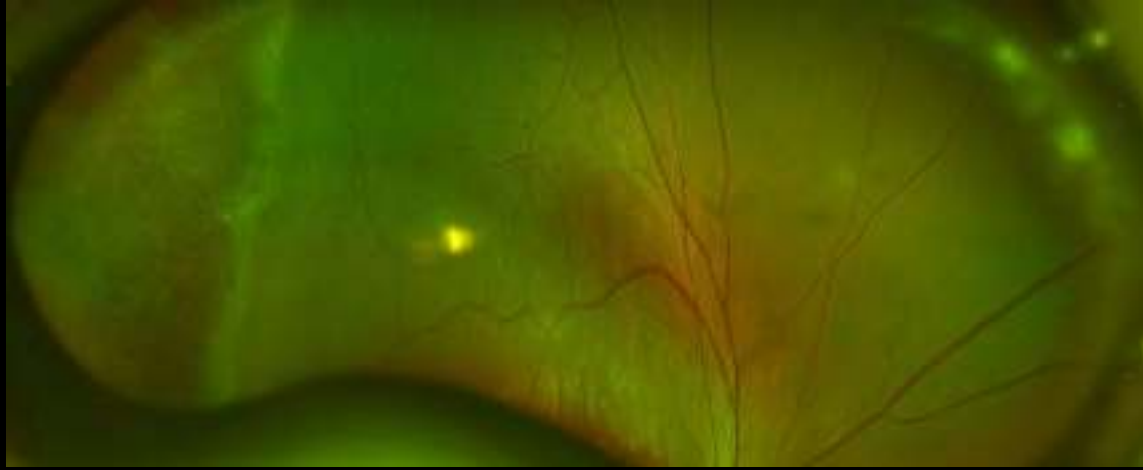
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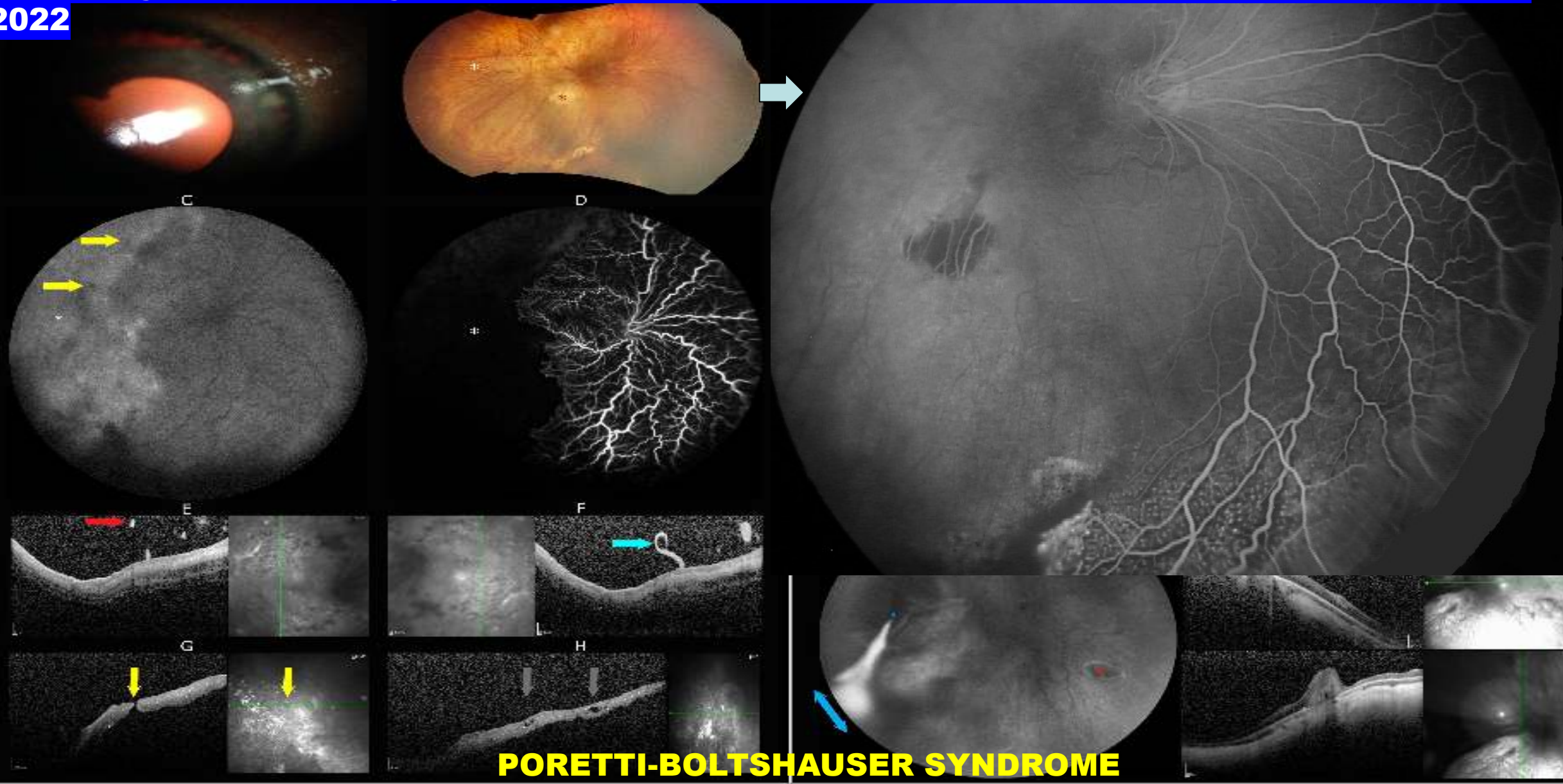
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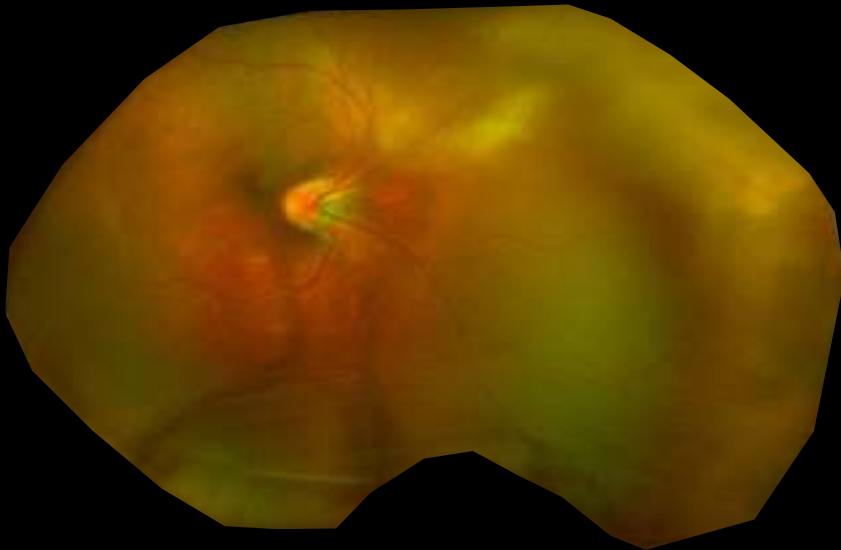
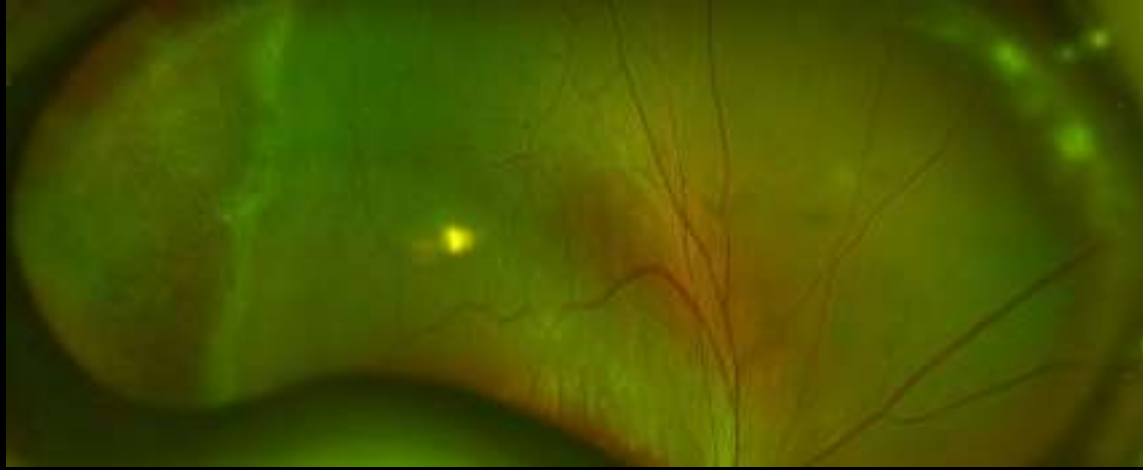
Is this eye at risk of genetic based Paed RRD ?



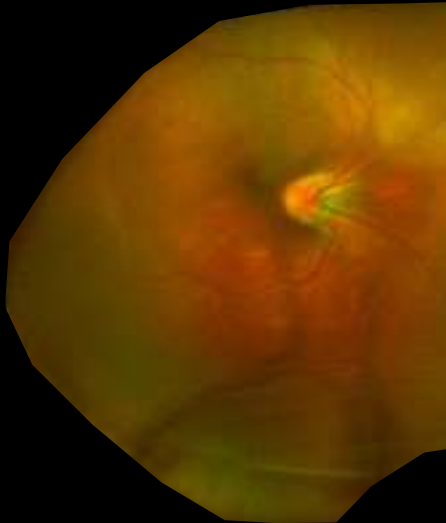
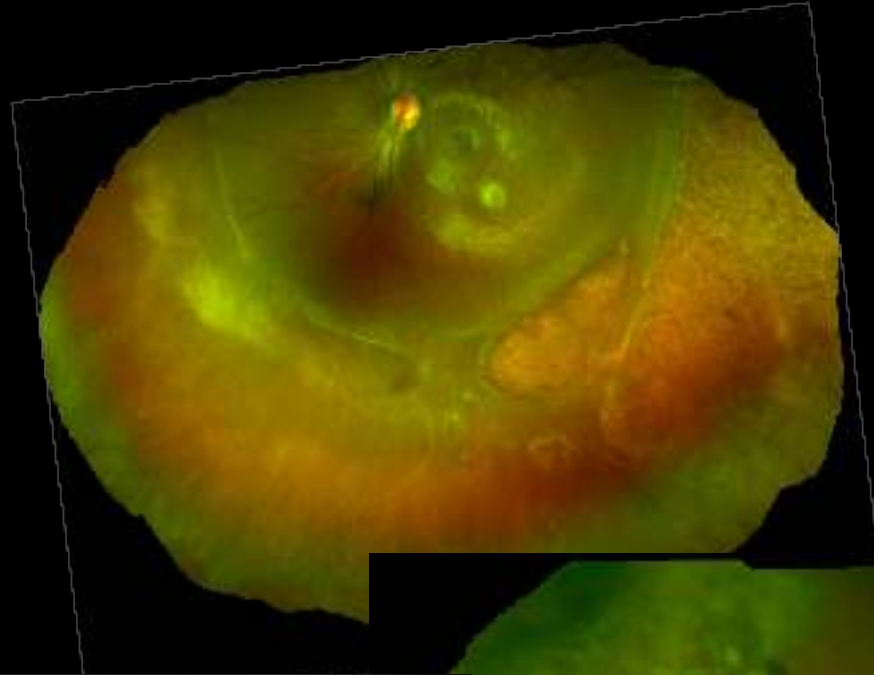
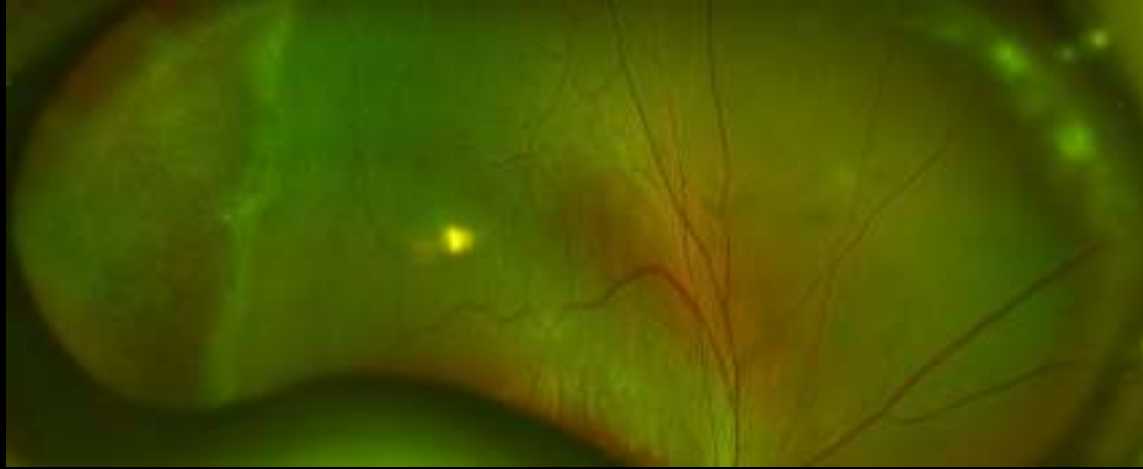
Variability of retinopathy consequent upon novel mutations in LAMA1: Ophthalmic Genetics 2022



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Is this eye at risk of genetic based Paed RRD ?



Wagner disease

Invariable:

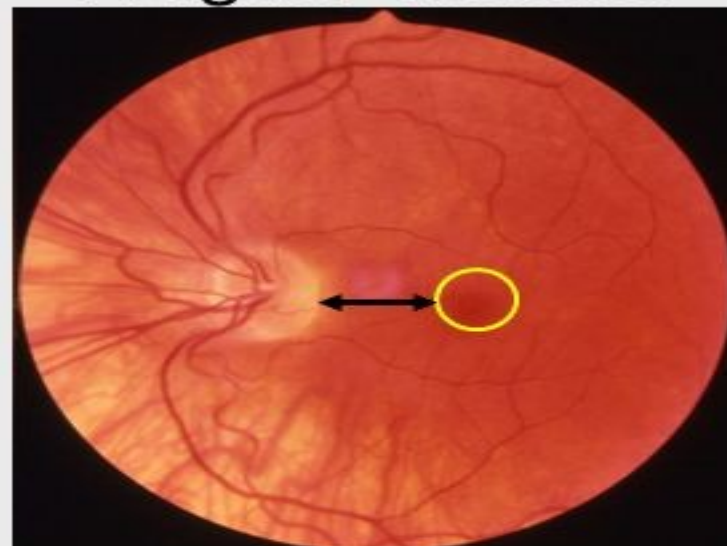
- “empty” vitreous
- preretinal membrane

Variable:

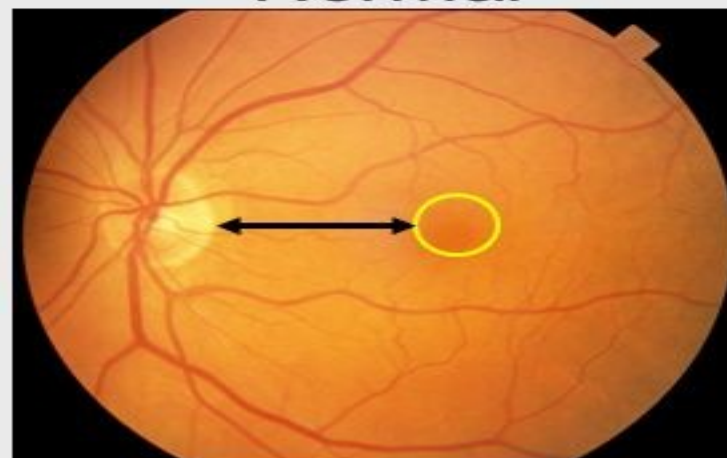
- abnormal vessel architecture
- fovea shifted
- cortical cataracts
- chorioretinal atrophy
- retinal detachment



Wagner disease



Normal



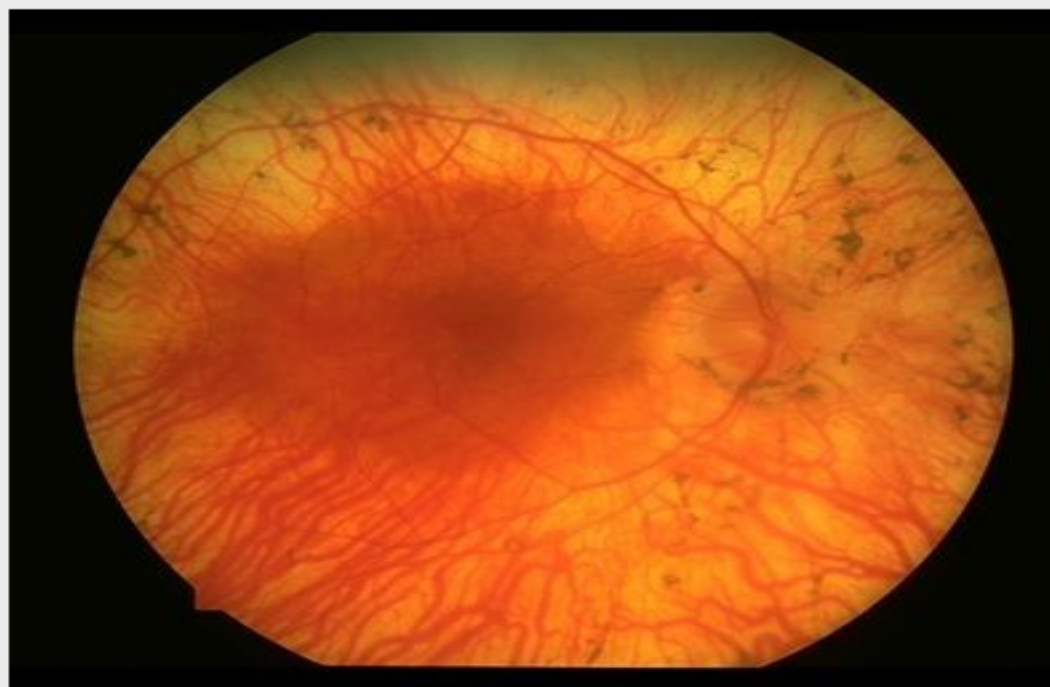
Erosive vitreoretinopathy

Invariably:

- “empty” vitreous
- night blindness
- RPE erosion (progressive)
- visual field loss

Variable:

- pigmentations
- retinal detachment



Courtesy: C.E. van Nouhuys

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Genetic diagnosis of RRD: fellow eye ?



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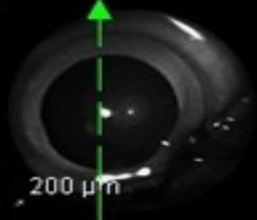
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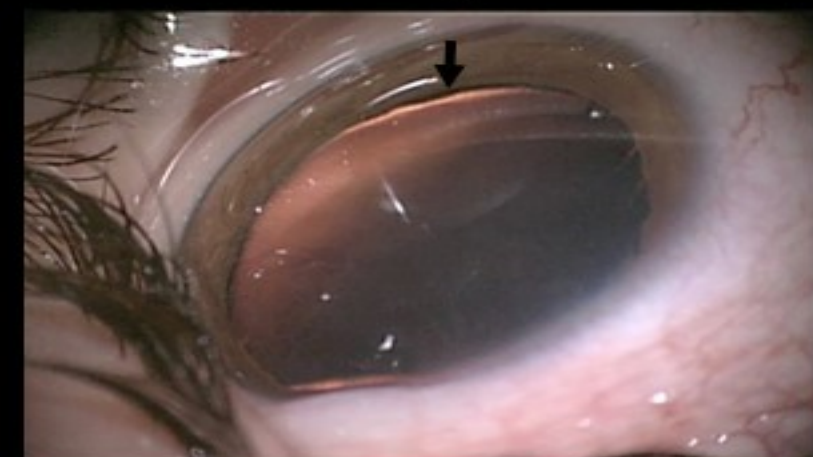
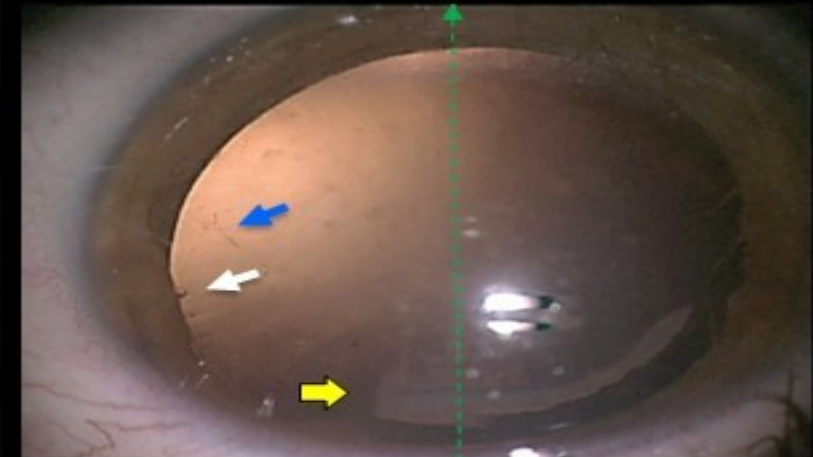
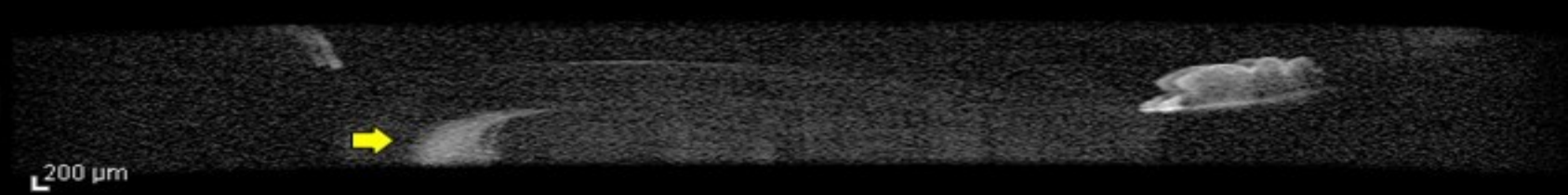
- **?**

Child with Nystagmus & RRD in fellow eye: Genetic diagnosis please?



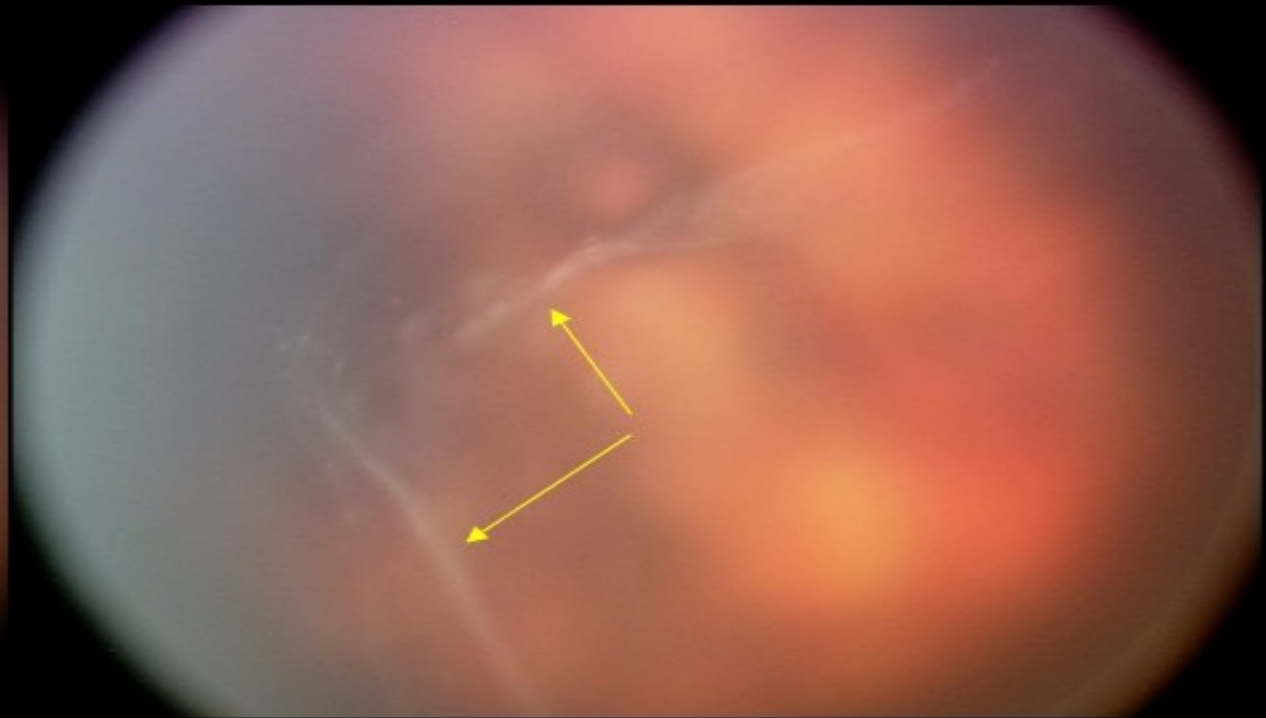
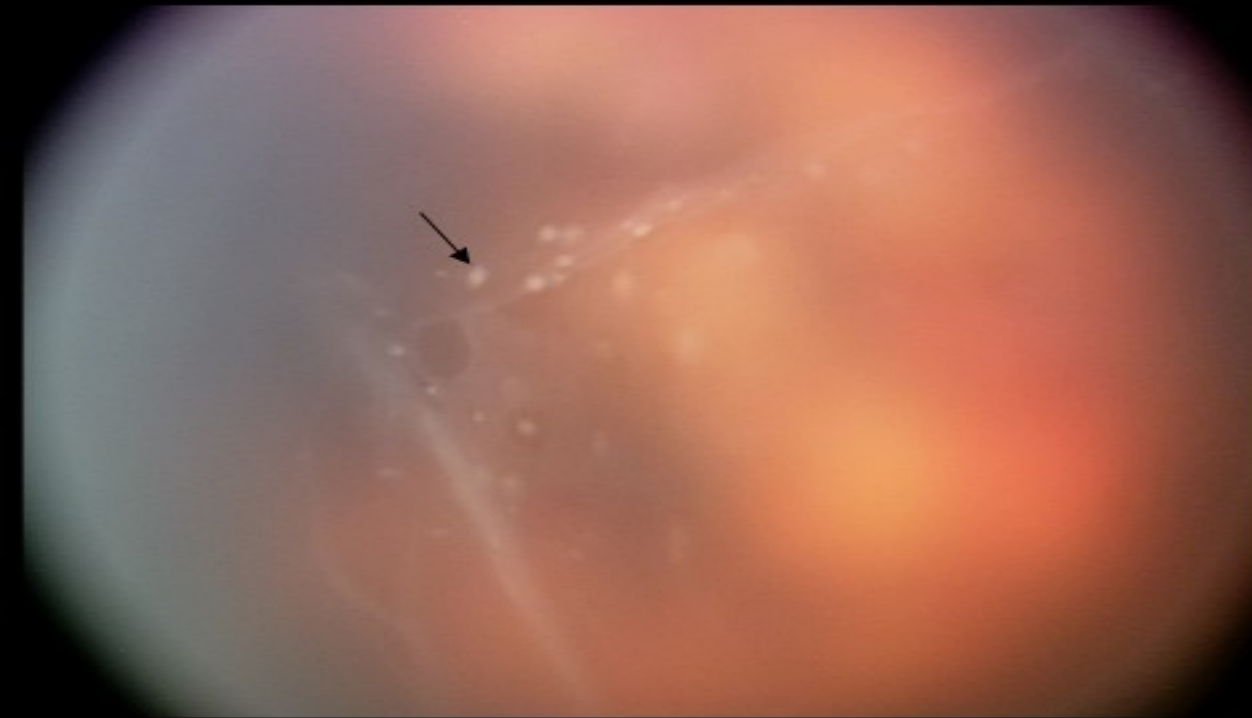


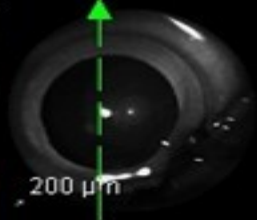
stevens-rowe, jay, 6/20/2004, #10152108



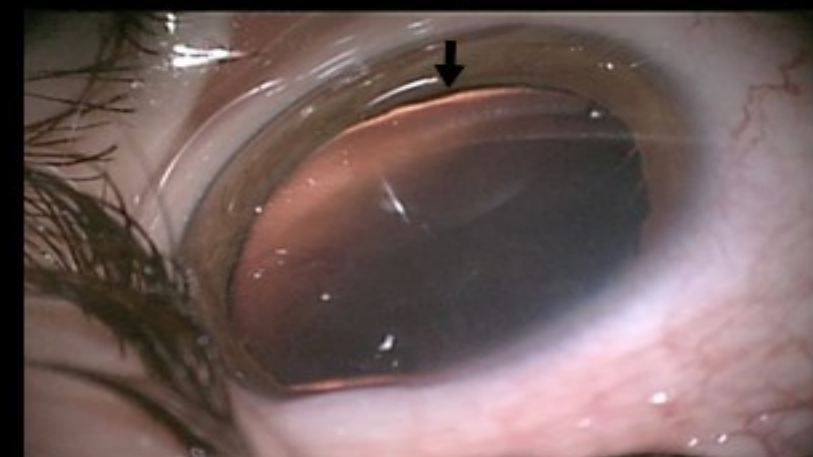
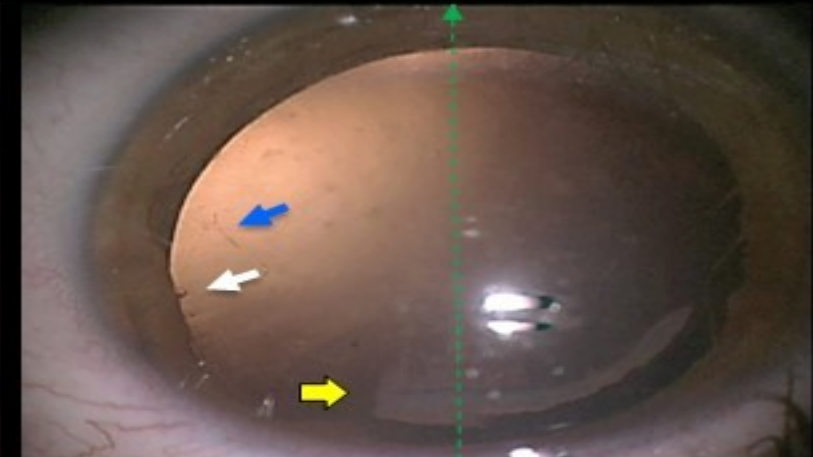
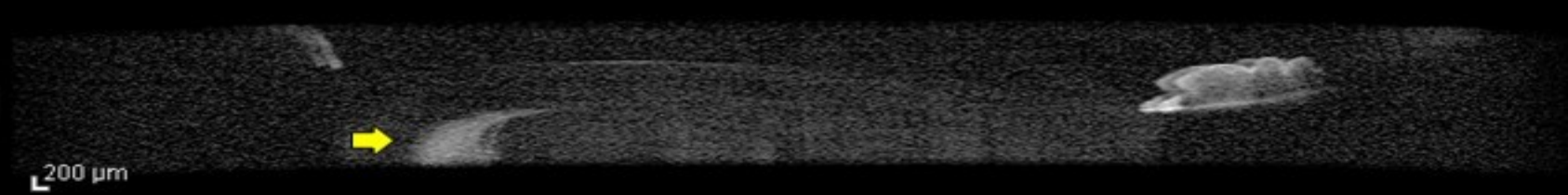
Have a portable slit lamp in your Operating suite

Hyaloid artery remnants: Muscae Volitantes

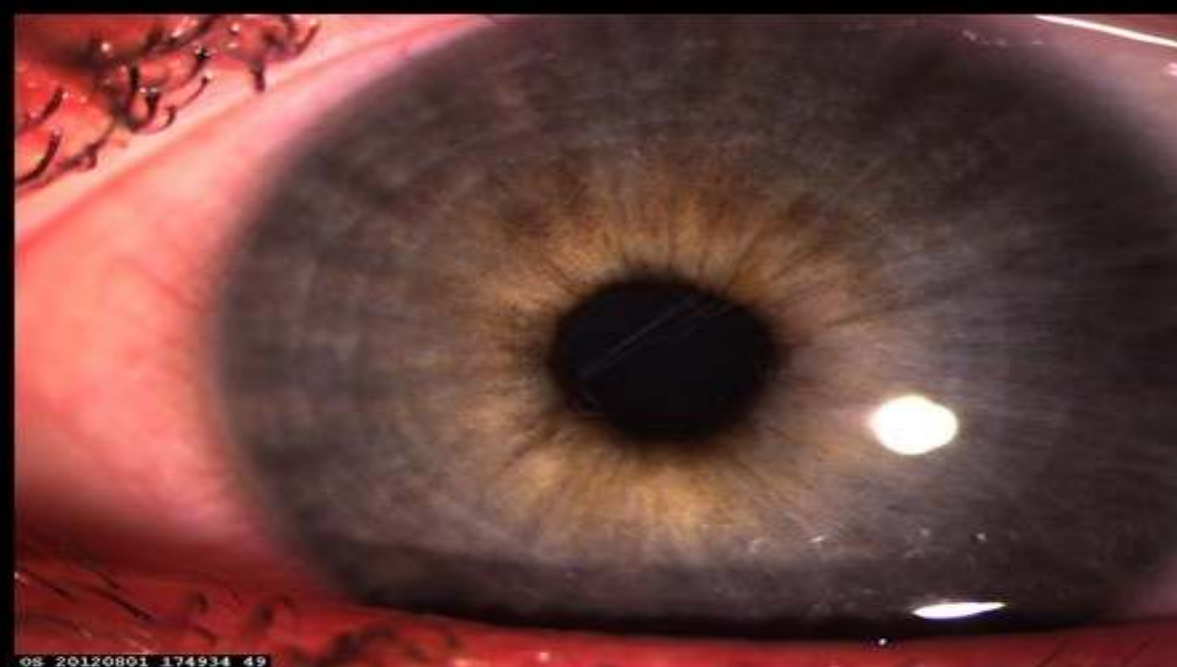
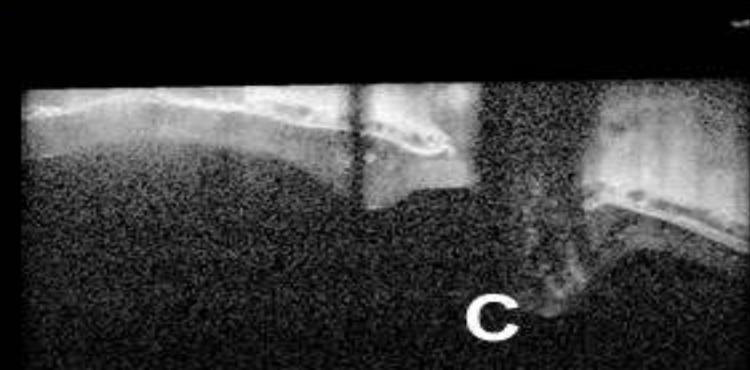




stevens-rowe, jay, 6/20/2004, #10152108



Have a portable slit lamp in your Operating suite

A**B****C****D**

WGS & Segregation Studies -to confirm a molecular diagnosis of Knobloch syndrome

Maternal: COL18A1, c.2864dupC; p.(Gly956ArgfsX20)

Paternal missense: c.5014 G > A; p.(Asp1672Asn): Hypomorphic allele



A



B



C



D



E



F

Surgical considerations

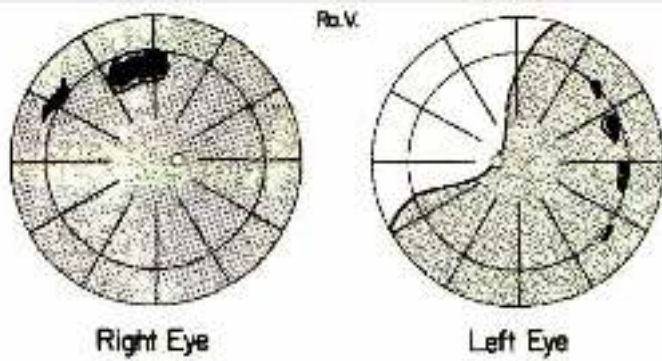
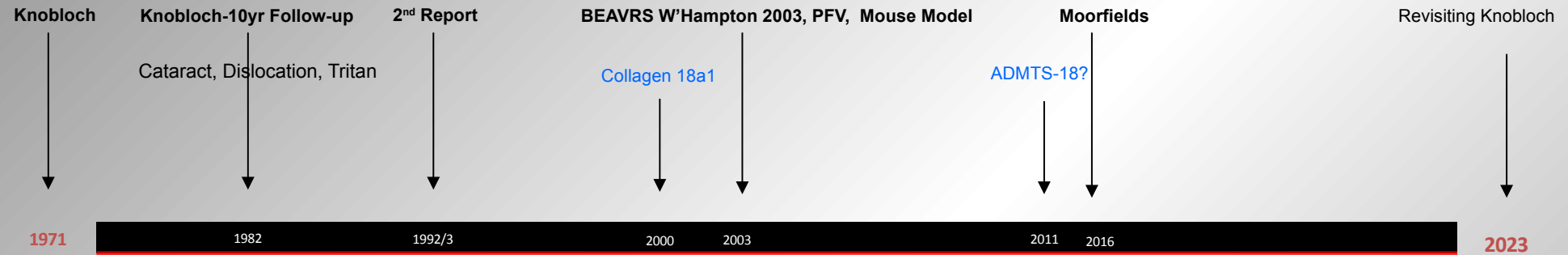


Fig. 2: Retinal detachments of pt III₁, OD, OS.

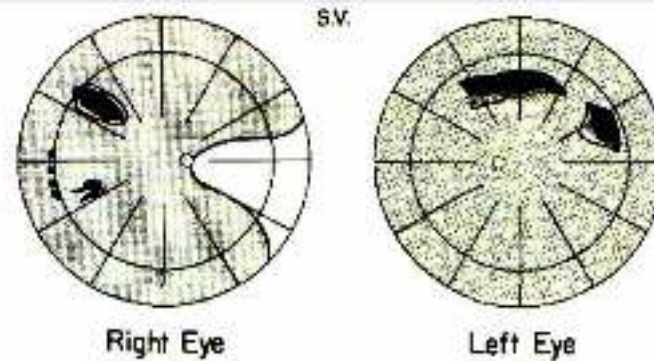


Fig. 3: Retinal detachments of pt III₁, OD, OS.

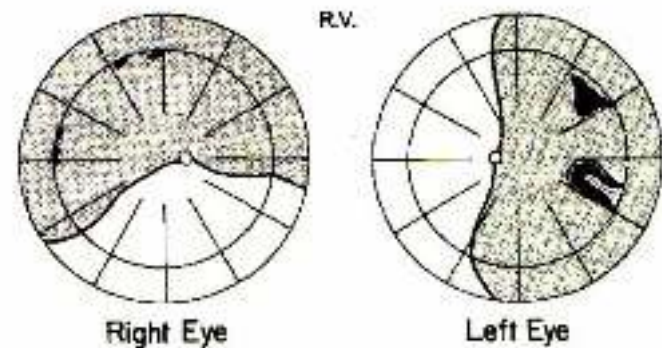
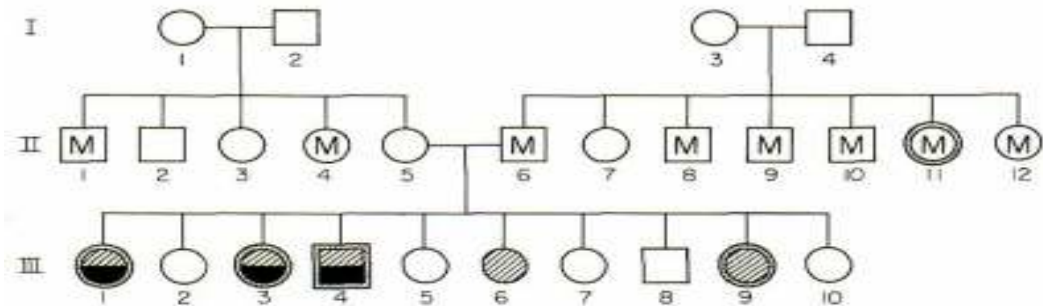
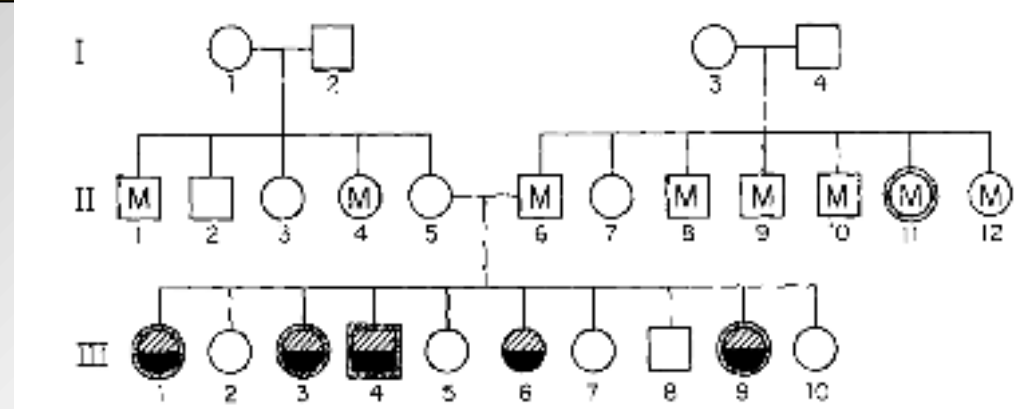
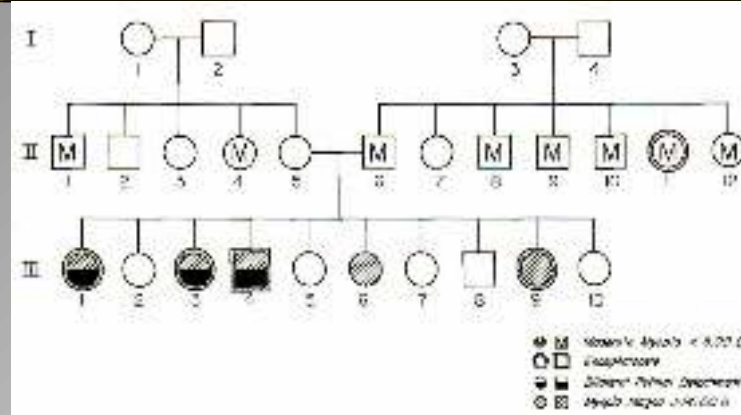


Fig. 4: Retinal detachments of pt III₄, OD, OS.



10 y Follow Up

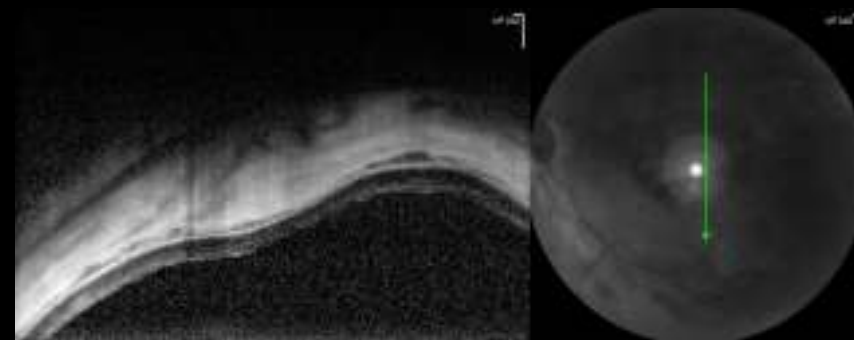
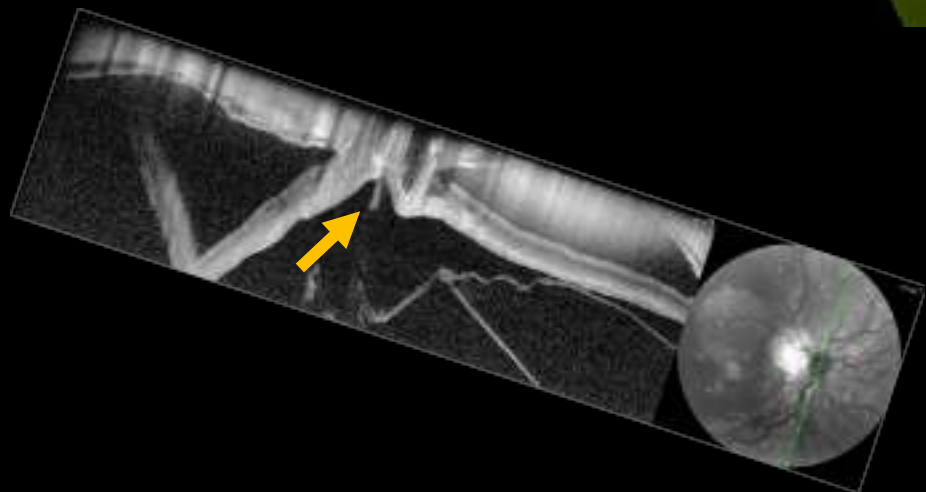
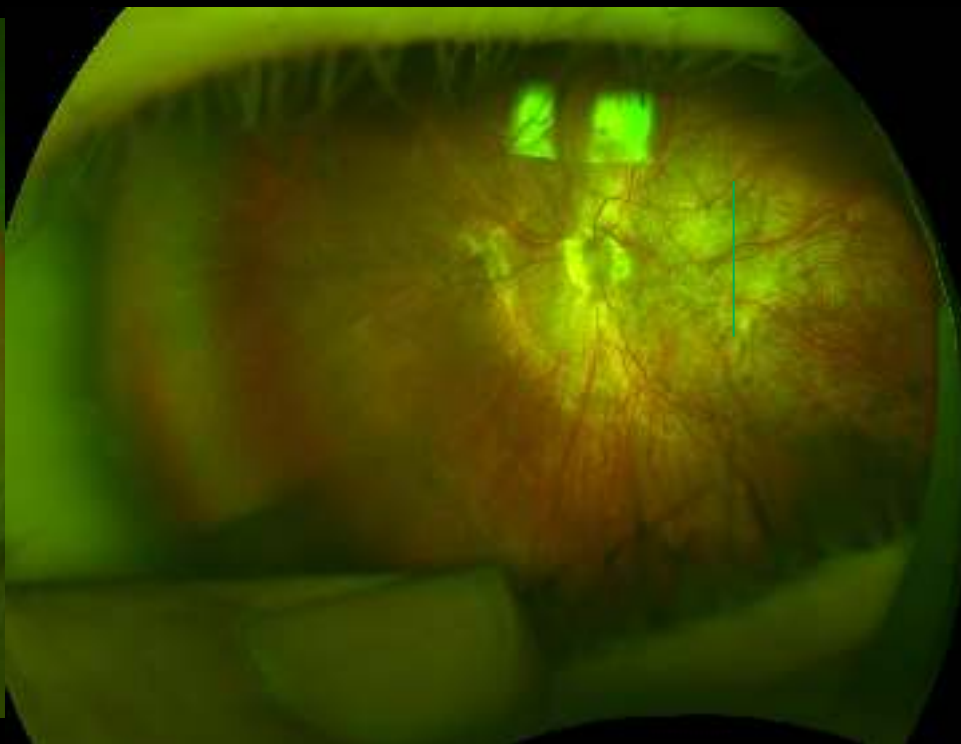


	Age(yrs)	VA OC	VA@10y		Ops RE	Ops LE	M-PVR	NVG	LOS/Dis
III(1)	11	NLP, 20/100	NLP, 20/200		M	M	PVR OD		Y-OS
III(3)	9	NLP, CF	NLP, NLP		M	M	PVR ou	Y	NA
III(4)	7	20/100, NLP	20/200, NLP		4	M	PVR OS		Y-OD
III(6)	4	20/200, 20/200	NLP, 20/200		3	5	PVR OD		Y-OU
III(9)	1....6	20/200, 20/400	AE, 20/400		2	3	PVR OD		Y-OS

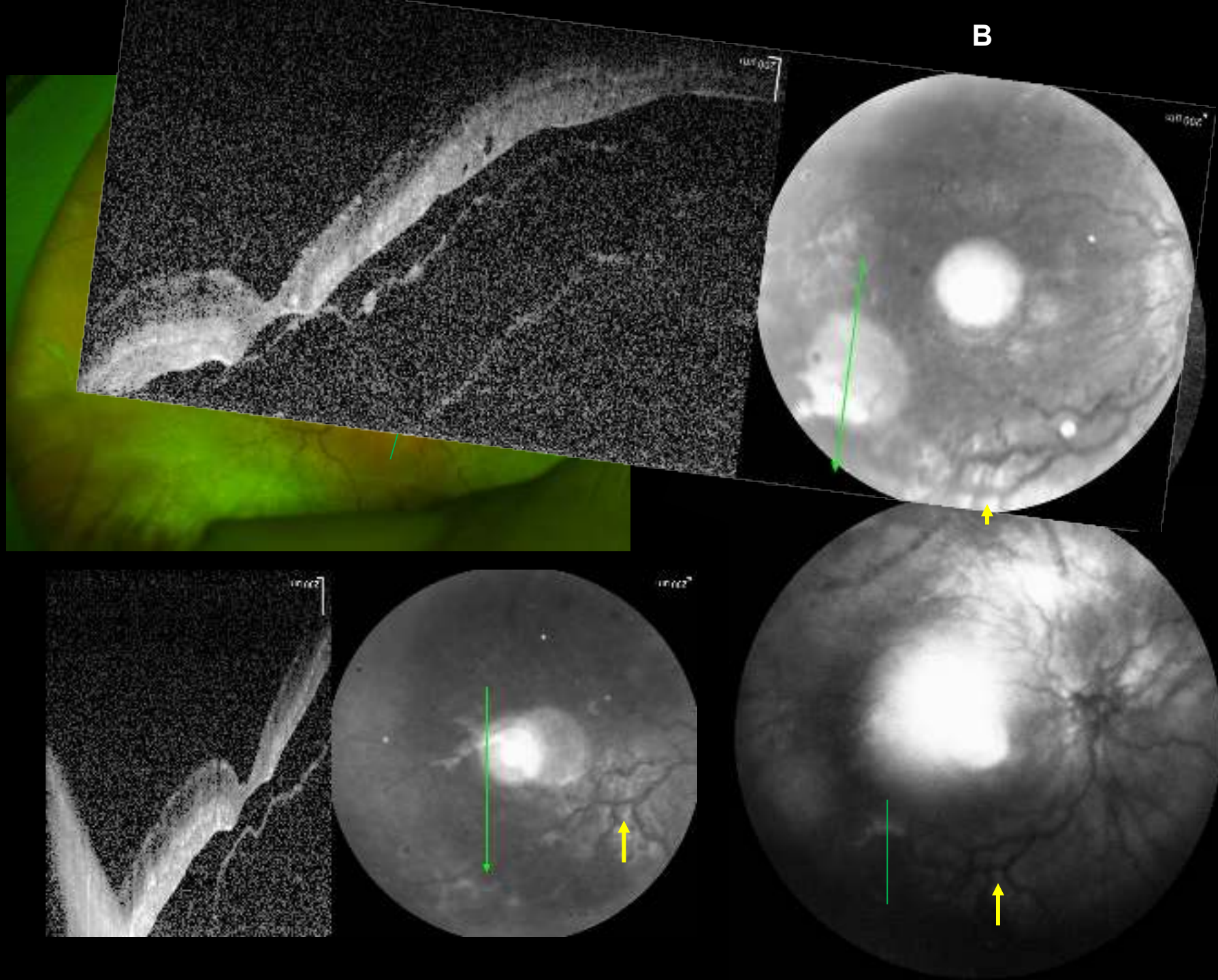
A

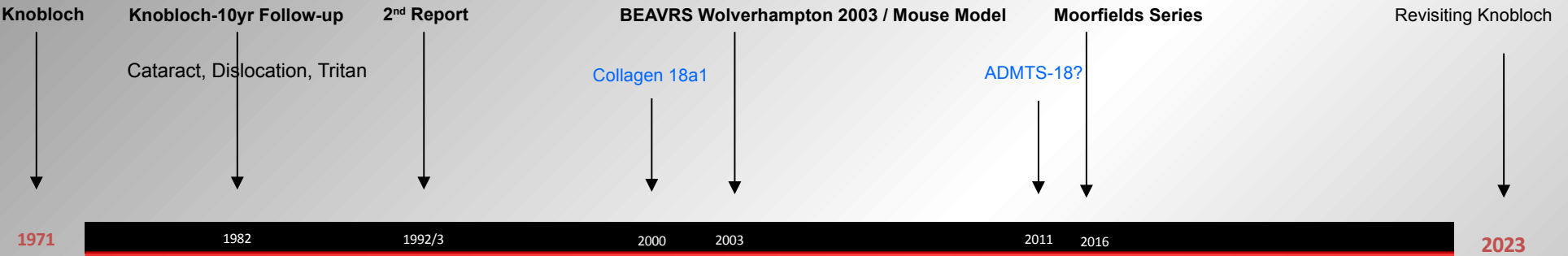


B



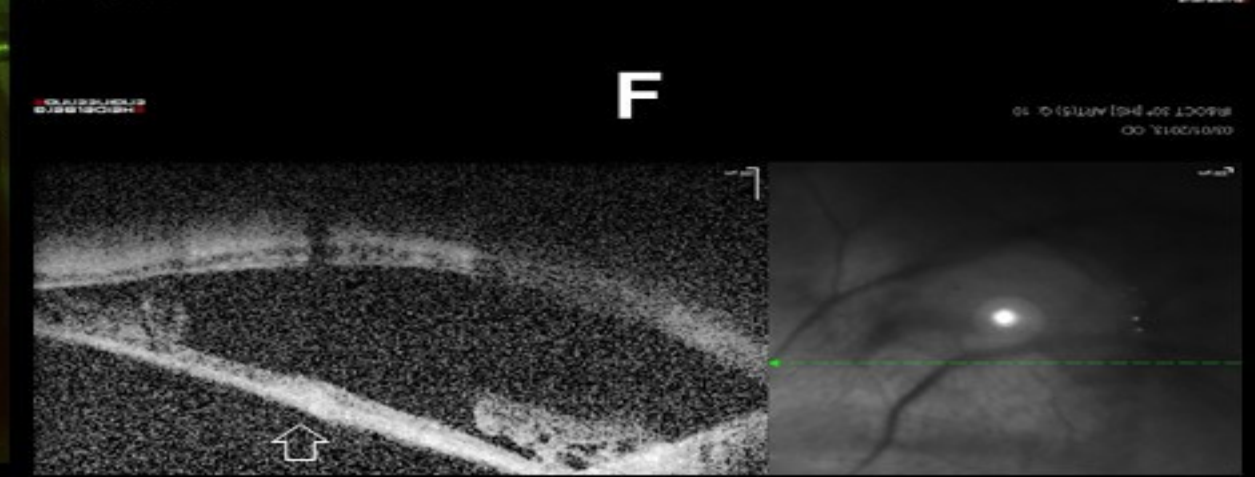
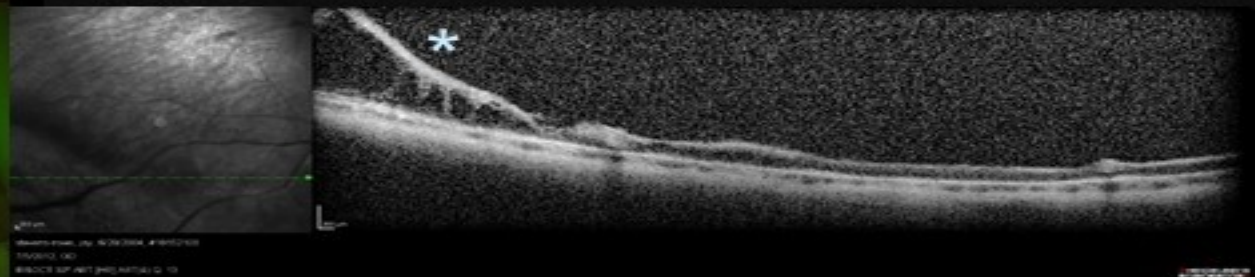
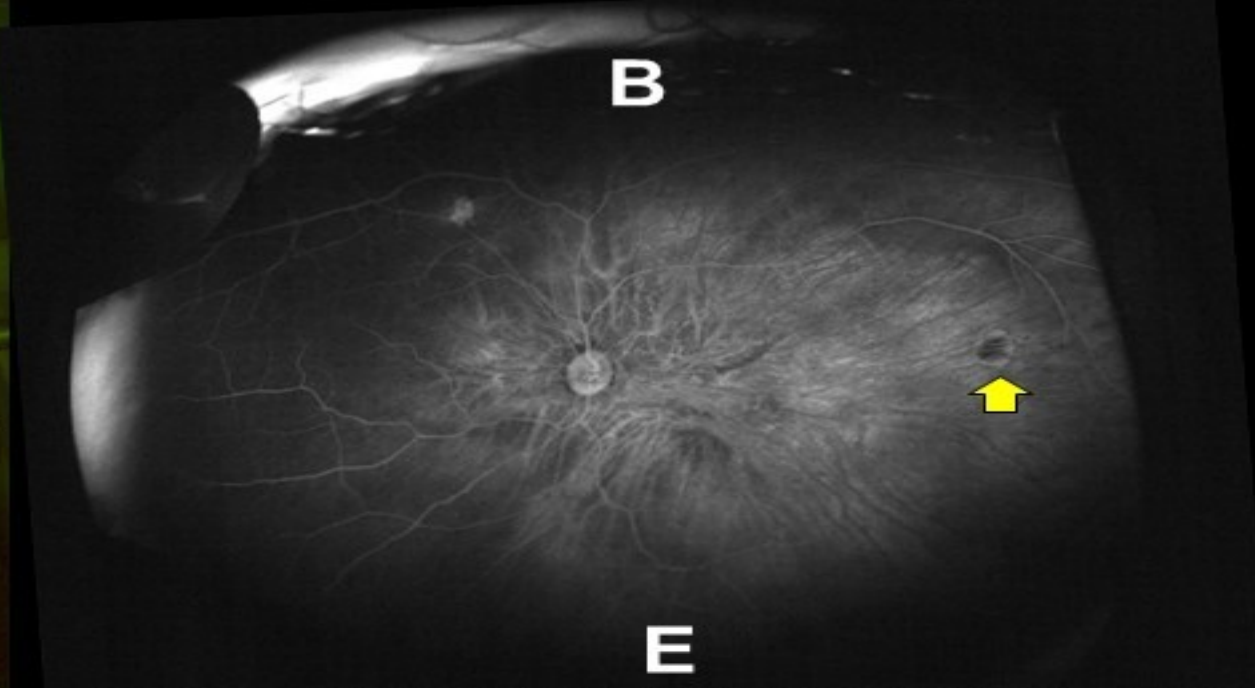
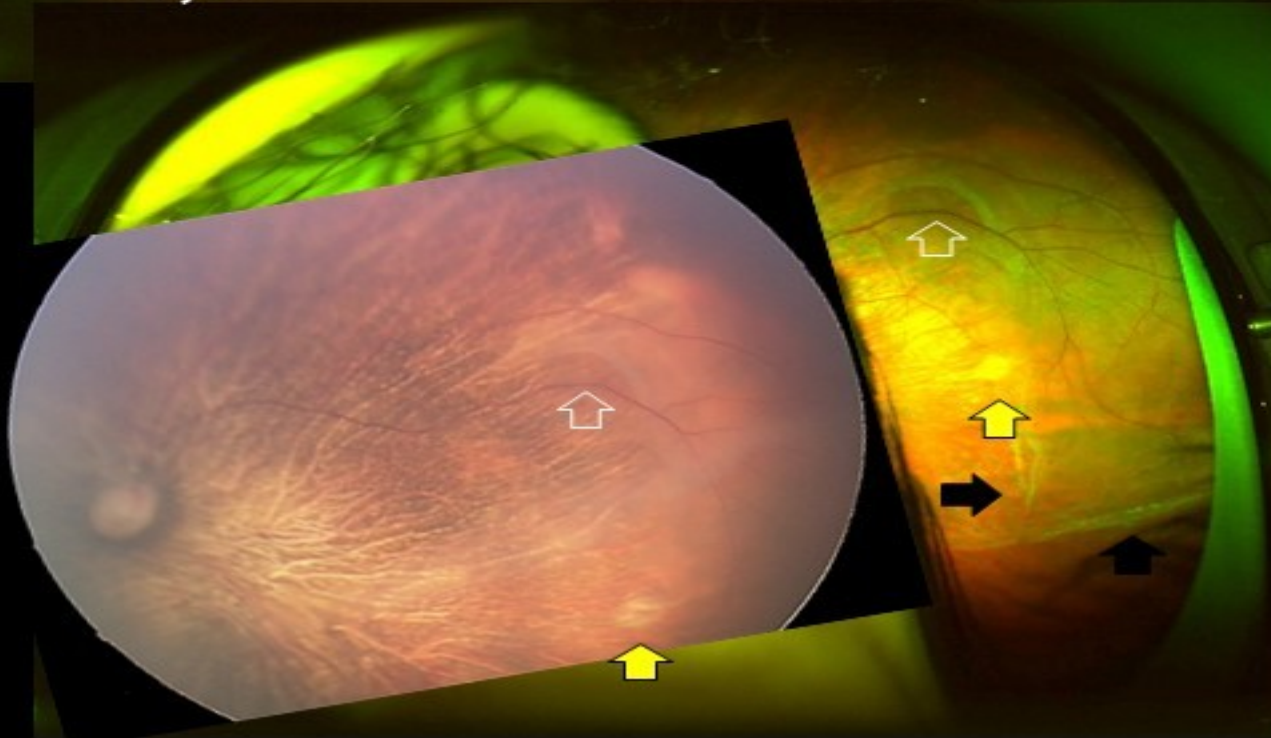
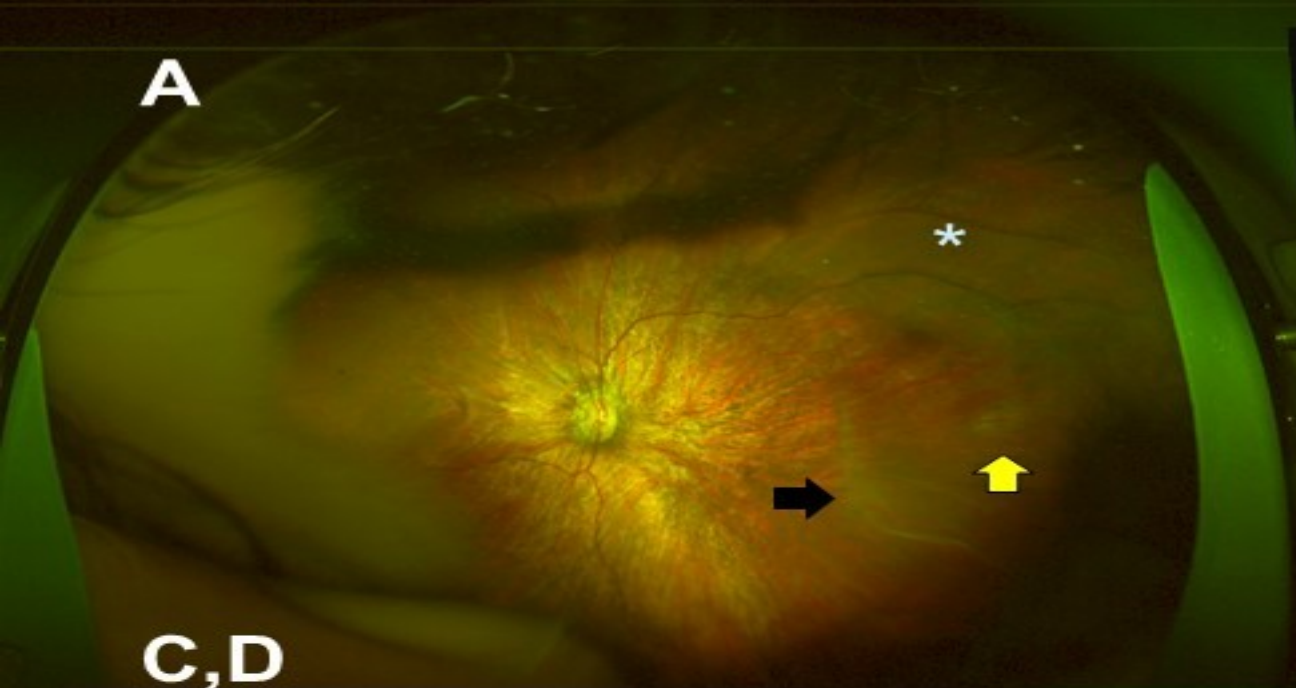
Operate? Buckle or Vitrectomy?





MEH-Series of 12 cases July 2016

- Cone-Rod Dysfunction
- Glaucoma with iris trans-illumination



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- ✗ **Knoblochs: COL18A1:Slax, Occipital Scalp(X) Serous RD, Schisis,**

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Is this eye at risk of RRD and should they have prophylactic laser?



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- ✓ Donnai-Barrow Syndrome: LRP2

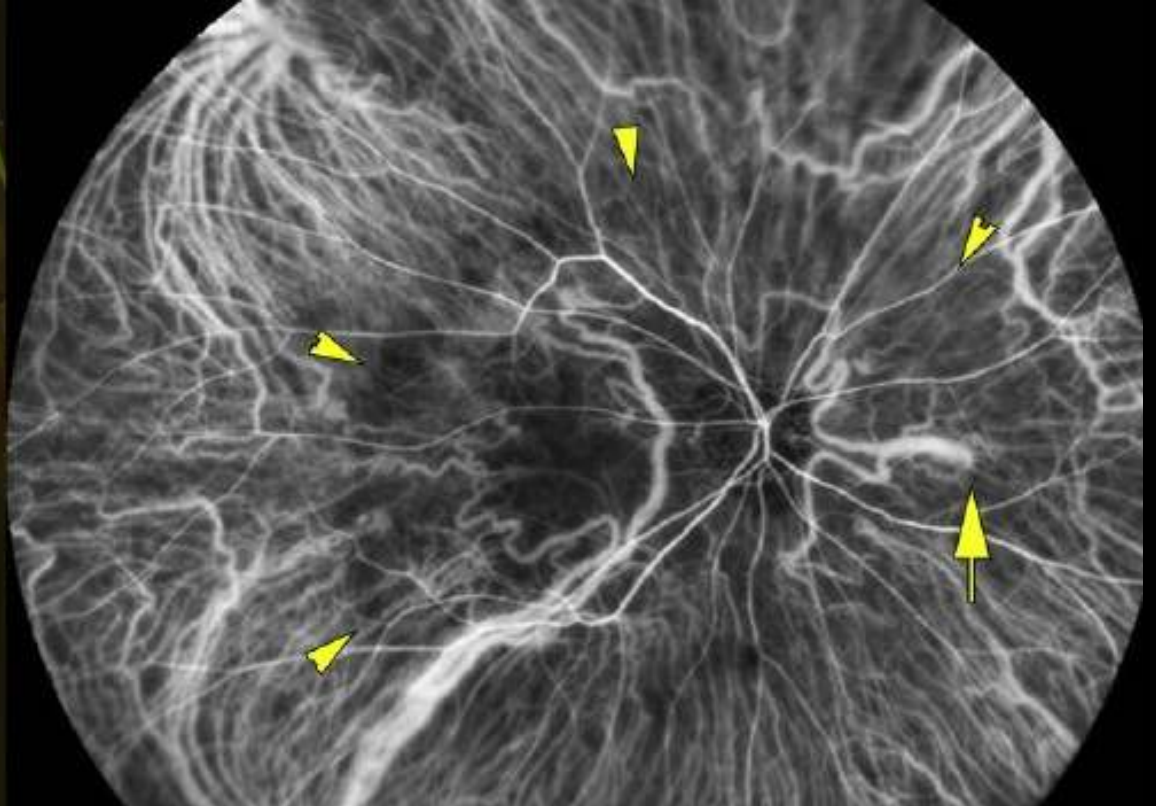
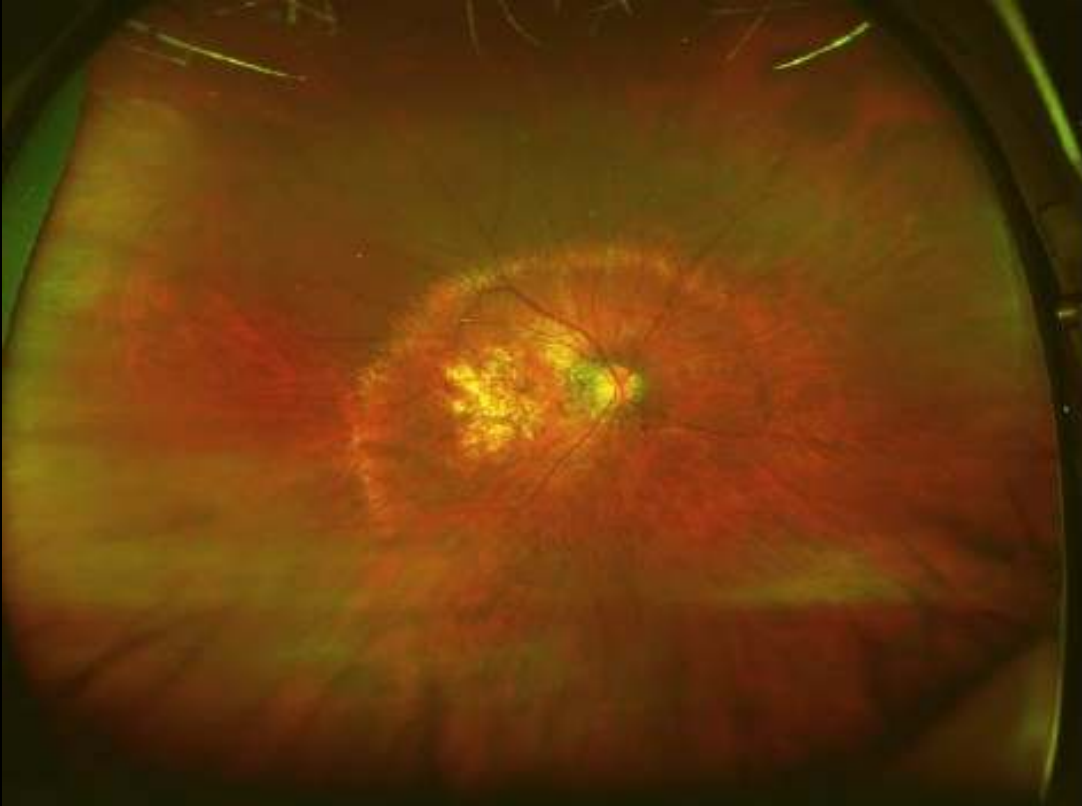


Ectopic vortex veins and varices in Donnai-Barrow syndrome: Pathognomonic?
OPHTHALMIC GENETICS 2021



Ectopic vortex veins in adult high myopia (25% > -8D) : Acquired secondary to staphyloma

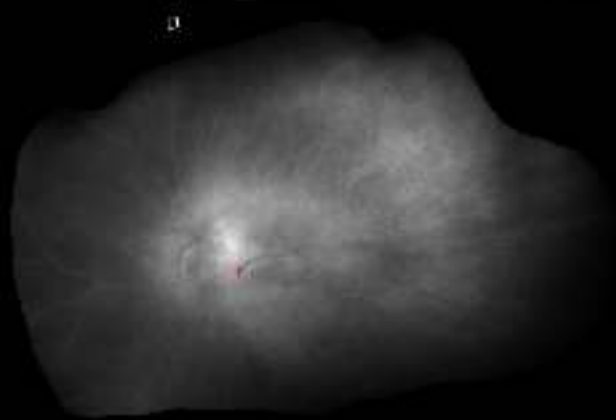
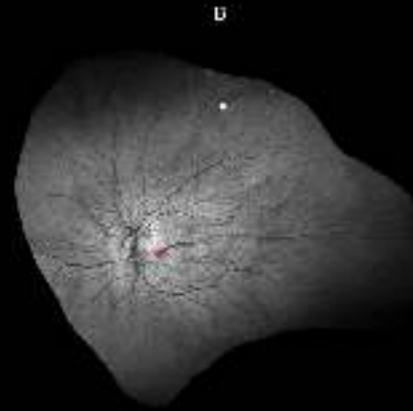
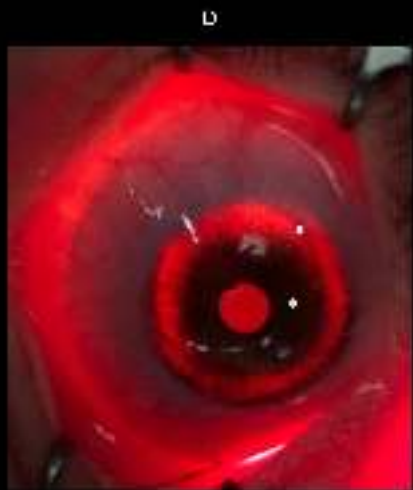
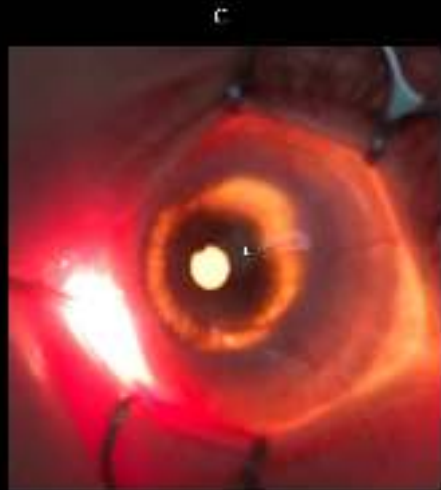
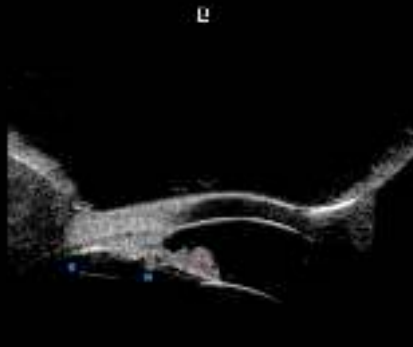
Moriyama M, et al. Br J Ophthalmol 2017;101:1179-1184



Ectopic vortex veins and varices in Donnai-Barrow syndrome

Prophylactic laser: Yes or NO?

OPHTHALMIC GENETICS 2021



Surgical considerations: Evidence Base

- **Population incidence lower than Sticklers**
 - **32 cases: 29 survived: 9(25%) RRD(1.5-9 years)**
- **Prophylaxis**
 - **Do no harm**
 - **GA risk-** multisystem anomalies- Lungs, Heart, Renal, CNS
 - **Ciliary body hypoplasia-** 360 laser
 - **RRD-** double sensory impairment
- **RD**
 - **Thin sclera:** *Perforation-* **Sutures, Needles, Cannulae, Cryo-probe**
 - **Ciliary body hypoplasia-** *Hypotony-* **local explant preferred?**
 - **Ectopic vortex veins-** *haemorrhage*

Childhood High Myopia-RRD- X Trauma

Marfans



Sticklers



Wagners



Knoblochs



Donnai-Barrow-FOAR

