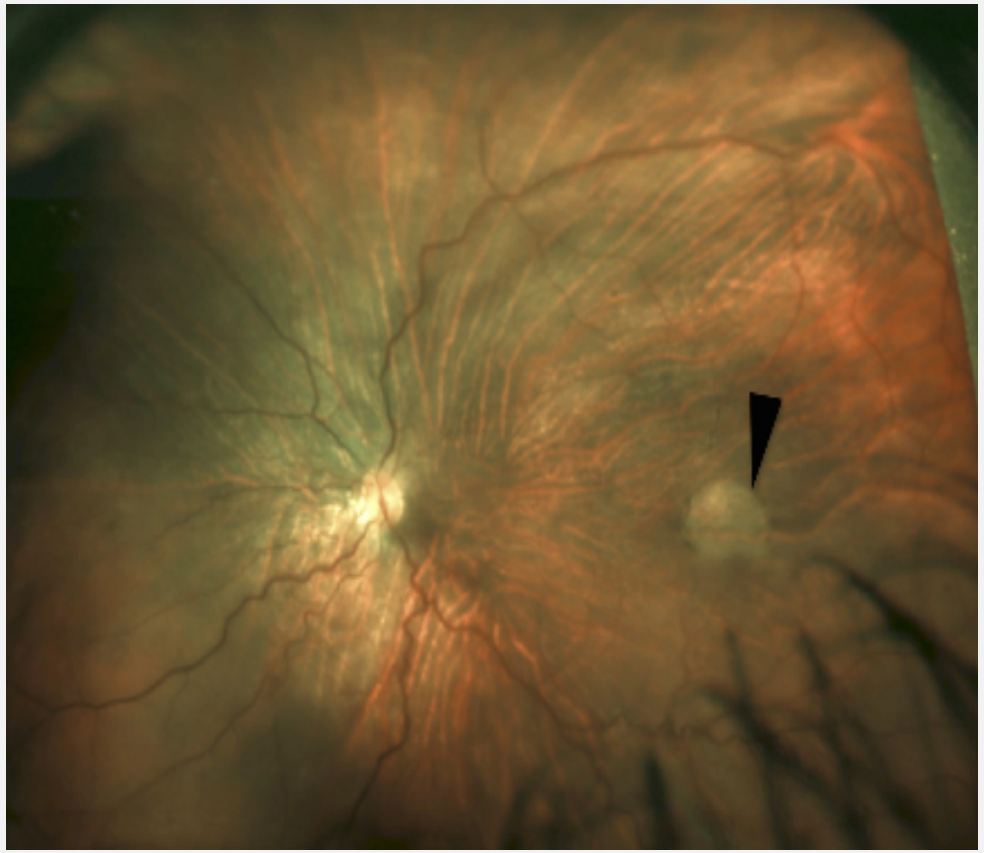
APPROACH TO SYNDROMIC RETINAL DISORDER

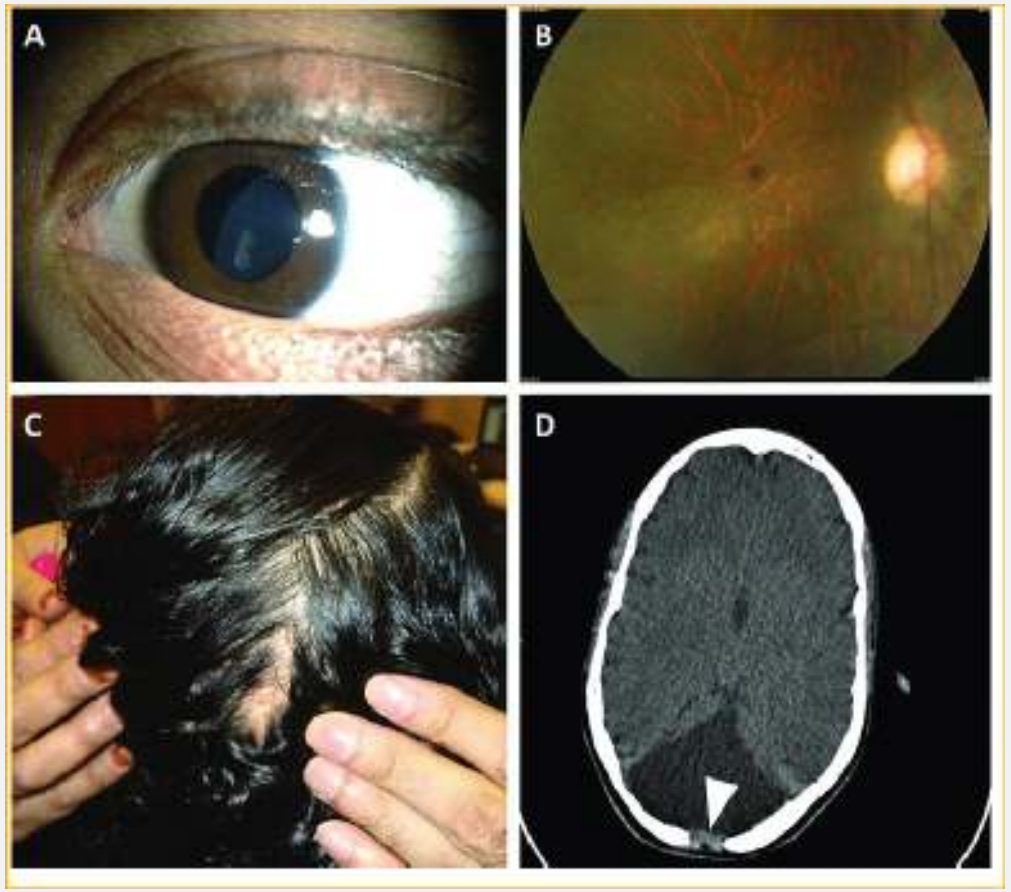
Abdulrahman Badawi, MD
KKESH, Riyadh, Saudi Arabia

INTRODUCTION

- Syndromic retinal detachment (SRD)is a significant ocular condition characterized by the separation of the retina from the underlying retinal pigment epithelium, which can lead to vision loss if not promptly treated.
- This type of retinal detachment is often associated with systemic syndromes, that it may occur in conjunction with other medical conditions that affect multiple systems of the body.
- Understanding the complex interplay between systemic conditions and retinal health is essential for clinicians to provide effective care and improve outcomes for patients with syndromic retinal detachment.

CASE 1





Knobloch syndrome

- Caused by an autosomal recessive mutation in the COL18A1 gene
- Associated with ophthalmic and neurological features.
- Usually have visual manifestation in first decade of life

❖ Ophthalmic features:

- High myopia , RD
- Sever chorioretinal atrophic changes with prominent choroidal vessel.
- Macular atrophic lesions with or without a punched out appearance
- Macular hypoplasia
- Lens subluxation, cataracts, strabismus.
- Smooth irides and posterior lens opacity and cataract

❖ Neurological features:

- Occipital skull defects with or without encephalocele

are the characteristic.

- Neuroradiologic imaging may show subependymal nodules, and cerebellar vermis atrophy.
- Delay in cognitive development.

> Ophthalmol Retina. 2020 May;4(5):498-503. doi: 10.1016/j.oret.2019.12.004. Epub 2019 Dec 12.

Macular Hole–Related Retinal Detachment in Children with Knobloch Syndrome

Sulaiman M Alsulaiman [1], Abdulelah A Al-Abdullah [1], Adel Alakeely [1], Hassan Aldhibi [1], Leonore Engelbrecht [1], Nicola G Ghazi [2], Marco Mura [3]

Affiliations + expand

PMID: 32111543 DOI: 10.1016/j.oret.2019.12.004

> Ophthalmol Retina. 2024 Sep;8(9):898-904. doi: 10.1016/j.oret.2024.03.020. Epub 2024 Mar 30.

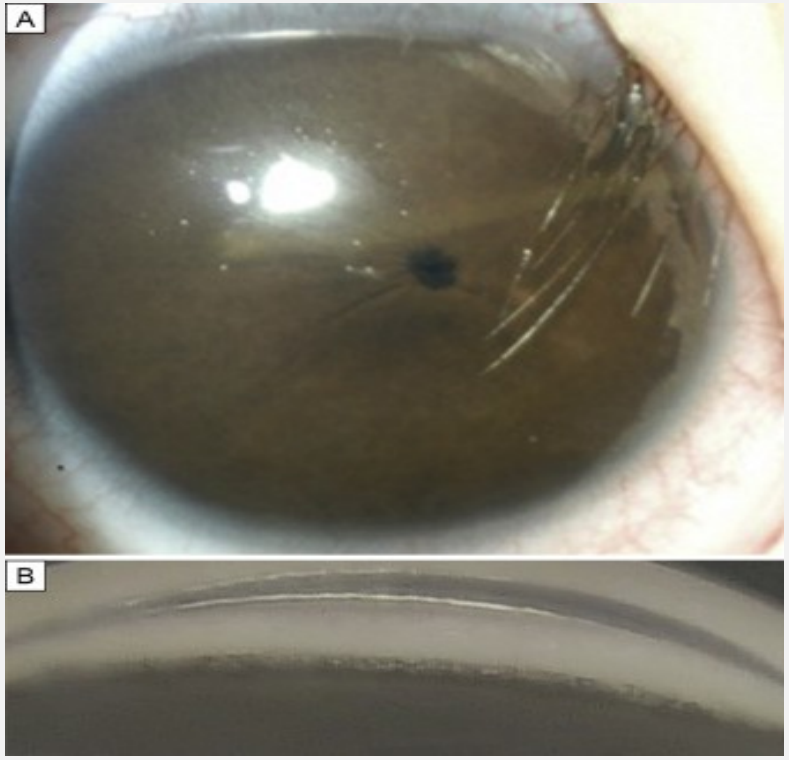
Surgical Outcomes of Retinal Detachment in Knobloch Syndrome

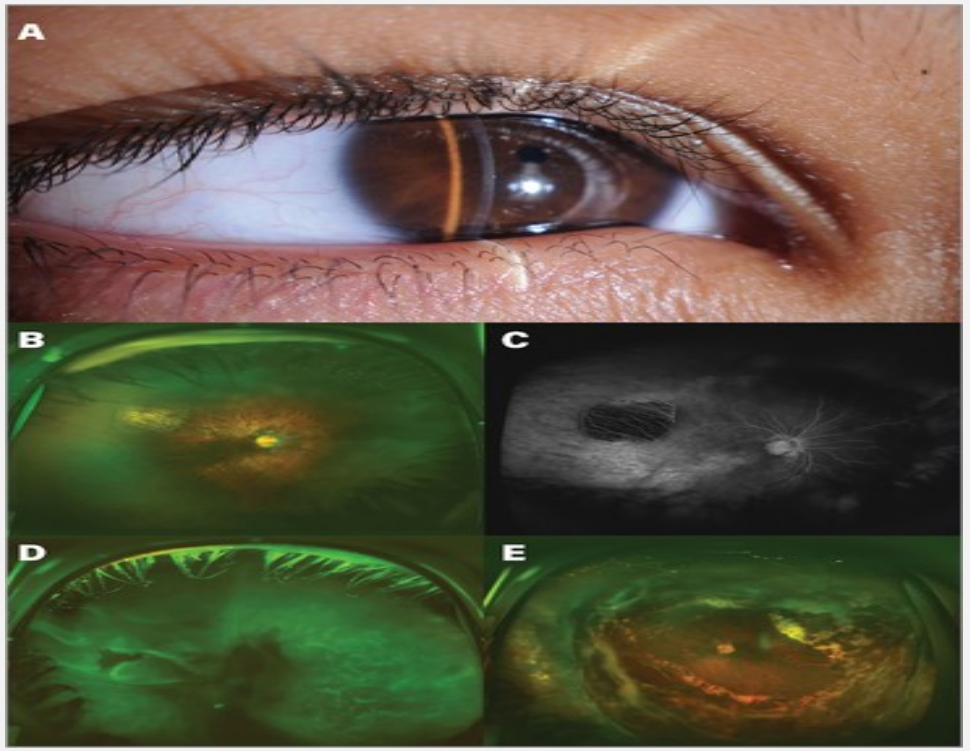
Khawlah A Alzaben [1], Ahmed Mousa [1], Lama Al-Abdi [2], Fowzan S Alkuraya [2], Sulaiman M Alsulaiman [3]

Affiliations + expand

PMID: 38556002 DOI: 10.1016/j.oret.2024.03.020

CASE 2





- **Pierson syndrome** is a rare genetic disorder caused by mutations in the **LAMB2** gene, which is responsible for producing a protein essential for **kidney** and **eye** development. It primarily affects infants and young children and is inherited in an **autosomal recessive** pattern.

- Key features of Pierson syndrome include:
- • **Congenital nephrotic syndrome**: severe kidney dysfunction present at birth, leading to excessive protein loss in the urine, swelling (edema), and kidney failure.
- • **Eye abnormalities**: such as **microcoria** (small, fixed pupils), which is a hallmark of the condition, along with High myopia, cataract and **RD**.
- • **Developmental delays** and other neurological complications may also occur in some cases.

Early-Onset Myopia and Retinal Detachment without Typical Microcoria or Severe Proteinuria due to a Novel LAMB2 Variant

Abdulaziz A Alshamrani [1], Moustafa Magliyah [2], Fowzan S Alkuraya [3], Lama Alabdi [3], Talal A Alfaadhel [4], Sulaiman M Alsulaiman [5]

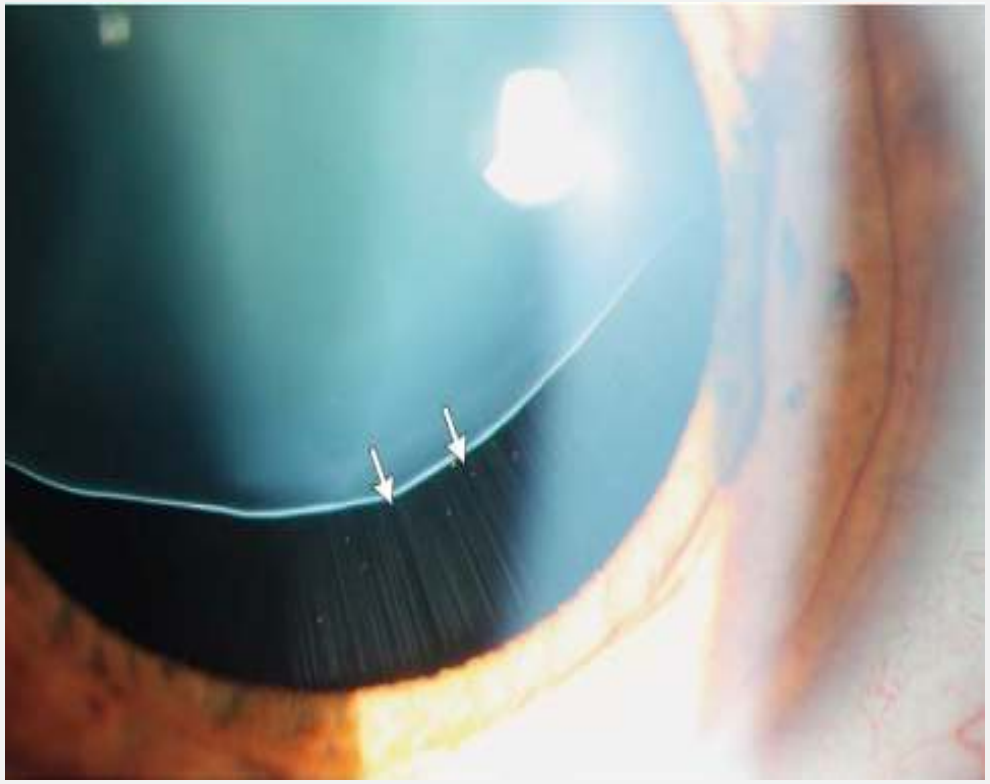
> Ophthalmic Surg Lasers Imaging Retina. 2020 Nov 1;51(11):618-627.
doi: 10.3928/23258160-20201104-03.

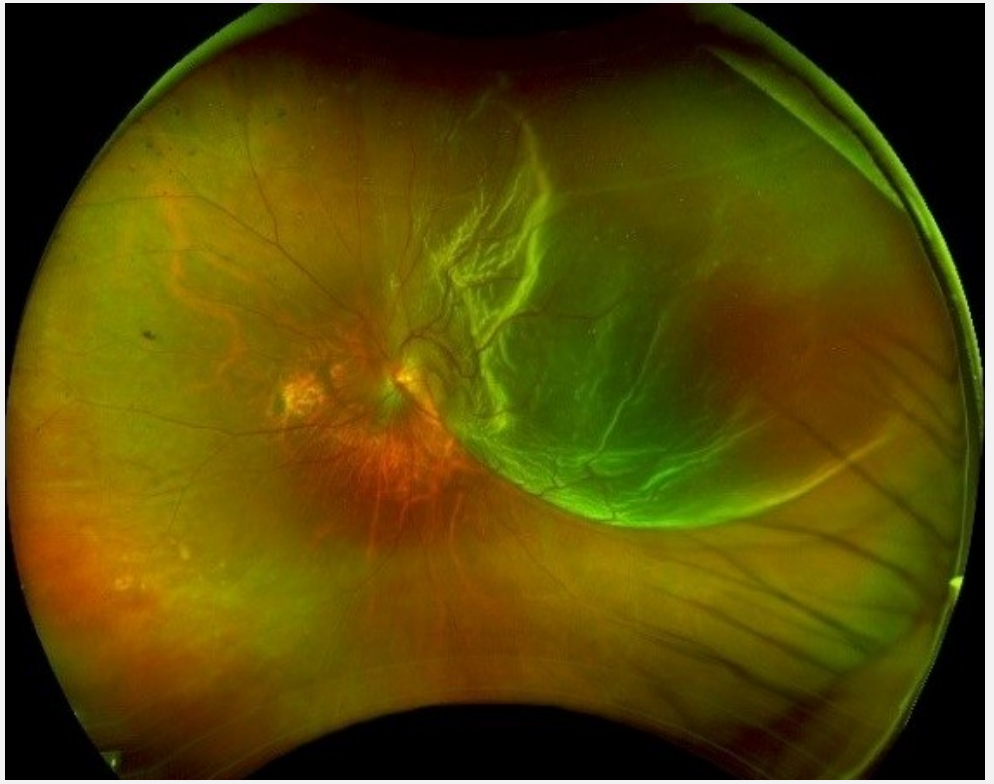
Posterior Segment Characterization in Children With Pierson Syndrome

Abdulaziz AlTaisan, Moustafa Magliyah, Marwan A Abouammoh, Ibrahim Taskintuna, Yahya Alzahrani, Emmanuel Chang, Sulaiman M Alsulaiman

PMID: 33231694 DOI: 10.3928/23258160-20201104-03

CASE 3





Marfan syndrome:

- An autosomal dominant disease associated with abnormal variants in fibrillin-1 (FBN1) gene or less commonly in tumor growth factor-beta receptor 2 (TGFBR2).

- Ocular manifestations of Marfan syndrome include ectopia lentis, rhegmatogenous retinal detachment (RRD), glaucoma, myopia, and corneal abnormalities.

- Surgical treatment outcomes for RRD in Marfan syndrome patients are variable and might be affected by several factors.

Table 1: Analysis of risk factors for RRD among Marfan Syndrome patients

Odds ratio	**P Value**	**Eyes without RRD (n=109)**	Eyes with RRD (n=54)
Male 0.9 Female 1.1	0.290	Gender Males 59 Females 50	Gender Males 26 Females 28
Yes 10.1	0.014	Prior Trauma Yes 0 No 110	Prior Trauma Yes 6 No 47
Yes 3.41	0.001	Previous Ocular surgery Yes 22 No 87	Previous Ocular surgery Yes 25 No 29
Lensectomy 2.65 Lensectomy + IOL 0.38	0.002	Type of previous surgery (n=23) Lensectomy 7 Lensectomy + intraocular lens implantation 16	Type of previous surgery (n=25) Lensectomy 18 Lensectomy + intraocular lens implantation 7
	0.001>	Lens status Phakic 86 (85 subluxated) Aphakia 7 PCIOL 5 SFIOL 7 ACIOL 4	Lens status Phakic 29 (24 subluxated) Aphakia 18 PCIOL 2 SFIOL 3 ACIOL 2
	0.001>	Axial Length mm 2.3 ± 25.9	Axial Length mm 2.7 ± 28.2

Conclusion

RRD can develop in as many as 33.1% of Marfan syndrome eyes. Risk factors for RRD development include high axial length, prior trauma, prior intraocular surgery and aphakic status. High anatomical success rates are achievable especially in eyes without PVR. Long duration of follow up is needed for these patients.

CASE 4

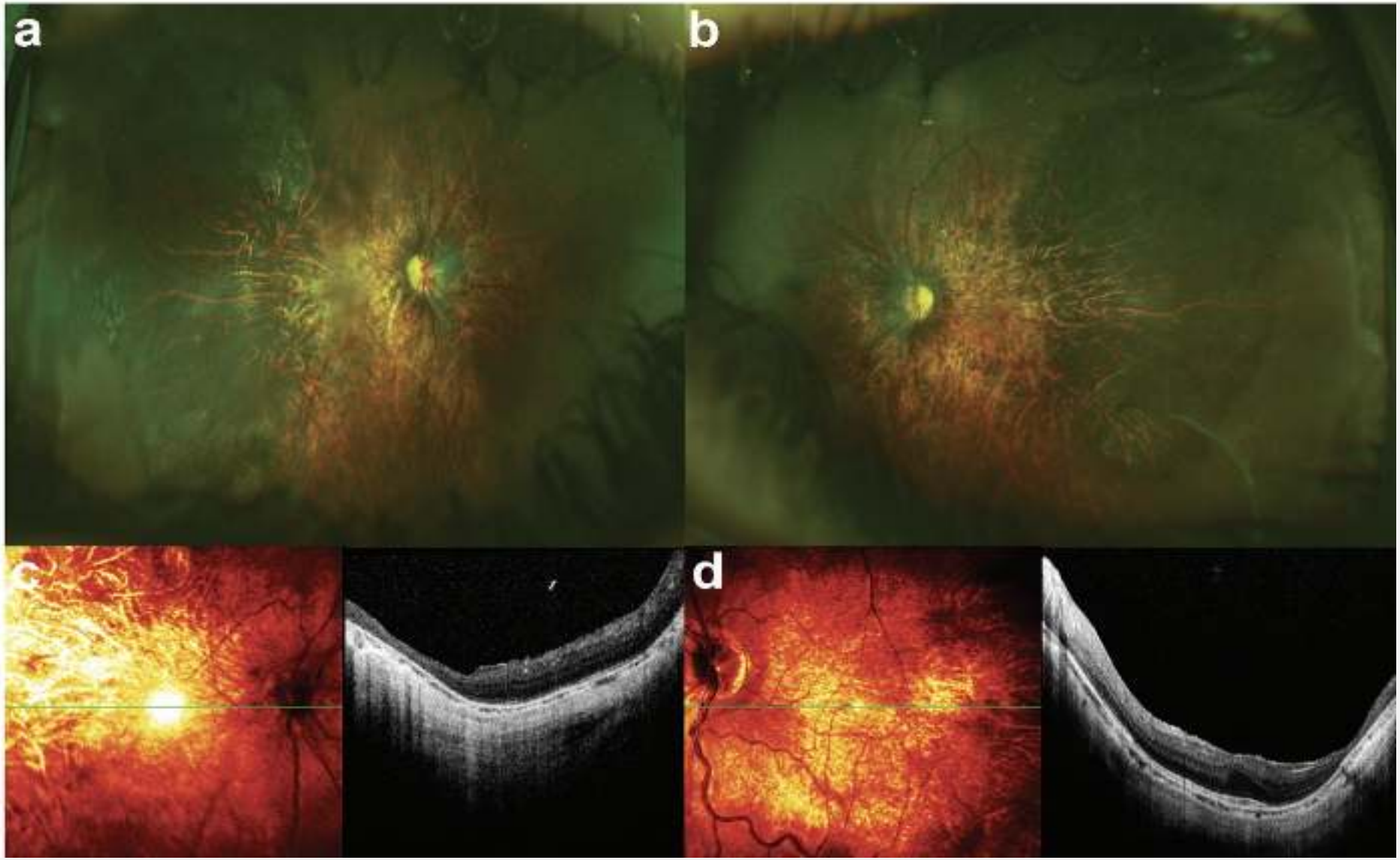
Case Reports > Ophthalmic Genet. 2021 Feb;42(1):96-98.
doi: 10.1080/13816810.2020.1849316. Epub 2020 Nov 29.

Poretti–Boltshauser syndrome: a rare differential diagnosis to consider in pediatric high myopia with retinal degeneration

Adel Salah Alahmadi [1], Abdulrahman H Badawi [1], Moustafa S Magliyah [1], Amani Albakri [1], Patrik Schatz [1 2]

Affiliations + expand

PMID: 33251915 DOI: 10.1080/13816810.2020.1849316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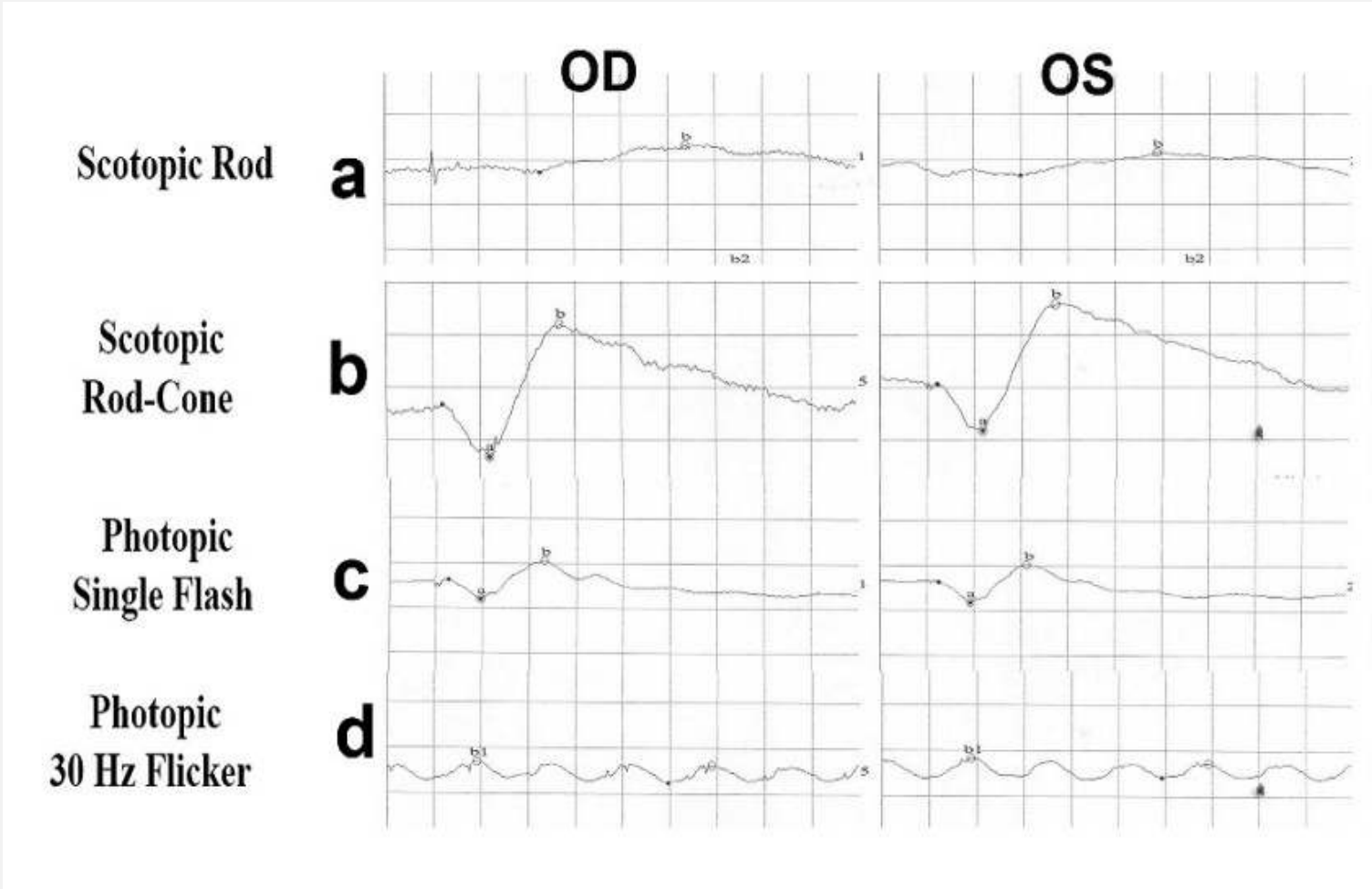
OD
OS
Scotopic Rod
a
Scotopic
Rod-Cone
b
Photopic
Single Flash
c
Photopic
30 Hz Flicker
d

Poretti-Boltshauser syndrome (PBS):

- **An autosomal recessive** disease associated with mutations in the gene **LAMA1 (laminin-1)** and typically causes neurological manifestations including non-progressive cerebellar ataxia, developmental delay, intellectual disability, ocular motor apraxia, and cerebellar dysplasia**.**

- **Ophthalmological findings** include nystagmus, strabismus, high myopia, and retinal manifestations including chorioretinal atrophy, nasalization of retinal blood vessels, and an abnormal ERG .


thank you