

Pigmented Paravenous Retinochoroidal Atrophy

Aditya Somisetty, BA; [Aaron D. Dotson, MD](#); [Ian C. Han, MD](#)

December 5, 2023



INITIAL PRESENTATION

Chief Complaint: "I was told that I have a retinal scar in my right eye."

History of Present Illness (HPI)

The patient is a 58-year-old African American woman with an ocular history of mild primary open angle glaucoma and age-related cataracts in both eyes who presents for the evaluation of a newly-noted retinal scar in her right eye. The scar was found incidentally during a routine exam by her optometrist. She is asymptomatic with good vision in both eyes and is unaware of any changes in her vision. She also denies episodes of flashes of light, floaters, visual field shadowing, pain with eye movements, light sensitivity, or night blindness.

Past Ocular History

- Primary open angle glaucoma, both eyes
- Age related cataracts, both eyes

Past Medical History

- Benign essential hypertension

Medications

- Amlodipine
- Gabapentin
- Latanoprost, both eyes

Allergies

- None

Family History

- Primary open angle glaucoma (Father)

Social History

- Non-contributory

Review of Systems

- Negative except for what is detailed in the history of present illness

OCULAR EXAMINATION

- Distance visual acuity with correction

- Right eye (OD): 20/20
- Left eye (OS): 20/20

- **Ocular Motility**

- OD: Full
- OS: Full

- **Intraocular Pressure (IOP) - Tonopen**

- OD: 17 mmHg
- OS: 16 mmHg

- **Pupils**

- OD: 4 mm in dark, 3 mm in light, no relative afferent pupillary defect (RAPD)
- OS: 4 mm in dark, 3 mm in light, no RAPD

- **Confrontation Visual fields - count fingers**

- OD: Full
- OS: Full

- **External**

- Normal OU

- **Slit lamp exam**

	OD	OS
Lids/Lashes	Normal	Normal
Conunctiva/Sclera	Limbal complex ion associated melanosis	Limbal complex ion associated melanosis
Cornea	Clear	Clear
Anterior Chamber	Deep and quiet	Deep and quiet
Iris	Normal architecture	Normal architecture
Lens	Trace nuclear sclerosis	Trace nuclear sclerosis

- **Dilated fundus examination (DFE)**

	OD	OS
Vitreous	No anterior vitreous cell	No anterior vitreous cell
Disc	No edema, pallor, or hemorrhages	No edema, pallor, or hemorrhages
Cup-to-disc ratio	0.45	0.45
Macula	Blunted foveal reflex	Blunted foveal reflex
Vessels	Normal	Normal
Periphery	Multifocal hyperpigmented chorioretinal scars and atrophy with perivenous predilection along branches of the superior arcade. Circular chorioretinal scar and atrophy along the supertemporal mid-periphery.	Normal

- **Dilated Fundus Photography**



[Enlarge](#) [Download](#)

Figure 1. Widefield montage color fundus photographs (1) and super peripheral sweep (2) of the right eye demonstrating multifocal hyperpigmented chorioretinal scars and atrophy with perivascular predilection along the superior arcade.



[Enlarge](#) [Download](#)

[Enlarge](#) [Download](#)

Differential Diagnosis

- Pigmented paravenous retinochoroidal atrophy
- Non-infectious inflammatory scarring due to autoimmune conditions (e.g., serpiginous choroidopathy, sarcoidosis)
- Scarring due to infectious inflammation (e.g., syphilis, tuberculosis choroiditis, toxoplasmosis)
- Congenital stationary night blindness
- Unilateral retinitis pigmentosa (e.g., manifesting carrier with mosaicism from X-linked retinitis pigmentosa)

CLINICAL COURSE

The patient was asymptomatic on presentation and was at her baseline visual acuity. Given the non-central location of the chorioretinal scarring and the lack of associated signs to suggest current or prior inflammation or other concomitant ocular disease, the patient was diagnosed with pigmented paravenous retinochoroidal atrophy, and reassurance was provided for annual monitoring for any progression.

DIAGNOSIS: Pigmented Paravenous Retinochoroidal Atrophy

DISCUSSION

Etiology

Pigmented paravenous retinochoroidal atrophy (PPRCA) is a rare condition characterized by paravenous pigment aggregations along the retinal veins associated with adjacent chorioretinal atrophy. Most cases of PPRCA are considered isolated or sporadic, with no known underlying inherited or inflammatory cause identified [1-3]. However, similar perivenular pigmentation can be seen in those with inherited retinal diseases [4-6], including CRB1-associated retinitis pigmentosa (RP) or Leber congenital amaurosis (LCA) [7,8], or in the context of abnormal embryonic ocular development such as macular coloboma [8,9]. Paravenous pigmentation can also be seen in inflammatory and infectious diseases, including tuberculosis, syphilis, sarcoidosis, Vogt-Koyanagi-Harada disease, Behcet's disease, measles, and rubella [10-13].

Pathophysiology and Natural History

PPRCA is highlighted by pigment aggregation and clumping along the retinal veins leading to zonal retinochoroidal atrophy, with resultant atrophic and eventual visual changes [11]. The exact pathophysiology of these changes remains poorly understood, but dysfunction of the retinal pigment epithelium (RPE) is thought to play a role in photoreceptor degeneration as well as pigmentary abnormalities [9,11].

Signs/Symptoms

PPRCA is often an incidental finding without associated symptoms. Some patients experience mild blurred vision, but severely reduced vision can be seen when the macula is involved. Fundusoscopic examination reveals coarse pigmentary clumps within the retina, fine pigmentary changes along the retinal veins, and bone spicule-like pigmentation, along with retinochoroidal atrophy, usually away from the optic nerve head. Typically, the pigmentation extends alongside the posterior pole of the retina and into the periphery. PPRCA is often bilateral but asymmetric [14], but unilateral presentations have also been reported [15-17]. While macular involvement is rare, some have reported associated pigmentary macular degeneration with RPE atrophy, lamellar macular hole formation, and cystoid macular edema [18].

Diagnosis

PPRCA is diagnosed by clinical examination and the exclusion of other potential underlying causes through a variety of different diagnostic tests that can be considered in workup, including fundus autofluorescence (FAF) [19], fluorescein angiography (FA) [19,20], indocyanine green angiography (ICG) [21], electroretinogram (ERG) [11], and optical coherence tomography (OCT) [22,23], and OCT angiography (OCTA)[24,25].

FAF can be helpful to demonstrate the extent of PPRCA, with areas of hypo-autofluorescence corresponding to areas of RPE atrophy, and hyper-autofluorescence corresponding to pigment clumping and areas of dysfunctional RPE. When feasible to obtain (as PPRCA is often peripheral), OCT may show areas of outer retinal and RPE loss corresponding to the pigmented lesions on clinical examination. FA and ICG may show hyperfluorescence from window defects in areas of retinochoroidal atrophy and can be considered in cases with suspicion for an underlying inflammatory condition (e.g., to evaluate for subclinical chorioretinal lesions or assess for vascular leakage). ERG may be considered in cases where an underlying photoreceptor degeneration may be present (e.g., retinitis pigmentosa). OCTA has been used to identify the extent of retinal and choroidal vascular injury related to PPRCA, with suggestion of vascular compromise to choriocapillaris and the deep capillary plexus playing a role [24,25].

Treatment

There is no current treatment for PPRCA.

<p>EPIDEMIOLOGY</p> <ul style="list-style-type: none"> • PPRCA is a very rare condition • Most cases are PPRCA isolated, but a genetic disposition has been suggested with some cases • Data is not available regarding the epidemiology of the disease 	<p>ETIOLOGY</p> <ul style="list-style-type: none"> • Unknown etiology; some cases are associated with inherited, inflammatory, and infectious diseases
<p>SYMPTOMS/SIGNS</p> <p>Symptoms</p> <ul style="list-style-type: none"> • Often asymptomatic and incidentally diagnosed • Infrequently mild blurred vision to severe reduced vision <p>Signs</p> <ul style="list-style-type: none"> • Coarse pigmentary clumps • Fine pigmentary changes along the retinal veins • Bone spicule-like pigmentation • Adjacent chorioretinal atrophy • Findings can be unilateral or bilateral and asymmetric 	<p>Diagnosis</p> <ul style="list-style-type: none"> • Numerous diagnostic tests can be considered, including to evaluate for potential underlying causes (i.e., inflammatory or inherited retinal disease) <ul style="list-style-type: none"> ◦ Fundus Autofluorescence (FAF) ◦ Fundus Fluorescein Angiography (FFA) ◦ Indocyanine Green Angiography (ICG) ◦ Electroretinogram (ERG) ◦ Optical Coherence Tomography (OCT) ◦ OCT Angiography (OCTA)

References

1. Strupaitė R, Ambrozaitytė L, et al. X-Linked Juvenile Retinoschisis: Phenotypic and Genetic Characterization. *Int J Ophthalmol*. 2018;11(11):1875-1878. doi: 10.18240/ijo.2018.11.22.
2. Sergeev YV, Vitale S, et al. Molecular modeling indicates distinct classes of missense variants with mild and severe XLRS phenotypes. *Hum Mol Genet*. 2013;22(21):4756-4767. doi: 10.1093/hmg/ddt323.
3. Molday RS, Kellner U, et al. X-linked juvenile retinoschisis: clinical diagnosis, genetic analysis, and molecular mechanisms. *Prog Retin Eye Res*. 2012;31(3):195-212. doi: 10.1016/j.preteyeres.2011.12.002.
4. Hu QR, Huang LZ, et al. Genetic analysis and clinical features of X-linked retinoschisis in Chinese patients. *Sci Rep*. 2017;7:44060. doi: 10.1038/srep44060.
5. Tolun G, Vijayarathy C, et al. Paired octamer rings of retinoschisin suggest a junctional model for cell-cell adhesion in the retina. *Proc Natl Acad Sci USA*. 2016;113(19):5287-5292. doi: 10.1073/pnas.1519048113.
6. Reid SN, Yamashita C, et al. Retinoschisin, a photoreceptor-secreted protein, and its interaction with bipolar and Muller cells. *J Neurosci*. 2003;23(14):6030-6040.
7. Chen J, Xu K, et al. Novel mutations of the RS1 gene in a cohort of Chinese families with X-linked retinoschisis. *Mol Vis*. 2014;20:132-139.
8. Thobani A, Fishman GA. The use of carbonic anhydrase inhibitors in the retreatment of cystic macular lesions in retinitis pigmentosa and X-linked retinoschisis. *Retina*. 2011;31(2):312-315. doi: 10.1097/IAE.0b013e3181e587f9.
9. Apushkin MA, Fishman GA. Use of dorzolamide for patients with X-linked retinoschisis. *Retina*. 2006;26(7):741-745. doi: 10.1097/01.iae.0000237081.80600.51.
10. Scruggs B, Chen C, et al. Efficacy of topical brinzolamide in children with retinal dystrophies. *Ophthalmic Genet*. 2019;40(4):350-358. doi: 10.1080/13816810.2019.1660381.
11. Turgut B, Demirel S, et al. Topical carbonic anhydrase inhibitors in the long-term treatment of juvenile X-linked retinoschisis. *Retina*. 2013;33(10):2131-2136. doi: 10.1097/IAE.0b013e3182a2cf05.
12. Cukras C, Wiley HE, et al. Retinal AAV8-RS1 Gene Therapy for X-Linked Retinoschisis: Initial Findings from a Phase I/IIa Trial by Intravitreal Delivery. *Mol Ther*. 2018;26(9):2282-2294. doi: 10.1016/j.ymthe.2018.05.025.
13. Pennesi ME, Yang P, et al. Intravitreal Delivery of rAAV2tYF-CB-hRS1 Vector for Gene Augmentation Therapy in Patients with X-Linked Retinoschisis: 1-Year Clinical Results. *Ophthalmol Retina*. 2022;6(12):1130-1144. doi: 10.1016/j.oret.2022.06.013.

Suggested citation format:

Gehrke E, Strampe MR, Drack AV. **Diagnosis and management of Juvenile X-linked Retinoschisis (JXLR)**. EyeRounds.org. Posted December 5, 2023; Available from <https://EyeRounds.org/cases/354-Morning-Glory-Disc-Anomaly.htm>

last updated: 12/05/2023

Image Permissions:

 [Creative Commons](https://creativecommons.org/licenses/by-nc-nd/3.0/)

Ophthalmic Atlas Images by [EyeRounds.org](https://www.eyerounds.org/), [The University of Iowa](https://www.theuniversityofiowa.edu/) are licensed under a [Creative Commons Attribution-NonCommercial-NoDerivs 3.0 Unported License](https://creativecommons.org/licenses/by-nc-nd/3.0/).

University of Iowa
Roy J. and Lucille A. Carver College of Medicine
Department of Ophthalmology and Visual Sciences
200 Hawkins Drive
Iowa City, IA 52242

Support Us

Copyright © 2019 The University of Iowa. All Rights Reserved

[Report an issue with this page](#)

[Web Privacy Policy](#) | [Nondiscrimination Statement](#)

Follow



[Receive notification of new cases, sign up here](#)

[Contact Us](#)

[Submit a Suggestion](#)