



DOI: 10.14744/eur.2025.60362
Eur Eye Res 2025;5(3):276–279

EUROPEAN
EYE
RESEARCH

CASE REPORT

Uncommon association: Bilateral astrocytic hamartoma in retinitis pigmentosa

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Abstract

Astrocytic hamartomas are benign glial tumors originating from the retinal nerve fiber layer. While commonly associated with tuberous sclerosis and neurofibromatosis, they can also coexist with retinitis pigmentosa (RP).

A 48-year-old RP patient presented for a routine examination with no systemic complaints. Best-corrected visual acuity was 0.8 bilaterally. Fundus examination revealed bone spicules, vascular attenuation, and elevated, nodular, cream-colored masses over both optic discs. Fundus autofluorescence showed hyperautofluorescent masses, while B-scan ultrasonography revealed hyperechogenicity with posterior shadowing. Optical coherence tomography angiography demonstrated a mass with moth-eaten cavities. Neurology and nephrology evaluations found no systemic pathology. Based on these findings, the lesions were suspected to be astrocytic hamartomas. No changes were observed over one year of follow-up.

The coexistence of RP and astrocytic hamartomas is rare. When optic disc masses are detected in RP patients, this possibility should be considered, and differential diagnoses should include optic disc drusen, meningioma, hemangioma, and papilledema.

Keywords: Astrocytic hamartoma; retinitis pigmentosa; optic nerve head drusen.

Retinitis pigmentosa (RP), a hereditary fundus dystrophy characterized by retinal photoreceptor degeneration, primarily affects rods and progressively leads to vision loss. Commonly associated ocular pathologies include posterior subcapsular cataract, glaucoma, myopia, optic nerve head drusen, and vitreous opacities.^[1] While most RP-related tumors are vasoproliferative, astrocytic hamartomas have also been reported.^[2]

Retinal astrocytic hamartomas are benign glial tumors arising from astrocytes in the retinal nerve fiber layer, occurring from the optic disc to the retinal periphery. They are commonly associated with tuberous sclerosis, less

often with neurofibromatosis type 1, and can also appear incidentally in healthy individuals.^[3]

In this study, we report findings related to an astrocytic hamartoma that was detected coincidentally on the optic disc of both eyes of a patient with RP.

Case Report

A 48-year-old female patient visited us for a routine check-up. We learned that she had been diagnosed with RP and had a history of hypertension and goiter. There are no known cases of RP, tuberous sclerosis, or neurofibromatosis type 1 in her family.



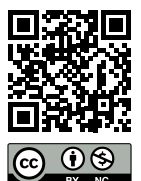
Cite this article as: Maden S, Kurt E, Erdogan M. Uncommon association: Bilateral astrocytic hamartoma in retinitis pigmentosa. *Eur Eye Res* 2025;5(3):276–279.

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Submitted Date: 03.03.2025 **Revised Date:** 27.05.2025 **Accepted Date:** 24.07.2025 **Available Online Date:** 17.12.2025

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During the medical examination, the best-corrected visual acuity was 20/25 (Snellen chart) bilaterally. In the biomicroscopic examination, a posterior subcapsular cataract was seen in both eyes. Fundus examination detected bone spicule pigmentary changes at the retinal midperiphery, along with creamy white, semitranslucent, elevated, mulberry-like lesions at the optic disc in both eyes (Fig. 1a, 1b).

In the optical coherence tomography (OCT) images (Optovue, Inc., Fremont, CA, USA), central macular thinning was observed in both eyes (Fig. 2a, 2b). Fundus autofluorescence (FAF) (Zeiss, Visucam, Oberkochen, Germany) imaging revealed hyperautofluorescent masses over the optic discs in both eyes (Fig. 1c, 1d). On B-scan ultrasonography (USG) (Ellex, Eye Cubed, Ohio, USA), hyperechogenicity causing posterior shadowing was noted. Optical coherence tomography angiography (OCTA) and radial OCTA imaging (Optovue, Inc., Fremont, CA, USA) of the optic disc demonstrated worm-eaten spaces within the masses (Fig. 2e, 2f and Fig. 2c, 2d). Full-field ERG showed significantly reduced rod responses, while cone function remained relatively preserved, a pattern typically seen in patients with RP (Fig. 3).

The patient was carefully evaluated by relevant specialties to assess the possibility of underlying systemic conditions such as tuberous sclerosis and neurofibromatosis. No characteristic skin findings of tuberous sclerosis, such as hypomelanotic macules or facial angiofibromas, were observed on dermatological examination. Similarly, neuroimaging revealed no signs of subependymal nodules or cortical tubers, and abdominal imaging showed no renal angiomyolipomas or cystic lesions. In the assessment for neurofibromatosis type 1, hallmark features, including café-au-lait spots, axillary or inguinal freckling, subcutaneous neurofibromas, optic gliomas, and Lisch nodules, were not present. Taken together, the absence of these clinical, dermatologic, and radiologic findings made a systemic syndromic diagnosis unlikely in this case.

In light of these findings, we conclude that the imaging of the mass on the optic disc is consistent with bilateral astrocytic hamartoma. The patient was placed under follow-up, and no changes in ophthalmologic findings were observed during a one-year follow-up period.

Discussion

Retinal astrocytic hamartomas are benign glial tumors that can arise anywhere from the optic disc to the peripheral retina.^[3] While commonly associated with phacomatoses such as neurofibromatosis and tuberous sclerosis, they may also occur in RP.^[4] Their appearance can mimic optic disc

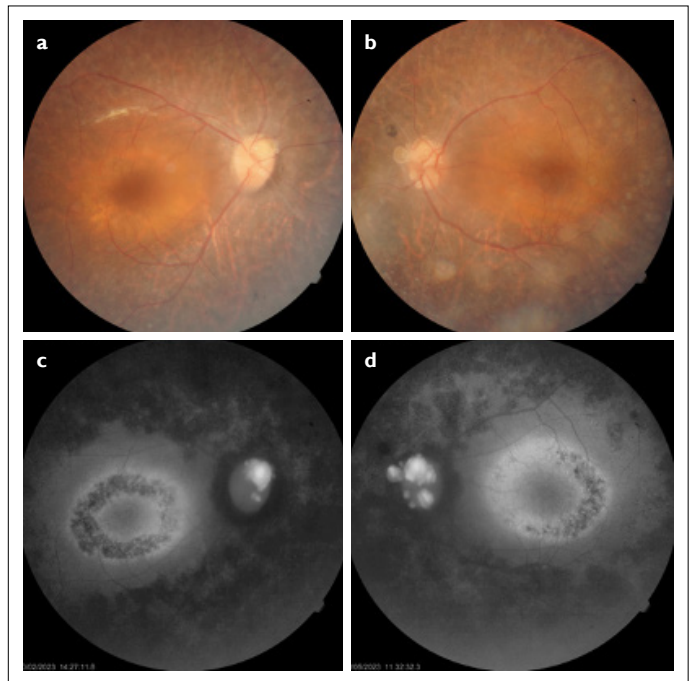


Fig. 1. (a, b) Color fundus: vessel narrowing is evident, along with creamy white, semitranslucent, elevated, mulberry-like lesions observed at the optic disc. (c, d) Fundus autofluorescence: masses exhibiting hyperautofluorescence on the optic disc.

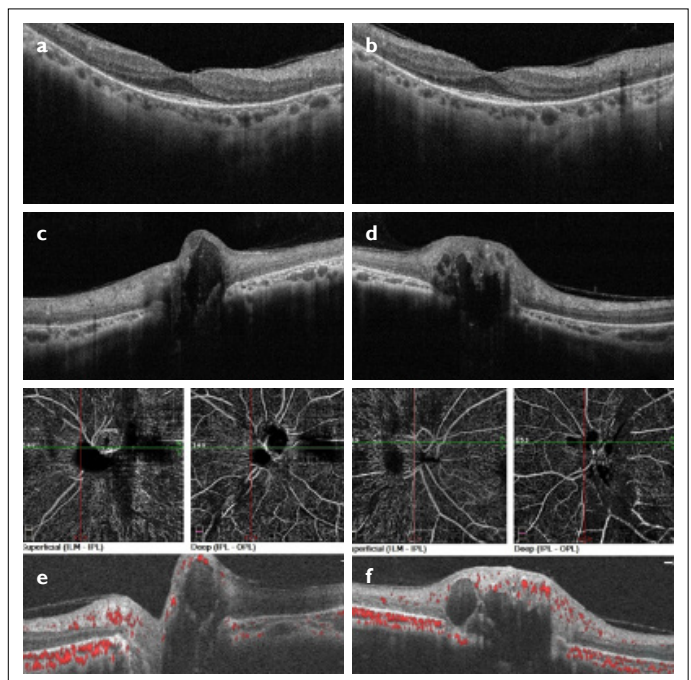


Fig. 2. (a, b) OCT: Macular thinning in both eyes. (c, d) OCTA radial: Moth eaten optically empty spaces. (e, f) In OCT angiography images of the 4.5 x 4.5 optic discs in the superficial plexus, no blood flow was detected within the tumors bilaterally.

head meningioma, hemangioma, papilledema, combined retinal hamartoma, or optic nerve head drusen (ONHD), which is frequently linked to RP.^[5]

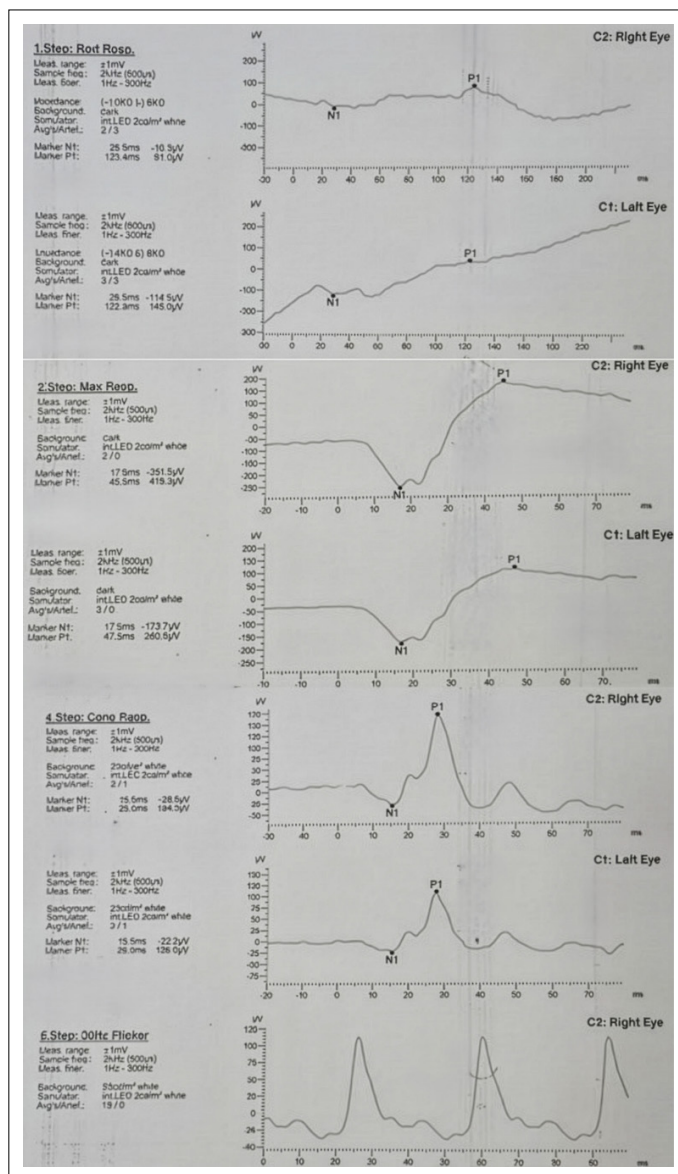


Fig. 3. Full-field ERG shows reduced scotopic responses with preserved photopic function, indicating selective rod dysfunction consistent with RP

ONHD consists of calcified materials originating in the nerve fiber layer of the optic nerve head, while disc hamartomas involve glial cell proliferation with calcification on the optic disc and peripapillary area.^[6] OCT findings of ONHD include RNFL thickening, scleral canal narrowing, prelaminar hyperreflective lines, central hyporeflexive, and peripheral hyperreflective oval mass-like structures (PHOMS).^[7]

Multimodal imaging methods such as B-scan USG, FAF, OCT, OCTA, and fluorescein angiography are used in the differential diagnosis of optic disc hamartomas.^[8]

Previous studies suggested that mass-like structures on the optic disc in RP patients were predominantly ONHD. However, advancements in multimodal imaging have

indicated that some cases may represent astrocytic hamartoma.^[5] Loukinaou et al. reported a 15-year-old RP patient initially diagnosed with ONHD, later reclassified as astrocytic hamartoma following lesion changes during a two-year follow-up.^[6] Similarly, Kinori et al. described a 24-year-old RP patient with multiple optic disc clusters, ultimately identified as an astrocytic hamartoma upon advanced imaging.^[9]

In our case, an astrocytic hamartoma was incidentally detected. We utilized multimodal imaging techniques in the differential diagnosis. The absence of OCT findings typical of ONHD, such as prelaminar hyperreflective lines, scleral canal narrowing, and PHOMS, supported the diagnosis of astrocytic hamartoma. Based on fundus imaging and OCT findings, the lesion was consistent with a type 3 astrocytic hamartoma.^[10] The optic disc lesions might have been misdiagnosed as ONHD in earlier assessments, possibly due to immature clinical features of astrocytic hamartoma.

RP patients should be closely monitored for mass development and timely diagnosis. Mass-like lesions on the optic disc require multimodal imaging for differential diagnosis. Although rare, astrocytic hamartomas in RP patients are often mistaken for ONHD. If imaging is inconclusive, regular follow-up is essential to confirm the diagnosis and address potential complications, such as vitreous hemorrhage, macular exudation, retinal detachment, epiretinal membrane, and visual field defects.

Conclusion

In conclusion, the detection of astrocytic hamartomas in RP patients should prompt careful differential diagnosis, especially considering the overlap in imaging features with ONHD. The use of advanced multimodal imaging techniques can help clarify the diagnosis and avoid misinterpretations. Regular monitoring of RP patients with optic disc lesions is crucial for detecting any changes and addressing potential complications that may arise over time.

Informed Consent: Written informed consent was obtained from the patient for the preparation of this work.

Peer-review: Externally peer-reviewed.

Author Contributions: Concept: E.K., M.E.; Design: E.K., M.E.; Supervision: E.K., M.E.; Resource: S.M.M.; Materials: S.M.M.; Data Collection and/or Processing: S.M.M., M.E.; Analysis and/or Interpretation: S.M.M., M.E.; Literature Search: S.M.M.; Writing: S.M.M., M.E.; Critical Reviews: E.K.

Conflict of Interest: None declared

Use of AI for Writing Assistance: Not declared.

Financial Disclosure: The authors declared that this study has received no financial support.

Acknowledgments: The authors would like to thank the patient for her consent and cooperation.

This case report was previously presented as a poster at the 57th National Congress of the Turkish Ophthalmological Association, held in Antalya from November 8-12.

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