Opening the Pandora Box of Retinal Dystrophies: Is it Malattia Leventinese?

Abrindo a Caixa de Pandora das Distrofias Retinianas: Será Malattia Leventinese?

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Recebido/Received: 2025-07-25 | **Aceite/Accepted:** 2025-08-19 | **Published online/Publicado online:** 2025-09-17 | **Published/Publicado:** 2025-09-29

DOI: https://doi.org/10.48560/rspo.42426

KEYWORDS: Choroidal Neovascularization; Optic Disk Drusen; Retinal Dystrophies.

PALAVRAS-CHAVE: Distrofias Retinianas; Drusas do Disco Óptico; Neovascularização de Coroide.

The photograph refers to a 50-year-old Indian male with a history of a retinal dystrophy that presents at our center with complaints of a sudden decrease in best corrected visual acuity (20/200) of his right eye and metamorphopsia. Slit lamp examination and Intraocular pressure measurement were unremarkable. Fundoscopic examination revealed massive hard drusen and hyperpigmentation in the macular region, bilaterally. On the right eye, one could observe an extensive premacular hemorrhage measuring 1-disc diameter both vertically and horizontally. Optical coherence tomography (OCT) revealed hyperreflective bumpy thickening of the retinal pigment epithelial (RPE)–Bruch's membrane complex, as well as areas with subretinal fluid in the right eye, suggesting the presence of a choroidal neovascular membrane (CNVM).

Initially described by Doyne in 1899, Doyne honeycomb dystrophy/malattia leventinese is a rare genetic inherited retinal dystrophy characterized by an autosomal dominant mutation in the *EFEMP1* gene. Patients can remain asymptomatic for decades. OCT can reveal focal dome-shaped, saw-tooth, or diffuse hyper-reflective deposits with elevation between the RPE and Bruch's membrane, as well as hypo-reflective fluid from a corresponding CNVM.

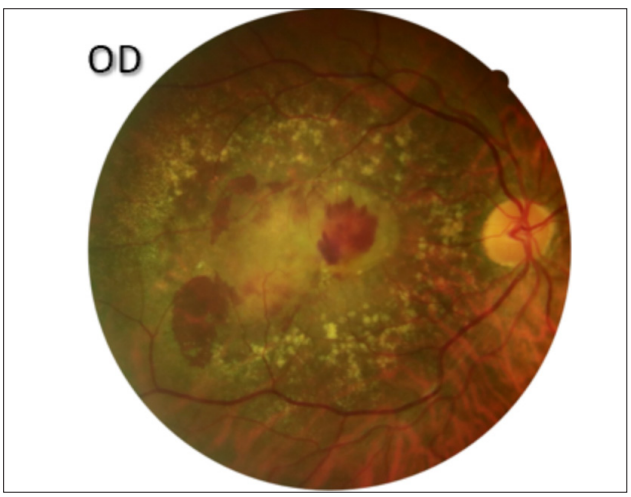

Figure 1. Color fundus photography showing premacular haemorrhage (OD).

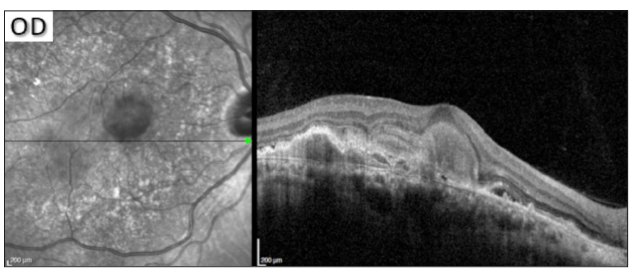

Figure 2. OCT showing revealing hyperreflective bumpy thickening of the retinal pigment epithelial (RPE)–Bruch's membrane complex.

CONTRIBUTORSHIP STATEMENT / DECLARAÇÃO DE CONTRIBUIÇÃO

All authors participated significantly in the conception, writing, critical revision, and final approval of the article.

Todos os autores participaram de forma significativa na conceção, redação, revisão crítica e aprovação final do artigo.

RESPONSABILIDADES ÉTICAS

Conflitos de Interesse: Os autores declaram a inexistência de conflitos de interesse na realização do presente trabalho.

Fontes de Financiamento: Não existiram fontes externas de financiamento para a realização deste artigo.

Confidencialidade dos Dados: Os autores declaram ter seguido os protocolos da sua instituição acerca da publicação dos dados de doentes.

Consentimento: Consentimento do doente para publicação obtido.

Proveniência e Revisão por Pares: Não comissionado; revisão externa por pares.

ETHICAL DISCLOSURES

Conflicts of Interest: The authors have no conflicts of interest to declare.

Financing Support: This work has not received any contribution, grant or scholarship.

Confidentiality of Data: The authors declare that they have followed the protocols of their work center on the publication of patient data.

Patient Consent: Consent for publication was obtained.

Provenance and Peer Review: Not commissioned; externally peer-reviewed.

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