HIV and ocular syphilis in a patient with retinitis pigmentosa

HIV e sífilis ocular em paciente com retinose pigmentar

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How to cite:
Souza AC, Ávila CO, Carvalho DA, Santos PR, Salomão HN. HIV and ocular syphilis in a patient with retinitis pigmentosa. Rev Bras Oftalmol. 2026;85:e0022.

doi:
https://doi.org/10.37039/1982.8551.20260022

Keywords:
Retinitis pigmentosa; Ocular syphilis; HIV infections; Highly active antiretroviral therapy; Diagnostic imaging

Descritores:
Retinite pigmentar; Sífilis ocular; Infecções por HIV; Terapia antirretroviral de alta atividade; Diagnóstico por imagem

ABSTRACT

The objective of this study was to report a case of retinitis pigmentosa in a patient with HIV and ocular syphilis, highlighting the complexity of diagnosis and treatment in overlapping systemic and ocular conditions. A descriptive, qualitative analysis of the patient's clinical evolution was conducted using medical records and patient interviews. A literature review was performed through PubMed® and SciELO. Data collection followed ethical standards, with informed consent obtained. Treatment with antiretroviral therapy and specific antibiotics for syphilis led to improved visual acuity, reduced VDRL titers (1:128 to 1:32), and immunological recovery. The case raised the suspicion of Usher syndrome due to the patient's clinical features. This case reinforces the importance of interdisciplinary collaboration between ophthalmologists, infectious disease specialists, and other professionals. A timely and targeted therapeutic approach resulted in favorable outcomes, demonstrating that coordinated care is essential in managing patients with co-infections and chronic or hereditary ocular diseases.

RESUMO

O objetivo deste estudo foi relatar um caso de retinose pigmentar em um paciente com HIV e sífilis ocular, destacando a complexidade do diagnóstico e do tratamento na sobreposição de condições sistêmicas e oculares. Foi realizada uma análise descritiva e qualitativa da evolução clínica do paciente usando prontuários médicos e entrevistas com o paciente. Foi realizada uma revisão da literatura por meio da PubMed® e da SciELO. A coleta de dados seguiu padrões éticos, com obtenção de consentimento informado. O tratamento com terapia antirretroviral e antibióticos específicos para sífilis levou à melhora da acuidade visual, à redução dos títulos de VDRL (1:128 para 1:32) e à recuperação imunológica. O caso levantou a suspeita de síndrome de Usher devido às características clínicas do paciente. Este caso reforça a importância da colaboração interdisciplinar entre oftalmologistas, especialistas em doenças infecciosas e outros profissionais. Uma abordagem terapêutica oportuna e direcionada gerou resultados favoráveis, demonstrando que o cuidado coordenado é essencial no manejo de pacientes com coinfecções e doenças oculares crônicas ou hereditárias.

Received on:
August 2,2025

Accepted on:
November 20, 2025

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Conflict of interest:
no conflict of interest.

Financial support:
no financial support for this work.

Data Availability Statement:
The datasets generated and/or analyzed during the current study are included in the manuscript

Associate editor:
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INTRODUCTION

The retina, a fundamental neurosensory structure for vision, can be affected by a variety of degenerative and infectious conditions, with significant impacts on visual acuity and visual field. Among these, retinitis pigmentosa stands out as a progressive hereditary dystrophy that initially affects rod cells and, in advanced stages, cone cells, resulting in progressive blindness. Its prevalence is estimated to range from 1:3,000 to 1:5,000 individuals, with clinical manifestations that include night blindness (nyctalopia), peripheral scotomas, and loss of visual field.

Ocular syphilis, known as the "great imitator", can manifest at any stage of infection, affecting various ocular structures. The increase in syphilis incidence in recent decades is a global concern, especially in vulnerable populations. In Brazil, there was a 74% increase in cases between 2015 and 2021, with a notable rise in HIV coinfections.[1] In this context, HIV-associated immunodeficiency compromises the inflammatory response and alters the natural course of syphilis, favoring atypical presentations.

The concomitance of retinitis pigmentosa and ocular syphilis in an immunocompromised patient represents a highly complex clinical challenge. The overlap between a chronic degenerative condition and an infectious inflammatory condition demands a prompt, precise, and integrative medical approach with multidisciplinary support. Furthermore, the presence of sensorineural hearing loss since childhood raises the hypothesis of Usher syndrome, a rare autosomal recessive genetic disorder characterized by the association of hearing dysfunction and retinitis pigmentosa.

Given the scarcity of reports addressing the intersection of these three conditions – retinitis pigmentosa, ocular syphilis, and HIV infection – this study aims to contribute to the literature by thoroughly describing a rare clinical case, highlighting diagnostic and therapeutic aspects and the role of imaging technologies. It also seeks to reflect on the importance of differential diagnosis and visual rehabilitation in contexts involving multiple chronic comorbidities.

This is a case report with a qualitative and descriptive approach, based on clinical and documentary analysis of a patient diagnosed with retinitis pigmentosa and coinfection with HIV and syphilis. The main objective was to describe the diagnostic and therapeutic challenges encountered, emphasizing the relevance of an interdisciplinary approach and continuous monitoring in patients with associated ophthalmologic and systemic conditions.

The participant was included at the end of the ophthalmologic appointment, in a private setting, respecting the ethical principles of autonomy, voluntariness, and confidentiality, in accordance with Brazilian Resolution CNS No. 466/2012. The Informed Consent Form was duly signed, authorizing access to the medical record, complementary tests, and clinical images, with approval from the Research Ethics Committee (REC) of the Universidade Federal de Catalão (approval No. 7.540.777, CAAE No. 83888024.7.0000.0164).

All sensitive information was anonymized, following the guidelines of Circular Letter No. 166/2018 – Comissão Nacional de Ética em Pesquisa/ Secretaria-Executiva do Conselho Nacional de Saúde/ Ministério da Saúde (Conep/SECNS/MS). The clinical images used in this article were expressly authorized by the patient.

Data collection was carried out during the first half of 2024, complemented by a comprehensive literature review. Inclusion criteria were articles published between 2005 and 2024, in Portuguese or English, addressing clinical, diagnostic, and therapeutic aspects of retinitis pigmentosa, ocular syphilis, and HIV coinfection. Exclusion criteria included duplicate studies, outdated narrative reviews, and publications lacking peer review.

The literature search was conducted using the PubMed®/MEDLINE®, SciELO, Cochrane Library, Google Scholar, *Biblioteca Virtual em Saúde* (BVS), and EBSCOhost databases. Health Sciences Descriptors (DeCS/MeSH) used were "Ocular Syphilis", "HIV", "Retinitis Pigmentosa", "Infectious Uveitis", and "Coinfection", combined with Boolean operators AND and OR to refine results. The time frame (2005 to 2024) was chosen due to technological advances in diagnostic tools and antiretroviral therapies over the past two decades.

CLINICAL CASE

In March 2023, a 38-year-old man, a sign language interpreter with hearing loss since age 4, sought ophthalmologic care due to progressive bilateral blurred vision that began in June 2022. He also reported peripheral vision loss and having difficulty seeing in low-light environments (nyctalopia), which worsened with prolonged light exposure in the studio. He had no complaints of eye pain, photophobia, or hyperemia. A preliminary diagnosis of toxoplasmosis had been suggested in January 2023 by a low vision specialist, with negative immunoglobulin M (IgM) and positive immunoglobulin G (IgG) serology. Treatment was discontinued due to diagnostic uncertainty. The patient reported a family history of retinitis pigmentosa (three paternal cousins).

Initial ophthalmologic examination showed visual acuity of counting fingers at 1 meter in the right eye and

20/40 in the left, with peripheral visual field loss. Pupils were isochoric, with a relative afferent pupillary defect in the right eye. Extraocular motility was preserved. Slit-lamp biomicroscopy revealed conjunctival hyperemia, a deep anterior chamber with 3+/4+ cell reaction, atrophic iris, and anterior vitritis graded 2-3+ in the right eye and 1+ in the left. Fundoscopy showed a waxy pale optic disc, hyporreflective macula, vascular attenuation, and peripheral bone spicule pigmentation.

The wide field retinography revealed changes consistent with retinal dystrophy: optic disc pallor, reduced macular sheen, and diffuse peripheral spots with pigment mobilization (Figure 1A). Optical coherence tomography (OCT) revealed hyperreflective spots in the inner retina, collapse of retinal layers, and disruption of the ellipsoid zone, indicating overlap between dystrophy and inflammatory process[2] (Figure 1B). Additionally, with the scan passing through the fovea, there is a collapse of the inner retinal layers in the mid-periphery, with a central thickness of 288 µm in the right eye (OD) and 297 µm in the left eye (OS), along with ellipsoid zone disruption and hyperreflective dots. The images support a pattern of rod-cone retinal dystrophy, showing retinal thinning in the mid-peripheral region. Furthermore, an inflammatory-infectious process is observed in the central retina (Figure 2).

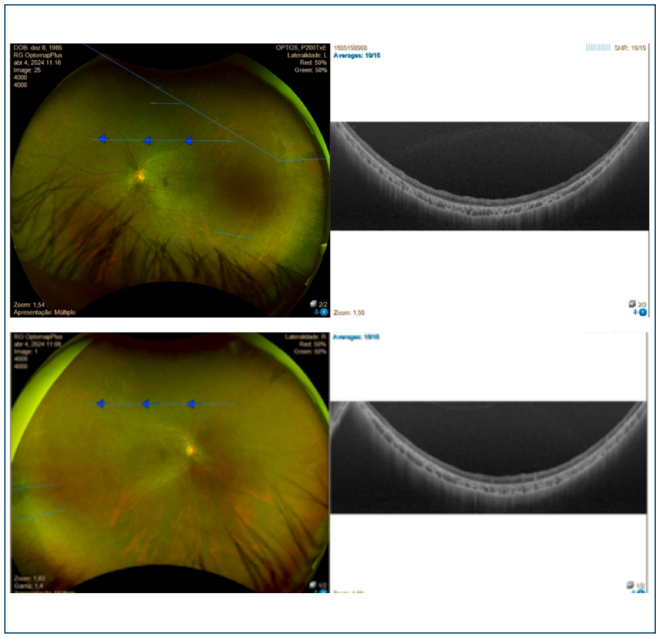


Figure 1. (A) Wide-Field Retinography. Performed with the Optos Daytona (ultra-widefield) equipment, showing optic disc pallor, vascular attenuation, and diffuse peripheral spots, findings consistent with retinal dystrophy. Image obtained with Optos Daytona, ultra-widefield mode. (B) Optical coherence tomography. Obtained with Spectralis Heidelberg engineering, horizontal raster mode (25 lines), highlighting hyper-reflective spots in the inner retina, disruption of the ellipsoid line, and collapse of the inner retinal layers. Exam performed with Spectralis Heidelberg, horizontal raster protocol (25 lines).

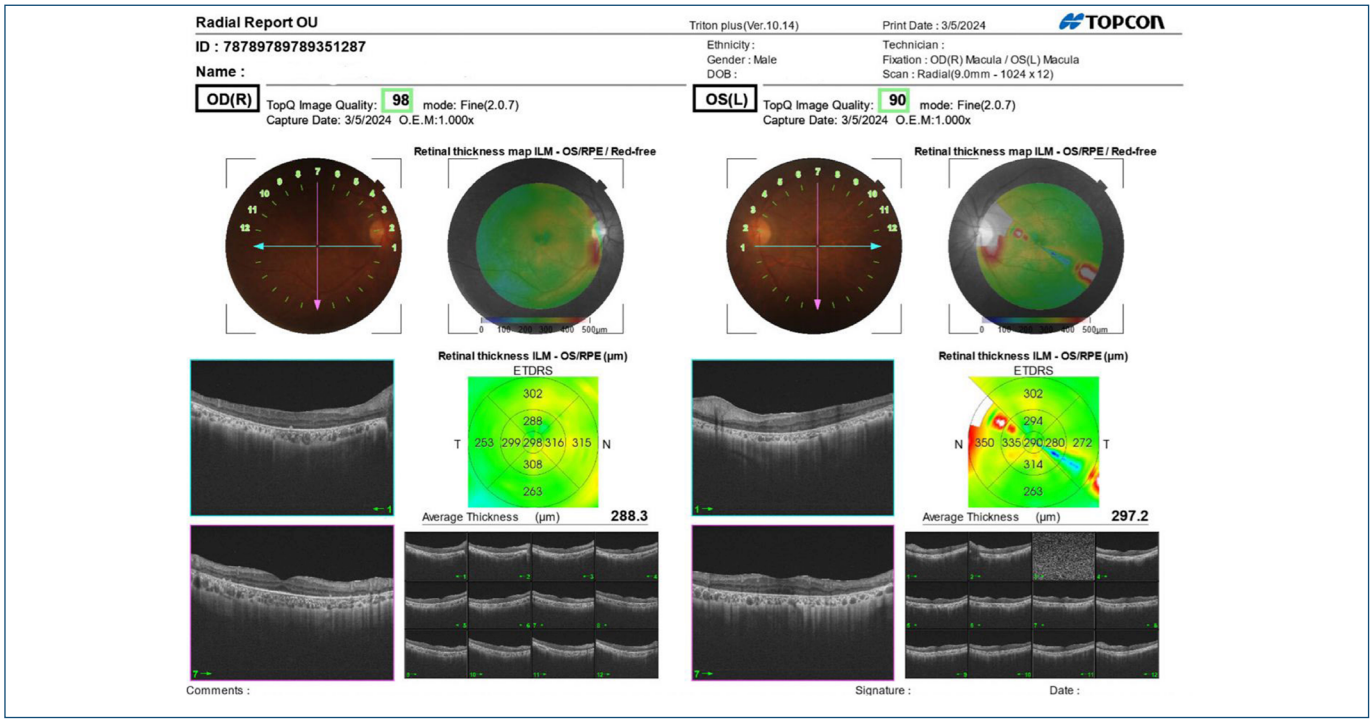


Figure 2. Macular optical coherence tomography with a scan passing through the fovea. Image captured with the autofluorescence module of Spectralis Heidelberg, showing areas of hypofluorescence with a peripheral hyperfluorescence halo, indicative of photoreceptor apoptosis. Image captured with the BluePeak module from Heidelberg Engineering.

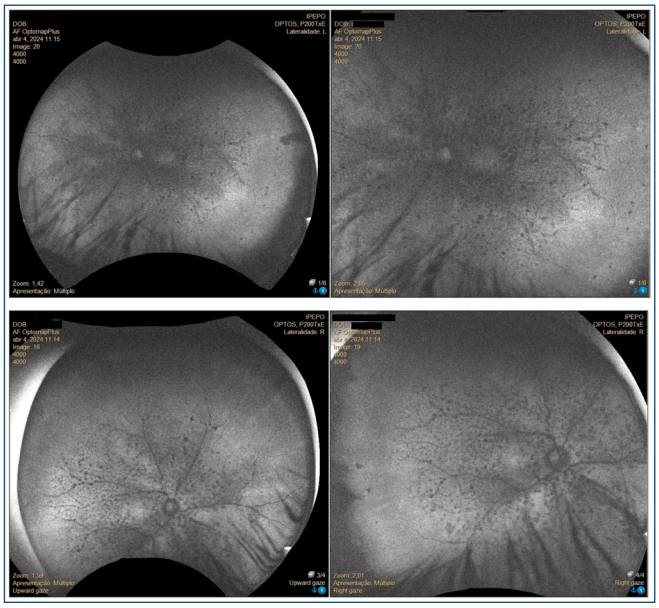


Figure 3. Fundus autofluorescence.

Fundus autofluorescence revealed hypofluorescent areas with a hyperfluorescent halo, suggesting photoreceptor loss and peripheral retinal stress[2] (Figure 3).

Automated perimetry (24-2) showed bilateral tubular visual fields, a typical pattern of hereditary dystrophies[4] (Figure 4).

Based on clinical history, findings, and imaging, serological testing for syphilis and HIV was performed. Both were positive, with a VDRL titer of 1:128. The initial viral load was 140,000 copies/mL (P6L32), with a CD4 count of 473 cells/mm³ (Figure 5). Treatment included antiretroviral therapy (ART) with dolutegravir, lamivudine, and tenofovir, as well as ceftriaxone 4g/day for 14 days, in accordance with evidence supporting its effectiveness in immunocompromised patients[5]. Prednisolone acetate eye drops were also prescribed to control uveitis.

After 15 days, visual acuity improved to 20/20 in both eyes and the VDRL titer decreased to 1:32. Peripheral visual field loss persisted. Six months later, visual acuity remained stable, with immunologic control (CD4 count increase) and no recurrence. The patient reported significant improvement in quality of life. They remain under regular follow-up, using lubricating eye drops and undergoing periodic examinations (OCT, autofluorescence, perimetry, and electroretinogram). Regular appointments with an infectious disease specialist were scheduled for antiretroviral regimen adjustments.

Table 1 presents the clinical timeline of key events during the patient's follow-up.

The hypothesis of Usher syndrome was considered given the history of sensorineural hearing loss and

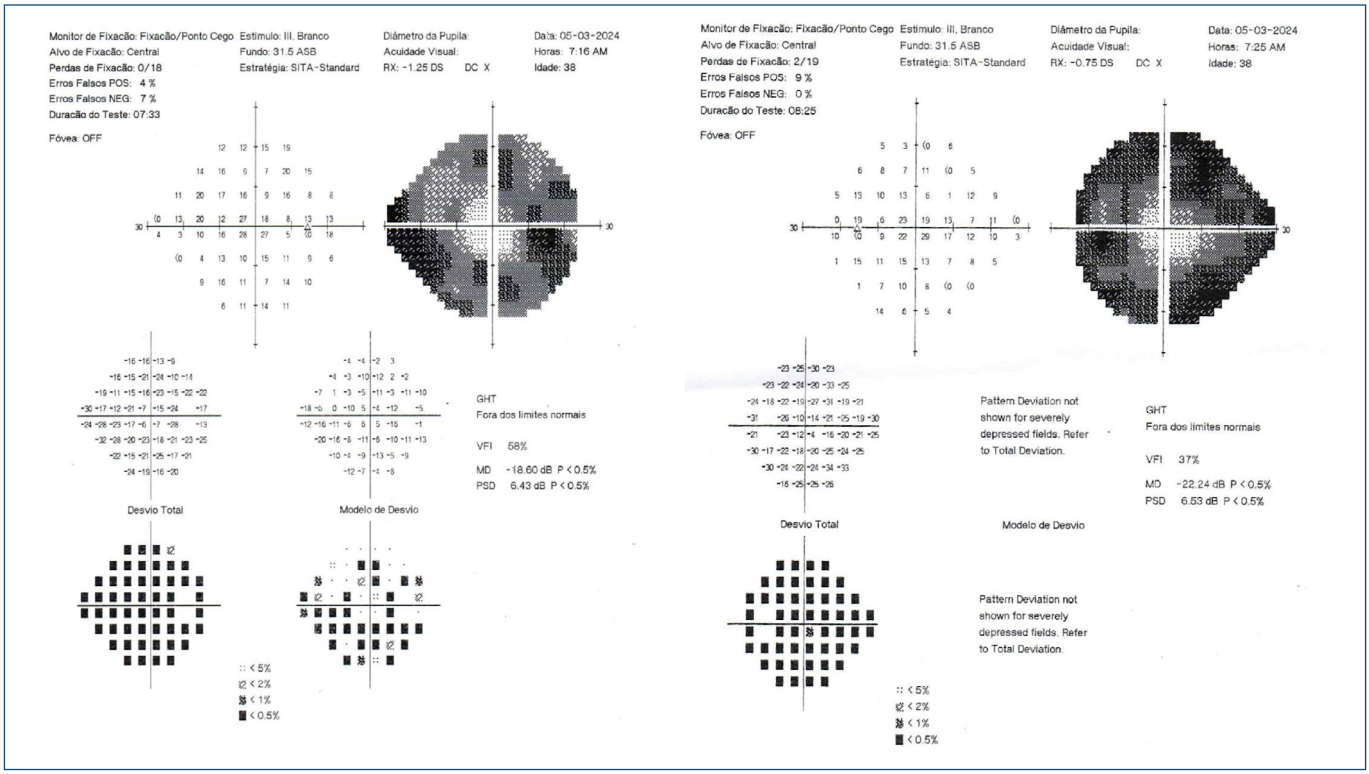
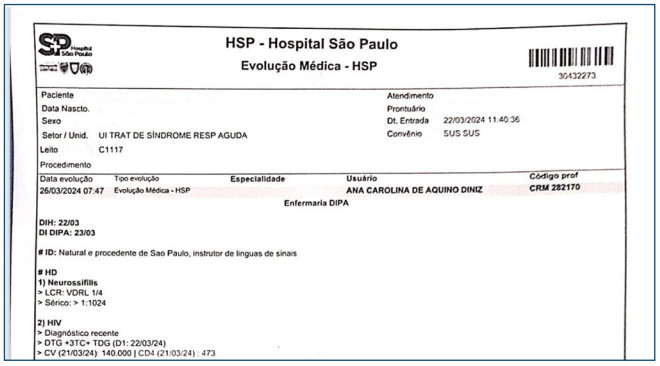


Figure 4. 24-2 Computerized perimetry. Exam performed with the Humphrey Field Analyzer II-i, showing a bilateral tubular visual field pattern with marked peripheral loss, typical of hereditary dystrophies. Exam performed on the Humphrey Field Analyzer II-i (Carl Zeiss Meditec).



HSP - Hospital São Paulo
Evolução Médica - HSP
30432273

Paciente
Data Nascto.
Sexo
Setor / Unid. UI TRAT DE SÍNDROME RESP AGUDA
Leito C1117
Procedimento
Atendimento
Prontuário
Dt. Entrada 22/03/2024 11:40:36
Convênio SUS SUS

Data evolução	Tipo evolução	Especialidade	Usuário	Código prof
26/03/2024 07:47	Evolução Médica - HSP		ANA CAROLINA DE AQUINO DINIZ	CRM 282170

Enfermaria DIPA

DIH: 22/03
DI DIPA: 23/03

ID: Natural e procedente de Sao Paulo, instrutor de linguas de sinais

HD
1) Neurossifilis
> LCR: VDRL 1/4
> Sérico: > 1:1024

2) HIV
> Diagnóstico recente
> DTG +3TC+ TDG (D1: 22/03/24)
> CV (21/03/24) 140.000 | CD4 (21/03/24) : 473

Figure 5. Record of hospital medical progress issued by a public reference institution for infectious diseases. The document confirms the diagnoses of neurosyphilis (CSF Venereal Disease Research Laboratory test 1:4; serum Venereal Disease Research Laboratory test >1:1024) and HIV infection with a high viral load (140,000 copies/mL) and a CD4 count of 473 cells/mm³. Notably, the initiation of antiretroviral therapy and intravenous ceftriaxone treatment in a unit for acute respiratory syndrome management is reported.

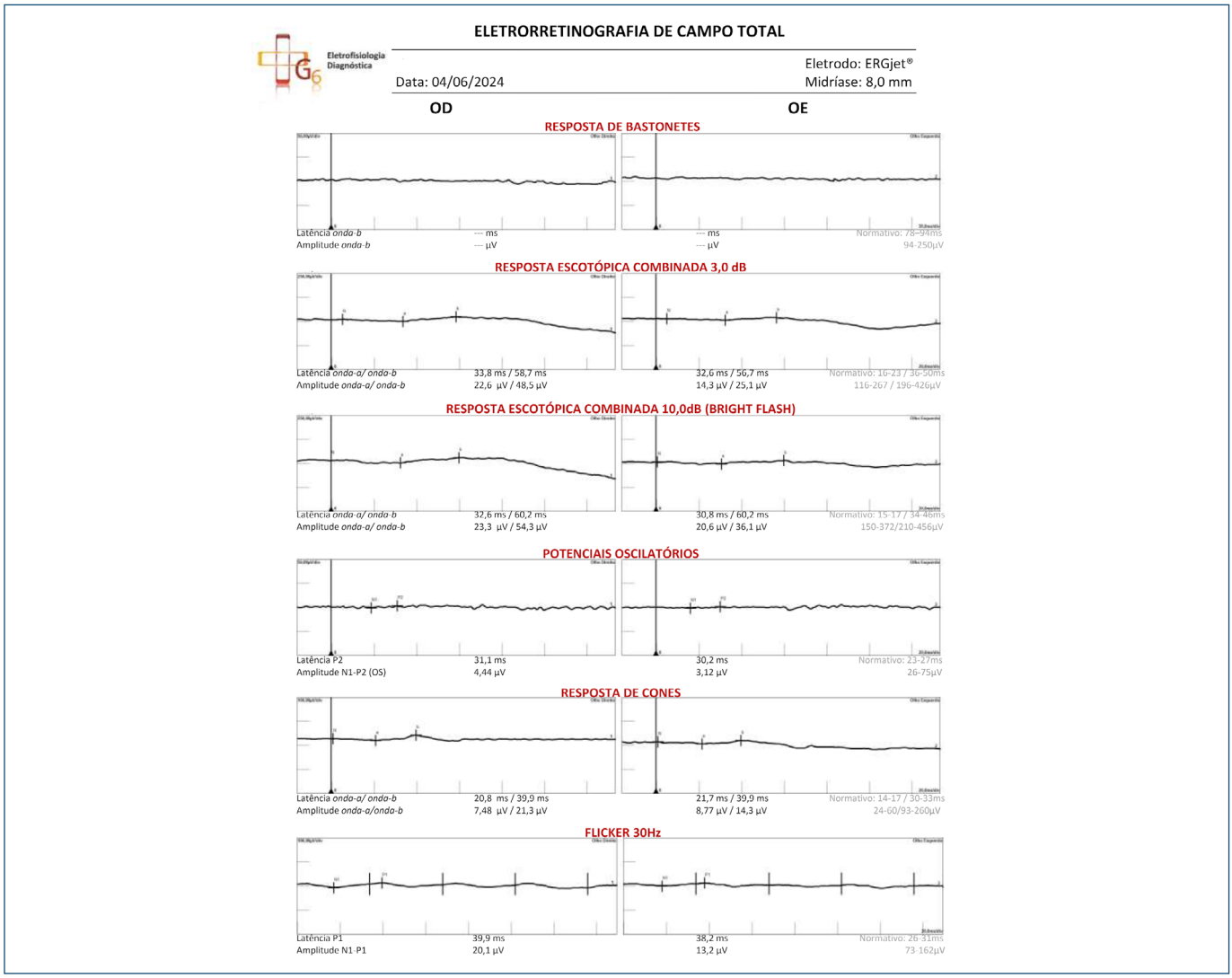
Table 1. Clinical timeline of the patient

Date	Clinical event
June 2022	Onset of visual blurring and nyctalopia
January 2023	Diagnostic hypothesis of toxoplasmosis with IgM-/IgG+
March 2023	Initial ophthalmologic consultation and diagnostic tests
April 2023	Start of treatment with ceftriaxone and ART
May 2023	Improvement in visual acuity and decrease in VDRL to 1:32
October 2023	Clinical stabilization and immunological control
November 2023	Scheduled genetic investigation for Usher syndrome

IgM: immunoglobulin M; IgG: immunoglobulin; ART: antiretroviral therapy; VDRL: Venereal Disease Research Laboratory test.

retinitis pigmentosa, and a genetic investigation was scheduled for diagnostic confirmation.

The diagnosis of retinitis pigmentosa was established through a functional (electrophysiological) examination, specifically the full field electroretinogram, which revealed a diffuse reduction in retinal function in both eyes, with greater impairment of the rod system findings consistent with a rod dystrophy, generally classified as retinitis pigmentosa (Figure 6).



ELETRORRETINOGRAFIA DE CAMPO TOTAL

Eletrofisiologia Diagnóstica

Data: 04/06/2024
Eletrodo: ERGjet®
Midríase: 8,0 mm

	OD	OE	
RESPOSTA DE BASTONETES			
Latência *onda-b*	--- ms	--- ms	Normativo: 78-94ms
Amplitude *onda-b*	--- µV	--- µV	94-250µV
RESPOSTA ESCOTÓPICA COMBINADA 3,0 dB			
Latência *onda-a/ onda-b*	33,8 ms / 58,7 ms	32,6 ms / 56,7 ms	Normativo: 16-23 / 30-50ms
Amplitude *onda-a/ onda-b*	22,6 µV / 48,5 µV	14,3 µV / 25,1 µV	116-267 / 196-426µV
RESPOSTA ESCOTÓPICA COMBINADA 10,0dB (BRIGHT FLASH)			
Latência *onda-a/ onda-b*	32,6 ms / 60,2 ms	30,8 ms / 60,2 ms	Normativo: 15-17 / 34-46ms
Amplitude *onda-a/ onda-b*	23,3 µV / 54,3 µV	20,6 µV / 36,1 µV	150-372/210-456µV
POTENCIAIS OSCILATÓRIOS			
Latência P2	31,1 ms	30,2 ms	Normativo: 23-27ms
Amplitude N1-P2 (OS)	4,44 µV	3,12 µV	26-75µV
RESPOSTA DE CONES			
Latência *onda-a/ onda-b*	20,8 ms / 39,9 ms	21,7 ms / 39,9 ms	Normativo: 14-17 / 30-33ms
Amplitude *onda-a/onda-b*	7,48 µV / 21,3 µV	8,77 µV / 14,3 µV	24-60/93-260µV
FLICKER 30Hz			
Latência P1	39,9 ms	38,2 ms	Normativo: 26-31ms
Amplitude N1-P1	20,1 µV	13,2 µV	73-162µV

Figure 6. Full field electroretinogram.

Considering the clinical and functional evidence, as well as financial limitations that precluded complete genetic sequencing, we decided not to perform genetic testing at this time. It is important to highlight that functional testing holds diagnostic value equivalent to or even greater than genetic testing in cases of retinitis pigmentosa, since the same clinical phenotype may be caused by different genetic variants, including some not yet cataloged as pathogenic. This case illustrates the importance of precise diagnosis, timely treatment, and multidisciplinary follow-up in patients with concomitant hereditary and infectious ophthalmologic conditions.

DISCUSSION

This report aims to describe the clinical progression of an HIV-positive patient with coinfection of ocular syphilis and retinitis pigmentosa, evaluate the efficacy of intravenous ceftriaxone as an alternative to crystalline penicillin for ocular neurosyphilis, demonstrate the value of advanced imaging tests (OCT, autofluorescence, and perimetry) for therapeutic monitoring, and highlight the importance of genetic diagnosis and a multidisciplinary approach in retinal dystrophies.

After combined therapy, central visual acuity was restored, but peripheral field loss persisted, consistent with irreversible photoreceptor damage in advanced stages of retinitis pigmentosa.[2,3] OCT revealed disruption of the ellipsoid zone and collapse of inner retinal layers, confirming active inflammation related to ocular syphilis.[4]

In recent decades, syphilis rates among people living with HIV have risen alarmingly,[1] and comparative trials have indicated that ceftriaxone 2 g IV/day for 14 days is equivalent to crystalline penicillin 18 to 24 MU/day in treating ocular neurosyphilis.[5,6] For retinitis pigmentosa, OCT and fundus autofluorescence are already standard tools for early structural change detection and prognosis evaluation.[2,3]

These findings support an integrated protocol that includes: annual serologic screening for syphilis in HIV-positive patients; biannual repetition of OCT, autofluorescence, and perimetry to monitor retinal damage; intravenous ceftriaxone use when crystalline penicillin is unavailable; early referral for genetic counseling in suspected cases of Usher syndrome; and coordinated involvement of ophthalmology, infectious disease, genetics, and visual and auditory rehabilitation teams to maximize functional autonomy.[2-6]

Ceftriaxone offers benefits comparable to penicillin without requiring prolonged hospitalization in specialized centers.[5,6] However, delayed diagnosis or poor ART adherence may perpetuate ocular inflammation and increase reinfection risk, worsening peripheral vision loss.[1-6] Retinitis pigmentosa maintains a progressive course, requiring continuous rehabilitation, and the inadvertent use of corticosteroids under an initial suspicion of toxoplasmosis could have worsened ocular syphilis.[1-6]

Professionals managing HIV-positive patients with visual complaints should routinely request syphilis serology, initiate intravenous ceftriaxone (2 g/day for 14 days) when crystalline penicillin is not viable, and implement biannual monitoring using OCT, autofluorescence, and perimetry. It is essential to ensure adherence to antiretroviral therapy, reinforce sexually transmitted infection (STI) prevention counseling, provide referrals to genetic services when signs of Usher syndrome or other retinal dystrophies are present, and integrate visual and auditory rehabilitation teams to optimize quality of life and functionality.

Finally, it is acknowledged that clinical overlap with other infectious uveitis led to delayed diagnosis and possibly greater retinal damage. The absence of molecular confirmation limited the genetic characterization of retinitis pigmentosa, and the short follow-up period does not allow for long-term progression or reinfection rate assessment, which indicates the need for prospective multicenter studies.

This report contributes to the literature by highlighting the importance of differential diagnosis in patients with retinal dystrophies, especially in atypical presentations in immunocompromised individuals. The overlap between retinitis pigmentosa, ocular syphilis, and HIV infection underscores the clinical complexity and the need for an integrated approach that considers not only ophthalmologic aspects but also systemic, infectious, and genetic factors.

Additionally, the case points to the importance of investing in research aimed at gene therapies for hereditary dystrophies such as Usher syndrome. Such innovative approaches may, in the future, change the natural course of these diseases. Personalized medicine, based on genetic and immunological profiles, represents a promising frontier in clinical ophthalmology.

ACKNOWLEDGMENTS

We thank patient JMS for their collaboration, willingness, and trust in sharing their clinical journey for scientific purposes, enabling deeper understanding of the conditions addressed. We also express our gratitude to Dra. Alline

Carvalho de Souza, volunteer at the Neuro-ophthalmology Department of the *Universidade Federal de São Paulo*, Dra. Heloísa Nascimento Salomão, volunteer at the Uveitis and Tumor Department of the same institution, and Professor Patrícia Roberta dos Santos, faculty member at the *Zarns*, whose clinical contributions and technical guidance were essential for case management and development of this report. We further thank the Neuro-ophthalmology Department of the *Universidade Federal de São Paulo* for their technical support, as well as the ophthalmic imaging laboratory team for performing and maintaining the exams that supported the analyses presented here.

AUTHOR'S CONTRIBUTION

Souza AC and Ávila CO contributed to the conception and design of the study, data analysis and interpretation, drafting of the manuscript, and critical revision of its content. Carvalho DA and Santos PR contributed to data acquisition and analysis, interpretation of the results, drafting of the manuscript, and critical revision of its content. Salomão HN contributed to data acquisition, initial drafting of the manuscript, and critical revision of its content. All authors approved the final version of the manuscript and agree to take responsibility for all aspects of the work, ensuring accuracy and integrity in every part of the research.

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