Choroideremia: a case report

Guilherme Thomé de Carvalho[1]; Bruno Vasconcelos Coimbra[1]; Daniella Pedra Vasconcellos[1]; Tamíres Able Carmona[1]; Vitor José Gonçalves Martins[1]; Ricardo Andreotti Paulo de Oliveira[1]; Patrick Ferreira da Rocha[1]; Kelma Macedo Pohlmann Simoes[2]

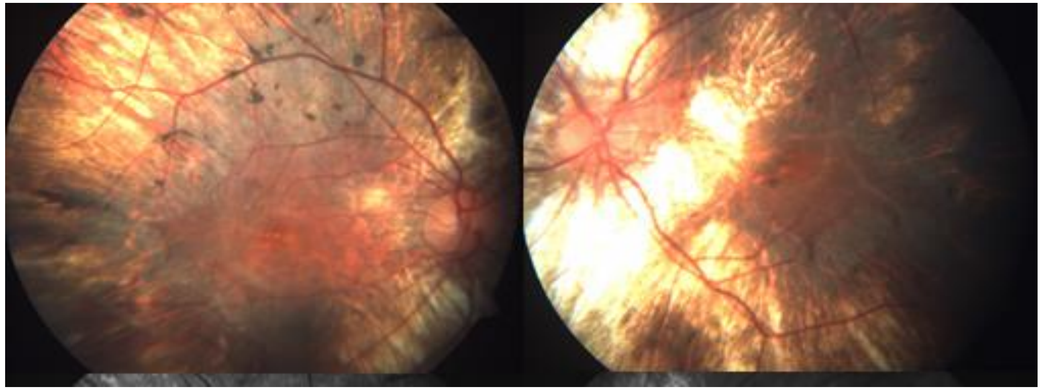
[1]Hospital Universitário Pedro Ernesto – UERJ; [2]Hospital Universitário Gaffrée e Guinle – UNIRIO – RJ

Purpose

This study aims to elucidate the clinical manifestations, genetic underpinnings, and diagnostic challenges of Choroideremia (CHM), a rare X-linked recessive retinal dystrophy[1], through the case study of a 15-year-old male patient. It also explores the current landscape of treatment options and the potential of gene therapy for CHM.

Methods

A comprehensive diagnostic approach was employed, including detailed clinical examination, genetic testing to identify mutations in the CHM gene, and advanced imaging techniques like Optical Coherence Tomography (OCT) to assess retinal degeneration. The study also reviews current literature on CHM to contextualize findings within the broader research field.



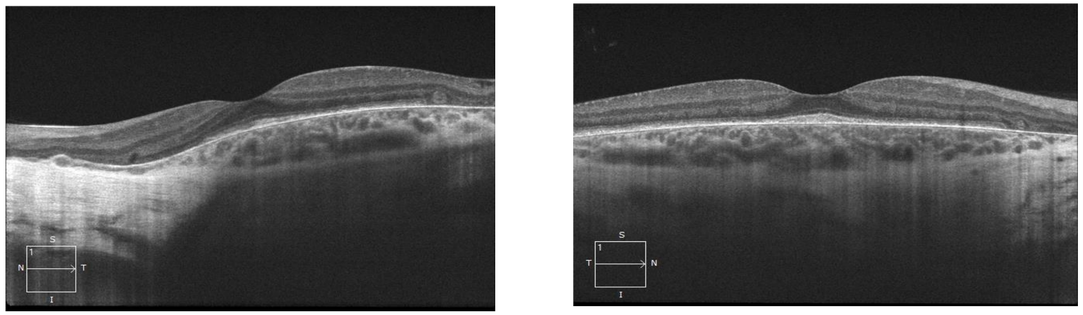
Retinography showing irregular areas of chorioretinal degeneration in the middle periphery and areas of hyperplasia of the retinal pigment epithelium in the vascular arcades

Results

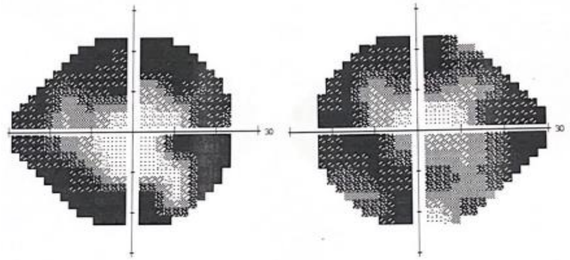
The patient exhibited typical CHM symptoms, including progressive night blindness and peripheral vision loss, with genetic testing confirming a mutation in the CHM gene. Imaging revealed extensive retinal degeneration, aligning with CHM's known pathophysiology. However, the study highlights the lack of effective treatments for CHM, with management strategies currently limited to symptomatic relief and low-vision aids.

Discussion

The case study underscores the urgent need for research into curative treatments for CHM, particularly gene therapy, which holds promise given the disorder's genetic basis[3]. It also emphasizes the importance of early diagnosis and regular monitoring to manage disease progression and optimize patient quality of life. The study calls for increased awareness among clinicians and researchers to facilitate early detection and encourage innovation in CHM treatment strategies.



Optical coherence tomography thinning of the outer retina, except in the foveal area. Presence of external retinal tubulations due to retinal injury



Computerized perimetry demonstrating generalized constriction of the visual field in both eyes

References

1 - Khan KN, Islam F, Moore AT, Michaelides M. Clinical and genetic features of choroideremia in childhood. Ophthalmology. 2016;123:2158–65.

2 - Aleman TS, Han G, Serrano LW, et al. Natural history of the central structural abnormalities in choroideremia: a prospective cross-sectional study. Ophthalmology 2017;124:359–373

3 - Mitsios A, Dubis AM, Moosajee M. Choroideremia: from genetic and clinical phenotyping to gene therapy and future treatments. Ther Adv Ophthalmol. 2018. 10 2515841418817490–2515841418817490. doi: 10.1177/2515841418817490