CHOROIDEREMIA: HEREDITARY CHORIORETINAL DYSTROPHY

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PURPOSE

Choroideremia is a progressive and diffuse degeneration of the choroid, RPE and photoreceptors. Inheritance is X-linked and is caused by mutation of the CHM gene. The report aims to highlight the importance of recognizing the pathology in order to improve the development of research aimed at gene therapy as a possible treatment in the future.

METHODS

This case report was obtained by literary review of scientific articles and evaluation of the patient's medical record.

RESULTS

Patient, male, 42 years old, reports low peripheral vision, especially at night for one year. There is no ophthalmological and personal history. On ophthalmologic examination, visual acuity (with correction) is 20:100 in both eyes. Fundoscopy shows atrophy of the RPE and choroid. Fluorescein angiography shows hypofluorescence with intact fovea and surrounded by window defect hyperfluorescence.

DISCUSSION

Choroideremia manifests in the first decade of life with the development of nyctalopia. Men are predominantly affected; however, women can be asymptomatic carriers. In affected men, the degeneration manifests primarily as mottled areas of pigmentation anterior to the equator and macula. The areas gradually degenerate to confluent shell-shaped areas by loss of RPE and choriocapillaris, but with preservation of the large vessels of the choroid. Changes on fluorescein angiography are even more pronounced, with cupped areas due to the lack of choriocapillaris showing hypofluorescent areas.