RETINA 2023 — 47th BRAVS MEETING
APRIL 28th to MAY 1st
WINDSOR BARRA HOTEL
RIO DE JANEIRO - BRAZIL

COMBINED HAMARTOMA OF THE RETINA AND RETINAL PIGMENT EPITHELIUM

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RETINA CURITIBA
CENTRO DE TRATAMENTO ESPECIALIZADO

IRMANDADE DA SANTA CASA DE MISERICÓRDIA DE CURITIBA 1852

PURPOSE

To report a clinical case about Combined Hamartoma of the Retina and Retinal Pigment Epithelium.

METHODS

Medical records, fluorescein angiography (FA) and Optical Coherence Tomography (OCT) analysis.

RESULTS

A 22-year-old man referred to the medical retina specialist for investigation of progressive vision loss in the right eye showed best corrected visual acuity of 20/70 on the right and 20/20 on the left eye. There was no other significant past medical history or family history of any phakomatoses or neurofibromatosis. Fundus examination of the right eye revealed a pigmented and elevated lesion located juxtapapillary and proliferative vitreoretinopathy over the optic nerve. FA revelled a blocked hypofluorescent area juxtapapillar, containing a hyperfluorescent spot. OCT presented epiretinal membrane (ERM). Further investigations were recommended to exclude neurofibromatosis type 2. The patient keeps routine follow-up semesterly and did not report any new or acute symptoms.

DISCUSSION

Combined hamartoma of the retina and retinal pigment epithelium (CHR-RPE) is an uncommon fundus benign tumor habitually diagnosed in young children with symptoms of painless decreased VA and strabismus. The tumor is usually presented as a unilateral solitary pigmented and elevated lesion. Vascular tortuosity and lipid exudate are frequently present, ERM formation is commonly associated with the disease. The majority of CHR-RPE occurs sporadically, most patients do not have systemic diseases. However, it has been reported to occur in association with neurofibromatosis Types 1 and 2, tuberous sclerosis and Gorlin syndrome. CHR-RPE patients may have neurofibromatosis investigation. CHR-RPE should be excluded in all young patients with epiretinal membrane or vitreomacular adhesion. Visual loss over time is still expected with this tumor. Surgery for associated ERM is still a subject of debate since visual acuity may not improve despite membrane removal. Patients should be monitored for decrease in vision, choroidal neovascularization, ERM formation, vitreous hemorrhage, and neovascularization periodically if non-surgical treatment is stablished.

IMAGES

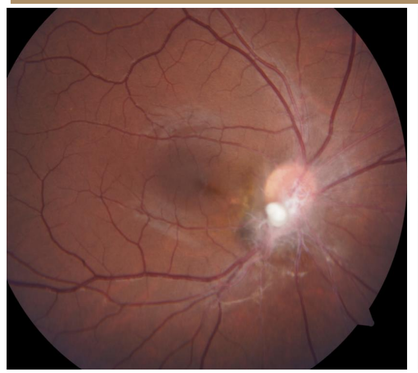

Image 1: Colour fundus photography

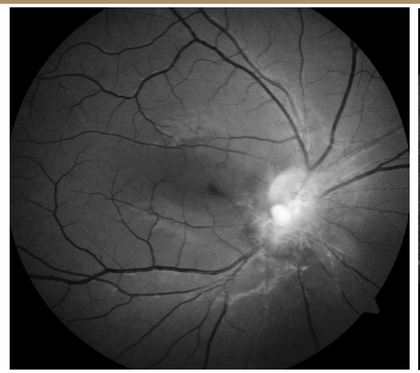

Image 2: Red free

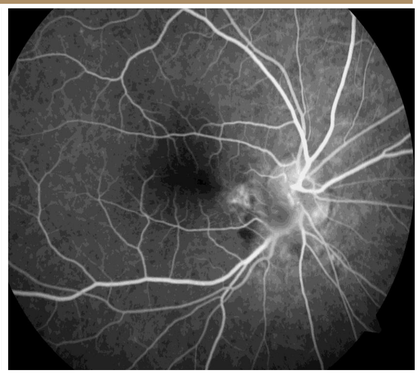

Image 3: Fluorescein angiography

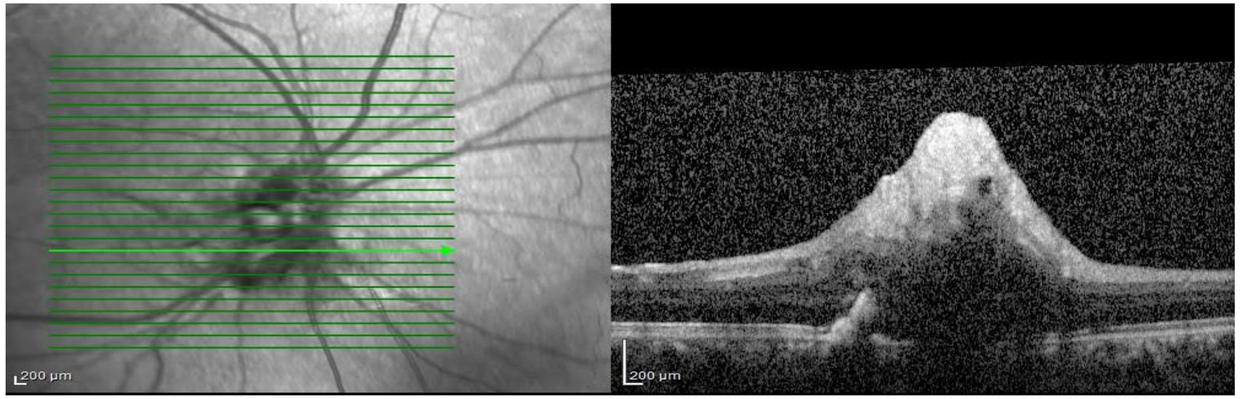

Image 3: Optical Coherence Tomography

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