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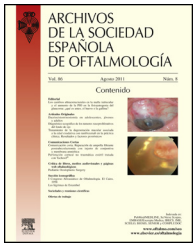

Short communication

Charcot–Marie–Tooth disease and bilateral vitritis☆

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ABSTRACT

Case report: We describe a patient diagnosed with Charcot–Marie–Tooth disease, with a 4 months history of bilateral decreased visual acuity and floaters. On examination, he had severe bilateral vitreous opacity and sectoral diffuse vascular sheathing. It could not be linked to some underlying etiology and did not respond to oral steroids.

Conclusions: Publications relating to ocular findings in patients with Charcot–Marie–Tooth disease exclude bilateral vitritis. In this case we were unable to test the association with another disease as the cause of vitritis.

Enfermedad de Charcot-Marie-Tooth y vitritis bilateral

Palabras clave:
Enfermedad de Charcot Marie Tooth
Polineuropatía motora-sensitiva
Vitritis bilateral
Vitrectomía diagnóstica
Vitritis no infecciosa

RESUMEN

Caso clínico: Se describe el caso de un paciente diagnosticado con la enfermedad de Charcot-Marie-Tooth que presentó disminución de la agudeza visual bilateral y miodesopsias de 4 meses de evolución. Se encontró una opacidad vítrea bilateral muy severa y un envainamiento vascular sectorial difuso, que no pudo ligarse a ninguna etiología de base y que no respondió a esteroides orales.

Conclusiones: Las publicaciones de hallazgos oculares en pacientes con enfermedad de Charcot-Marie-Tooth no incluyen vitritis bilateral. No se pudo comprobar la asociación con otra enfermedad como causa de la vitritis.

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Introduction

The Charcot–Marie–Tooth (CMT) disease is a multi-sensitive polyneuropathy caused by alterations in the genes that produce the proteins related to the axon or the myelin layer, causing chronic segmented demyelinization of the peripheral nerves and hypertrophic changes caused by remyelinization. Ocular findings associated to CMT include pupil abnormalities, Leber optic neuropathy, ischemic neuropathy, retina pigment degeneration, optic atrophy and glaucoma. In addition, one case was described with intermediate uveitis although in the framework of a concomitant thyroid disease.[1–5]

Case report

A 47-year-old male visited the ophthalmological practice due to diminished visual acuity (VA) with onset 4 months before, with insidious and progressive development in both eyes simultaneously, accompanied by occasional myodesopsiae. The patient did not exhibit red eye clinic or refer pain, photophobia or other associated symptoms during development of the condition.

In addition, the patient did not refer relevant family history, recent travels, contact with animals, pulmonary or gastrointestinal conditions. However, in a specialized hospital he was recently confirmed the CMT disease diagnostic (Fig. 1).

Ocular exploration determined VA in both eyes of 20/400. Very severe 4+ bilateral vitreous opacity was observed, as well as glint (Fig. 2). The rest of the examination in both eyes was completely normal.

General analyses (hemogram, blood chemistry, etc.) were normal. Explorations for STORCH profile, toxocara, HIV, ANA, ANCA, anti-DNA, anticardiolipin antibodies, rheumatoid factor, C-reactive protein, VSG, ECA and HLA-B27 were all negative.

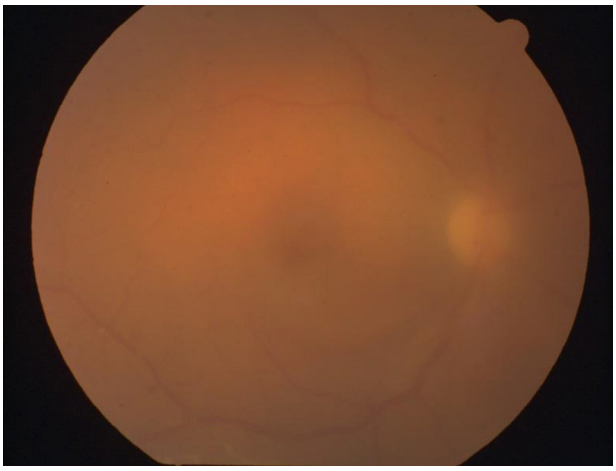

Fig. 2 – Photograph of right ocular fundus prior to vitrectomy. Severe vitreous opacity with glint and normal looking posterior pole. Optic nerve can be seen with well defined limits, free of edema or hemorrhage with both temporal vascular arches without hemorrhage or retinal inflammatory sites.

In addition to the tomographic and magnetic resonance findings in the CMT disease, CSF and imaging studies for abdomen, chest and cranium were normal.

In the absence of evidence of infection process, prednisone during 4 weeks was established without exhibiting improvement. Conventional 23G dry pars plana vitrectomy was considered first in the right eye without being able to process the sample in the lab. In addition to vitreous opacity, a segmented yellowish-whitish sheathing was

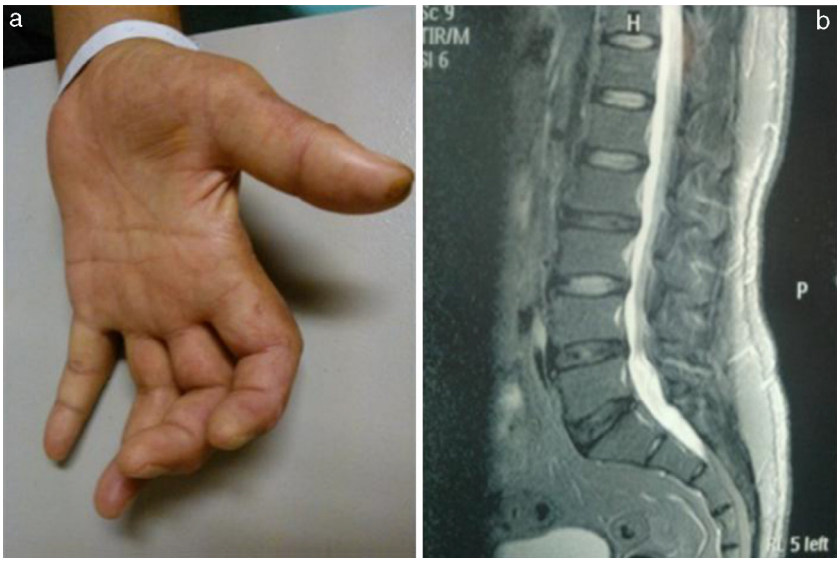

Fig. 1 – Some findings of CMT disease. (a) Claw hand. Note hyperextension of metacarpophalangeal joints and flexion of interphalangeal joints. (b) Lumbar sacral nuclear magnetic resonance, showing diffuse thickening of the horsetail and nerve roots.