Short communication

A combination of topical and systemic carbonic anhydrase in the treatment of chromosome X-linked retinoschisis

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ABSTRACT

Case report: A 17-year-old male patient, who since 2000, has referred to a progressive bilateral decrease in visual acuity. A “bicycle wheel” macula pattern was observed in his retina.

The electroretinogram showed a decrease in the b-wave amplitude. The visual evoked potentials were normal. Optical coherence tomography showed bilateral macular edema. All this supported the diagnosis of X-linked retinoschisis.

Discussion: Genetic counseling was given and the pattern of X-linked inheritance was explained.

A significant improvement of the macular thickness was observed after treatment with topical dorzolamide and oral acetazolamide.

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Combinación de la anhidrasa carbónica tópica y sistémica en el tratamiento de la retinosquisis ligada al cromosoma X

RESUMEN

Caso clínico: Varón de 17 años que desde el año 2000 refiere disminución progresiva de agudeza visual bilateral. Se observan en la retina máculas en patrón de “rueda de bicicleta”.

El electroretinograma informa de disminución en la amplitud de la onda b. Los potenciales evocados visuales son normales. La tomografía de coherencia óptica muestra edema macular bilateral. Todo ello compatible con el diagnóstico de retinosquisis ligada al cromosoma X (RLX).

Discusión: Se realiza consejo genético y se explica el patrón de herencia ligada al X.

Se instaura tratamiento con dorzolamida tópica y acetazolamida oral evidenciando una mejora significativa del grosor macular.

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Introduction

X chromosome-linked retinoschisis is a retinal dystrophy caused by mutation of the RS1 gene located in the Xp22.1 chromosome, leading to outer plexiform retina layer schisis; it primarily affects males and occasionally it is autosomal dominant.\(^1\)

It was linked for the first time to the X chromosome in 1913, and the term “X chromosome-linked retinoschisis” was used by Jaeger in 1953 along with juvenile retinoschisis.

The gene causing RS1, identified in 1997, provides instructions for producing a protein called retinoschisin.\(^2\)

Even when two individuals have the same RS1 causative mutation, there is a wide range in disease severity,\(^3\) without correlation between mutation type and disease severity or progression.

This disease may be detected years after birth and consists of star-shaped cystic changes in the fovea and retinoschisis in the periphery.

Peripheral retinoschisis occurs in 50% and carries an increased risk of retinal complications.

Diagnosis relies on clinical examination, microperimetry and electrophysiology with visual evoked potentials (VEP) and electroretinogram (ERG).

Our report aims to demonstrate the usefulness of carbonic anhydrase-inhibiting drugs for treating X chromosome-linked retinoschisis.

Case report

A 17-year-old male seen at the ophthalmology department for the first time in 2000 due to reduced visual acuity and headaches. No relevant family history. Corrected visual acuity is 0.5 in both eyes; binocular vision tests positive, normal biomicroscopy and mottled ocular fundus at foveal level (Fig. 1). The requested ERG shows slight decrease in b-wave amplitude in maximum or mixed response with white light. Cone test under photopic conditions and Flicker 30 Hz also show a slight decrease in b-wave amplitude. VEP are normal. Everything supports X chromosome-linked retinoschisis.

Subsequently, since 2010, a slight decrease occurred in visual acuity (0.3 in both eyes), and a color vision test (Ishihara) was normal. Ophthalmoscopic examinations show macules with “bicycle wheel” pattern without peripheral retinoschisis (Fig. 2), microperimetry with absolute scotoma in schisis areas and a central cystoid macular thickening (Panozzo E2 class), confirmed by optical coherence tomography (OCT) (Cirrus® HD-OCT 4000-4661 version 5.1.1.6), 407 μm in the right eye (OD) and 397 μm in the left eye (OS) (Fig. 3). Fluorescein angiography shows no intraretinal cyst filling (Fig. 4).

Genetic test is positive for RS1 mutation.

Therefore, topical treatment is implemented with dorzolamide eye drops 3 times daily and oral acetazolamide 500 mg daily; at 3 months slight improvement in visual acuity is detected in his OS changing to 0.4, and noticeable macular thickness reduction, i.e., 266 μm in OD and 269 μm in OS (Fig. 5).

Genetic counseling was provided, to be considered regarding his future offspring; a genetic test was requested for the rest of the family, which was negative for the mutation.

Discussion

Foveal retinoschisis in a male with b-wave reduction in ERG leads most likely to this diagnosis. This can be confirmed by molecular genetic tests. Retinoschisis usually stabilizes...
from adolescence, and sometimes in midlife, having varying degrees of unpredictable disease severity.

Differential diagnosis must be performed for Goldmann–Favre syndrome, acquired retinoschisis, pars planitis, retinal cysts, retinitis pigmentosa, Stargardt disease, cystoid macular edema and exudative retinal detachment, among others.

Many children suffering from this disease may benefit from refractive correction, reduced vision aids and educational support.

Genetic counseling should be provided to families with a member affected by X chromosome-linked retinoschisis, explaining the linked inheritance pattern and possible risks of transmission to future children. Likewise, look for possibly affected siblings who may have various degrees of severity.

The topical dorzolamide and systemic acetazolamide treatment prescribed to our patient produced changes in macular thickness, maintained over time, possibly due to resorption of foveal cysts, mainly by retinal pigment epithelial cells. Early diagnosis and treatment with carbonic anhydrase-inhibiting drugs, in the future, may represent a significant improvement for patients.

Fig. 3 – Macular OCT of both eyes. Note cystoid macular center thickening: 407 μm in OD and 397 μm in OS.

Fig. 5 – OCT after dorzolamide and acetazolamide prescribed treatment, with macular thickness of 266 μm in OD and 269 μm in OS.

Fig. 4 – Angiography at 1:30 min in both eyes showing that no filling occurs in intraretinal cysts.
Conflicts of interest

The authors declare that they have no conflicts of interest.

REFERENCES


