DOI 10.5935/0034-7280.20170018



# Optical coherence tomography and congenital retinoschisis: three case reports

## *Tomografia de coerência óptica e retinosquise congênita: relato de três casos*

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### *Abstract*

*Congenital retinoschisis is an X-linked recessive inherited disease. It causes the splitting of the retina's neurosensory layers from the remaining of the sensory retina, presenting itself as a "stellate" or "bicycle-wheel" maculopathy, vitreous hemorrhage and retinal detachment. We report three cases of congenital retinoschisis, two of them brothers. optical coherence tomography was used when evaluating the cases. It was impossible to differentiate retinoschisis from retinal detachment in one of the cases through optical coherence tomography due to lack of patient collaboration. We then performed laser photocoagulation to mark and follow-up the affected area.*


***Keywords:*** *Retinoschisis; Retinoschisis/congenital; Maculopathy; Retina/pathology; Retinal detachment; Case reports*


### Resumo

A retinosquise congênita é uma doença autossômica recessiva ligada ao X. Resulta em separação da camada de fibras nervosas do restante da retina sensorial, e manifesta-se como maculopatia estriada, hemorragias vítreas e descolamento de retina. Relatamos três casos de retinosquise congênita, sendo dois deles irmãos. Utilizou-se a tomografia de coerência óptica na avaliação dos casos. Em um, não foi possível efetuar o diagnóstico diferencial com descolamento de retina através da tomografia de coerência óptica, devido a não cooperação no exame, optando-se pela realização de fotocoagulação com laser para demarcação e seguimento da área.


**Descritores**: Retinosquise; Retinosquise/congênito; Maculopatia; Retina/patologia; Descolamento de retina; Relatos de casos

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Funding: No specific financial support was available for this study.
Disclosure of potential conflicts of interest: None of the authors have any potential conflicts of interest to disclose.

Recebido para publicação em 27/03/2016 - Aceito para publicação em 07/07/2016.

## Introduction

Congenital Retinoschisis is an X-linked recessive inherited disorder. It is defined by the splitting of the retina's neurosensory layers from the remaining of the sensory retina, being the most common cause of juvenile macular degeneration in men.[1-3] Affected patients may present strabismus, nystagmus, peripheral retinoschisis, vitreous hemorrhage, vitreous membranes, retinal detachment (RD), "stellate" or "bicycle-wheel" maculopathy. Optical coherence tomography (OCT) helps in the evaluation and differential diagnosis between congenital retinoschisis and RD. Treatment is based on management of complications. Congenital retinoschisis has an unpredictable prognosis, being reported cases of regression, stabilization and late progression.

**Case Reports**

**Case 1**

PBO, male, 11 years old, presented a diminished visual acuity in both eyes (oculus uterque - OU). Visual acuity was 20/200 in the right eye (OD) and 20/70 in the left eye (OS). Slit lamp biomicroscopy revealed peripheral retinoschisis in the inferotemporal quadrant reaching the inferotemporal vascular arcade and a pseudohole aspect maculoschisis OD (Figure 1). It also revealed small maculoschisis and peripheral vitreous membranes OS. (Figure 2)

OCT showed cysts on the macular area, which coalesced to a bigger cyst in the foveal area OD (Figure 3) and a symmetrical presentation OS. These findings were consistent with Bilateral Macular Retinoschisis (BMR)

Photocoagulation of the posterior edge of the right eye's Retinoschisis was performed to mark and follow-up the affected area. A control-OCT, performed one year later, showed no alterations. It was impossible to identify the marked area due to lack of patient cooperation. Ophthalmoscopy follow-up revealed no disease progression.

**Case 2**

EHCA, male, 23 years old, presented a diminished peripheral visual field and low visual acuity OD. Best corrected visual acuity (BCVA) levels of 20/200 OD and 20/30 OS. Ophthalmoscopy revealed vitreous membranes OU. Campimetry showed nonspecific alterations. OCT revealed BMR of symmetrical aspect OU. (Figure 4)

**Case 3**

LHCA, male, 15 years old, case 2 patient's brother, presented a diminished visual acuity OU. BVCA of 20/25 OU. Ophthalmoscopy revealed vitreous membranes, altered fovea appearance and beaten-bronze appearance of the macula. OCT showed BMR of symmetrical aspect OU. (Figure 5)

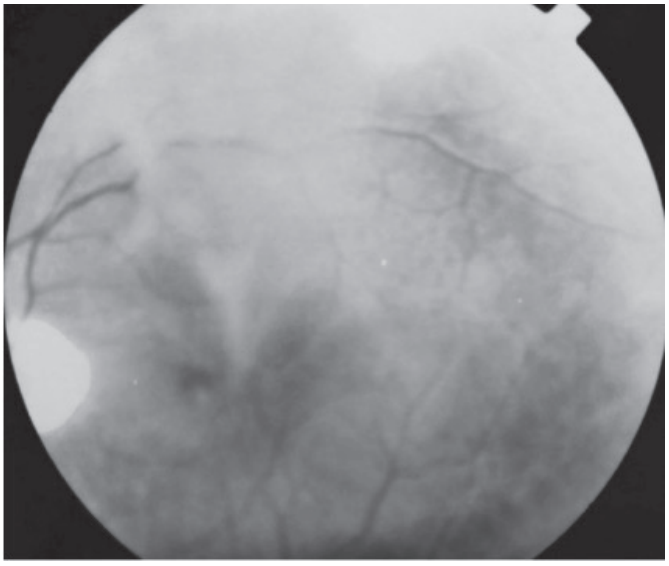

**Figure 1** - Fundography OD: Peripheral retinoschisis on the inferotemporal quadrant reaching the inferotemporal vascular arcade and maculoschisis with a pseudohole aspect

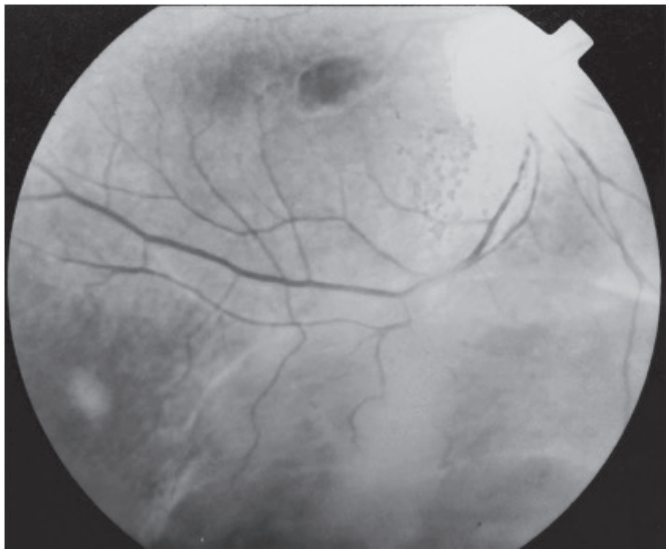

**Figure 2** - Fundography OS: Maculoschisis and peripheral vitreous membranes

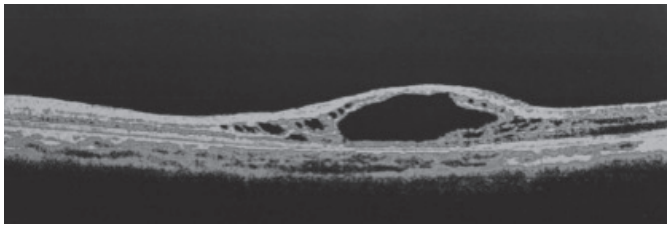

**Figure 3** - OCT OD: Various cysts in the macular area situated on the inner nuclear and plexiform layers that suggest coalescence to a bigger macular cyst at the foveal area

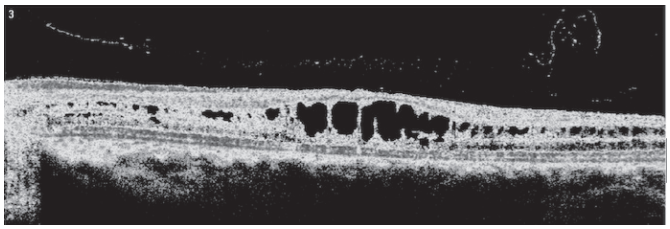

**Figure 4** - OCT OS: Macular retinoschisis

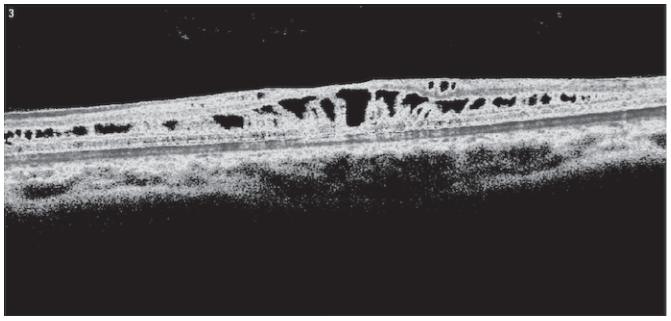

Figure 5 - OCT OS: Macular retinoschisis

## Discussion

Congenital Retinoschisis is a rare, bilateral, asymmetric X-linked inherited disorder that presents itself on the first decade of life.[1,2] It affects primarily young males.[4,5]

The estimated prevalence of the disease is 1:5000-25000 individuals[6]. The anomaly was identified in the Xp22 chromosome's gene XLRS1, causing more than 130 mutations. Therefore, there are many diverse clinical manifestations, even in the same family. The translated protein (retinoschisin) is related to the adherence of the cells during the retinal development.[4-7] The disorder results on the splitting of the retina's neurosensory layers from the remaining of the sensory retina. [1,8]

The patients can exhibit strabismus, nystagmus or diminished visual acuity due to the maculopathy. The disease presents itself as a foveoschisis with macular edema and bicycle wheel-like cystoid spaces, the latter being a typical finding of the pathology. [9-11] Some other observed signs are peripheral retinoschisis - mostly on the inferotemporal quadrant - and oval holes on the retinoschisis' inner layer, which originate vitreous veils. [1,2,6]

The diagnosis is made by indirect binocular ophthalmoscopy. OCT can be of aid on the diagnosis of atypias and advanced stages of the disease, in which the late phenomena can make the diagnosis difficult. The OCT identifies different phases of the disorder: (a) isolated foveoschisis, (b) foveoschisis associated with lamellar cysts without a peripheral ophthalmoscopic disease, (c) foveoschisis associated to lamellar cysts and peripheral disease, and (d) foveoschisis associated to peripheral disease in the absence of lamellar cysts. [4,6,11]

Retinal detachment, pigmentary retinitis, Eales disease, sickle cell retinopathy, retinopathy of prematurity and Goldmann-Favre vitreoretinal degeneration and the Stellate non familial retinoschisis[12] are differential diagnosis of Congenital Retinoschisis.

The pathology's progression is fast during the first years of life, stabilizing around early adulthood. It may improve or worsen during the fifth decade of life, due to macular degeneration. Its possible complications are RD and vitreous and intra-schisis hemorrhage.

Treatment consists on the management of complications. Pars plana vitrectomy is recommended for persistent vitreous hemorrhage or RD, aiming to close the retinal fractures of the peripheral retinoschisis external layers. Laser photocoagulation or vitrectomy are indicated in cases of recurrent or non absorbing vitreous hemorrhage in patients with severe low visual acuity due to this cause. [1,2,10] Carbonic anhydrase inhibitors may be effective for cystic foveal lesions. 10

This series of cases indicate the importance of OCT. Due to the early age of onset and consequent lack of cooperation, its execution may be difficult. It may have little value on peripheral retinoschisis, but it remains a useful tool to differentiate between retinoschisis and RDs that may harm the macula.

## References


1. Ryan SJ. Retina: basic science and inherited retinal disease. 4th ed. Philadelphia: Mosby; 2006. Vol. 1
2. Vilela MA, Colossi CG. Retinosquises. Rev Bras Oftalmol. 2011; 70(2):125-32.
3. Caye LM, Barbosa PH, et al. Uso da dorzolamida tópica em paciente portador de retinosquise juvenil ligada ao X. Rev Bras Oftalmol. 2010; 69(2):129-131.
4. Forsius H, Vainio-Mattila B, Eriksson A. X-linked hereditary retinoschisis. Br J Ophthalmol. 1962;46(11):678-81.
5. Sauer CG, Gehrig A, Warneke-Wittstock R, Marquardt A, Ewing CC, et al. Positional cloning of the gene associated withX-linked juvenile retinoschisis. Nat Genet. 1997;17(2):164-70.
6. Bastos AL, Freitas BP, Villas Boas O, Ramiro AC. Uso da dorzolamida tópica em pacientes com retinosquise juvenil ligada ao X. Arq Bras Oftalmol. 2008; 71(2): 286-90.
7. Kanski JJ, Bowling B. Clinical ophthalmology: a systematic approach. 7th ed. Edinburgh: Elsevier Saunders; 2011.
8. Nelson textbook of pediatrics. 19th ed. Philadelphia, PA; Elsevier/ Saunders; c2011. Disorders of the eye. Part 29.
9. Hofling-Lima AL, Moeller CT, Freitas D, Martins EN. Manual de condutas em Oftalmologia. São Paulo: Atheneu; 2008.
10. Audo I, ,lder GE, Moore A T. Inherited stationary disorders of the retina. In: Puech B, De Laey J-J, Holder GE, editors. Inherited chorioretinal dystrophies. A textbook and atlas. 2014.p. 77-98
11. Apushikin MA, Fishman GA. Use of dorzolamide for patients with X-linked retinoschisis. Retina. Retina. 2006; 26(7):741-5. Erratum in: Retina. 2007;27(1):128.
12. Ober MD, Freund KB, Shah M, Ahmed S, Mahmoud TH, Aaberg TM Jr, Zacks DN, GaoH, Mukkamala K, Desai U, Packo KH, Yannuzzi LA. Stellate nonhereditary idiopathic foveomacular retinoschisis. Ophthalmology. 2014;121(7):1406-13.