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FAMILY CASE OF X-LINKED RETINOSCHISIS WITH OCULAR COMPLICATIONS

Author: *Larisa Bucinscaia*
Scientific adviser: *Angela Corduneanu, PhD, assoc. prof., Chair of Ophthalmology*

Introduction Juvenile retinoschisis or X-Linked Retinoschisis (XLR) is a rare congenital disease of the retina, caused by mutations in the RS_1 gene, which encodes retinoschisin, a protein, which is found throughout the retina, and is thought to be involved in cell-cell adhesion and intercellular matrix retinal architecture development through interactions with αβ crystallin and β2-laminin. XLR is characterized by bilateral maculopathy, with associated peripheral retinoschisis in 50%. Complications include vitreous haemorrhage (4% - 40%), subretinal exudation, neovascularization, and rhegmatogenous or tractional retinal detachment (5%–22%).

Keywords Retinal degeneration, congenital retinoschisis.

Purpose To emphasize the clinical manifestations and particularities of X-linked retinoschisis.

Material and methods We have evaluated the case of 3 patients, men, first degree relatives (brothers) with progressive decrease of visual functions.

Results *First patient*, 26 years old, complained of progressive loss of vision from childhood (Fig.1).
VA OD/OS – 0,4 n.c./ CF at 30 cm. IOP OD/OS – 14/15 mmHg.
Diagnosis: *OU X-Linked Retinoschisis. Complicated cataract.*
OD Vitreoretinal tractional membrane.
Barrage laser was performed for the vitreoretinal tractional membrane.

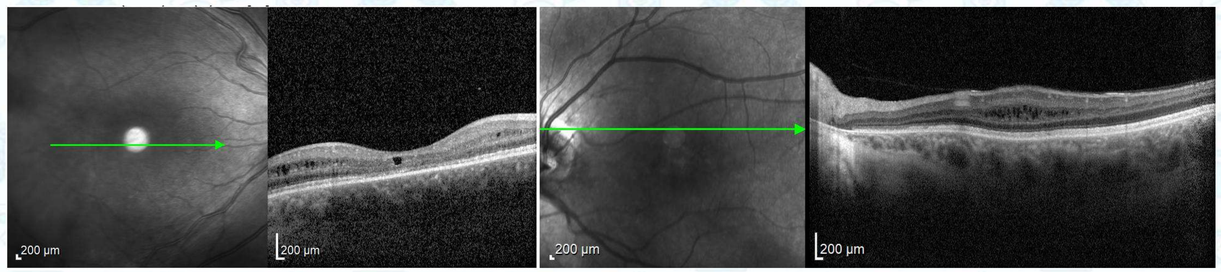

Fig.1 Linear macula OCT reveals cystic spaces in the inner nuclear and outer plexiform layers in the both eyes

Second patient, 33 years old, complained of progressive loss of vision from childhood (Fig.2).
VA OD/OS – 0,6 n.c./ 0,12 n.c. IOP OD/OS – 18/17 mmHg.
Diagnosis: *OU X-Linked Retinoschisis.*

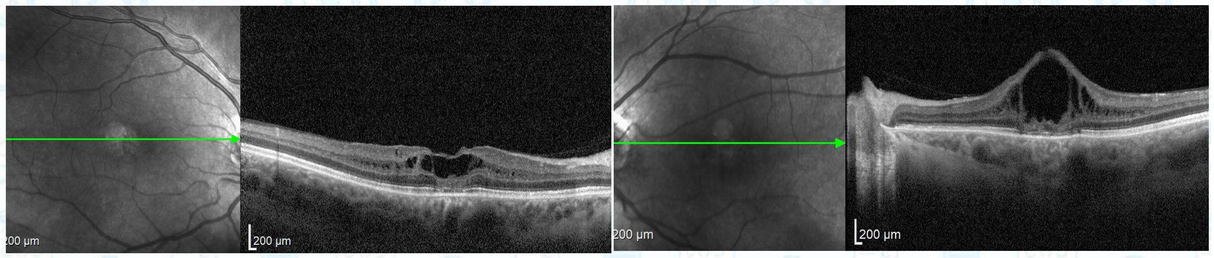

Fig.2 Linear macula OCT

Third patient, 37 years old, complained of progressive loss of vision from childhood (Fig.2).
VA OD/OS –PLC/ 0,4 n.c. IOP OD/OS – 14/18 mmHg.
Diagnosis: *OU X-Linked Retinoschisis. OD Rhegmatogenous retinal detachment, vitreous hemorrhage.*
Surgical treatment (vitrectomy) was performed on OD.

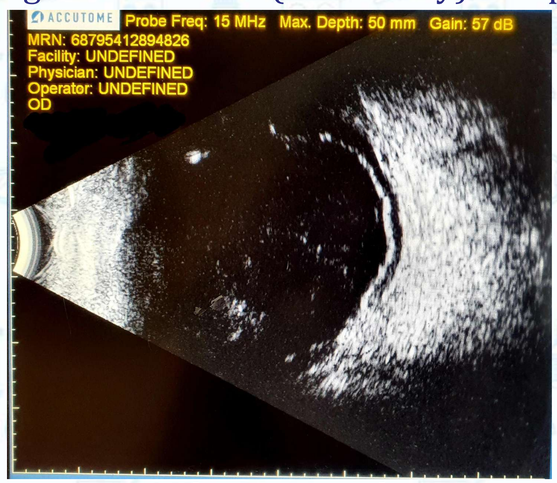

Fig.3 Ultrasonography OD

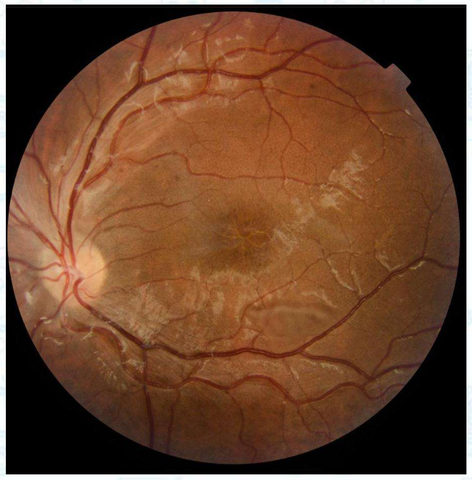

Fig.4 Fundus photo OS

Conclusions Patients with X-linked retinoschisis need to be monitorised for indication of an appropriate treatment and prevention of serious complications, which may lead to significant impairment of visual function.