Translation from Romanian

X-linked Retinoschisis

DEFINITION:

→ X-linked retinoschisis (XLRS) is an eye genetic disease characterized by reduced visual acuity in men due to juvenile macular degeneration.

PREVALENCE: 1/5,000-1/25,000

CLINICAL ASPECTS:

→ Clinically, XLRS is a symmetrical bilateral macular disorder that begins in the first decade of life.

- It is manifested by poor eyesight and reading difficulties.
- Nystagmus can be seen in severe cases.
- Severe cases may involve complete retinal detachment leading to dramatic vision impairment or blindness.
- In the more advanced stages of the disease, it can be observed: vitreous hemorrhage, retinal detachment and neovascular glaucoma, pathologies that can induce a severe loss of vision.
- Pregnant women rarely have visual impairments.

GENETICAL ASPECTS

→ The disease is caused by mutations in the **RS1 gene (Xp22.2-p22.1). RS1** encodes **retinoschisine,** an adhesive protein that is thought to participate in the structural and functional integrity of the retina.

→ XLRS is transmitted **X-linked,** a female carrier having a 50% chance of transmitting the mutation to her offspring. Carrier tests for high-risk female relatives and prenatal diagnosis for high-risk pregnancies are possible if the mutation has been identified in an affected family member.

→ There is also a family history consistent with X-related transmission. Molecular genetic analysis by direct sequencing of the RS1 gene detects mutations in approximately 90% of patients.

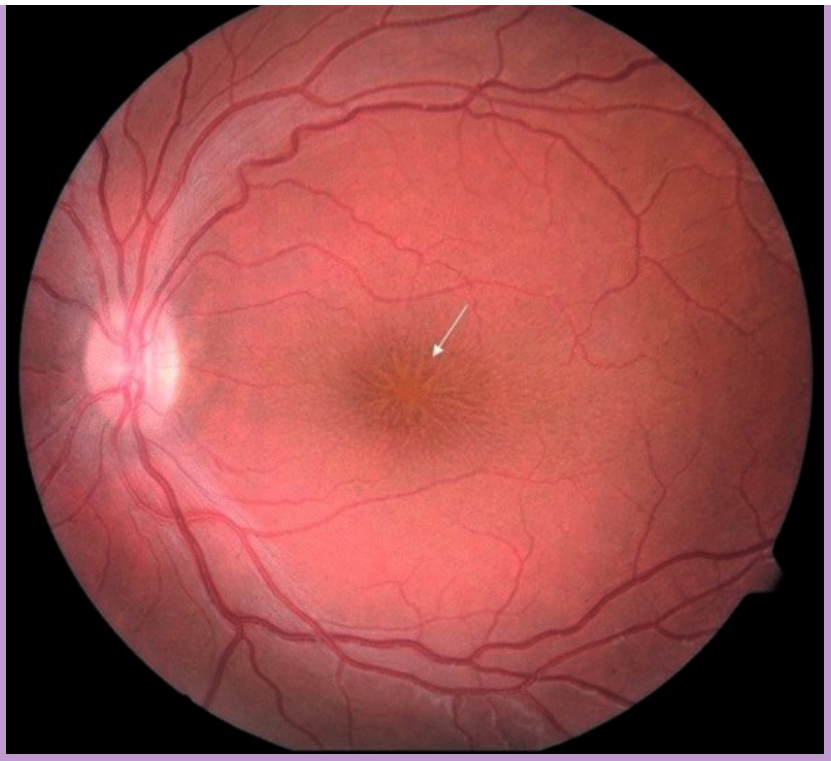
The **DIAGNOSIS of XLRS** can be made clinically, based on the ophthalmoscopic appearance.

- Examination of the fundus of the eye shows microcystic changes in the macular region of the retina and areas of division in the layer of nerve fibers, or schisis (model "in spokes of the wheel") and vitreene flanges.
- The electroretinogram (ERG) shows a reduction in beta wave amplitude and a relative conservation of the alpha negative wave in scotopic ERG (electronegative rods and mixed ERG) and normal photopic ERG.
- Optical coherence tomography (OCD) shows the sketch areas in the macular region.

MANAGEMENT includes periodic eye examination to monitor the evolution of XLRS. In addition, patients are informed of possible ophthalmic complications that can be treated

surgically (ie, retinal detachment, in vitro hemorrhage, cataracts, or strabismus). Therefore, patient education and close follow-up are the only clinical alternatives to the early identification and treatment of complications that may endanger vision.

→ In XLRS, vision decreases slowly until adolescence, and then in most patients it remains relatively stable in adulthood. The disease does not progress until the fourth or fifth decade of life, when there is usually a significant decline in visual acuity.



Img. 1: Ophthalmoscopic appearance in juvenile retinoschisis - model "in spokes of the wheel"

Source: https://ghr.nlm.nih.gov/condition/x-linked-juvenile-retinoschisis#resources

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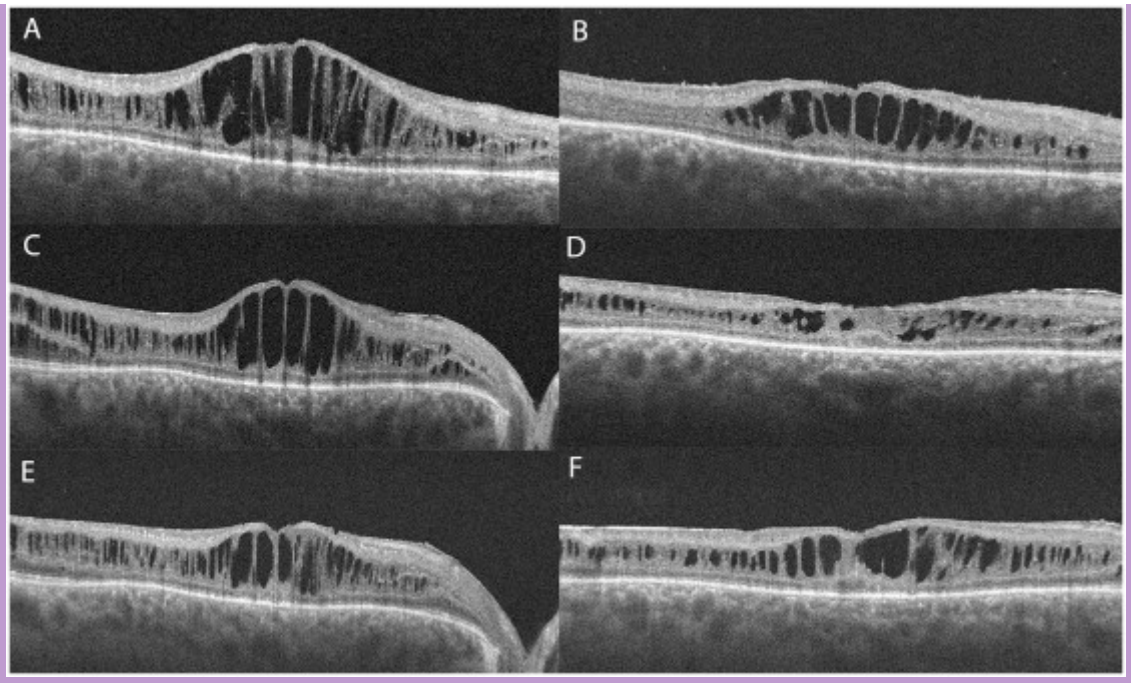

Img. 2: OCT aspect in X-linked juvenile retinoschisis

Source: https://www.sciencedirect.com/science/article/pii/S2451993616301256#fig3

I, BURA TEODORA - NATALIA, certified interpreter and translator for English, by virtue of the authorization no. 35530/20.03.2013, issued by the Ministry of Justice of Romania, hereby certify the accuracy of the translation from Romanian into English.

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