JUVENILE MACULAR RETINOSCHISIS - CASE REPORT

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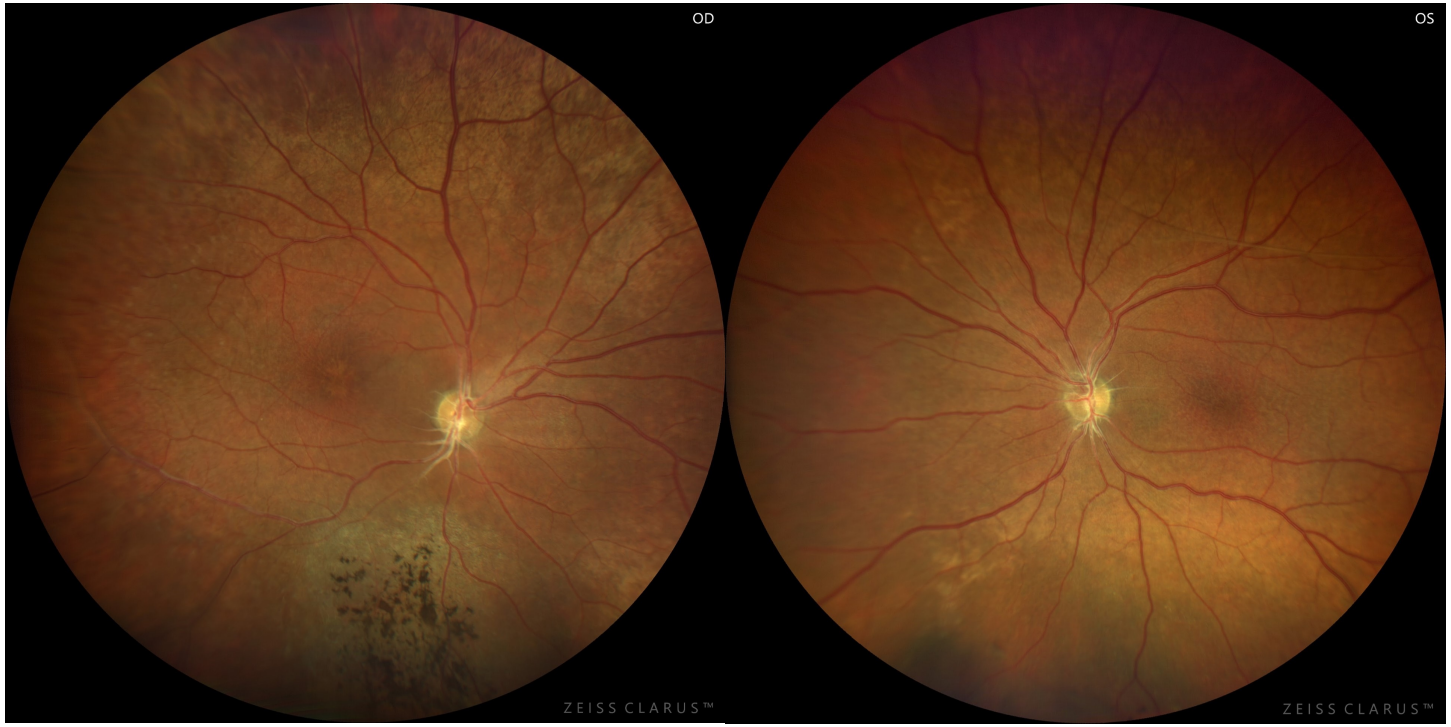
PURPOSE

We report a case of a 34-year-old male, with low vision since childhood, (worse in OS) with no conclusive diagnosis. Had undergone strabismus surgery as a child. Refers low progressive visual acuity for 4 years, especially in central areas.



METHODS

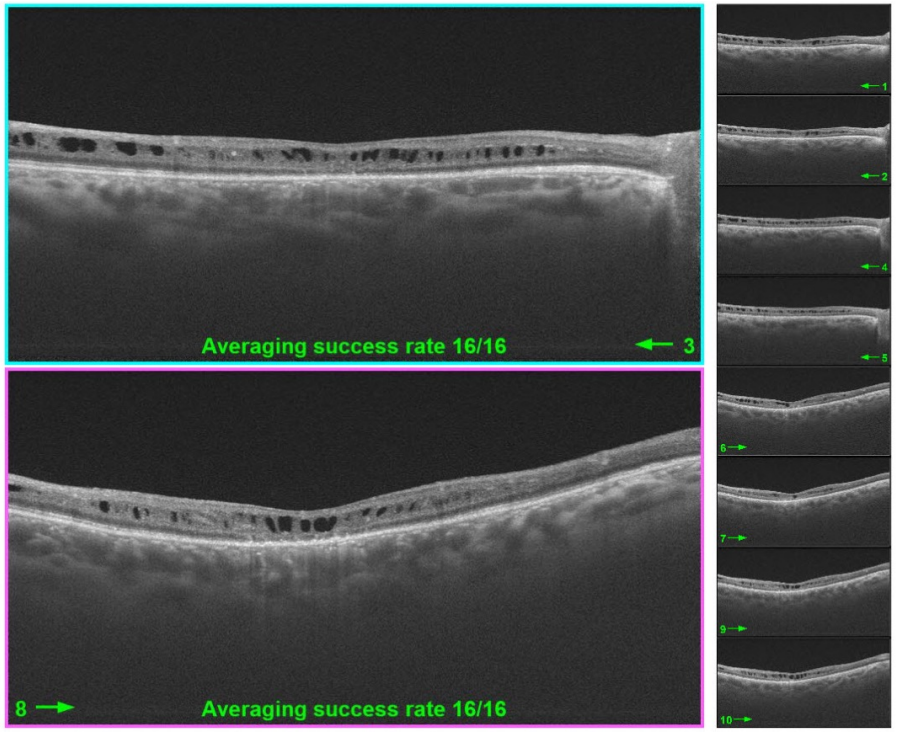
On examination, the anterior segment was unremarkable in both eyes.

In the fundus examination of the right eye was found 0,4 exavation, pale optic nerve, myelin sheath persistency, reduced macular glow, macular pleasing and applied retina.

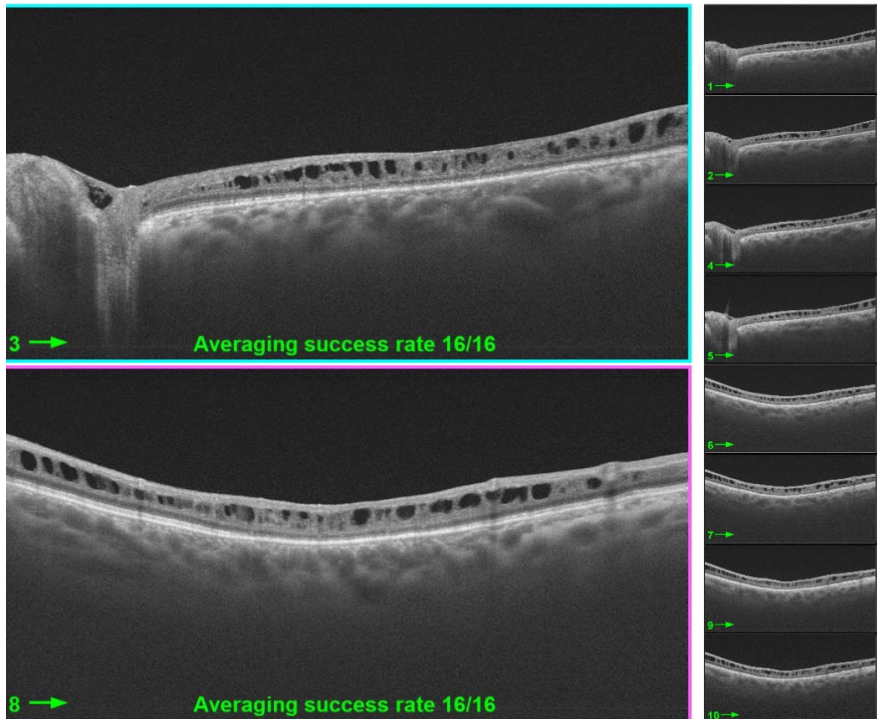
Whereas in the left eye was found an 0,4 excavation, pale optic nerve, persistency of the myelin sheath and macular glow reduction and applied retina.

RESULTS

The patient was instructed on periodic ophthalmologic follow-up and warning signs.



Right Eye



Left Eye

DISCUSSION

X-linked juvenile retinoschisis (XLRS) is an inherited, recessive, vitreoretinal degeneration caused by mutations in the RS1 gene, encoding the retinoschisin protein which leads to schisis (splitting) of the neural retina.

XLRS is one of the most common macular degenerations in young male. Characterized by bilateral maculopathy with peripheral retinoschisis in 50% of patients. However, clinical diagnosis can be challenging due to the highly variable presentation. The evolution is aggravated by the occurrence of vitreous hemorrhage and retinal detachment.