**Information about diagnostic tests and therapeutic medical interventions**

[Ophthalmology clinic]

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<th>No.</th>
<th>Question</th>
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<tbody>
<tr>
<td>1.</td>
<td>What disease (health condition) is described?</td>
<td>X-linked juvenile retinoschisis (XJR ORHA792) caused by mutations of the RS1 gene, which encodes the protein retinoschisin. This results in retinoschisis, or splitting of the retina's layers, usually in the outer plexiform layer. This condition is also known as vitreous veils, congenital vascular veils in the vitreous, and congenital cystic retinal detachment.</td>
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<td>2.</td>
<td>What are the reasons for the development of this disease?</td>
<td>X-linked juvenile retinoschisis (XJR) is an X-linked recessive disease, caused by mutations of the RS1 gene, which encodes the protein retinoschisin. Retinoschisin is a secreted protein that is involved in cellular adhesion and cell-cell interactions within the inner nuclear layer as well as synaptic connection between photoreceptors and bipolar cells. Defective or absent retinoschisin may reduce adhesion of the retinal layers, resulting in the creation of schisis cavities.</td>
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<td>3.</td>
<td>What factors increase development of the disease or exacerbation of the risk (eg. age, sex, heredity)?</td>
<td>It almost exclusively affects young males. The prevalence is estimated to be about 1:15,000-1:30,000. Peripheral retinoschisis is seen in about half of patients. Early in life, the central vision usually is mildly impaired because of a cyst in the fovea. Later, the central vision can become impaired more markedly, resulting in symptoms similar to those of macular degeneration. More seriously, retinal detachments can occur when holes in the inner and outer retinal layers are present. The incidence rate is 5-22% of individuals affected. Other complications include neovascular glaucoma, vitreoretinal traction with secondary macular dragging, spontaneous vitreous haemorrhage and secondary optic atrophy.</td>
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<td>4.</td>
<td>What are the symptoms?</td>
<td>Symptoms of X-linked juvenile retinoschisis may include poor eyesight, detachment of all or part of the retina from the rest of the eye, and eventually complete retinal atrophy (wasting away) with hardening of the choroid (the membrane between the white part of the eye and the retina).</td>
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<td>5.</td>
<td>How is disease diagnosed?</td>
<td>Electoretinography measures the response of the cones and rods (photoreceptor cells of the retina) under light- and dark-adapted conditions. Electrodes, in the form of contact lenses, are placed in the eye while other electrodes are applied to the forehead and earlobe. The patient looks at a flashing light and the electrodes measure the retinal responses.</td>
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Electroretinography abnormalities in XJR may be variable: electronegative waveform to the dark-adapted maximal response, a reduced a/b ratio have been reported.

Optical coherence tomography creates high-resolution color images of the parts of the eye. It is a non-invasive technique that measures the thickness of the macula and/or the nature of the nerve fibers and/or reflections of any particles that may be in the jelly-like substance that fills the interior of the eyeball. In cases of XJR optical coherence tomography reveals typical cystic macular changes.

| 6. | **What are the available treatment options?** | Treatment is generally symptomatic and supportive. Genetic counseling may be of benefit for patients with X-linked juvenile retinoschisis and their families. |
| 7. | **What are the side effects of treatment, and the risk of suffering it?** | - |
| 8. | **What are the consequences of refusing treatment, and the risk of them suffering it?** | - |
| 9. | **What could be subsequent stages of treatment?** | - |
| 10. | **How patient can mitigate experiencing discomfort and reduce the risk of disease progression?** | Magnifying glasses and other aids can be used. |
| 11. | **What are the additional instructions (eg. tips on how not to distribute the disease, etc.)?** | Head trauma and high-contact sports to reduce risk of retinal detachment and vitreous hemorrhage. |
| 12. | **Who will answer additional questions if, there will be any?** | Your family doctor, ophthalmologist. |


3. Byrne LC, Oztürk BE, Lee T, Fortuny C, Visel M,
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