



Juvenile retinoschisis is a genetic condition that causes vision loss. It almost always affects boys usually starting in childhood. Juvenile retinoschisis causes a loss in central vision. In some cases, a person's peripheral vision may also be damaged.

Juvenile retinoschisis should not be confused with senile retinoschisis. This is a condition of aging, which affects roughly equal numbers of men and women.

Medical Description

Retinoschisis literally means splitting of the retina. In this condition, tiny splits appear in the retina between the photoreceptor cells and the next layer of nerve cells which relay their signals to the brain. Cysts form in these splits often making a wheel-like pattern in the centre of the retina. This damage causes a loss of central vision. These splits and cysts also make the retina abnormally thick.

Retinoschisis may also cause similar splits in the farthest edges of the retina, causing a loss of peripheral vision. This happens in about half of people with juvenile retinoschisis.

Over time, damage to the retina builds up causing progressive loss of vision.

Early Symptoms

The symptoms of juvenile retinoschisis are usually noticed about the time that a boy enters public school. In families where the disease is inherited, early testing is often done, so young children can be diagnosed.

The most common symptoms are a general loss of vision and difficulty seeing fine detail. Some boys may also have nystagmus (rapid involuntary movements of the eye from side to side) and strabismus (eyes turned in or cross-eyed). Because of this, sometimes retinoschisis is initially diagnosed incorrectly as amblyopia (lazy eye).

Diagnosis

An ophthalmologist may suspect retinoschisis after a simple eye examination, particularly if a wheel-like pattern of splits is visible in the macula. Several tests can clarify and confirm the diagnosis:

- **ERG** (electroretinography) measures the electrical responses of the retina. During this test, a special contact lens is placed on the eye and the eye is exposed to flashes of light.
- **OCT** (optical coherence tomography) makes digital images of the retinal layers.
- **Genetic testing** (starting with a blood sample) can identify the gene defects that cause juvenile retinoschisis.

What to Expect

Boys with retinoschisis usually have vision loss in grade school and may continue to lose vision into their teens. Once they are adults, their vision most often stabilizes until they are in their 50s or 60s.

Men with retinoschisis almost never lose their vision completely. However, they are at risk of serious eye complications such as retinal detachment (5-20% have

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this complication) and bleeding into the vitreous body (jelly) of the eye (4-40%).

Women who are carriers of a damaged gene may also develop some vision loss, particularly peripheral vision, later in life.

Genetic Causes

Juvenile retinoschisis is an X-linked condition. It is caused by defects in a gene, RS1 (retinoschisin 1), on the X chromosome. Several different changes (mutations) within this gene can cause the disease. Genetic testing can identify the specific mutation within the RS1 gene almost 95% of the time.

What is meant by “X-linked”?

Chromosomes are complex strings of genes. Each person has 23 pairs of chromosomes. One of each pair comes from your biological mother, the other from your biological father. Thus, we have duplicate copies of most genes. One pair is different. These are the X and Y chromosomes that make us female and male. These sex chromosomes carry many different genes. Most of the genes on X are missing from Y. RS1 is one of these.

Women have two X chromosomes, but men have an X and a Y. Thus men have no protective back-up if a gene on the X chromosome is damaged.

Thus usually only boys inherit retinoschisis. Rarely, a girl may inherit damaged genes on both X chromosomes and have the disease. More often, a girl inherits just one damaged gene, which she may pass on to her children without having the disease herself.

Treatment

No treatments are currently approved for retinoschisis. However, dorzolamide eye drops, developed and approved to treat glaucoma, may help reduce the cysts and thickening of the retina, particularly in younger patients. Although the drops may help reduce the early changes associated with retinoschisis, the effect on vision is usually modest. Side effects of dorzolamide eye drops include stinging or burning in the eye. Some children may also be allergic.

Patients of all ages should have regular eye exams even if their vision is not changing, to avoid serious but treatable complications such as retinal detachments.

Research

The Foundation Fighting Blindness supports scientists working to understand the causes of vision loss and to develop treatments for it. A Canadian researcher, Dr. Robert Molday, is a world leader in research on juvenile retinoschisis. He has been funded by the Foundation Fighting Blindness to study retinoschisis, and currently leads an international team of researchers funded by the Foundation Fighting Blindness and the Canadian Institutes of Health Research, which aims to develop gene treatments for several genetic conditions. To subscribe to our print and e-newsletters on the latest vision research, call **1.800.461.3331** today or email **info@ffb.ca**.

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Updated April 5 2012: Reviewed by Dr. Kevin Gregory-Evans, Professor in Ophthalmology, University of British Columbia.

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