RETINA australia

Juvenile X-linked retinoschisis

Juvenile X-linked retinoschisis is a rare genetic disease of the retina and primarily affects boys and young men. Retinoschisis is a condition in which an area of the retina (the tissue lining the inside of the back of the eye that transmits visual signals to the optic nerve and brain) has separated into two layers. The part of the retina that is affected by retinoschisis will have poorer vision, however very few people with retinoschisis lose all of their vision.

What are the symptoms of retinoschisis?

Affected boys are usually identified in primary school, but occasionally are identified as young infants. Most boys with retinoschisis present with a mild decrease in central vision in primary school that may be subtle and not perceptible. They may continue to lose vision into their teens. Once they are adults, their vision often stabilises until they are in their 50s or 60s.

Retinoschisis patients are more susceptible to retinal detachment and eye haemorrhage (bleeding) than other people and they should have regular examinations with an eye doctor. When detected early, a complicating retinal detachment can be treated surgically.

What is the cause of retinoschisis and how is it inherited?

The exact prevalence of retinoschisis is currently unknown, but it is thought to affect between one in 5,000 to 25,000 people and is therefore a rare condition. X-linked retinoschisis is usually caused by mutations (defects) in the RS1 (retinoschisin 1) gene, which is located, as the name suggests, on the X chromosome. Men have only one X chromosome, while women have two. Therefore, because women almost always have another functioning X chromosome, they typically retain normal vision, even as carriers.

However, in rare circumstances, due to the phenomenon of 'non-random X chromosome inactivation' some female carriers may have symptoms. Men, on the other hand, will develop sub-optimal vision if they have an affected X chromosome. Affected males cannot pass on the disease to their sons, because they pass on their Y chromosome. Men who have retinoschisis must pass on the disease gene to all of their daughters who in turn become carriers of the condition.

What treatments are available?

Maximising an individual's remaining vision is a crucial first step to take, and there are many new low vision aids including telescopic and magnifying lenses. Currently, there are no medical or surgical treatments available for retinoschisis.

Glasses may improve the overall quality of vision in a patient with retinoschisis who is also near-sighted or farsighted but will not repair the nerve tissue damage from the retinoschisis. In 1997, researchers identified mutations in the RS1 gene on the X chromosome that cause retinoschisis.

Scientists then studied the gene to determine its function in the retina. These included some in a 2014/2015 collaboration between research institutes in NSW, Victoria, Tasmania and Western Australia who researched X-linked retinoschisis in females. Various groups worldwide are now working on developing gene therapies for retinoschisis.

These innovative therapies aim to deliver healthy copies of the RS1 gene by a non-toxic virus, therefore replacing defective copies at the back of the eye. This type of gene therapy has been shown to be safe in other conditions, and a trial is currently underway in the United States to determine safety of this approach in retinoschisis.

General eye check-ups are important for people with retinoschisis, as these men and boys are still at risk for other kinds of eye problems that can affect the general population and may be treatable. They are also more vulnerable to retinal detachments and haemorrhages. Regular visits to your eye doctor can also make you aware of current advances as we learn more about retinoschisis.