WHAT YOU SHOULD KNOW ABOUT X-LINKED RETINOSCHISIS

WHAT IS X-LINKED RETINOSCHISIS (XLRS)?

X-linked retinoschisis (XLRS) is an inherited retinal disease that causes loss of central and peripheral vision due to degeneration of the retina. About 35,000 people in the United States and Europe have the condition.

The retina is a thin piece of tissue lining the back of the eye. Rod and cone photoreceptors in the retina convert light into electrical signals that the brain interprets as vision.

XLRS leads to vision loss due to the splitting of retinal layers, which leads to degeneration of photoreceptors.

WHAT TO EXPECT WITH X-LINKED RETINOSCHISIS:

One of the first signs of XLRS is loss of visual acuity that is not correctable.

Cystic macular lesions (like blisters) in the retina are the hallmark feature of XLRS, exacerbating vision loss.

Cystic macular lesions can be treated with medications including oral or topical carbonic anhydrase inhibitors.

HOW IS X-LINKED RETINOSCHISIS INHERITED?

XLRS is caused by mutations in the RS1 gene. The condition is passed down in families by the X-linked pattern of inheritance.

Female carriers have a 50% chance of passing the disease gene to their daughters, who become carriers, and a 50% chance of passing the gene to their sons, who are then affected by the disease.

Males with X-linked diseases pass their Y chromosome to their sons, and therefore will never pass an X-linked disease to their sons. However, affected males will always pass their affected chromosomes to their daughters who will always become carriers.

To learn more about living with X-linked retinoschisis, finding a specialist, genetic testing or clinical trials, visit FightingBlindness.org or call 888-332-3667.