

# **What You Should Know About Marfan Syndrome**

*By David J. Browning MD, PhD*

Marfan syndrome is a hereditary disease of connective tissue that causes abnormalities of many organ systems. Patients may have any or all of the following systemic findings: aortic aneurysm, long fingers (arachnodactyly), joint laxity, curved spine (scoliosis), and a sunken chest (pectus excavatum). In addition to these, there are many abnormalities of the eye in Marfan syndrome. Patients can have any or all of the following: dislocated lens, thin areas of the retina (lattice degeneration), retinal tears and detachments, glaucoma, elongated eyes (axial myopia), and in-turned or out-turned eyes (strabismus). Because the eyes are so often involved in Marfan syndrome, all affected patients require regular eye examinations.

## **What Causes Marfan Syndrome?**

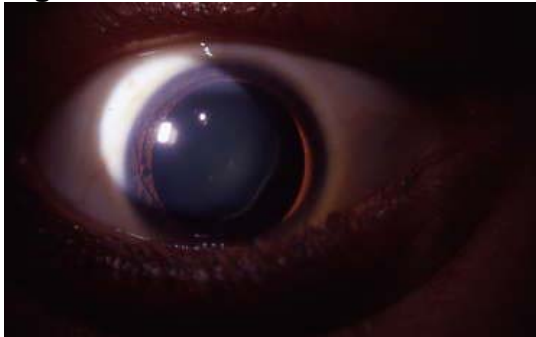
Patients with Marfan syndrome have a DNA mutation in the fibrillin gene found on chromosome 15. The fibrillin gene codes for a protein involved in collagen cross linking. Its absence leads to collagen abnormalities that are manifest as thin sclera and flawed zonules (the small cables that secure the natural crystalline lens in position). Patients also have abnormal pupillary dilator muscles, and generally are difficult to dilate for examination. This mutation affects approximately 1 in 20,000 newborns in the United States.

## **How Are Dislocated Lenses (Ectopia Lentis) Managed?**

The natural crystalline lens in Marfan syndrome begins to move temporally (toward the ear) and superiorly in 50-80% of affected patients, often during childhood. An example of this is shown in figure 1. As long as good visual acuity can be achieved with glasses or contact lenses, conservative follow up is chosen. Once good vision cannot be achieved, surgery to remove the dislocated lens is performed, usually with a vitrectomy approach, involving 3 one-millimeter incisions and controlled intraocular pressure. In children, no intraocular lens is implanted, but rather a contact lens is fitted after surgery to provide focus. In adults, an intraocular lens may be

implanted, especially if previous experience was notable for contact lens intolerance.

**Figure 1. Dislocated Lens**

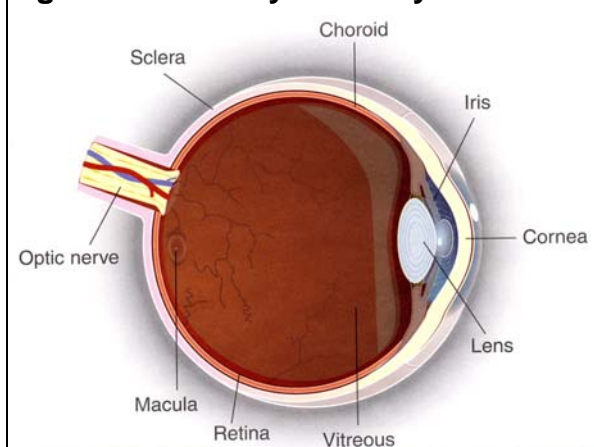


### **How Are Retinal Detachments Managed?**

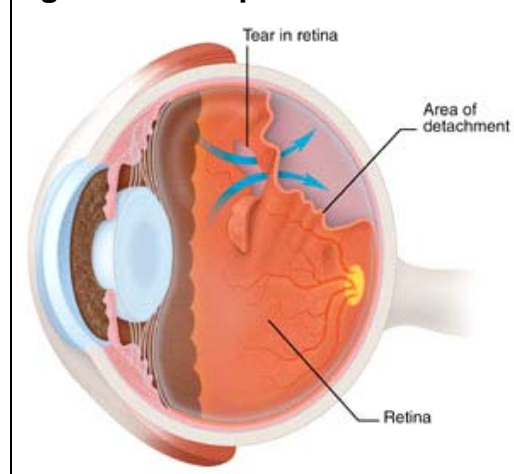
The retina is the thin neural lining of the back of the eye that turns light into a neural signal. Figure 2 shows its location.

Retinal detachments occur in 10-25% of all patients with Marfan syndrome. They are repaired with surgery using a silicone band around the eye, often with removal of the vitreous gel (vitrectomy), and sometimes simultaneous lens removal (lensectomy), especially if the lens is dislocated and makes retinal surgery more complicated. Sometimes prophylactic laser or cryotherapy is given to the fellow eye depending on examination findings. Bilateral retinal detachments occur in 34-69% of patients who have retinal detachment. Figure 3 shows a schematic of a retinal detachment.

**Figure 2. Anatomy of the Eye**



**Figure 3. Example of RD**



### **Final Comments**

Marfan syndrome frequently produces ocular abnormalities, but with careful screening examinations, most patients can retain excellent vision throughout life as any abnormalities are detected and

treated. Appropriate genetic counseling is necessary for affected patients, as the probability of a child of an affected patient being affected is 50%.

After you read this brochure, we encourage you to browse our website, including the Frequently Asked Questions section and the Forums, where patients may share their experiences with one another. If you have a focused question for which you cannot find an answer, we welcome you to ask Dr. Browning at: [ask@theretinaexchange.com](mailto:ask@theretinaexchange.com). Also, an excellent resource for medical literature is Pubmed, on the National Library of Medicine website, accessible via a link on our website, or directly at [www.pubmed.com](http://www.pubmed.com).