

THE EYES IN MARFAN SYNDROME

Marfan syndrome and some related disorders can affect the eyes in many ways, causing dislocated lenses and other eye problems that can affect your sight. Except for dislocated lenses, these eye problems also occur in the general population, which is why doctors do not always realize they are caused by Marfan syndrome. It is important to know that, even though these problems occur in the general population, they are much more common in people who have Marfan syndrome.

About 6 in 10 people with Marfan syndrome have dislocated lenses in one or both eyes.



People with Marfan syndrome should see an ophthalmologist (a medical doctor who takes care of the eyes) to find out if they have any eye problems and learn how to care for their eyes.

What are the common types of eye problems in people with Marfan syndrome?

Some features of the eye related to Marfan syndrome that can cause vision problems include:

Dislocated lenses

About 6 in 10 people with Marfan syndrome have dislocated lenses in one or both eyes. This means the lens, located at the front of the eye, has slipped out of place because the connective tissue that holds the lens in place (called zonules) is weak. When this happens, the lens can slip in any direction—up, down, to the side, or back. It can slip a little or completely out of place, and anywhere in between. With the lens out of place, the eye can't focus properly and vision is blurry.

Symptoms of lens dislocation depend on severity and may include mild to severe nearsightedness, blurred vision, and fluctuating vision. Lens dislocation can only be confirmed by an ophthalmologist using a slit-lamp eye examination after fully dilating the pupil.

For most people, dislocated lenses occur before age 20, although lenses can dislocate at any age. Babies and children can have dislocated lenses. Dislocated lenses are rare in the general population, so people with dislocated lenses should be tested for Marfan syndrome if there is not another known cause.

Retinal Detachment

Retinal detachment is a separation of the light-sensitive membrane in the back of the eye (the retina) from its supporting layers. Symptoms that may indicate a retinal detachment include:

- Flashing lights
- New floaters
- A gray curtain moving across your field of vision

These symptoms do not always mean a retinal detachment is present, but you should see your eye doctor immediately if any of these signs occur.

Head trauma can cause retinal detachment in anyone, and those who are highly myopic (near-sighted) are always at risk for retinal detachment. For people with Marfan syndrome, however, retinal detachment can happen spontaneously. See your doctor immediately if you see flashing lights, new floaters or a gray curtain moving across your field of vision.

Severe Myopia

Myopia is nearsightedness. Objects in the distance are blurred and only objects close to the eye are in focus.

Astigmatism

Astigmatism is blurred vision caused by an irregular curve of either the lens or the cornea. The cornea is the layer of tissue covering of the very front of the eye.

Amblyopia

Amblyopia is when the vision in one of the eyes is reduced because the eye and the brain are not working together properly. The eye looks normal, but it is not being used normally because the brain is favoring the other eye. This condition is also sometimes called “lazy eye.”

Strabismus

Strabismus is when the two eyes do not focus on the same object simultaneously. One eye may drift in or out compared to the other eye. The eyes may alternate fixating on an object or one eye may fixate more. It occurs in people with Marfan syndrome at a higher rate than in the general population. In the general population, the deviating eye more commonly turns inward, toward the nose. In people with Marfan syndrome, the deviating eye usually turns outward. The eyes may also deviate vertically. Strabismus can cause loss of depth perception, double vision, and amblyopia. However, amblyopia is often present first and leads to strabismus, but once amblyopia is successfully treated, the strabismus may correct itself.

Glaucoma

A disease of the eye caused by increased pressure inside the eye. Glaucoma develops in about 35 percent of people with Marfan syndrome, often at an earlier age than the general population. Untreated glaucoma can cause blindness.

Pre-senile Cataracts

This is a clouding of the eye lens before age 60. Cataracts are common in older people who do not have Marfan syndrome, but people with Marfan syndrome can get cataracts at younger ages—even before age 40.

Some other eye features often occur in people who have Marfan syndrome that do not usually cause vision problems, but they can help doctors decide whether or not a person has Marfan syndrome. They include:

- Flattened curve of the cornea. This feature may make it more difficult to fit contact lenses.
- Larger than normal corneas
- Difficulty in completely dilating (opening) the pupils when the doctor does an eye exam
- Obvious sunken eyeballs (enophthalmos)

What are the considerations for vision problems in children with Marfan syndrome?

There are many causes of vision problems in children with Marfan syndrome. Often, the eyeball is large or too long, which keeps light rays from focusing on the retina and causes myopia or near-sightedness. In other cases, the position of the lens or the shape of the eye (if it is shaped like a football) may cause astigmatism.

In either case, it is important for the child's eyes to be checked regularly to ensure the proper prescription. From a diagnostic and treatment standpoint, it is essential to evaluate the eyes of all children as early as possible because if the brain has not perceived good vision in both eyes by the age of ten or 12 at the latest, achieving good vision might not be possible.

It is essential to evaluate the eyes of children as early as possible—do not wait until they are of school age.

How are the common types of eye problems treated in people with Marfan syndrome?**Lens dislocation**

Eyeglasses and contact lenses

Eyeglasses or contact lenses can usually correct blurred vision caused by dislocated lenses. To find glasses that work well, your doctor first needs to decide whether your glasses should use the dislocated lens or ignore it. It takes extra time and effort for your doctor to find out whether to use or ignore the lens and to find the best glasses for a person with dislocated lenses. It also matters whether or not the dislocation is in one eye or in both eyes. For these reasons, you may want to choose an eye doctor who has experience treating patients with Marfan syndrome.

Your doctor may prescribe eye drops to use every day. The drops cause the pupil of your eye to stay dilated (wide open). This makes glasses that ignore your dislocated lens work better.

Lens removal and replacement

Lens removal and replacement is another treatment for lens dislocation, but people with even a total lens dislocation may not need surgery for decades, depending on their age and other factors.

In the largest research study of people with Marfan syndrome, the most common reasons for surgery were lens dislocation with fluctuating vision, progressive cataracts, and uncorrectable vision.

There are some good reasons for people with Marfan syndrome to think carefully about lens removal and replacement, particularly in young children.

- Although a dislocated lens is not able to adequately perform its primary function and fine-tune visual acuity, it may still be able to help balance the pressures in the eye between the anterior and posterior chambers..
- Lens removal in young children raises the risk for retinal detachment and makes lens replacement surgery later in life, when the eye is fully grown, more risky and technically difficult.

Experts recommend delaying, if medically possible, lens removal and artificial lens implantation surgery until the later teen years, once the eye is more likely to have finished growing. The more stable the ocular parameters, such as a healthy retina and an eye that has stopped growing, the better the calculations for the lens implant.

If artificial lens implantation surgery is recommended, posterior, as opposed to anterior, chamber lenses are usually the better choice for people with Marfan syndrome because they tend to have large eyes and deep anterior chambers. Standard anterior chamber lenses are often too small, and may result in complications such as excess movement of the lenses (windshield wiper effect), inflammation of the iris, glaucoma, pain, and the need for corneal transplantation. Posterior chamber lenses should always be sutured in place.

How do you care for dislocated lenses if glasses do not work?

When glasses do not work well to correct blurred vision, some doctors recommend removing dislocated lenses. Not all doctors agree on the reasons to remove dislocated lenses. Here are some of the issues you and your doctor need to talk about to help you decide whether or not to remove your lenses:

- Is it really impossible to find the right glasses to correct your vision? Remember it takes extra time and care to find glasses that either use or ignore dislocated lenses.
- Once the lens is removed, to see anything clearly you will need corrective lenses—either very thick glasses or contact lenses—or an artificial lens placed in your eye. Which type of corrective lenses does your doctor suggest you use?
- People are not always happy using thick glasses because they often distort peripheral vision, that is, cause vision to the side to be wavy and unclear. Also, thick glasses are often heavy and uncomfortable. Finally, it may be hard to buy the special glasses because many eyeglass stores cannot make them.
- You may need to decide which is better for you: keeping your lenses and not seeing quite so well or using thick glasses.

Using Artificial Lenses

One way to avoid thick glasses is to have an artificial lens placed in your eye. Will an artificial lens work for you? Not all doctors agree that people with Marfan syndrome can use artificial lenses, but we know many people who see well and are happy with their artificial lenses.

There are many issues about artificial lenses you need to talk about with your doctor. Because operating on patients with Marfan syndrome can be difficult and complications (unwanted results) can occur, you should try to find a doctor who has experience operating on the eyes in people with Marfan syndrome.

Here is some information to help you talk with your doctor about artificial lenses:

Artificial lenses are put in either the anterior (front) or posterior (rear) chambers of the eye. There are pros and cons to using either kind of lens in people with Marfan syndrome.

- Using an anterior chamber lens

Reasons to use anterior chamber lenses include:

- It is easier to put a lens in the anterior chamber.
- When using an anterior chamber lens, there is less chance of causing a retinal detachment as a complication of the surgery.

Reasons not to use anterior chamber lenses include:

- Anterior chamber lenses may be too small to fit well in the eyes of people with Marfan syndrome. This is because many people with Marfan syndrome have extra large anterior chamber spaces in their eyes.
- As a result, anterior chamber lenses may move around and cause poor vision and pain.

- Using a posterior chamber lens

Reasons to use posterior chamber lenses include:

- Posterior chamber lenses come in sizes that can better fit in a patient with Marfan syndrome.
- Using a posterior chamber lens that is sewn (sutured) into place can help prevent unwanted movement of the lens.

Reasons not to use posterior chamber lenses include:

- Posterior chamber surgery is more difficult and delicate than anterior lens surgery.
- Posterior chamber surgery takes more time than anterior lens surgery.
- There is greater chance of causing a retinal detachment as a complication of the surgery.
- There is a chance the suture can break over time and the artificial lens dislocate.

It is important to remember that not all doctors agree on which kind of lens to use for people with Marfan syndrome. You should ask enough questions so you understand the reasons for your doctor's choice. You should ask your doctor about the features of your eyes so you can understand which kind of lens is best for you. These features include the size of your anterior chamber and the nature

of your retina. Some people have features in their retina that raise the risk of retinal detachment. Ask your doctor if you have those features.

Myopia (nearsightedness)

Eyeglasses or contact lenses

Various types of nearsightedness can be corrected with glasses or contact lenses (special flat contacts may be required for proper correction). A thorough measurement of the refractive condition of a patient's eyes (retinoscopy) is essential for adequate visual correction, and full correction is desirable. Corneal refractive surgery (laser keratotomy) is not recommended for most people with Marfan syndrome if the corneas are already markedly flattened.

Laser correction of myopia

Laser correction can correct myopia up to -10 diopters. Most people with Marfan syndrome do not qualify because their nearsightedness is greater than -10 diopters.

Minor cases of myopia can be corrected via laser surgery in people with Marfan syndrome if they do not have lens dislocation. If a person does have a lens dislocation, laser surgery is not recommended because it will make the dislocation worse. Additional information is needed about the experiences of those with Marfan syndrome who have had laser correction of myopia to help determine if this procedure is worth the risk for affected people.

Cataracts

Implant surgery is quite routinely performed and new techniques for lens removal and implant have produced very high success rates. It is essential that the surgery is performed at an ophthalmology center that recognizes the surgical complications that may occur in people with Marfan syndrome. These complications include vitreous loss, rupture of the residual zonules, and extension of the capsulotomy.

Glaucoma

Glaucoma is more prevalent among people with Marfan syndrome than people who do not have the disorder. Low-tension glaucoma is also seen in people with Marfan syndrome. If medical management (drug therapy) is not successful, surgery may be recommended. The surgery usually has very good results.

Amblyopia (decreased vision or "lazy eye")

When treating amblyopia, the goal is to stimulate use of the weaker eye by blurring the vision of the better eye and helping the part of the brain that manages vision to improve function. Special attention must be paid to identifying children with amblyopia. It is essential that treatment be started prior to the age of 6 or 7 years so that sight can be recovered in the weaker eye.

There are two principal treatments:

Eye drops

This approach uses eye drops of a long-acting dilating agent, such as atropine, in the stronger eye to blur the vision and provoke use of the weaker eye. It is important to make sure that the vision in the medically blurred eye is worse than the vision in the amblyopic eye.

Eye patch

Using an eye patch is another option. An opaque, adhesive patch is worn over the stronger eye for several hours a day for weeks to months. Before starting amblyopia therapy, the prescription of eyeglasses has to be maximized.

Strabismus (misalignment of the eye)

About half of people with strabismus also have amblyopia. About 95 percent of people with strabismus can attain excellent visual acuity if they are wearing the proper glasses and are treated for amblyopia.

How do you handle eye emergencies such as retinal detachment?

Marfan syndrome significantly increases a person's risk of retinal detachment, a serious condition that should be treated as an emergency. Because of the risk of retinal detachment, people with Marfan syndrome should avoid activities that can result in sudden or severe blows to the head.

Retinal detachment is a separation of the light-sensitive membrane in the back of the eye (the retina) from its supporting layers. Early symptoms that may indicate a retinal detachment are:

- Bright flashes of light, especially in peripheral vision
- Translucent specks of various shapes (floaters) in the eye
- Blurred vision
- Gray curtain moving across your field of vision

The symptoms may occur gradually or quite suddenly. They do not always mean a retinal detachment is present, but they do require seeing an eye doctor immediately.

Surgery is required to repair a retinal detachment. Immediate treatment is recommended usually within the first 24 hours; the longer the wait, the greater the chance that the retinal detachment will become more severe or even total. The more severe the detachment, the more complicated the surgery is and the less vision may be recovered.

Your vision after a retinal detachment depends on the location and extent of the detachment, and how quickly you receive treatment. If the macula was not damaged, the outlook after surgery can be good. Any surgery has its risks; however, an untreated retinal detachment usually results in permanent, severe vision loss or blindness.

As in any emergency, it is best to be prepared in advance. Complete our Emergency Preparedness Kit before an emergency occurs and keep it handy. You can find it on our website at marfan.org.

Do you have questions? Would you like more information?

- Call our help center, 800-862-7326, ext. 126 to speak with a nurse who can answer your questions and send you additional information.
- Visit our website at marfan.org. You can print information that interests you and ask questions online.

Figure 1. The Eye

