Marfan and the eye

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What is Marfan Syndrome?

Marfan syndrome (MFS) is a genetic disorder of the body's connective tissue that affects any gender, race or ethnic group. Connective tissue helps provide structure to the body, binding skin to muscle and muscle to bone. It provides the stretchy strength of tendons and ligaments around joints and in blood vessel walls. It also supports the internal organs. This tissue is made of fine fibres and 'glue'. One fibre is called fibrillin.

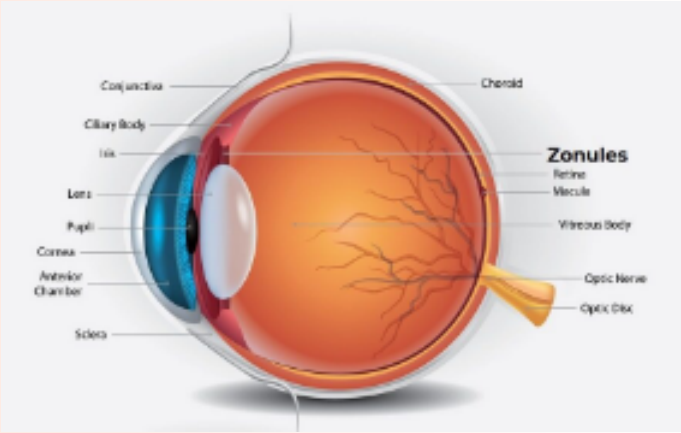
In MFS, a change in the fibrillin-producing gene, fibrillin-1, means that this protein is deficient in connective tissue throughout the body, creating an unusual stretchiness and weakness of tissues. This has far-reaching implications and can affect the eyes, lungs, gut, nervous system, skeleton and, most dangerously, the cardiovascular system. Symptoms can vary widely from person to person with people experiencing mild to severe manifestations.

MFS affects roughly 1 in 3,000 people which means that approximately 18,000 people in the UK have MFS. We estimate that half remain undiagnosed. 75% of patients inherit the condition whilst 25% develop it as a result of a spontaneous (new) gene change. Each child of an affected parent has a 50% chance of inheriting Marfan syndrome.

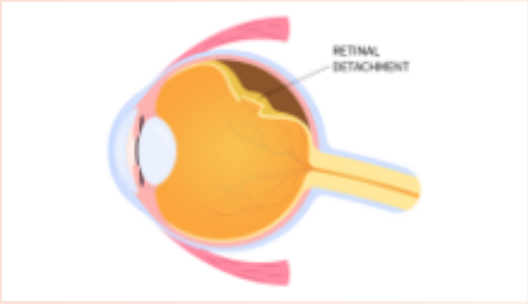
Every year, on average in the UK there are over 200 new cases of Marfan diagnosed.

Marfan and the eye

Vision is vital for daily functioning. Impaired vision hampers the ability to carry out everyday activities smoothly. Patients with Marfan Syndrome are advised to have their eyes examined by an ophthalmologist starting as soon as possible on diagnosis.



Checks of visual acuity and refractive error (any condition wherein your eye doesn't focus light on the retina and corrected usually by glasses, or contact lenses) should be carried out at least once annually by an optometrist or ophthalmologist to detect any development of lens abnormalities for timely intervention in all Marfan patients without eye involvement. Myopia (Short-sightedness) is more common in Marfan Syndrome owing to the increase in the length of the eye, the curvature of the lens, and the likelihood of dislocation of the lens that leads to a focus of light in front of rather than on the retina. Objects in the distance are blurred and only objects close to the eye are in focus.



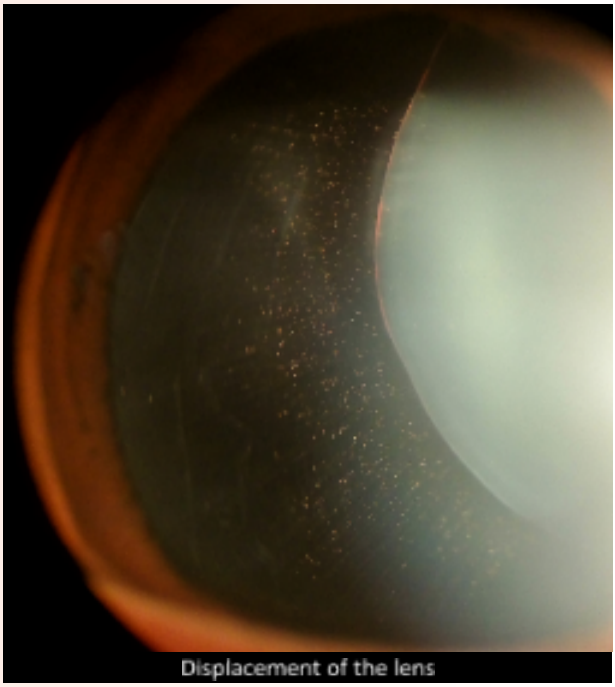

Strabismus (squint)

Retinal Detachment: Retinal detachment occurs when the retina, the light-sensitive layer at the back of the eye, pulls away from its normal position, causing vision loss. It can occur as a complication following previous surgery of a dislocated lens or in the absence of it.

Other ophthalmic problems include strabismus (squint), amblyopia (lazy eye), and glaucoma. Glaucoma occurs due to an increase in the pressure of your eye following the dislocation of the lens into the anterior chamber (the area between your iris and cornea in the front of your eye) and resulting blockage of the outflow of the aqueous (the clear liquid present in the front of the eye). Glaucoma may cause eye pain, vision loss, and redness, however, it can be totally without symptoms so it is important to be assessed for it.

Ectopia lentis

It is well established that Marfan Syndrome is associated with the displacement of the lens from its normal position in the eye. It is the most common heritable cause of ectopia lentis.



Displacement of the lens

The displacement of the lens can range from a subtle tilt to a complete dislocation and usually involves both eyes symmetrically. Displacement occurs due to the disruption of the zonular filaments, the tiny, thread-like structures rich in fibrillin that act like little strings holding the lens of your eye in its natural position.

Ectopia Lentis can present as Redness, Pain, Poor near vision, and Monocular diplopia (Double vision with one eye open). Ectopia Lentis is the abnormal location of the lens. It may be asymptomatic, particularly if mild. In children particularly, Ectopia Lentis may be identified incidentally. Symptoms depend on the extent and direction of the dislocation. Dislocated lenses may develop cataracts earlier. A cataract is the cloudiness of the otherwise clear lens that results in hazy vision.

Assessment of your eye

Before your appointment: It will be helpful if you can bring someone along to your appointment. Lots of information will be shared with you and it can be useful to have someone else listening too. Moreover, the use of eye drops to dilate your pupil before examination will temporarily blur your vision due to which you cannot drive yourself back home. Write a list of questions before you go to the hospital. Bring a list of all your medications and doses. Let the team know about any allergies you have.

Examination of your eye

Your doctor will first examine your eyes for any visible deformities and evaluate your squint (if present). This is then followed by:

Assessment of visual acuity

Refraction and Retinoscopy: Refraction will help assess the correction required for short-sightedness or astigmatism. Astigmatism refers to a condition causing blurry eyes due to an irregularly curved cornea or lens. Refraction is performed by using various lenses and having you read from a Snellen chart to determine your ideal prescription, and/or using retinoscopy. In retinoscopy, light is shone into your eye and its reflection is used to determine your eyeglass or contact lens prescription. Dilating drops may be used for retinoscopy.

Slit Lamp Examination: The slit lamp is an instrument microscope that allows for a thorough examination of the front and back of your eyes after dilation of the pupils.

Keratometry: Keratometry measures the curvature of your cornea. It determines whether astigmatism (if present) is primarily corneal or due to a dislocated lens.

Tonometry: A test that involves gently pressing on your cornea with a puff of air and/or by a probe on a slit-lamp to measure the pressure in your eye, which helps doctors check for glaucoma, a condition where high pressure can damage your vision. You may experience mild discomfort.

A and B-Scan Ultrasonography: This is an ultrasound of your eye. Sound waves are used to create images of the inside of your eye. It is painless and helps doctors assess short-sightedness due to a lengthy eyeball or to rule out retinal detachment.

Slit Lamp Examination

Refraction and tonometry will be first done in the same setting, usually within 30 minutes. Dilating drops will be used after that. Once your pupils are adequately dilated, the remaining tests, notably the slit-lamp examination, will be done. Not all tests will be done necessarily.

Management

A displaced lens can be managed conservatively. Visual correction with appropriate glasses or contact lenses may be sufficient. It is important to note that the best form of visual correction varies among individuals with Marfan Syndrome and can be discussed with your ophthalmologist and optometrist.

Eyeglasses and Contact Lenses: Your doctor will consider the following before prescribing your eyeglasses or contact lenses:

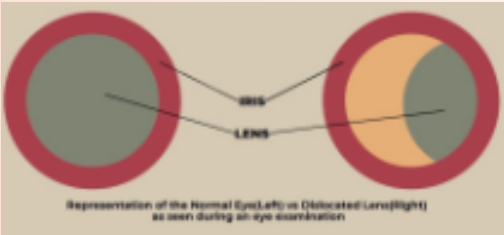
Whether your glasses should use the dislocated lens or ignore it

Whether or not the dislocation is one or both eyes.

You may be prescribed eye drops to use daily to keep your pupils dilated. This makes glasses that ignore your dislocated lens function better.

When is surgery required?

- Severely dislocated lens
- The required level of vision is no longer achievable using glasses or contact lenses
- Troublesome fluctuation of vision caused by the instability of the lens
- For prevention of amblyopia (lazy eye)
- Dislocation of the lens into the anterior chamber
- Lens-induced glaucoma or uveitis
- Visually significant cataract



Representation of the Normal Eye(Left) vs Dislocated Lens(Right) as seen during an eye examination

Reasons for avoiding lens removal and replacement, despite complete lens dislocation, particularly in young children are:

1. A dislocated lens, though lacking in its visual function, is still essential for balancing the pressures in the eye between the anterior and posterior chambers.

2. Removing the lens in young children increases the likelihood of retinal detachment and complicates lens replacement surgery later in life, when the eye has reached full maturity, making it more hazardous and technically challenging.

A healthy retina and an eye that has stopped growing enable better measurements and calculations for the lens implant, hence, lens removal and replacement is best delayed until the later teenage years. Thus, the course of treatment varies among individuals with MFS and is best discussed with your ophthalmologist.

It is normal to feel conflicted about the removal of the lens. Here are some issues you can discuss with your doctor about removing it:

- Finding the right eyeglasses for correction of vision - It takes more time and effort to prepare and find glasses that either make use of or ignore the dislocated lens

- The need for special thick glasses which are not made in most stores and often distort peripheral vision, that is, cause vision to the side to be blurry and wavy

Not all patients may have the option to keep their lens and it would be best to remove and replace it, especially in the presence of other eye conditions. Your doctor will discuss the management alongside the existing condition.

Intraocular lenses (IOLs)

Intraocular lenses (IOLs) are artificial lenses that serve to replace the refractive power of the natural crystalline lens removed. Various approaches are available for lens replacement. Not all will be suitable for an individual. The appropriate one for you will be best discussed and explained by your doctor.

The indications for surgery and ocular and medical comorbidities will affect the choice of IOL and its placement. The methods of IOL fixation are as follows:

1. Capsule-Fixated:

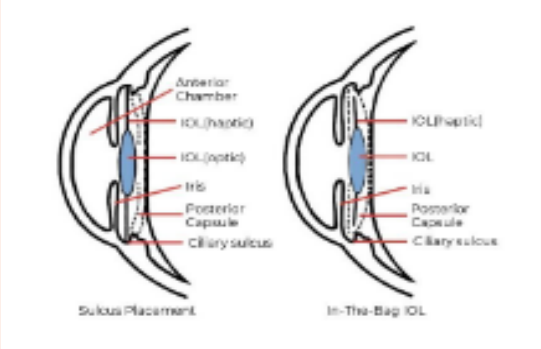
   – In-the-bag
   – Sulcus placement
   – Passive sulcus fixation

2. Scleral Fixated:

   – Sutured
   – Sutureless

3. Iris Fixated

4. Anterior Chamber IOLs



The choice of IOL is done on an individual basis as there is no consensus on the IOL-of-choice for the replacement of the natural lens in MFS patients. Choosing the appropriate IOL will be a mutual decision made with your surgeon. They will be able to give you their recommendations based on your visual goals and the surgical risks involved. It is important to ask questions and discuss your concerns so the surgeon can provide all the information you need to make an informed decision about the surgery you need.

Capsule-Fixated

In-The-Bag: It can be done when the capsular bag is intact or salvageable. The natural bag that held the human lens will now hold the man-made lens. Though the capsule is preserved, gradual loss of zonular function may continue and can be exacerbated by the surgery.

Sulcus Placement: An IOL is placed in the ciliary sulcus. This can be done when there is a defect in the capsule over the back of the lens that would prevent its placement inside the bag or due to loose zonules.

IOLs can be placed in the sulcus with an optic capture to prevent them from getting stuck behind the iris, coming out of place, and causing problems with vision. Passive sulcus fixation, however, requires an intact anterior capsule and complete 360-degree zonular support.

Scleral-Fixated

Scleral-fixated IOLs can be divided into sutured and sutureless techniques. Scleral-sutured lenses can be sutured from inside the eye or by passing either needles or forceps from the outside of the eye to the inside.

The sutureless technique involves embedding the haptics (the arms/loops of the lens that hold it in place within the eye) of an IOL within the sclera. There are currently two main methods of sutureless intrascleral fixation: flanged and “glued” techniques.

Iris-Fixated Posterior Chamber IOL

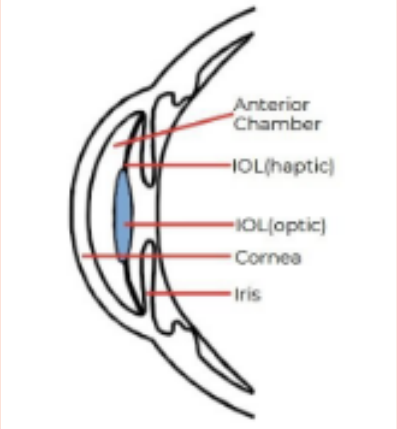
The IOL is placed behind the iris and is secured to it by enclavation in which the “claws” of the IOL entrap small snips of the iris tissue.

Iris fixation is falling out of favour as scleral-fixated IOL placement becomes more popular. This technique is useful if an intraocular lens is in the sulcus and dislocated or if the conjunctiva needs to be spared.

Anterior Chamber IOL

The IOL is placed in the anterior chamber of the eye, that is, the front part of the eye between the cornea and the iris:

Selecting the correct size of the ACIOL is essential for decreasing complication rates. ACIOLs are implanted with additional caution or avoided in the following scenarios:



– Younger patients, particularly in children
– Those with decreased corneal endothelial cell counts
– Chronic ocular inflammatory disease
– Abnormal irides
– Shallow anterior chamber
– Glaucoma

Patient anatomy, such as availability or integrity of iris/capsule support, narrow angles, and thickness of sclera strongly influence the placement of a secondary IOL. Patients with certain comorbidities such as uveitis or glaucoma may not be the best candidates for certain techniques.

Knowledge Is Power!

It is important to remember that not all doctors agree on which kind of lens to use for people with Marfan syndrome. Ask your doctor about the features of your eyes and any questions relevant to helping you understand the reasons behind your doctor's lens choice. These features include the depth of your anterior chamber and the nature of your retina. Some people are at a greater risk of retinal detachment while some have too small an anterior chamber for an ACIOL. Ask your doctor if you have those features.

Before your procedure begins...

Your doctor will numb your eye with eye drops or an injection around the eye to ensure you don't feel pain during the procedure. Mild sedation may also be given to help you relax during the surgery. Surgery is usually done in under 30 minutes. Following surgery, you will be observed for an hour, following which you can head home, on the same day of your surgery.

Postoperative Period

Dos after surgery for a smooth recovery:

1. Using eye drops as prescribed

- These help to prevent infection and aid recovery
- Be sure to administer them with clean hands to the treated eye
- Use them as advised: stop per your consultant's advice

2. Resting and avoidance of strenuous activities

- It is important to give adequate time for your eyes to heal
- Housework or exercise is best avoided during the period of rest advised by your consultant. It is usually the first two or three days after surgery.
- You can still enjoy reading, watching television, or using your computer

3. Wearing eye shield or glasses outside

- You should use these for at least a week following the procedure to prevent irritants from entering your eyes. Your consultant will be able to advise you further.
- Make sure to wear your eye shield during sleep to prevent you from rubbing your eyes

4. Taking painkillers if you need them

- It is normal for patients to experience feelings of soreness, grittiness, and watering in their eye
- You may use Paracetamol or ibuprofen along with your prescribed eye drops for pain relief

5. Showering and bathing as usual after surgery

- Avoid any water or irritants, such as shampoo or soap, getting into your eye
- It is generally advised to wipe your face with water rather than splashing water in the first few weeks
- Use cotton wool or gauze dipped in cooled, boiled water to wipe your eyes from the inner to the outer corners
- Avoid wiping the inside or putting pressure on your eye while wiping
- Always ensure hand hygiene when wiping

Don’ts after surgery

1. Don’t drive home after your surgery

- Do arrange for someone to drive you home after your operation
- You won’t be able to drive after cataract surgery until you meet the DVLA requirements for vision (being able to read a number plate at 20.5 meters with both eyes open)
- Your eyes should take around three to four days to recover to this point
- Your consultant will let you know when you can start driving again during your postoperative appointment

2. Don’t rub your eyes while they are healing

3. Don’t wear eye makeup for the first few weeks

4. Don’t swim, use a hot tub or sauna

- This is because water can carry bacteria which can potentially lead to an infection in your eye
- It’s recommended that you don’t swim for between four and six weeks following your operation
- You can ask your consultant further when you can resume swimming after the procedure

Eye Emergencies

Retinal Detachment

Retinal detachment is a separation of the light-sensitive membrane in the back of the eye (the retina) from its supporting layers. Marfan syndrome significantly increases a person’s risk of retinal detachment. 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
- A gray curtain moving across your field of vision

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

Surgery is required to repair a retinal detachment. Immediate treatment is recommended within the first 24 hours; the longer the wait, the greater the chance that the retinal detachment will become more severe and the more complex the surgery required.

References

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- Chandra, A., Charteris, D.G. (2016). Ophthalmic Abnormalities in Marfan Syndrome. In: Child, A. (eds) Diagnosis and Management of Marfan Syndrome. Springer, London. https://doi.org/10.1007/978-1-4471-5442-6_11
- Chandra A, Charteris D. Molecular pathogenesis and management strategies of ectopia lentis. Eye (Lond). 2014 Feb;28(2):162-8. doi: 10.1038/eye.2013.274.
- Garg, S., & Koch, D. (Eds.). (2023). Steinert's Cataract Surgery (4th ed.).Elsevier.

Useful Links

https://www.marfantrust.org/articles/lucy-s-new-focus

https://www.marfantrust.org/articles/dr-child-s-casebook-out-of-focus-

https://www.marfantrust.org/articles/dr-child-s-casebook-delayed-focus

https://www.youtube.com/watch?v=jeM0P0sNtos&t=32s

https://www.youtube.com/watch?v=YbH9xhO3huY

About the Marfan Trust

Co-founded in 1988, the Marfan Trust is the sole charity in the United Kingdom dedicated to improving and saving the lives of those with Marfan syndrome. It is estimated that approximately 18,000 people are living with Marfan syndrome in the United Kingdom and around half of these remain dangerously undiagnosed.

The Marfan Trust's three main objectives are to:

- Provide personalised support and medical guidance through its helpline;
- Conduct cutting-edge medical research through its self-funded Sonalee Laboratory, named after a young doctor who tragically died of complications from MFS during her ward round;
- Continue to provide educational information and raise awareness of the condition.

How you can help

Marfan Trust
Supporting Research into Inherited Aortic and Related Disorders

Loeys Dietz

You can help to secure the Marfan Trust's future by becoming a member today for just £3 per month:

www.marfantrust.org/pages/10-membership

JustGiving

Just Giving – **http://bit.ly/3Scj51w**

PayPal

PayPal Giving – **https://bit.ly/45NCuwQ**

By donating to the Marfan Trust, you are contributing to an ever-growing body of knowledge on the condition, allowing more doctors and medical specialists to deliver the best possible treatment to patients affected by Marfan syndrome.

- BANK: Charities Aid Foundation (CAF) ACCOUNT NAME: Marfan Trust
- SORT CODE: 40-52-40
- ACCOUNT NUMBER: 00017677
- REFERENCE: Your Name (plus campaign name if relevant)

You can also help to fund a piece of equipment in Dr José Aragon-Martin's Sonalee Laboratory. Email info@marfantrust.org to find out more.

Registered charity number 1198847

THE POWELL FAMILY FOUNDATION