# Ophthalmic Abnormalities in Marfan Syndrome

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The ocular manifestations of Marfan Syndrome (MFS) are varied. They include myopia, corneal flattening, ectopia lentis (EL), and retinal detachments. The prevalence of these in MFS is unclear, though a recent survey of the UK Marfan Trust, suggested that 30 % of patients with MFS had EL, and 15 % had had retinal detachments [1]. We will discuss the most important of these features below.

## Ectopia Lentis (Lens Subluxation/Dislocation)

Ectopia Lentis (EL) is the most diagnostic ophthalmic abnormality in Marfan syndrome [2]. Alternate conditions may cause EL [3, 4], thus careful genetic investigations must be undertaken to definitively diagnose or exclude MFS in the presence of EL [5]. The subluxation is due to weaknesses of the lens zonular fibres secondary to abnormal fibrillin. Fibrillin-1 is a critical structure in these zonules, and thus it is unsurprising that these are affected in MFS. Subluxation of the lens is usually bilateral and symmetrical. Occasionally, complete dislocation of the lens into the vitreous cavity or into the anterior chamber can also occur. The subluxation of the lens usually occurs in the first two decades and, in general, is stable once an individual reaches their twenties. The subluxed lens itself can however develop a cataract, which can lead to visual impairment. Occasionally a subluxed or dislocated lens

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A.H. Child (ed.), *Diagnosis and Management of Marfan Syndrome*,
DOI 10.1007/978-1-4471-5442-6_11

will become unstable resulting in fluctuating vision [6, 7]. Furthermore, anterior subluxation of the lens may compromise the drainage angle of the eye, resulting in raised intraocular pressure and secondary glaucoma.

Often visual acuity will remain relatively good, despite subluxation of the lens. Optical correction with appropriate glasses or contact lenses (often to correct around the lens itself in the 'aphakic' part of the pupil) will be sufficient to maintain adequate vision.

If the vision becomes significantly impaired by a cataract, if there is troublesome fluctuation of vision caused by the instability of the lens, or if an individual is intolerant to aphakic glasses or to contact lenses then surgical correction should be considered [3]. The current surgical technique for lens dislocation/subluxation in Moorfields Eye Hospital is the operation of vitreolensectomy. This uses an approach through the pars plana of the eye using automated ultrasonic fragmentation and cutting to remove the dislocated lens and the vitreous gel. Removal of both the lens and the vitreous gel minimises the chances of subsequent retinal detachment, which has been a significant complication of previous forms of surgery for lens dislocation in Marfan syndrome. At the same time that this procedure is performed a detailed examination of the retina is undertaken and laser or cryotherapy is applied to surround any areas of lattice retinal degeneration or retinal tears which might predispose to retinal detachment. This form of surgery can also be applied to eyes which have EL and a concomitant retinal detachment; the detachment itself can be repaired simultaneously often using intraocular expansile gas or silicone oil tamponade.

Our results using this technique at Moorfields Eye Hospital [8] demonstrated that only two of a total of 40 eyes did develop a retinal detachment following such a procedure and these were successfully managed with further retinal surgery. Following the surgery itself, correction with contact lenses is often adequate (many individuals will have been contact lens wearers prior to the surgery). For certain individuals a lens implant may be considered. This procedure differs from that employed in standard cataract surgery where the lens capsule and lens zonular fibres are intact. Where vitreolensectomy has been performed a lens implant may be sutured into the posterior chamber, captured in the sclera, clipped to the iris, or placed in the anterior chamber [9]. Surgical correction following removal of the lens can be undertaken simultaneously with the vitreolensectomy procedure, or as a secondary procedure at a later date.

## Myopia

Myopia is more common in Marfan syndrome patients and is regarded as a minor diagnostic feature of MFS [2]. However, myopia itself is very common, and for this reason we do not feel that it serves any diagnostic utility in MFS.

In many cases myopia is of low to moderate degree in MFS and can easily be corrected with glasses or contact lenses. Where there has been substantial subluxation or dislocation of the crystalline lens the Marfan patient may be rendered hypermetropic (long-sighted) and occasionally the refraction may be somewhat

unstable. The above refractive errors can often be adequately corrected with glasses or contact lenses. Occasionally, however, particularly where there is a large difference in refraction between the eyes, vitreolensectomy may be performed to correct a refractive error.

## Retinal Detachment

As outlined above, retinal detachment can occasionally occur as a complication of surgery to remove a lens which is dislocated or subluxed. This complication is now much less common using the modern viteolensectomy approach. Retinal detachment may occur in the absence of prior lens surgery, with our survey suggesting this to be the case in up to 80 % of RD cases in MFS [1]. Surgical management of this may involve removal of the lens by a vitreolensectomy procedure if EL is concomitant. Other manoeuvres would include a combination of laser or cryotherapy, intraocular gas injection (occasionally silicone oil may be used as a tamponade) and a silicone plastic explant placed around the eye. Generally such techniques will be successful, although in 30–40 % of cases detachment surgery will require subsequent procedures to stabilise the retina [1].

In a small percentage of cases the detachment will be caused by a giant retinal tear (defined as a retinal tear greater than 25 % of the circumference of the eye). Again, this will require vitrectomy procedure and may require silicone oil to be used as the tamponading agent.

## Other Ophthalmic Problems

Strabismus (squint) is more common in Marfan syndrome individuals [10] and may in some cases be associated with amblyopia. Abnormalities of the anterior chamber can rarely be associated with raised ocular pressure (and sometimes glaucoma [11]) and occasionally dislocation of the lens into the anterior chamber can produce acute glaucoma. This type of lens dislocation can result in an acutely painful eye and requires urgent ophthalmic management. Finally, the cornea of patients with MFS is reported to be thinner and flatter than the normal population [12]. It is therefore not advisable for these patients to consider corneal refractive surgery.

## General Ophthalmic Management in Marfan Syndrome

It is advisable that an ophthalmologist examines individuals with MFS or a family history of MFS at an early age. Regular checks of visual acuity and refractive error should also be carried out to detect any development of lens abnormalities in children. An optometrist or an ophthalmologist may carry these out. It is probably advisable that children refrain from sports where there is excessive head contact or deceleration, such as boxing or bungee jumping, but most sports such as football

and athletics are acceptable. Parents should maintain a degree of awareness for any loss of vision that the child may experience and seek prompt ophthalmic advice should this occur.

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