Eye Issues and Treatment in Marfan Syndrome and Stickler Syndrome

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The Marfan Eye

Marfan Patients could sustain all eye diseases as in general population, but specifically:

**Major problems**
- Dislocated lenses – 60%
- Pre-senile cataract – 10-20 years earlier than general population
- Retinal detachment – 10%
- Glaucoma – 30%

**Minor problems**
- Refractive errors
- Amblyopia
- Strabismus
- Iris hypoplasia and poor pupil dilation
Dislocated Lens
Dislocated Lens

Mild

Severe
Cataract removal

IOL implantation
Difficulty with cataract surgery in Marfan patients
Complications of retinal detachment
Glaucoma

Vascular etiology

Biomechanical etiology
Glaucoma is a progressive disease
Refractive Errors
Myopia

Normal Eye

Marfan Eye
Corneal Astigmatism

Lenticular Astigmatism

Normal Eye

Astigmatism

Point of Focus

Focal Point

Light

Iris

Cornea

Lens

Retina

Optic Nerve

sciera

Normal cornea

Cornea with astigmatism

Corneal Astigmatism

Lenticular Astigmatism
Amblyopia: lazy eye

Amblyopia: Early Detection Can Help Reduce Risk of Impaired Vision
Strabismus
Common Features between Marfan Eye and Stickler Eye

**Marfan** Patients could sustain all eye diseases as in general population, but specifically:
• Dislocated lenses – 60%
• Pre-senile cataract – 10-20 years earlier than general population
• Retinal detachment – 10%
• Glaucoma – 30%

**Stickler** Patients could sustain all eye diseases as in general population, but specifically:
• Myopia – 83%
• Retinal detachment – 45%
• Cataract – 36% (STL1) and 59% (STL2)
• Glaucoma – 10%
Ocular complications and prophylactic strategies in Stickler syndrome: a systematic literature review.

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Abstract

Background: Stickler syndrome is a collagenopathy caused by mutations in the genes COL2A1 (STL1) or COL11A1 (STL2). Affected patients manifest ocular, auditory, articular, and craniofacial manifestations in varying degrees. Ocular symptoms include myopia, retinal detachment, cataract, and glaucoma. The aim of this systematic review was to evaluate the prevalence of ocular manifestations and the outcome of prophylactic treatment on reducing the risk of retinal detachment. Method: A systematic literature search was performed in the PubMed database. Information on the cross-study prevalence of myopia, retinal detachment, cataract, glaucoma, visual impairment, severity and age of onset of myopia and retinal detachments. Studies that reported on the outcome of prophylactic treatment against a control group were explored. Results: 37 articles with 2324 individual patients were included. Myopia was found in 83% of patients, mostly of a moderate to severe degree. Retinal detachments occurred in 45% of patients. Generally, the first detachment occurred in the second decade of life in STL1 patients and later in STL2. Cataracts were more common in STL2 patients, 59% versus 36% in STL1. Glaucoma (10%) and visual impairment (blind: 6%; vision loss in one eye: 10%) were rare. Three studies reported on the effect of prophylactic treatment being protective. Conclusion: Ocular manifestations are common in Stickler patients, but the comparison between studies was difficult because of inconsistencies in diagnostic and inclusion criteria by different studies. Sight-threatening complications such as retinal detachments are common but although prophylactic therapy is reported to be effective in retrospective studies, evidence from randomized trials is missing.
Goals of ocular management

- Excellent and equal vision in both eyes
- Straight eyes
- Controlled glaucoma
- Prevention or treatment of retinal detachments
- Informed patients and families
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