VANDERBILT UNIVERSITY
MEDICAL CENTER

Eye Issues and Treatment in Marfan Syndrome and Stickler Syndrome

Rachel Kuchtey, M.D., Ph.D.
Vanderbilt Eye Institute
Vanderbilt University Medical Center

The Marfan Eye

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

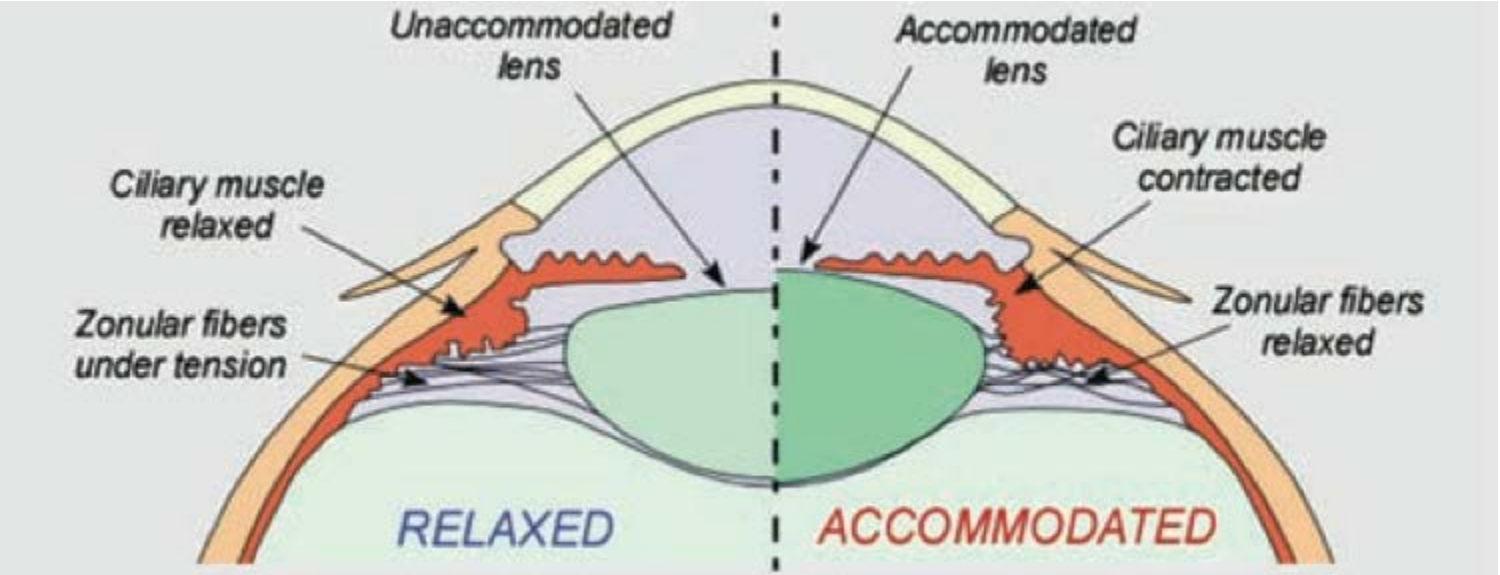
Major problems

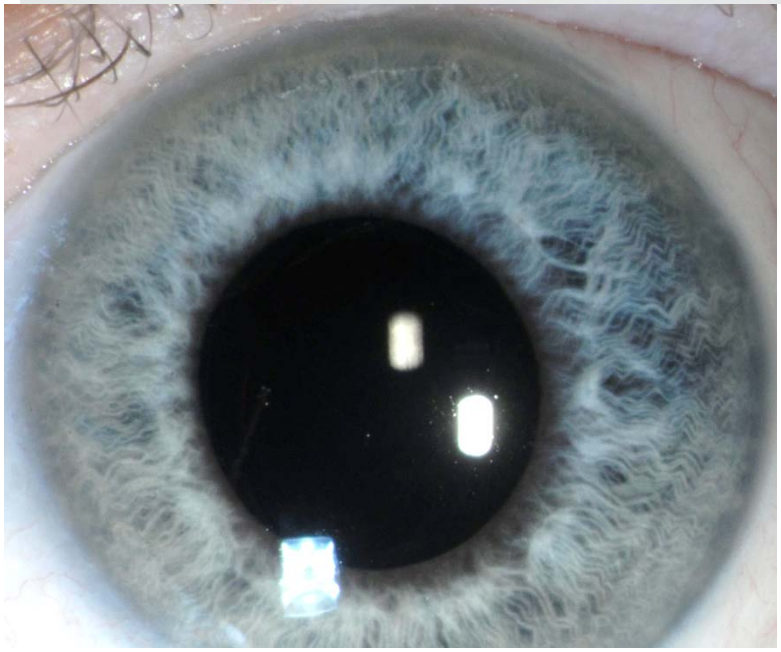
- Dislocated lenses – 60%
- Pre-senile cataract – 10-20 years earlier than general population
- Retinal detachment – 10%
- Glaucoma – 30%

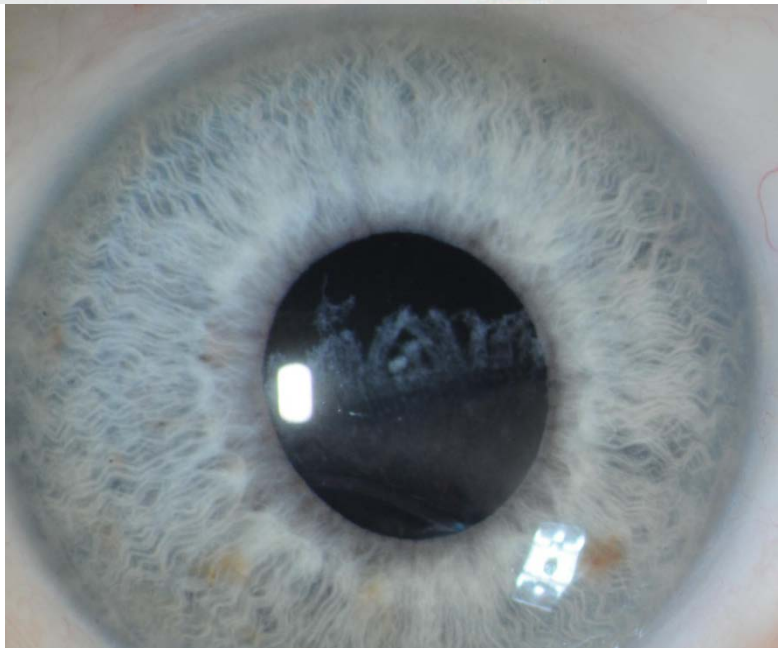
Minor problems

- Refractive errors
- Amblyopia
- Strabismus
- Iris hypoplasia and poor pupil dilation

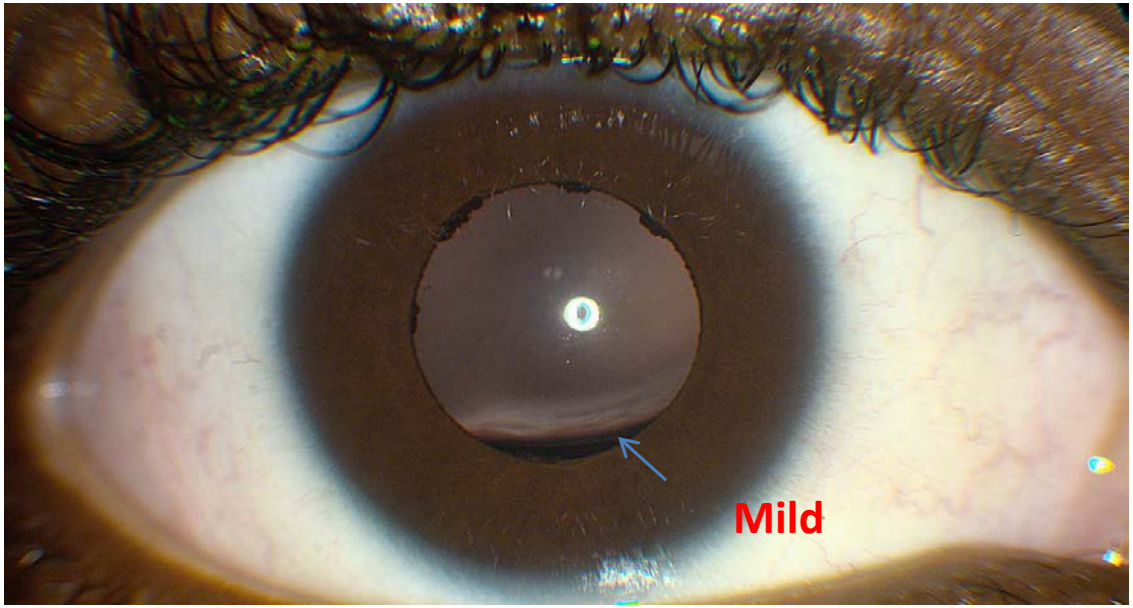
Dislocated Lens

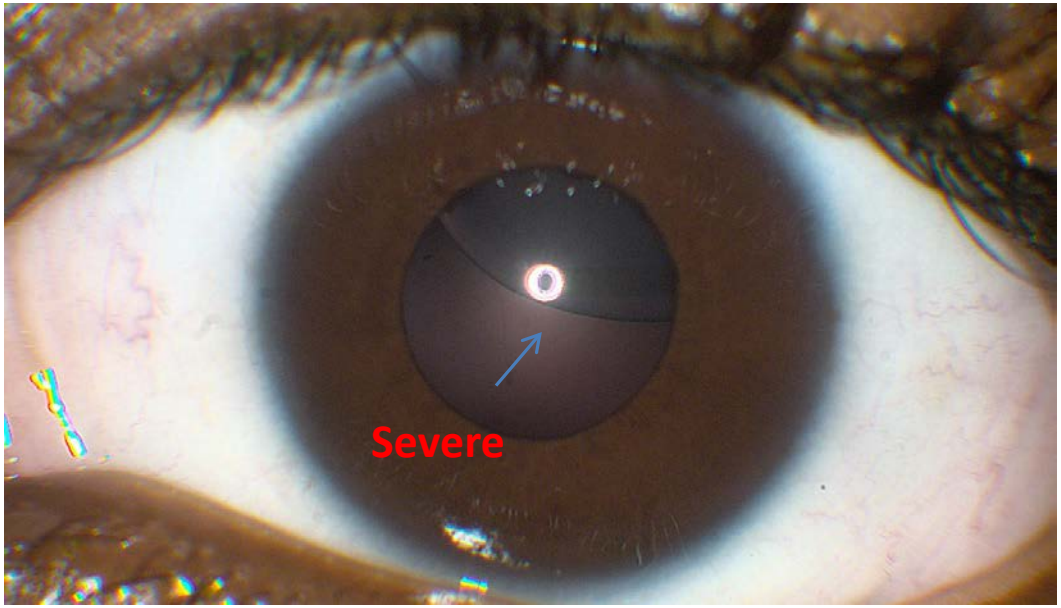




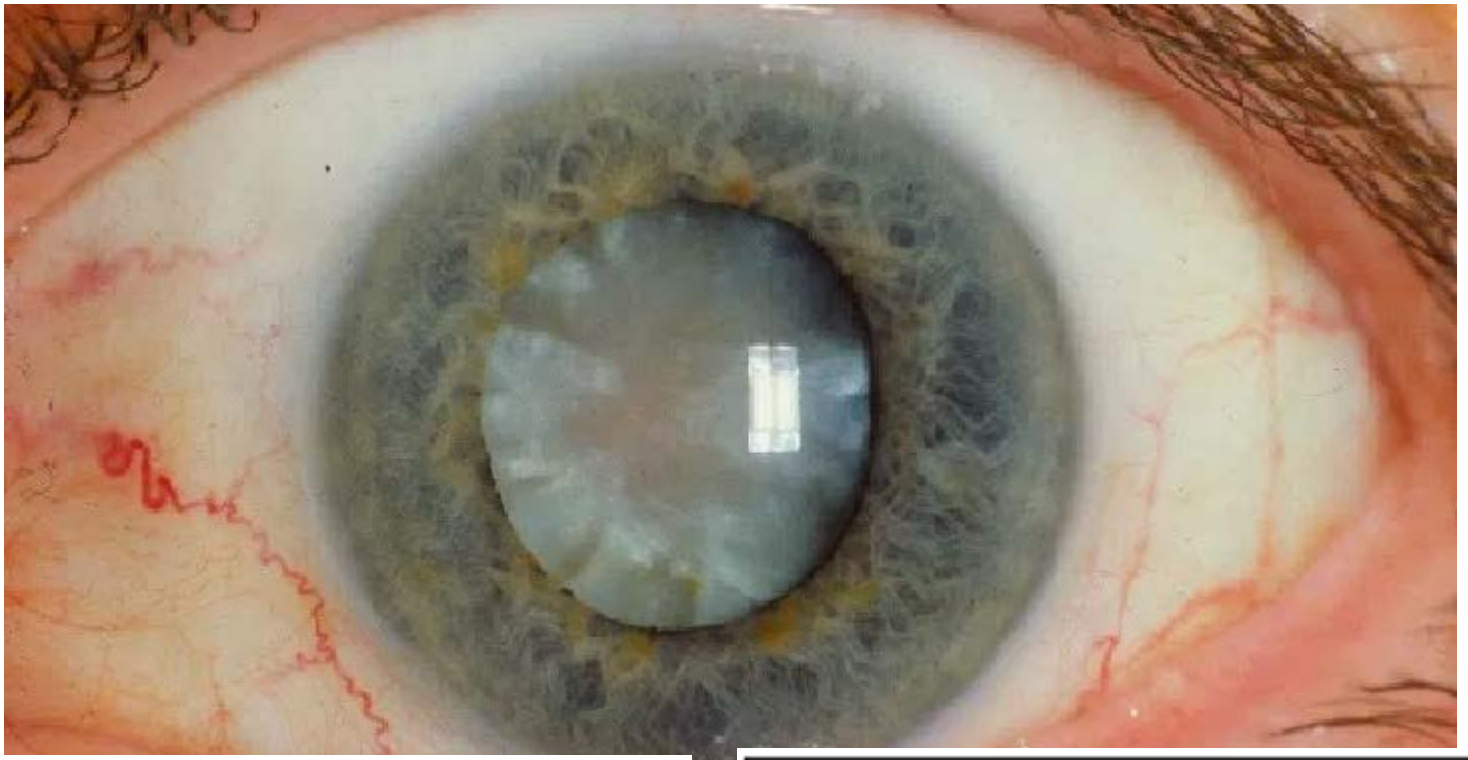


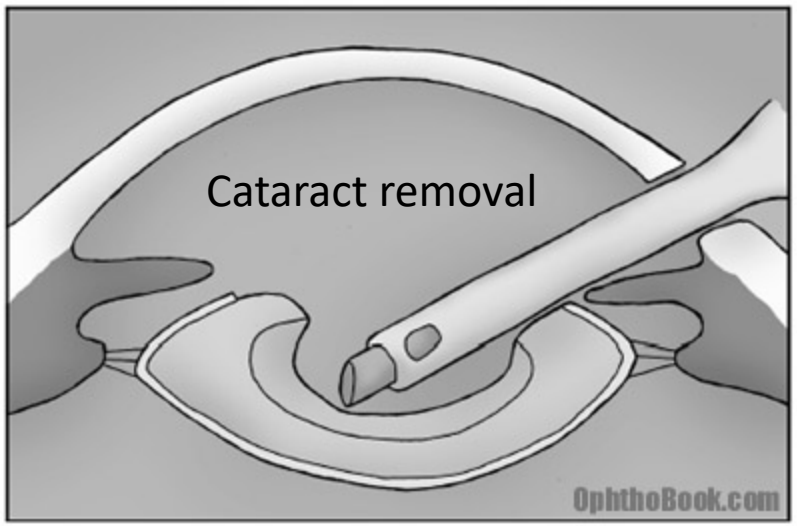
Dislocated Lens

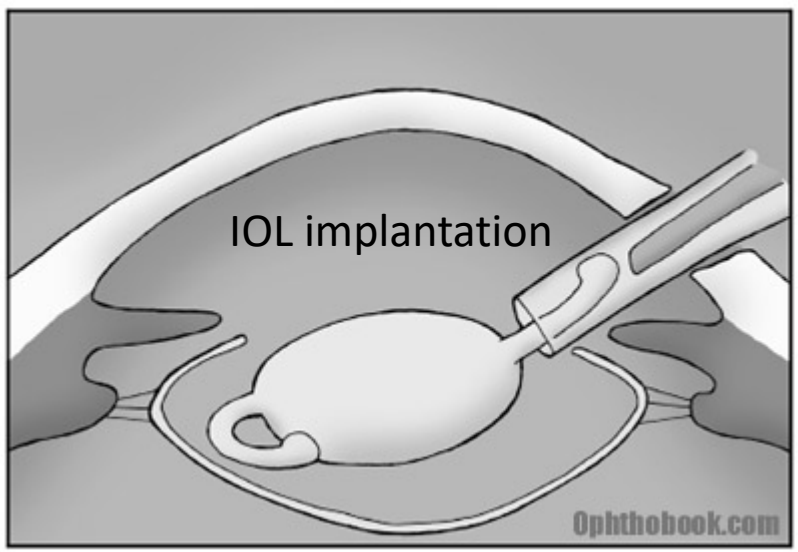




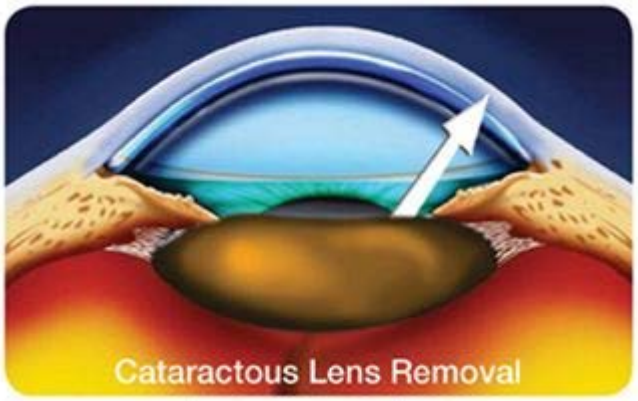
Cataract

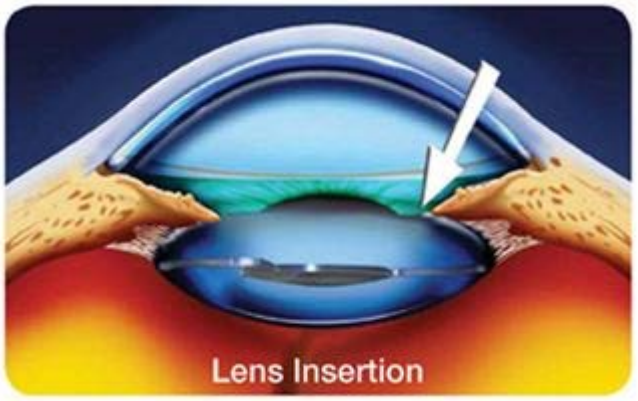


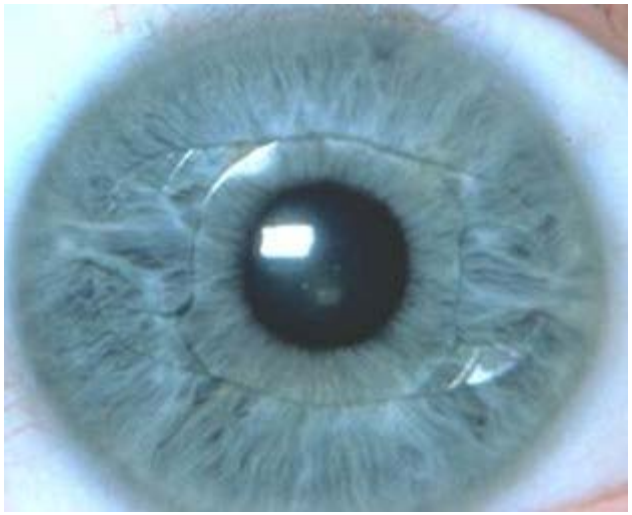

Cataract removal
OphthoBook.com

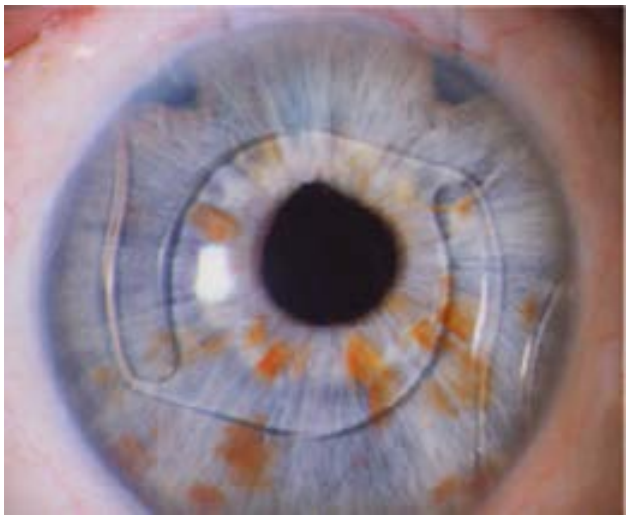

IOL implantation
Ophthobook.com

Difficulty with cataract surgery in Marfan patients

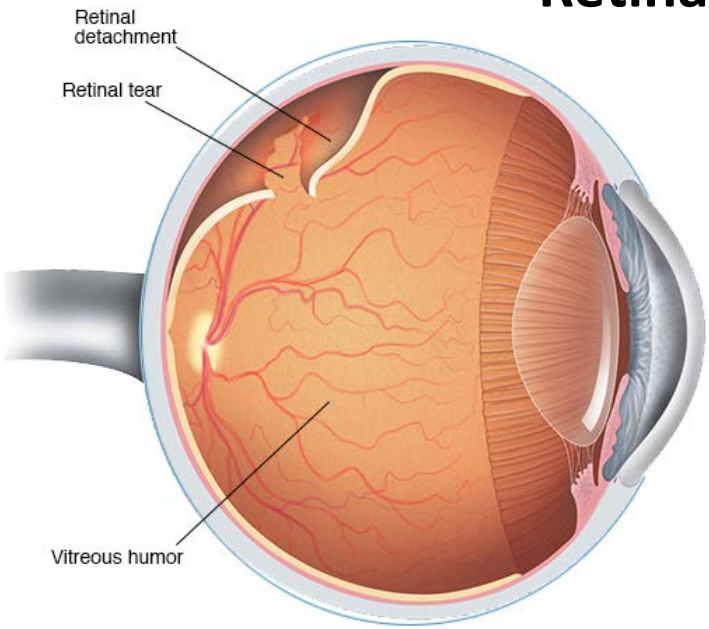


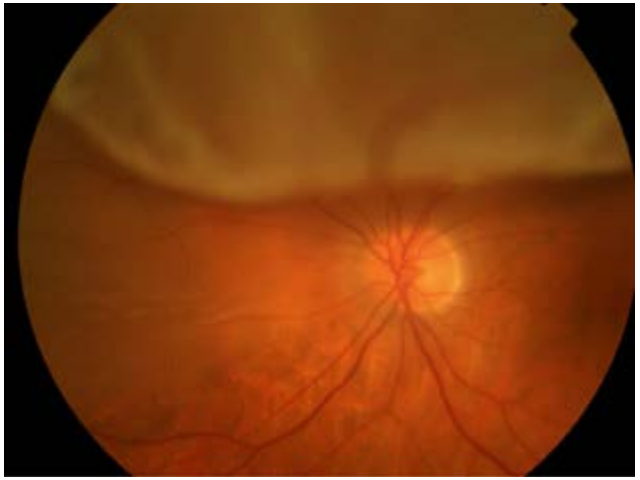




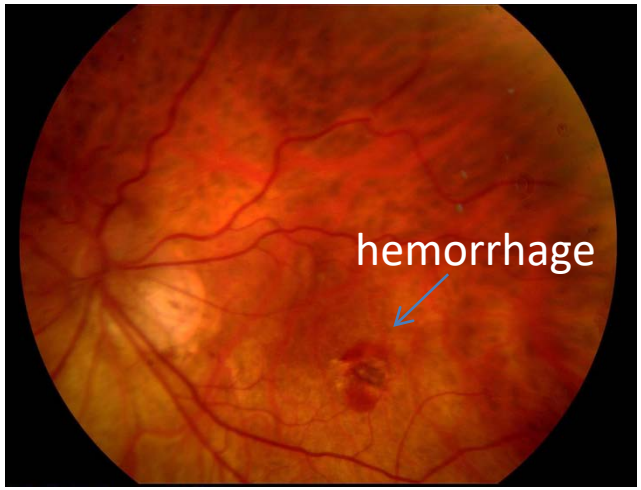


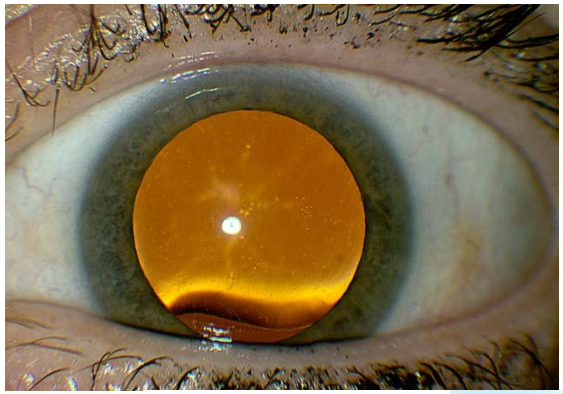
Retinal Detachment

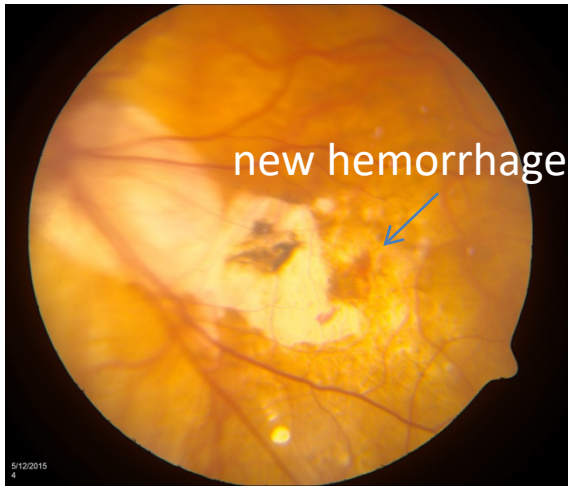




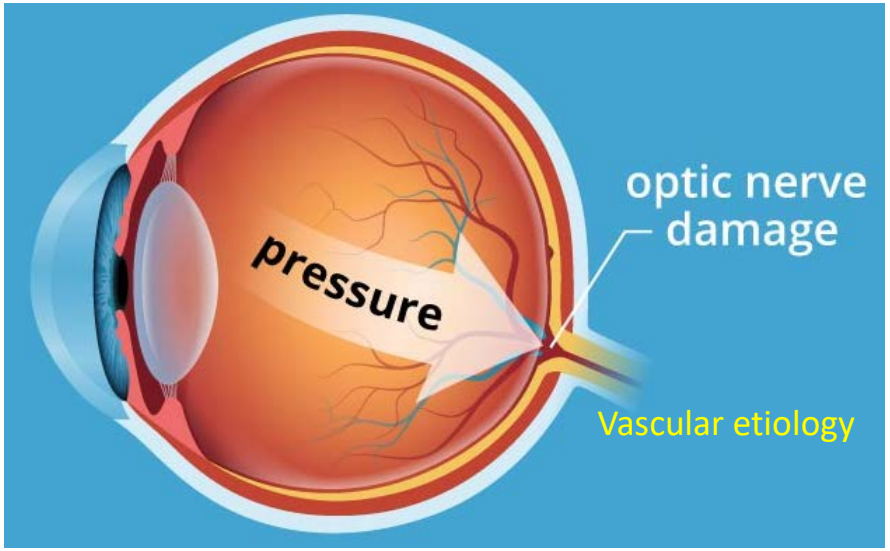
Complications of retinal detachment

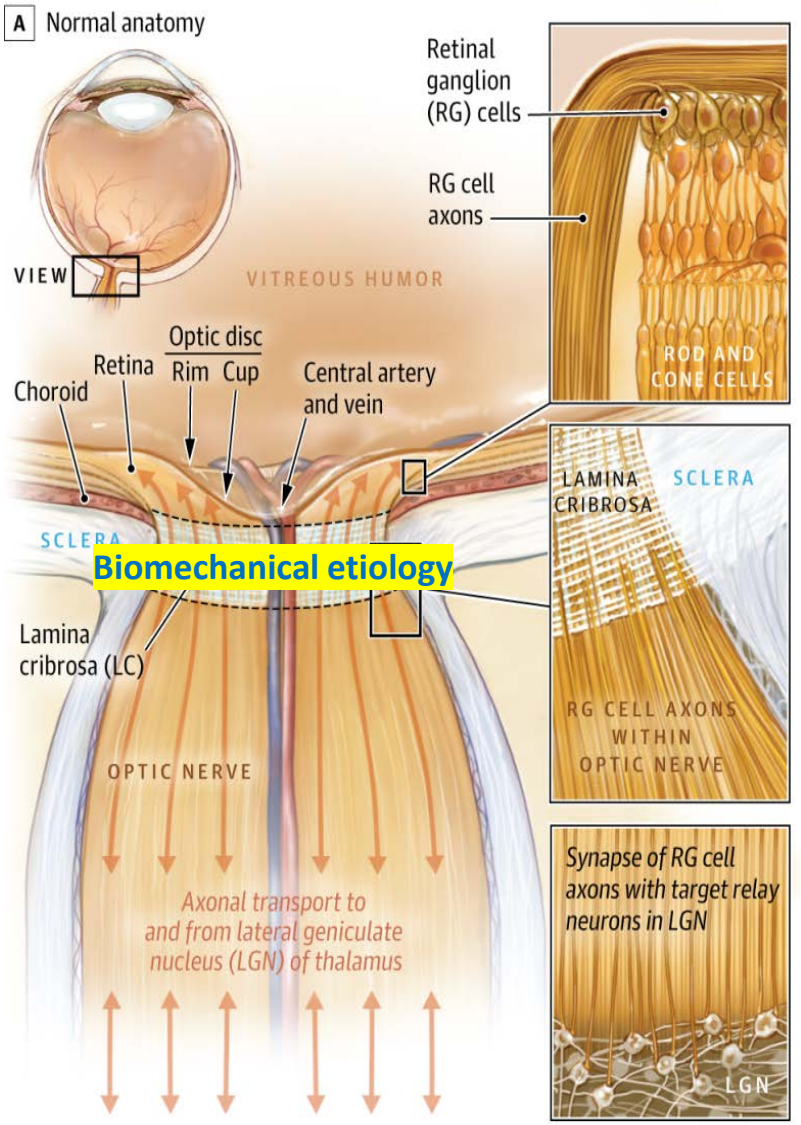




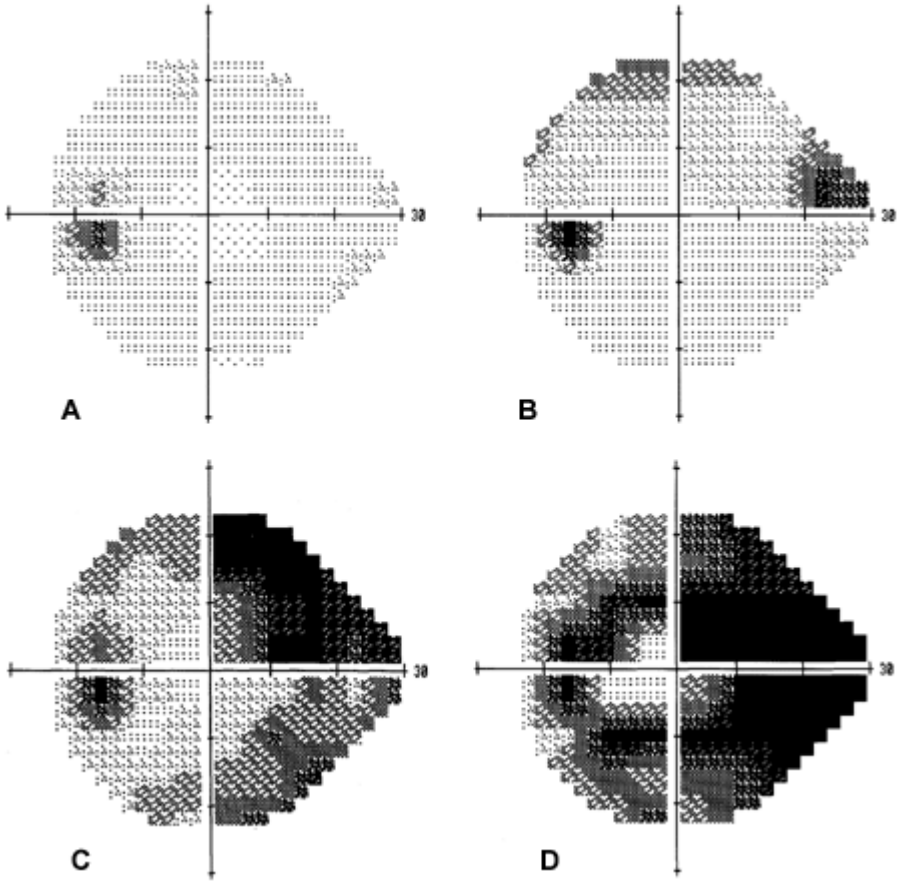


Glaucoma


pressure
optic nerve damage
Vascular etiology

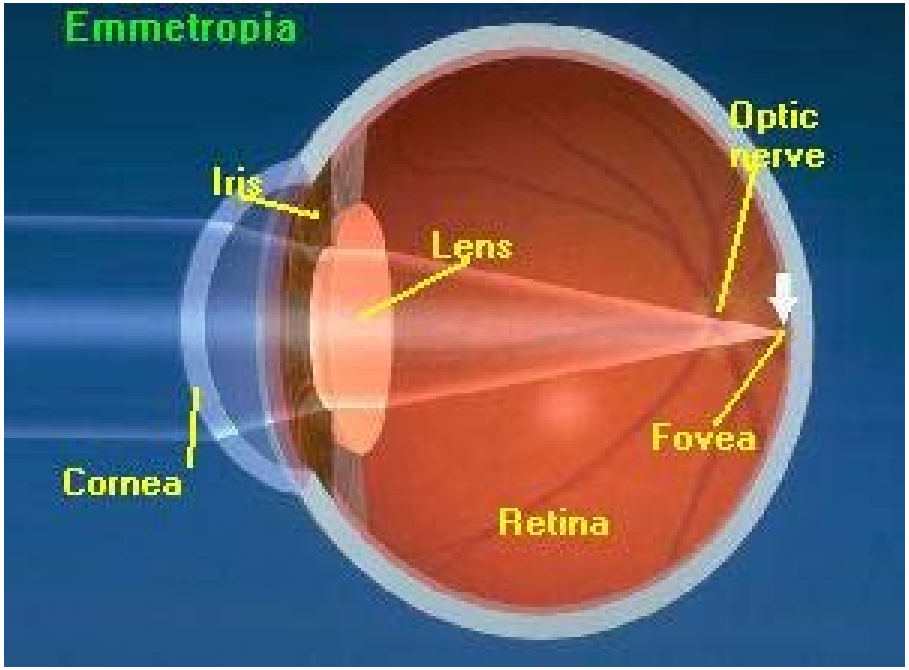

A Normal anatomy
VIEW
VITREOUS HUMOR
Retinal ganglion (RG) cells
RG cell axons
ROD AND CONE CELLS
Optic disc
Retina
Rim
Cup
Choroid
Central artery and vein
SCLERA
Biomechanical etiology
Lamina cribrosa (LC)
LAMINA CRIBROSA
SCLERA
RG CELL AXONS WITHIN OPTIC NERVE
OPTIC NERVE
Axonal transport to and from lateral geniculate nucleus (LGN) of thalamus
Synapse of RG cell axons with target relay neurons in LGN
LGN

Glaucoma is a progressive disease

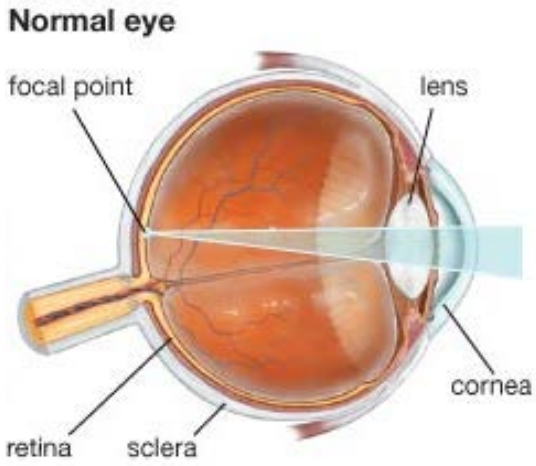


Humphrey Visual Field testing

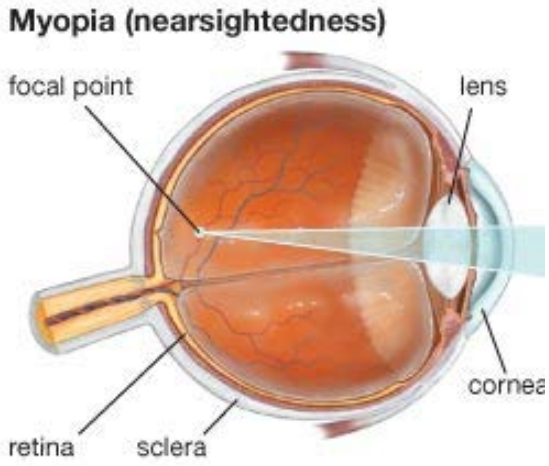
Refractive Errors



Myopia

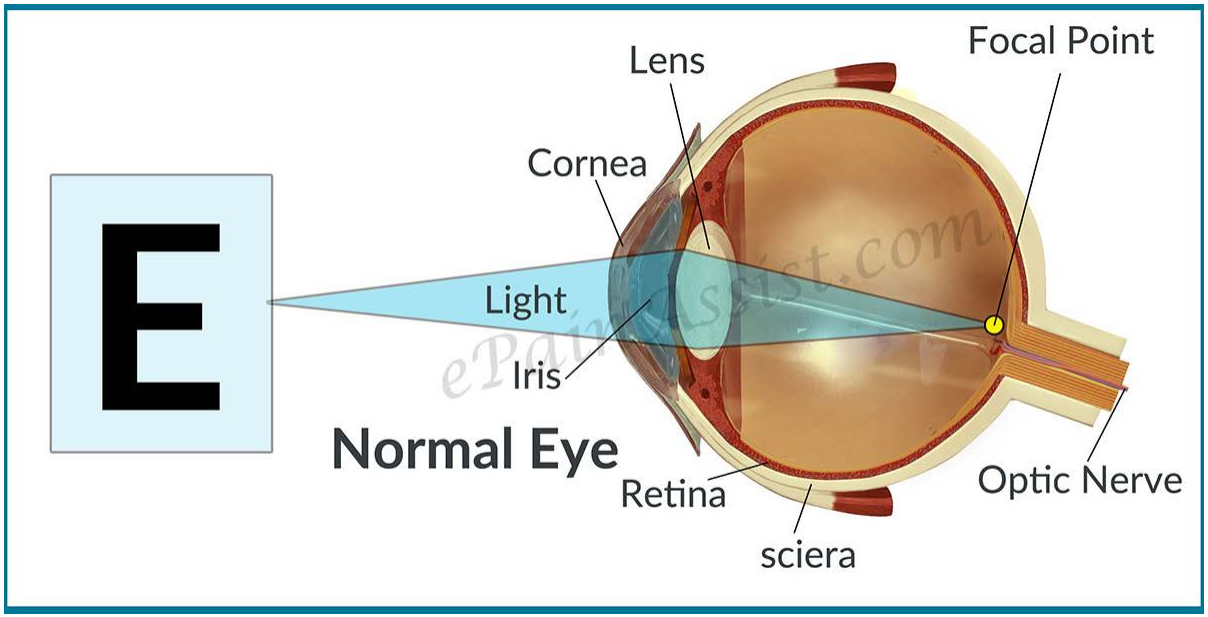


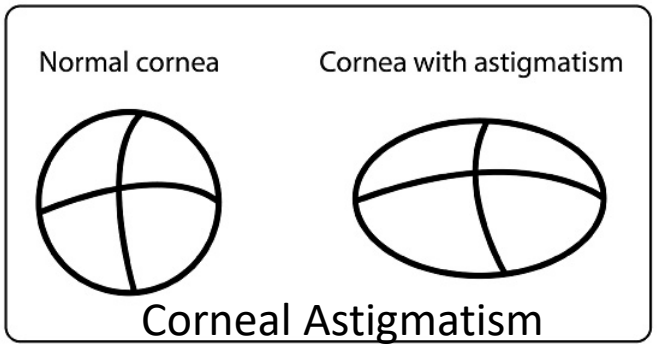
Normal Eye

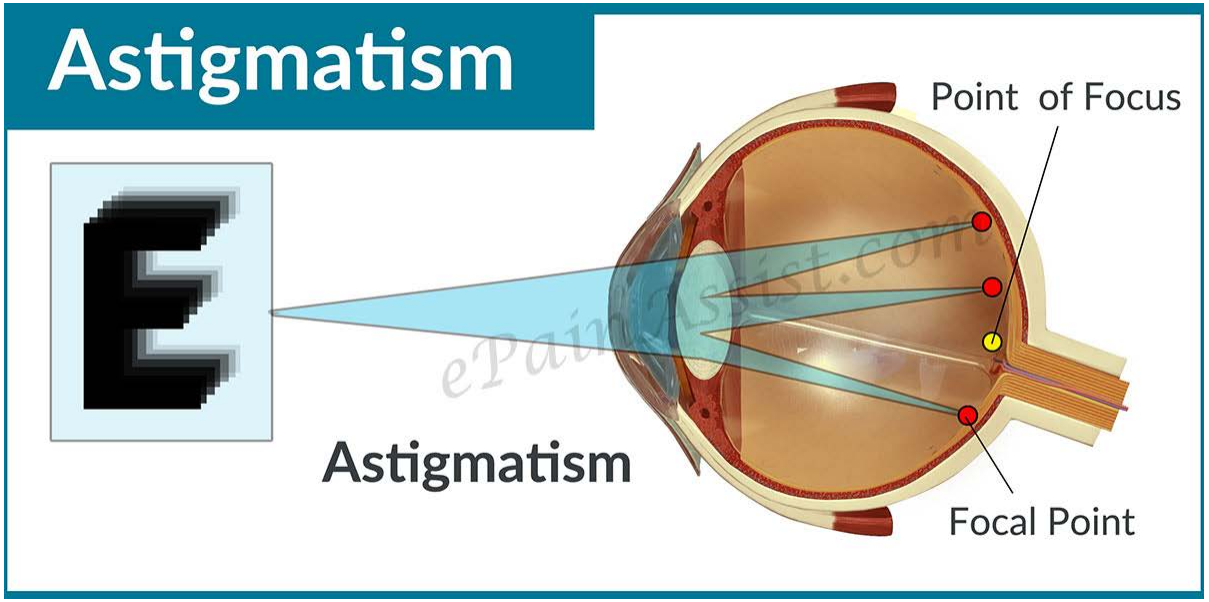


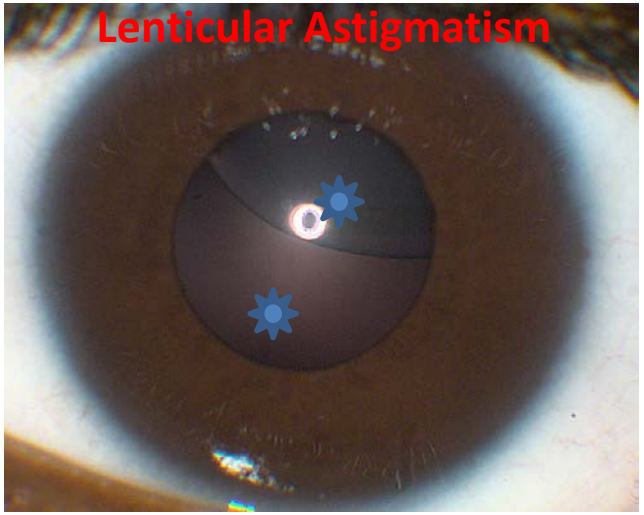
Marfan Eye

Astigmatism



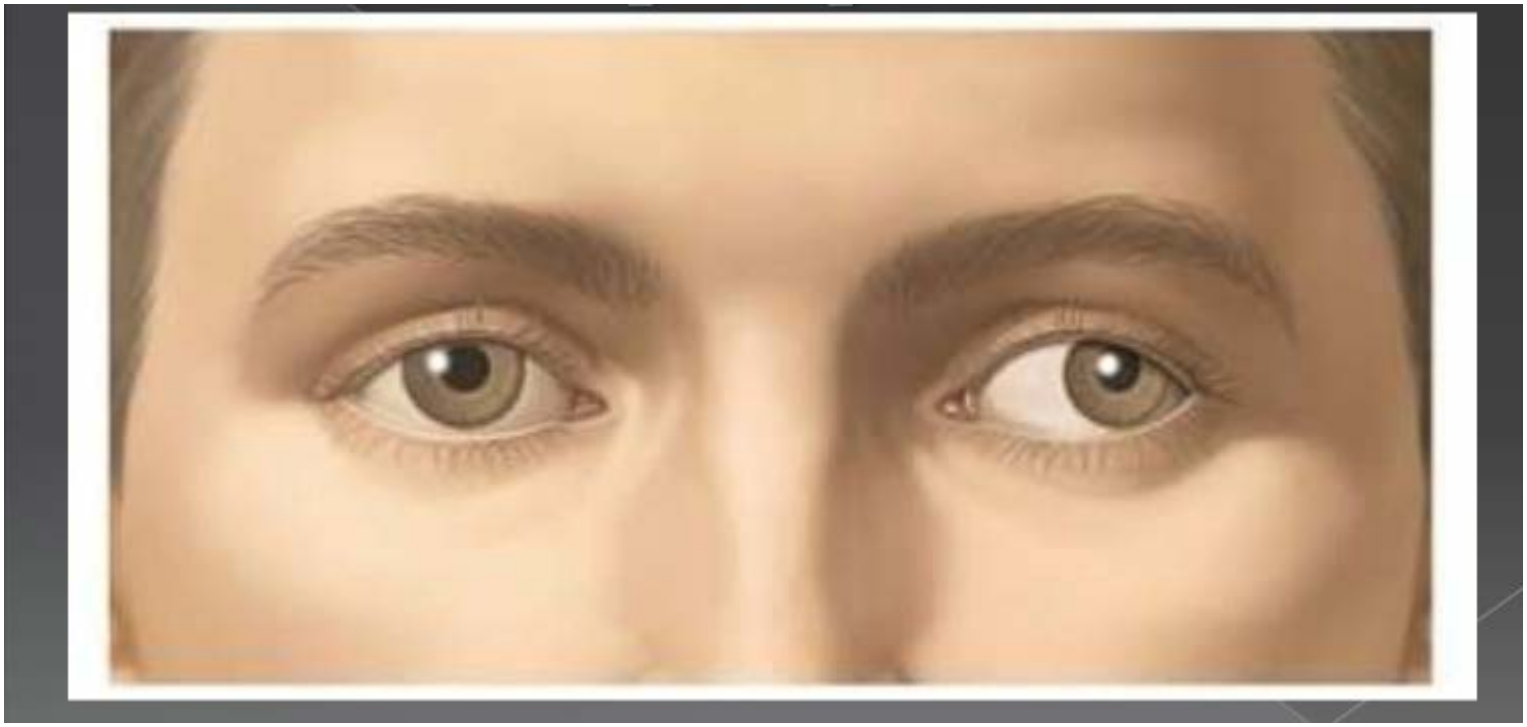






Amblyopia: lazy eye

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%

The Stickler Eye

Ophthalmic Genet. 2020 Apr 21:1-12. doi: 10.1080/13816810.2020.1747092. [Epub ahead of print]

Ocular complications and prophylactic strategies in Stickler syndrome: a systematic literature review.

Boysen KB[1], La Cour M[1,2], Kessel L[1,2].

Author information

1 Department of Ophtalmology, Rigshospitalet, Glostrup, Denmark.
2 Department of Clinical Medicine, University of Copenhagen, Copenhagen, Denmark.

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

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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Rachel Kuchtey, M.D., Ph.D.
Vanderbilt Eye Institute
Vanderbilt University Medical Center
Rachel.Kuchtey@vumc.org