Stickler Syndrome: Effects on the Eye

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Stickler Involved People

Las Vegas

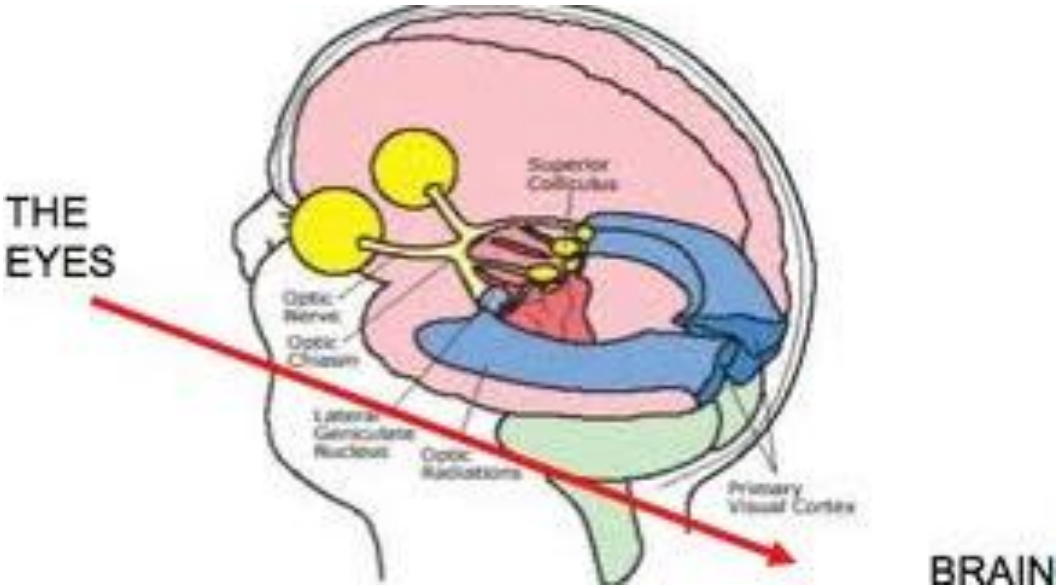
13 July 2013

Overview

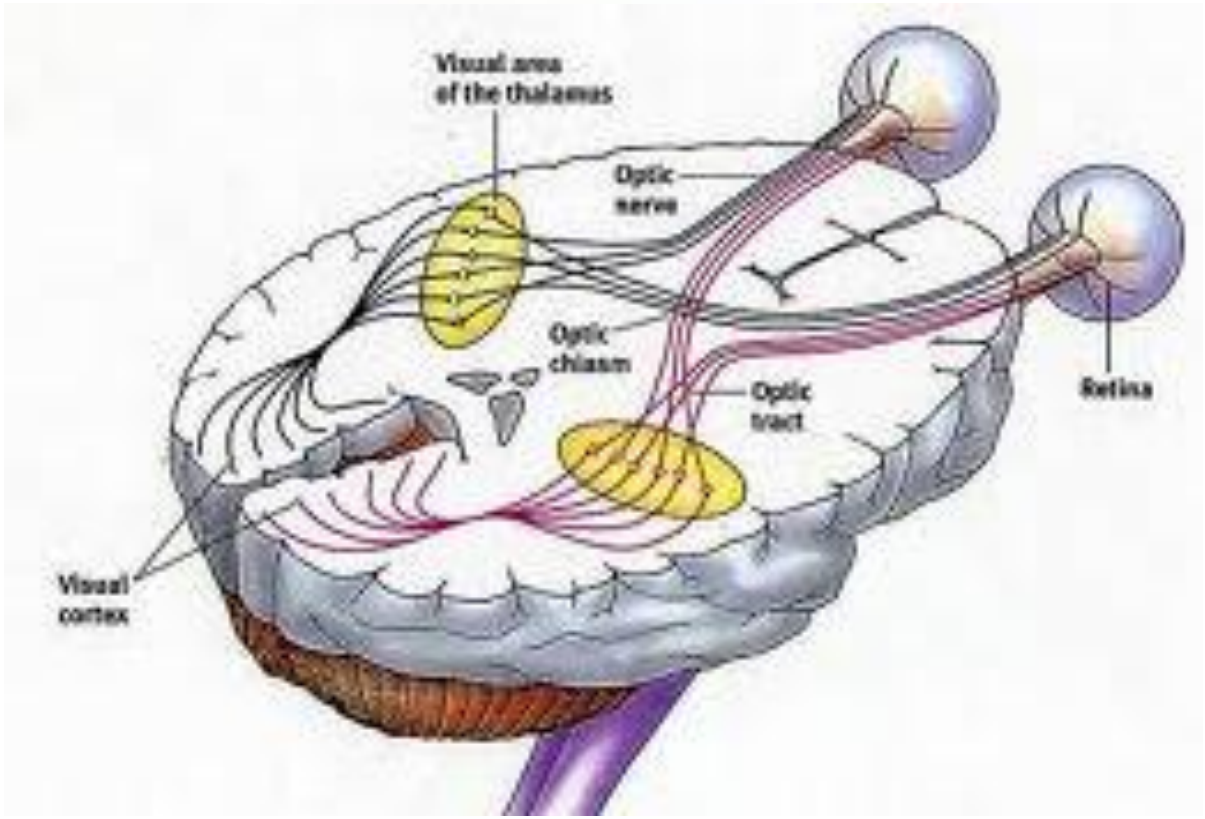
- Vision and the Eye
- Retinal Detachment basics
- Challenges of Stickler Syndrome
- Future ideas

Vision

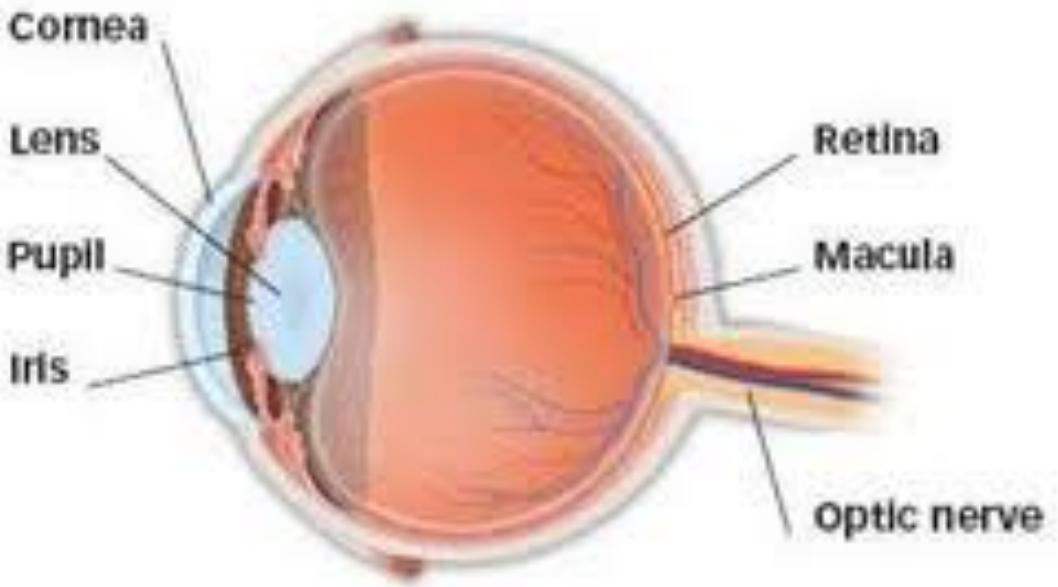
- The Eyes are sensory organs
- The Brain “sees”

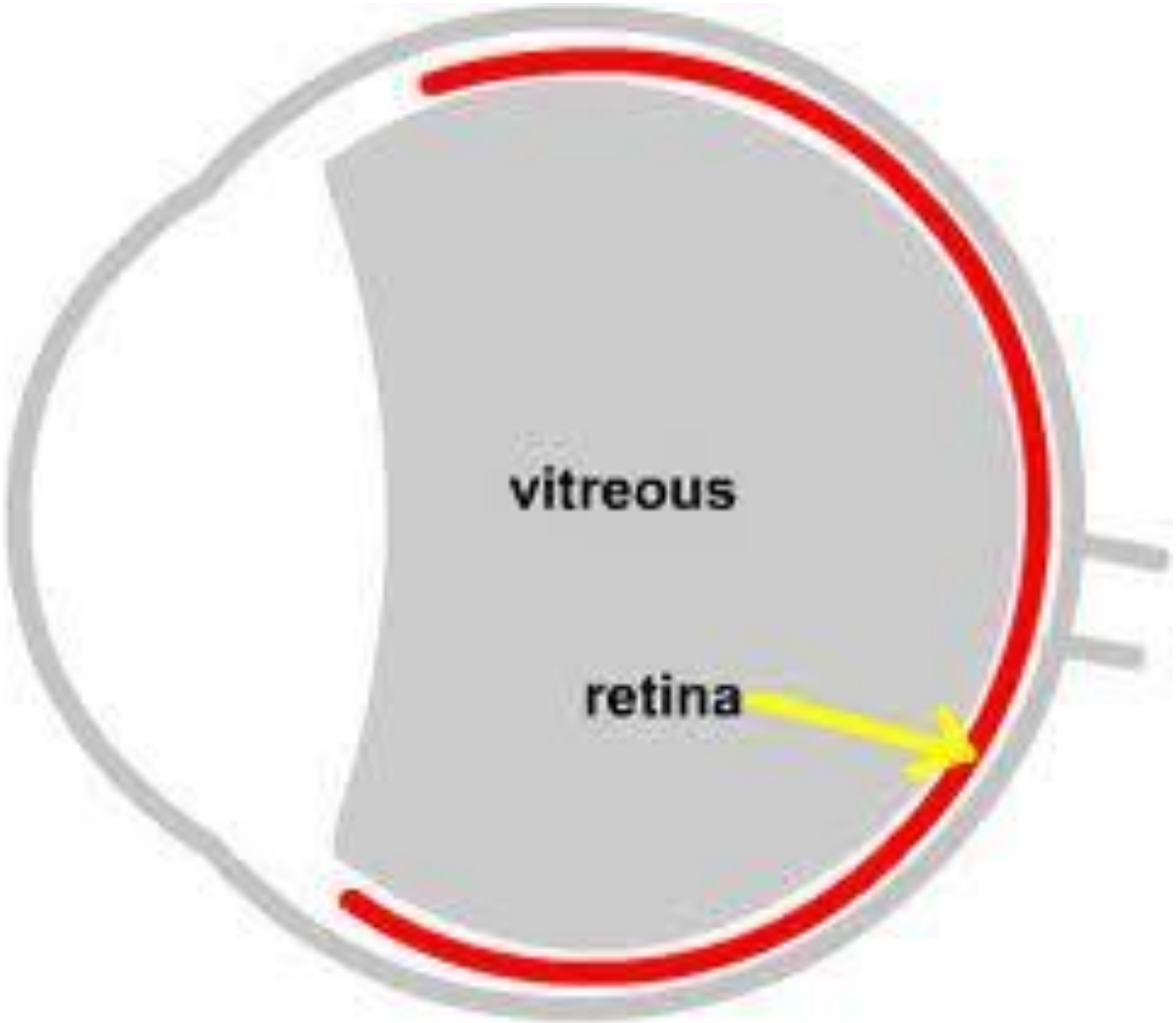


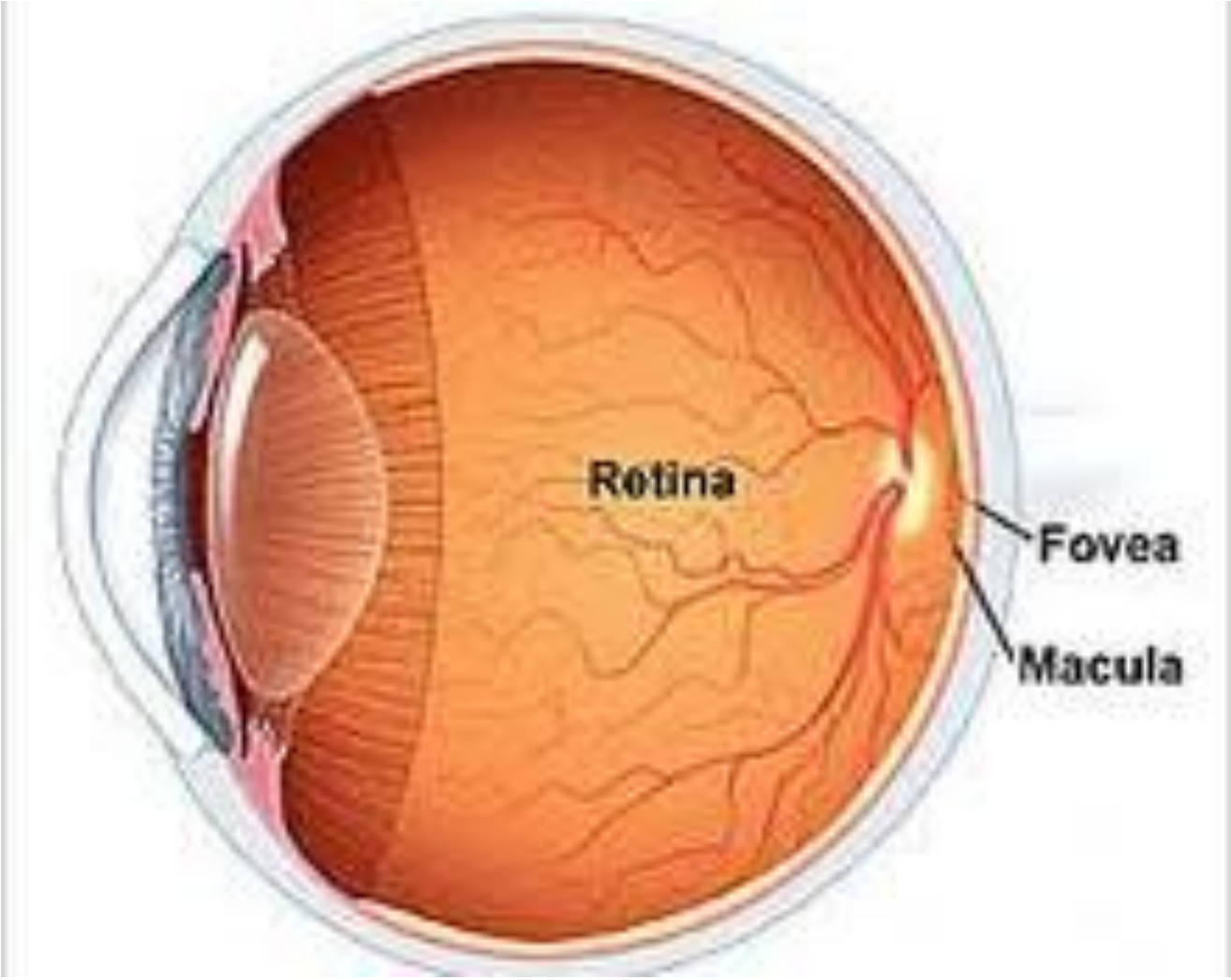

VISION

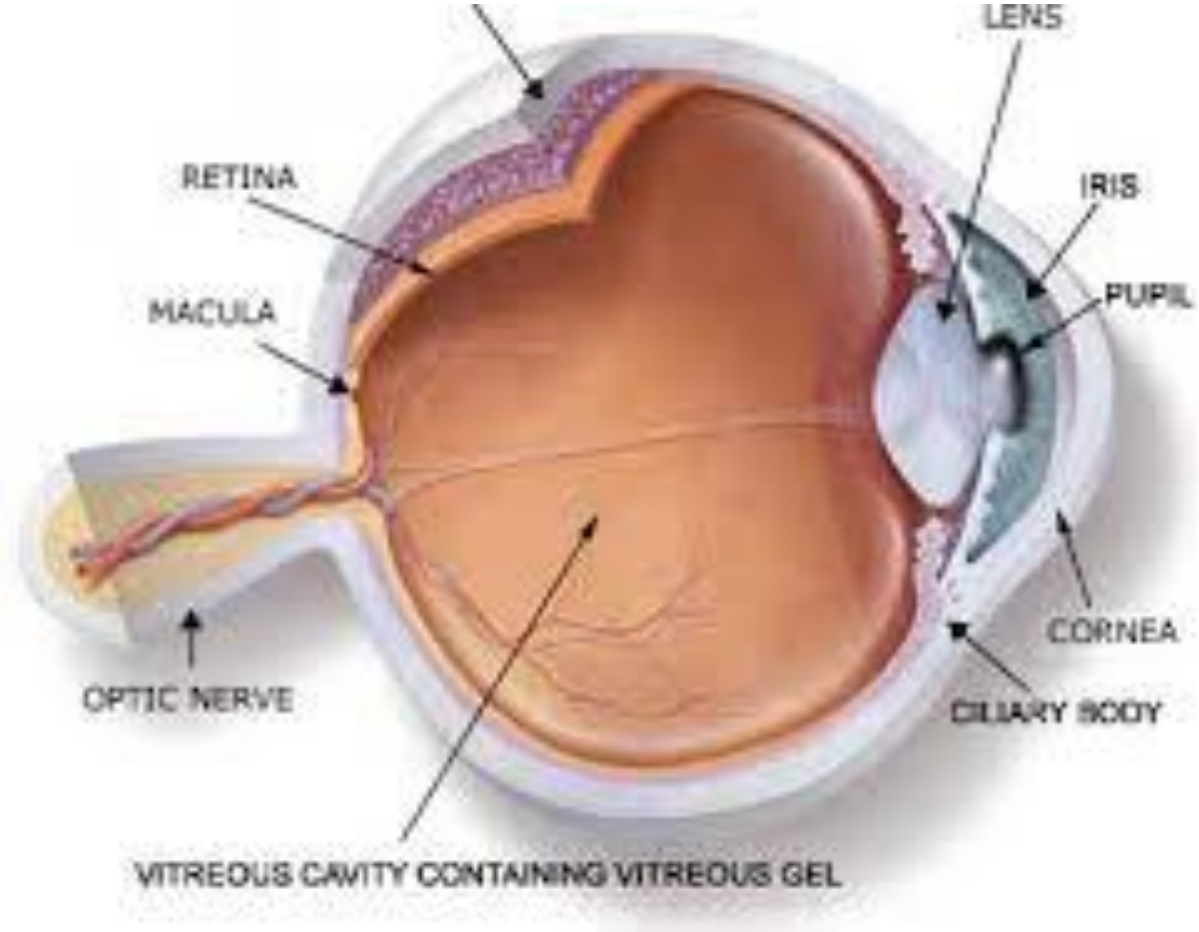

Visual area
of the thalamus
Optic
nerve
Optic
chiasm
Optic
tract
Retina
Visual
cortex

The Eye Anatomy




vitreous
retina


Retina
Fovea
Macula

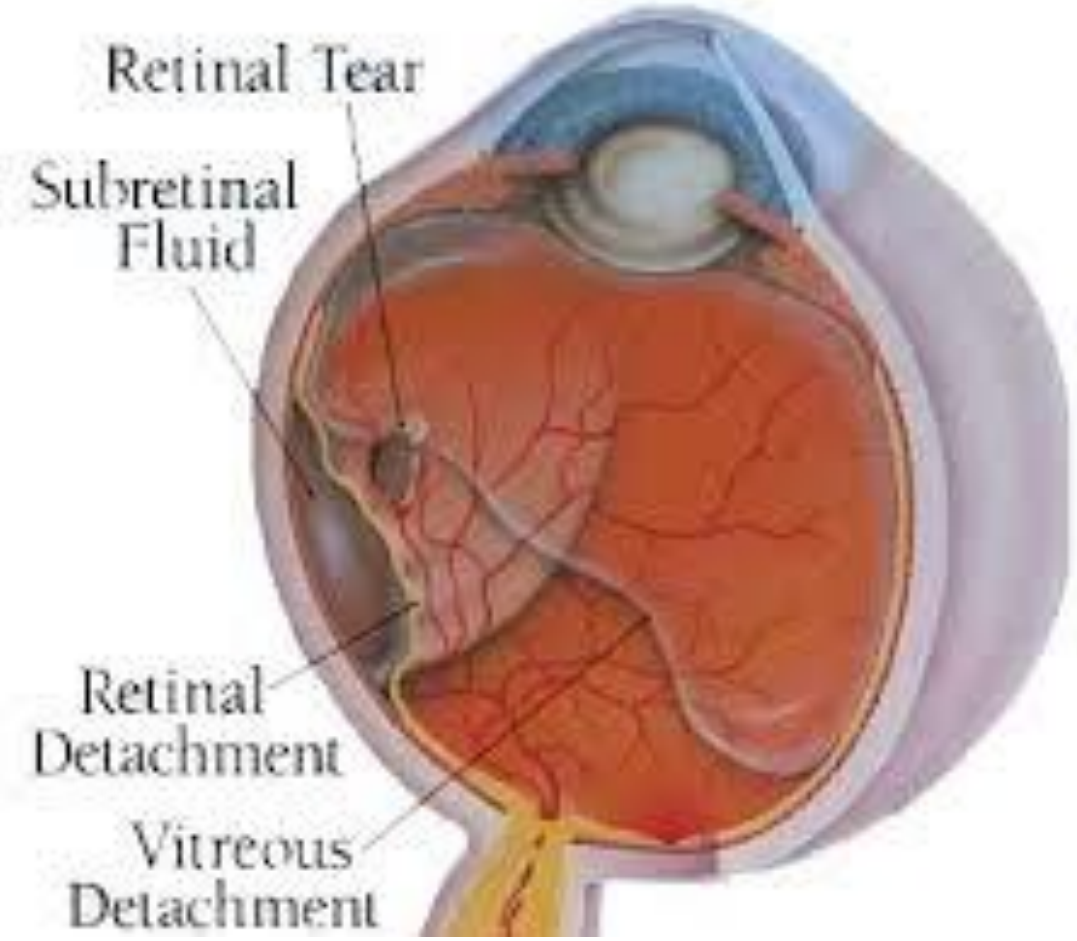

LENS
RETINA
IRIS
MACULA
PUPIL
CORNEA
OPTIC NERVE
CILIARY BODY
VITREOUS CAVITY CONTAINING VITREOUS GEL

Vitreous fills the eye

Retinal detachment basics

- Vitreous causes hole in retina
- Vitreous jelly attached to the retinal surface
  - A solid gel when younger
  - Gradually becomes more liquid, moves
  - Whip like motion of vitreous stresses retina at points of attachment

Vitreous tearing retina



Symptoms

- Flashes
- Floaters
- Shadow in periphery

Flashes

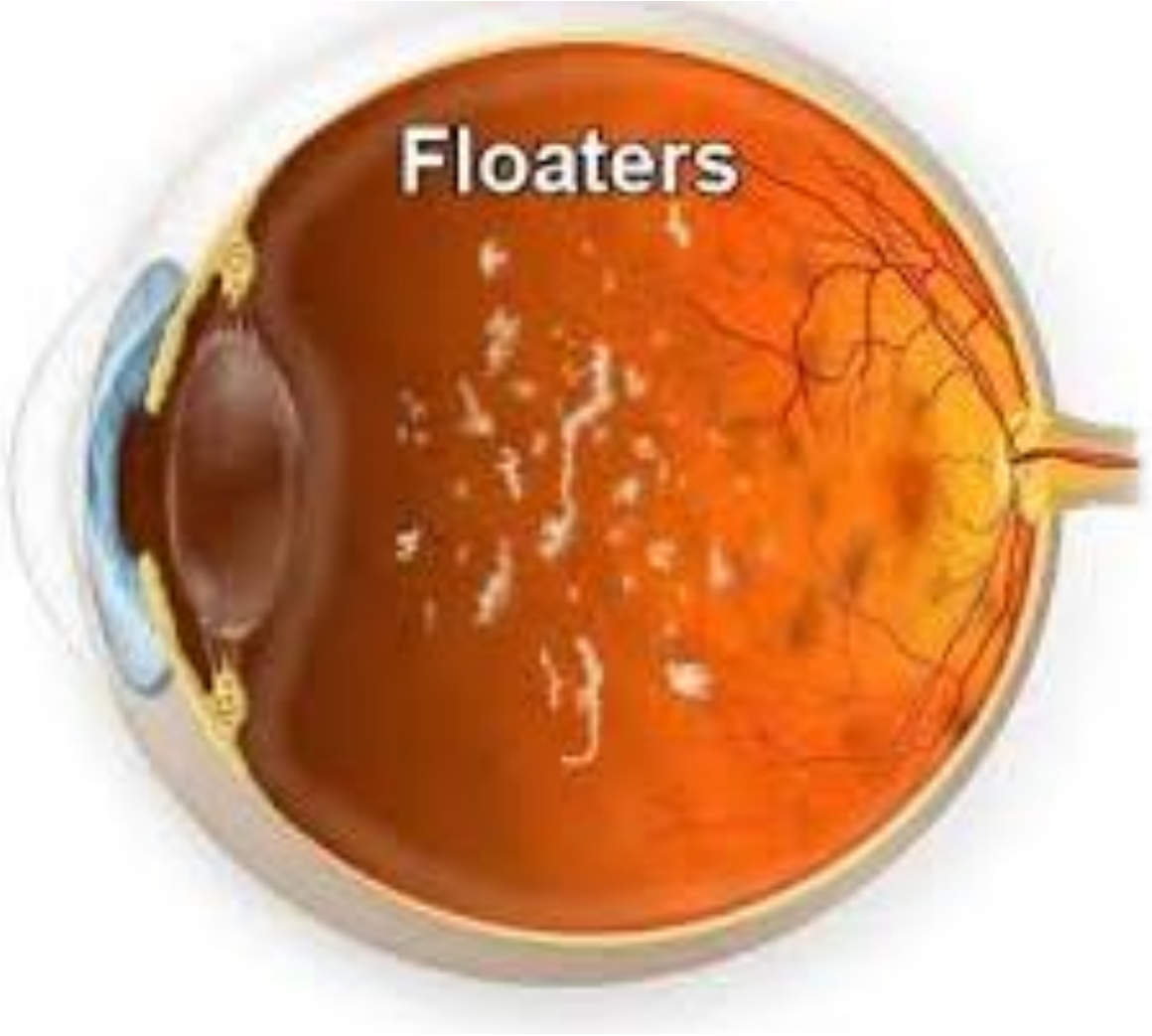
- The sudden onset of flashes in the vision are usually seen in the periphery.
- Flashes are the direct result of the vitreous jelly tugging on the retina.
- If the vitreous is too strong, (or the retina too weak) the retina will tear.

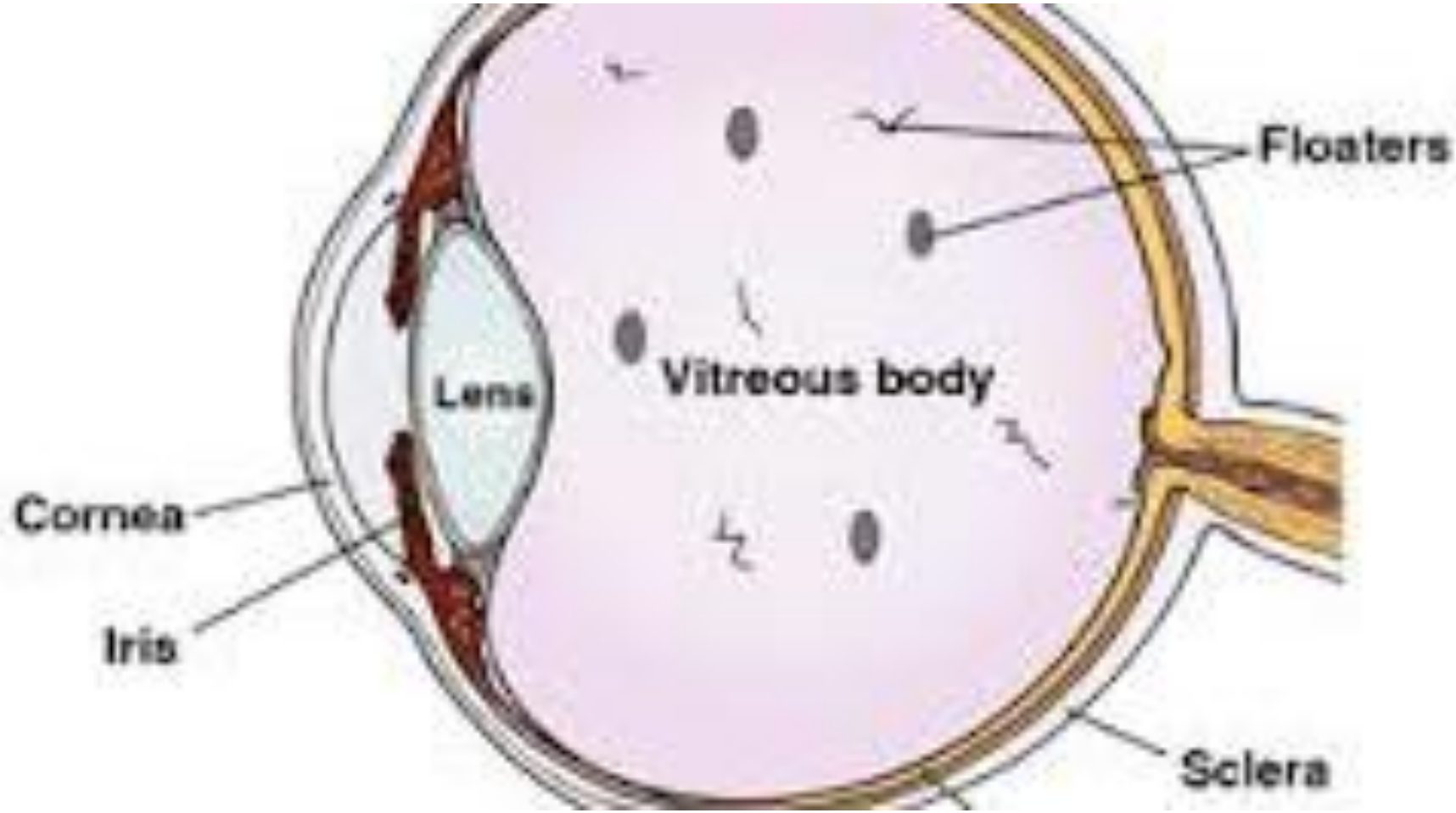
Floaters

- The sudden onset of floaters is an early symptom that a retinal detachment is going to happen.
- Sudden floaters mean the vitreous has had a sudden change and that the retina may have been torn.

Floaters

Floaters are in the vitreous




Floaters
Lens
Vitreous body
Cornea
Iris
Sclera

Shadow

A shadow is probably a retinal detachment!

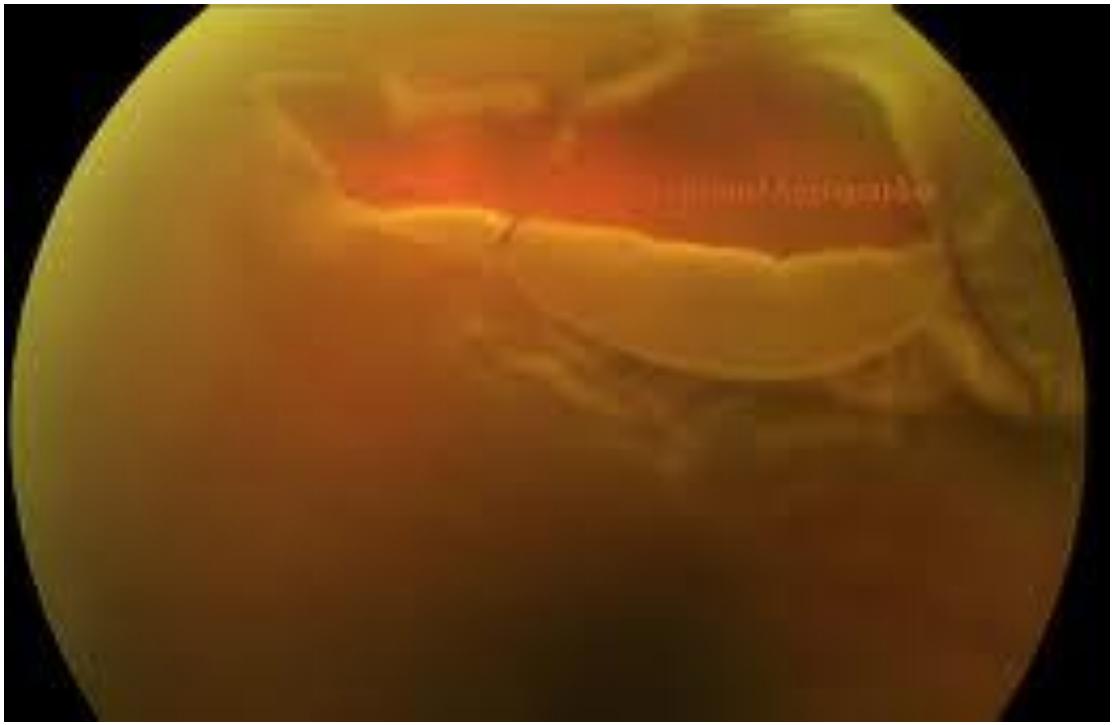
Normal changes

Myopia a risk factor for retinal detachement

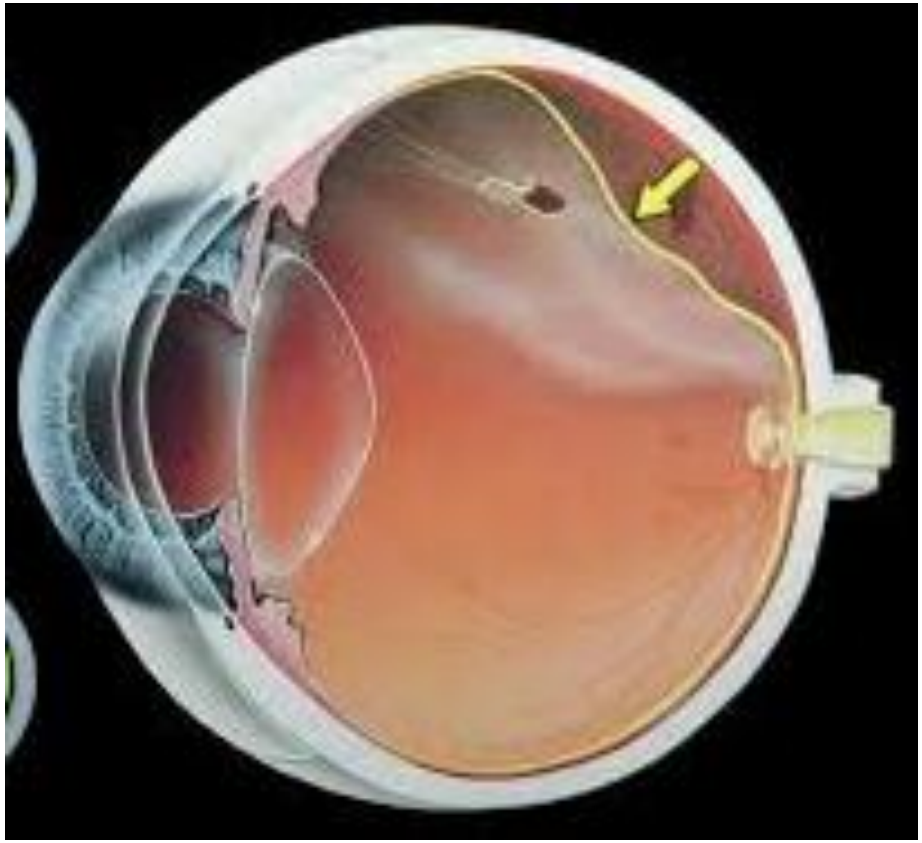
Retinal Detachment repair

- Find and fix the hole

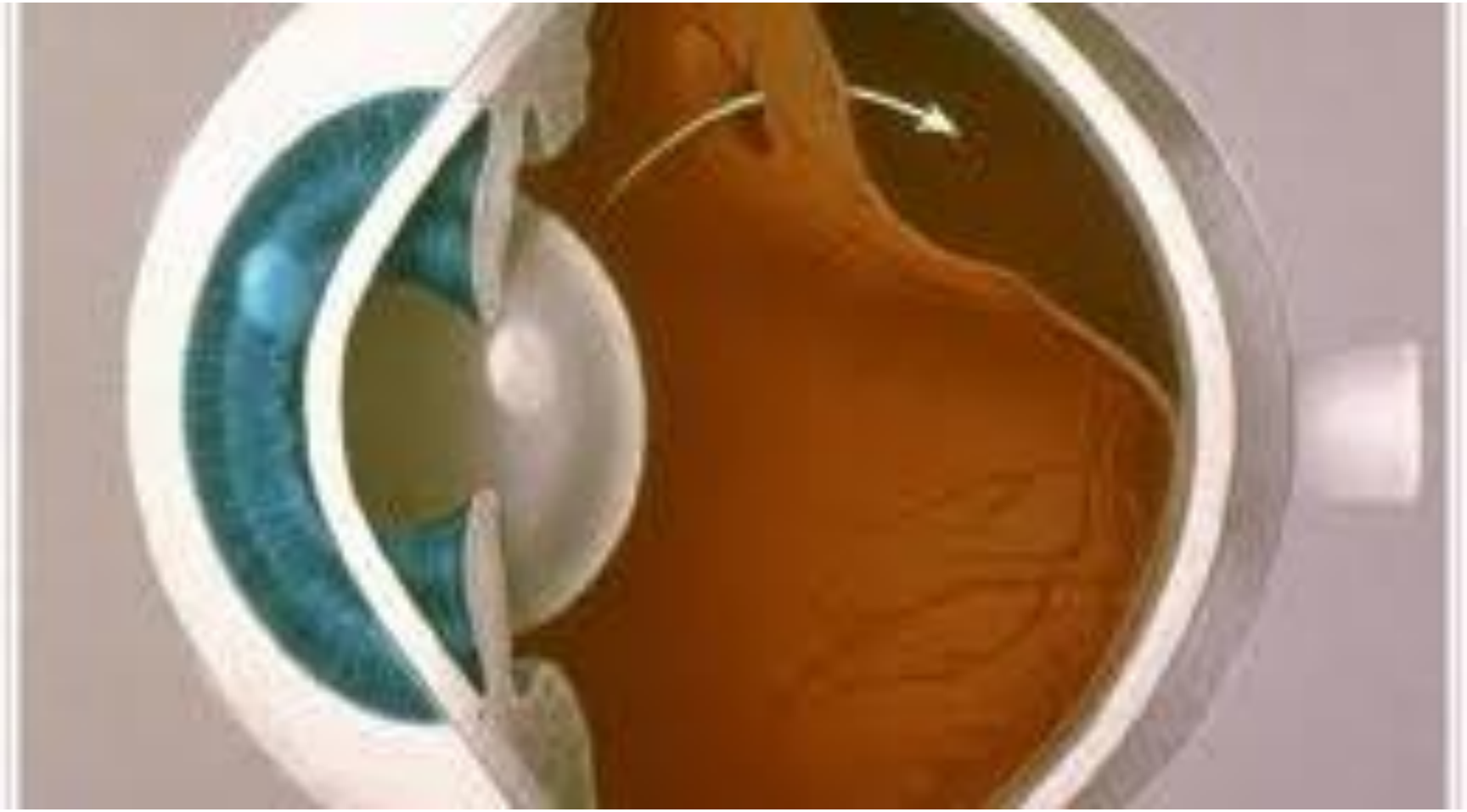
A large retinal tear



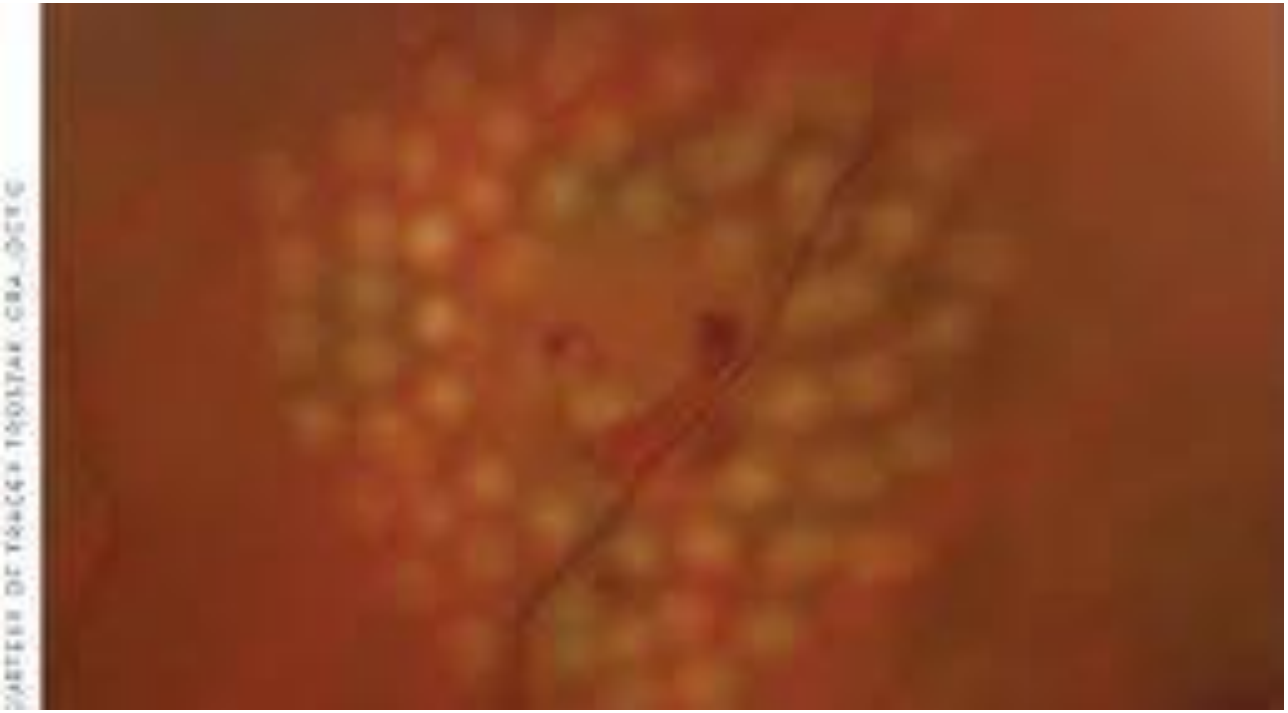
Retinal hole and detachment



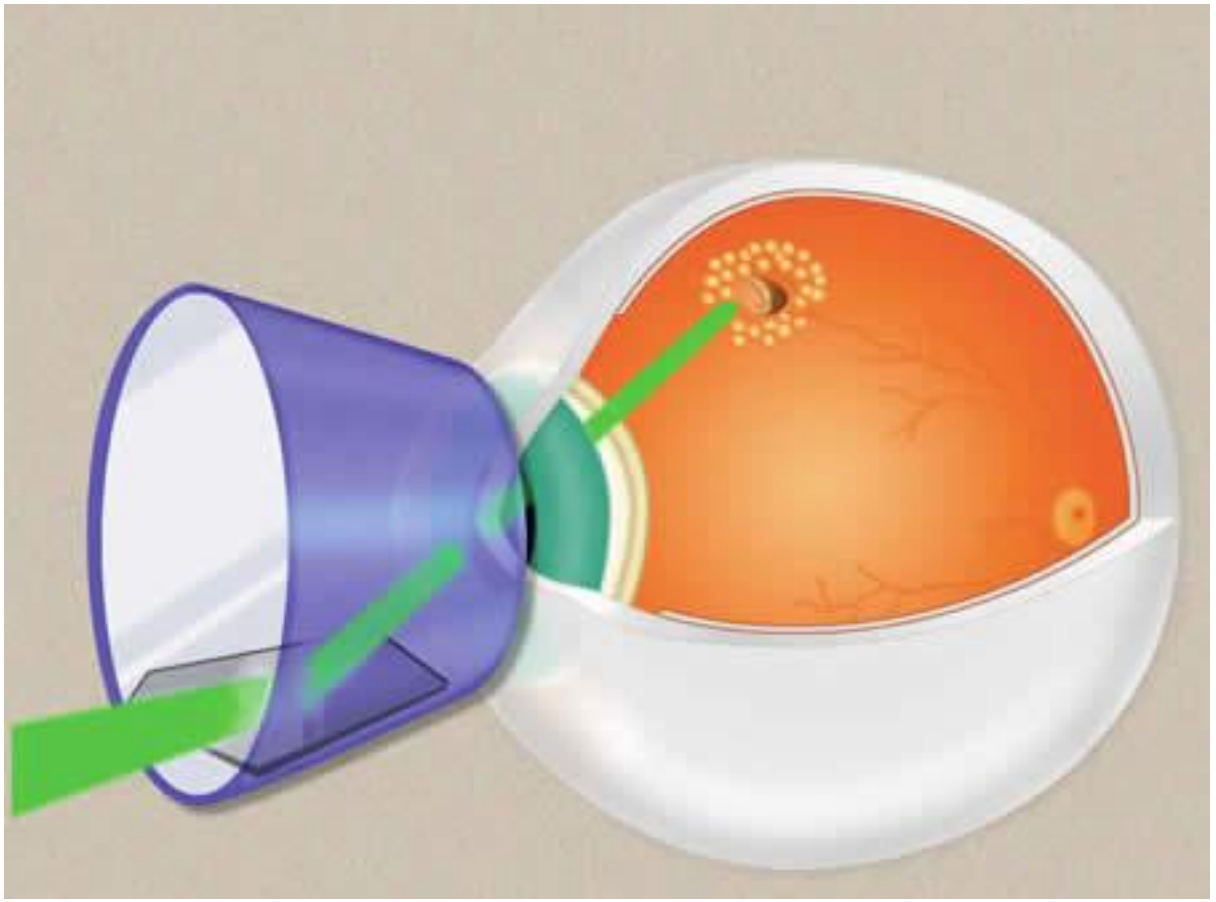
Fluid passes through hole in retina



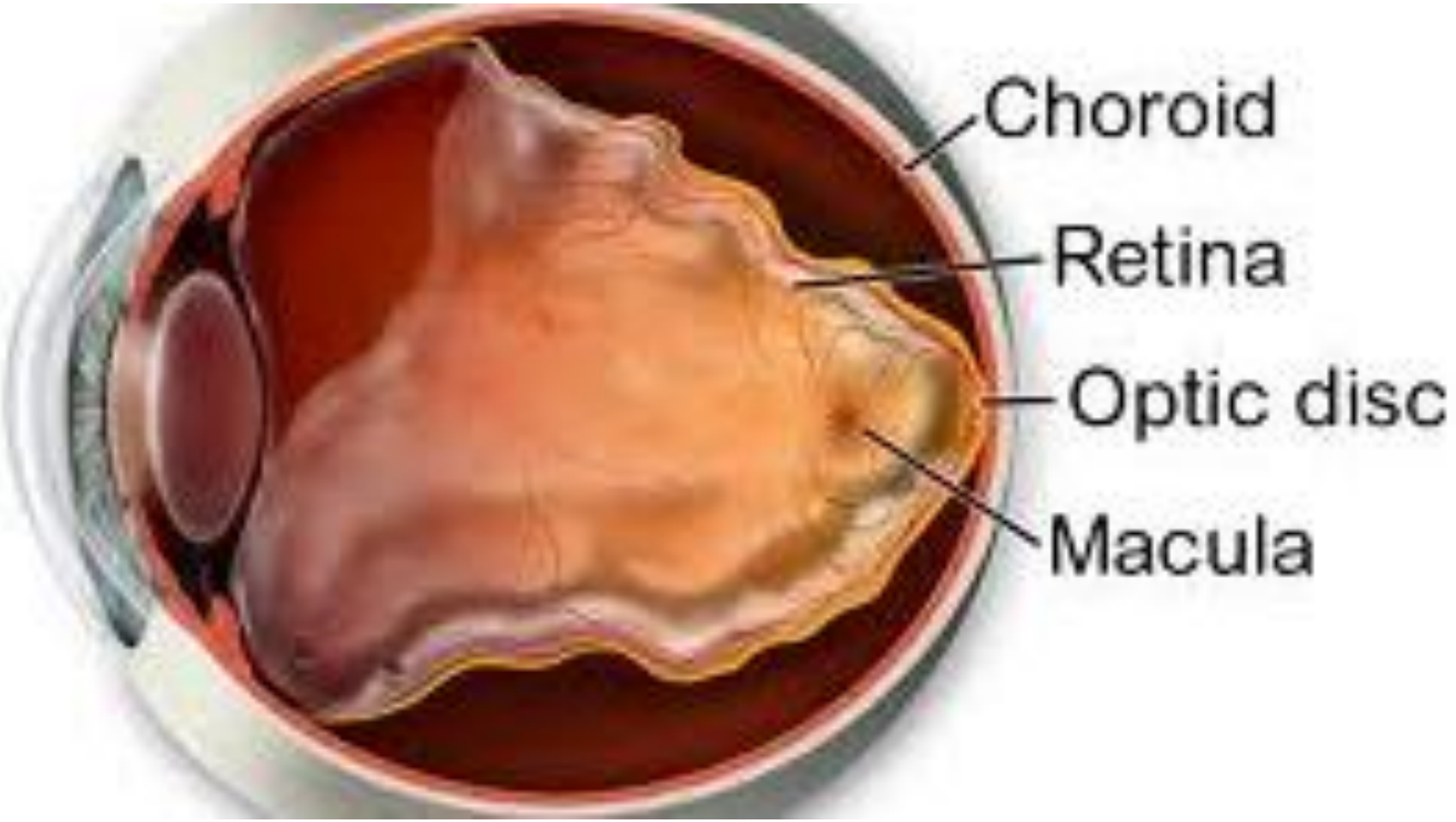
A retinal hole treated with laser



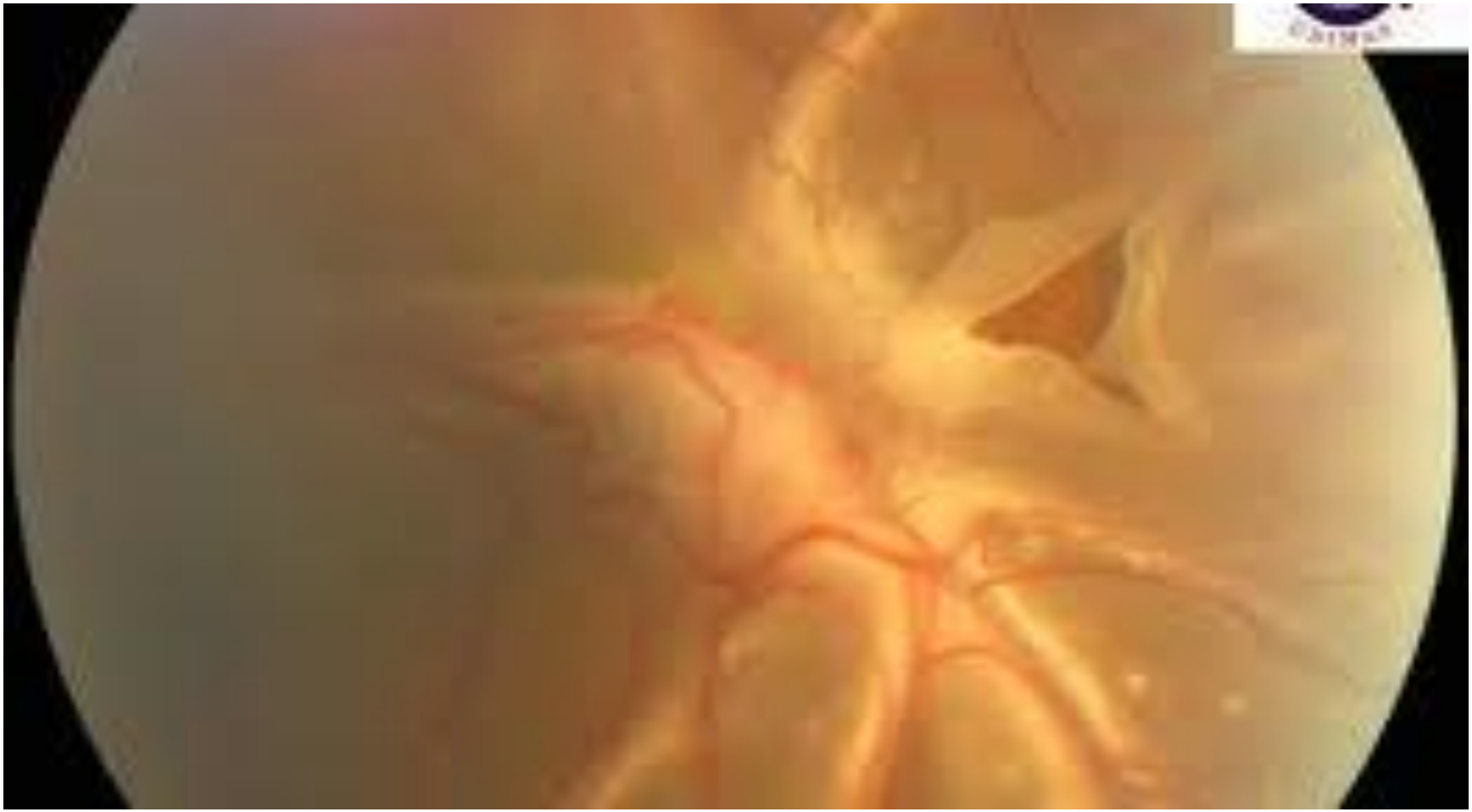
Laser with a lens



Total retinal detachment

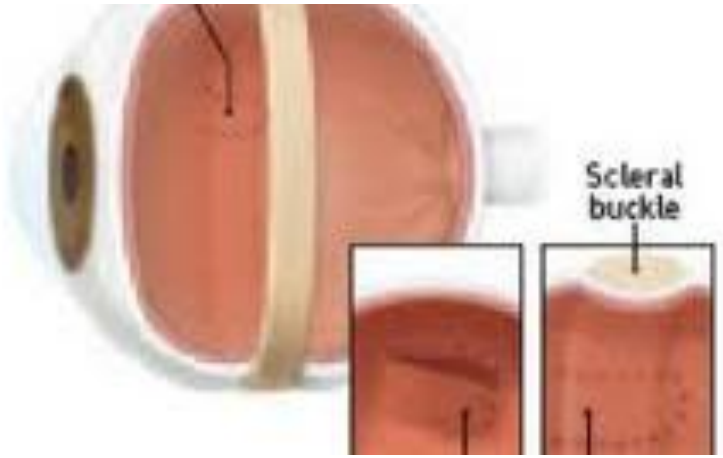


Starfold scarring

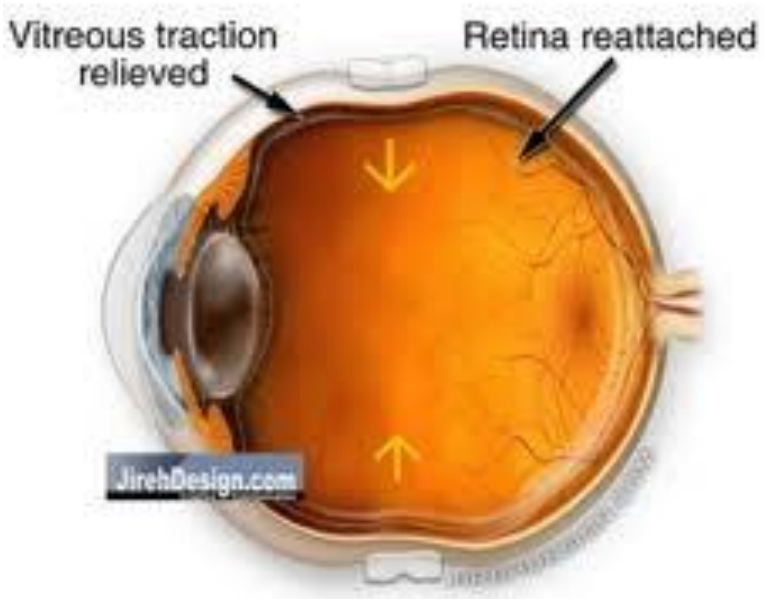


Scleral Buckle

- Early successful surgical technique
- Developed in the 1960's
- Dr. Schepens at Harvard

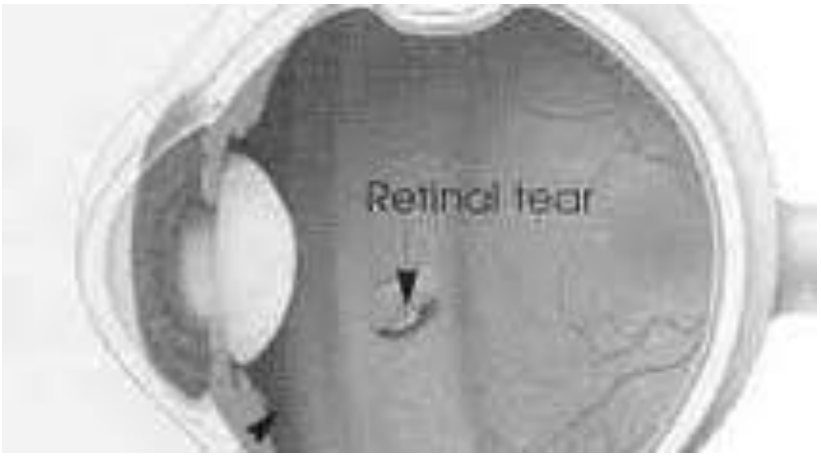


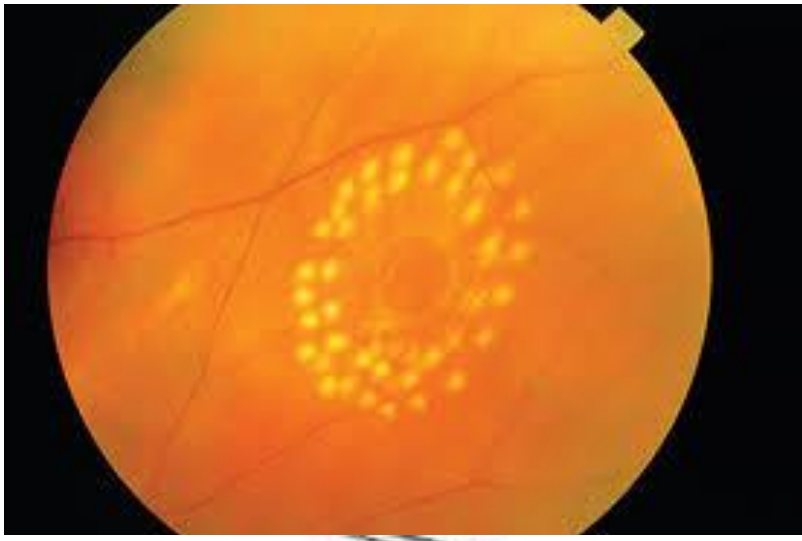
Scleral Buckle
for Retinal Detachment

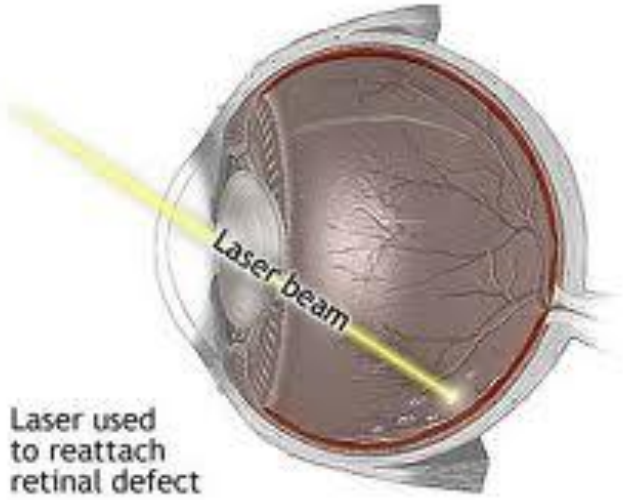


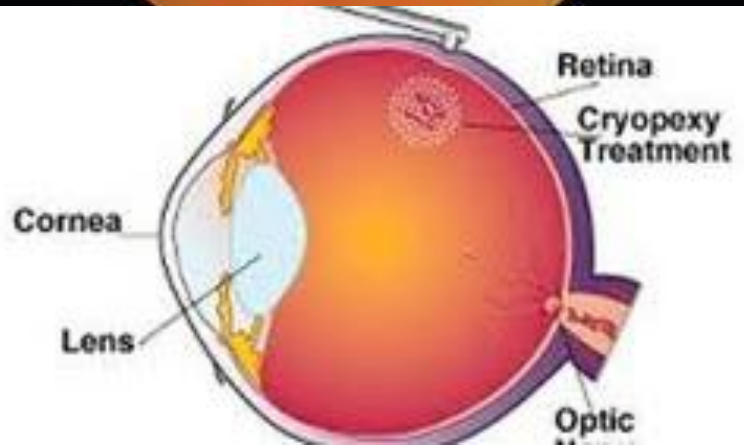
Scleral buckle

Lasers and cryothermy

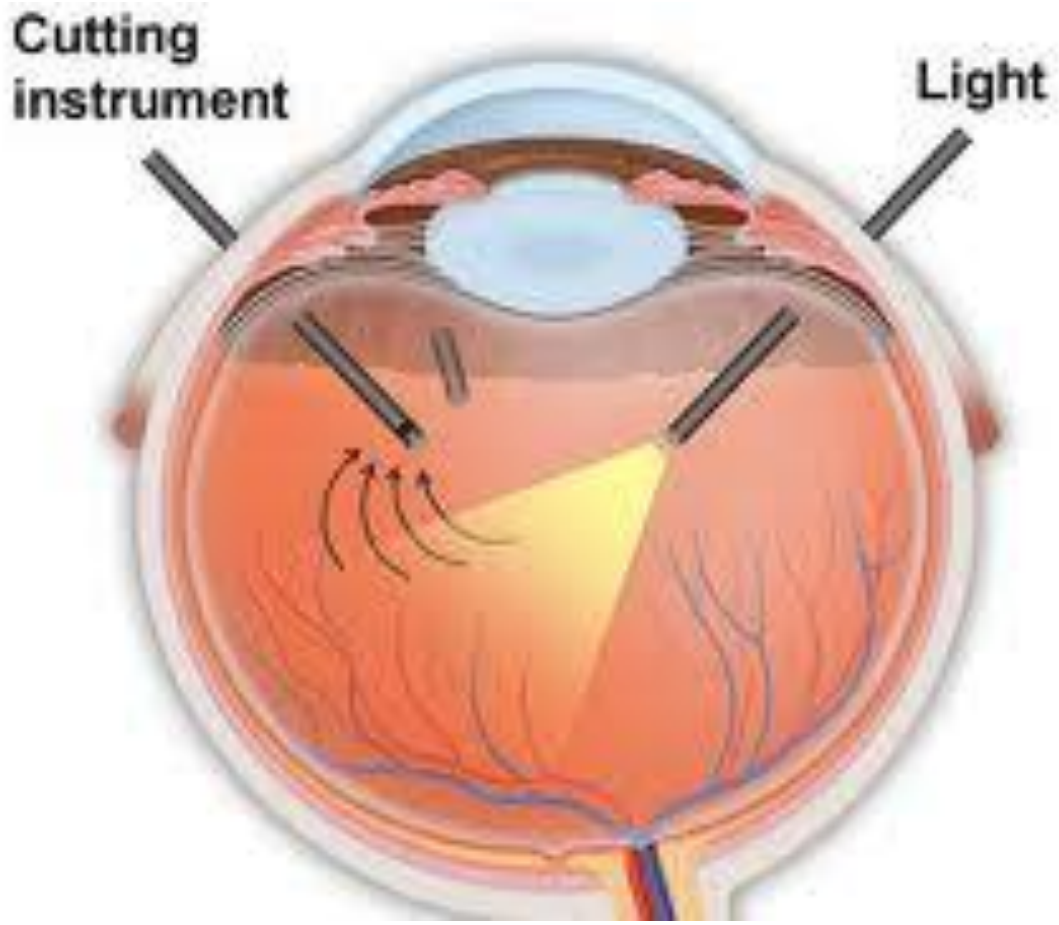




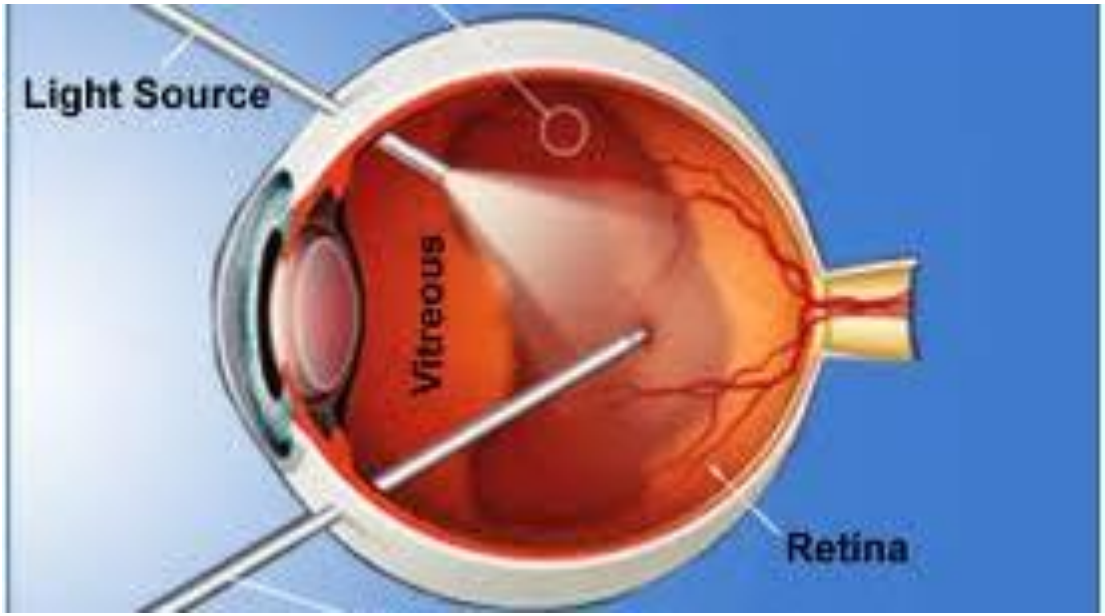


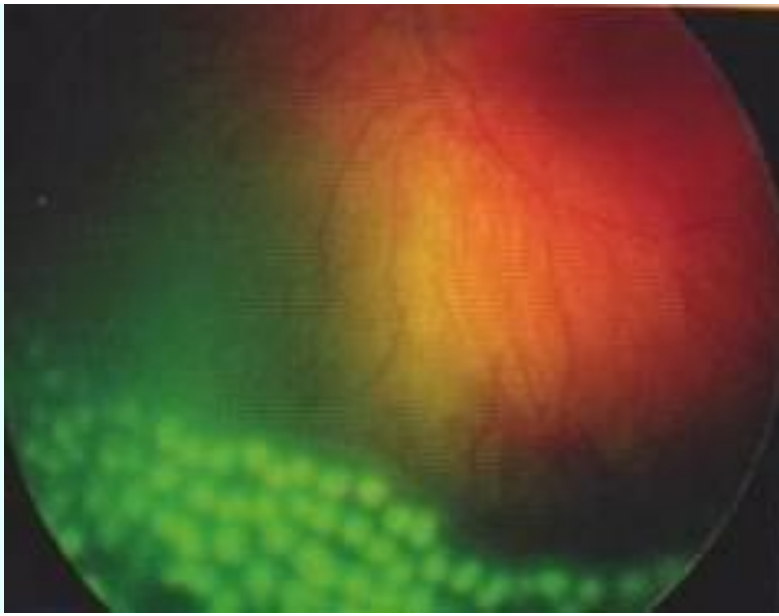


Vitrectomy



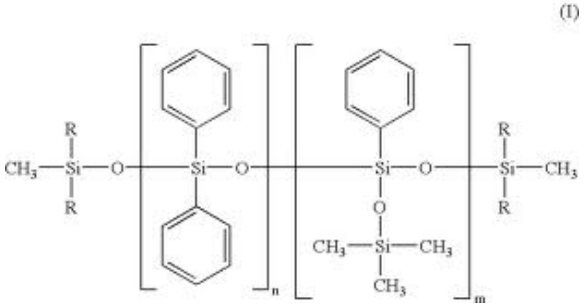
Vitrectomy with endolaser



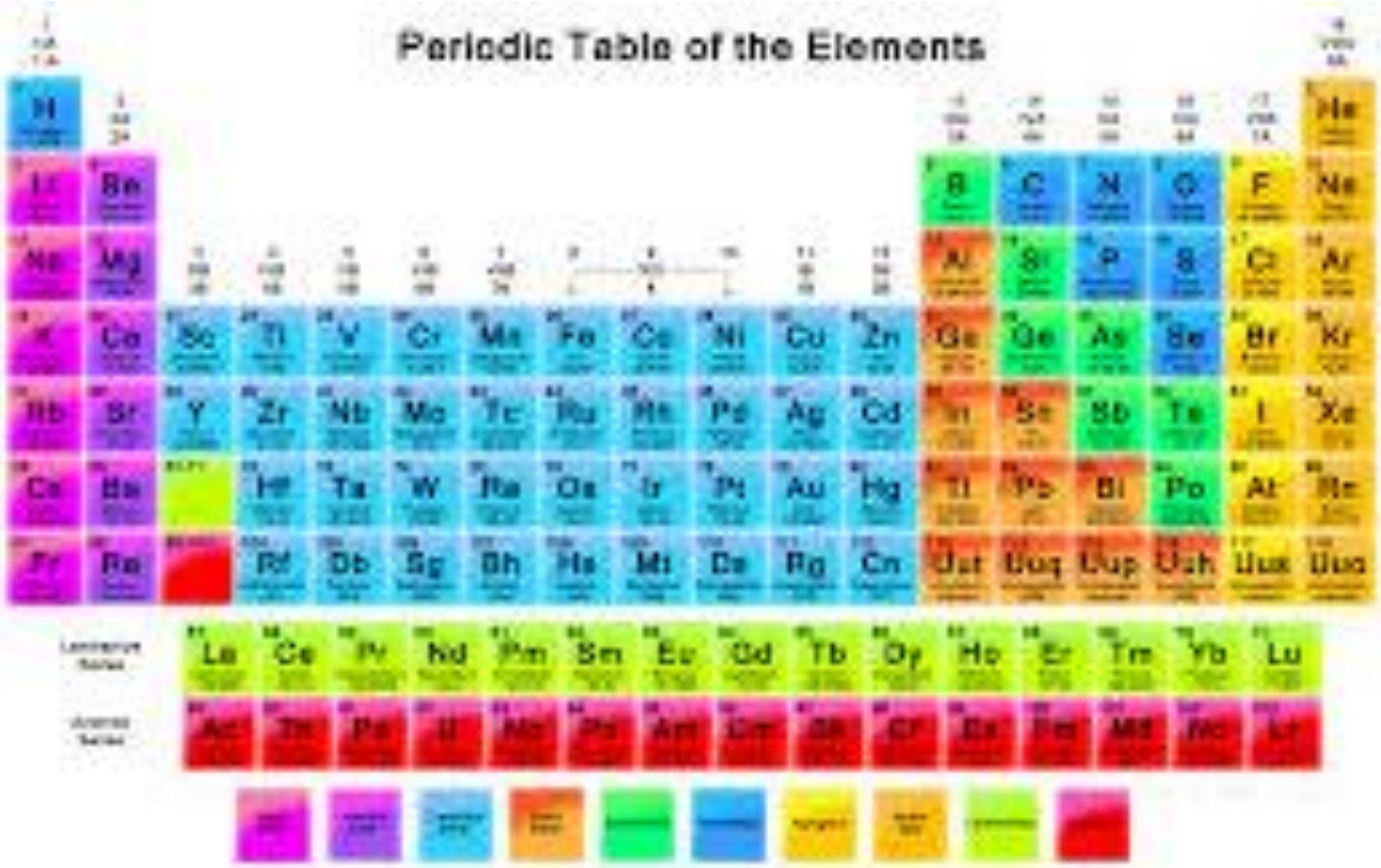


New techniques

Silicone Oil

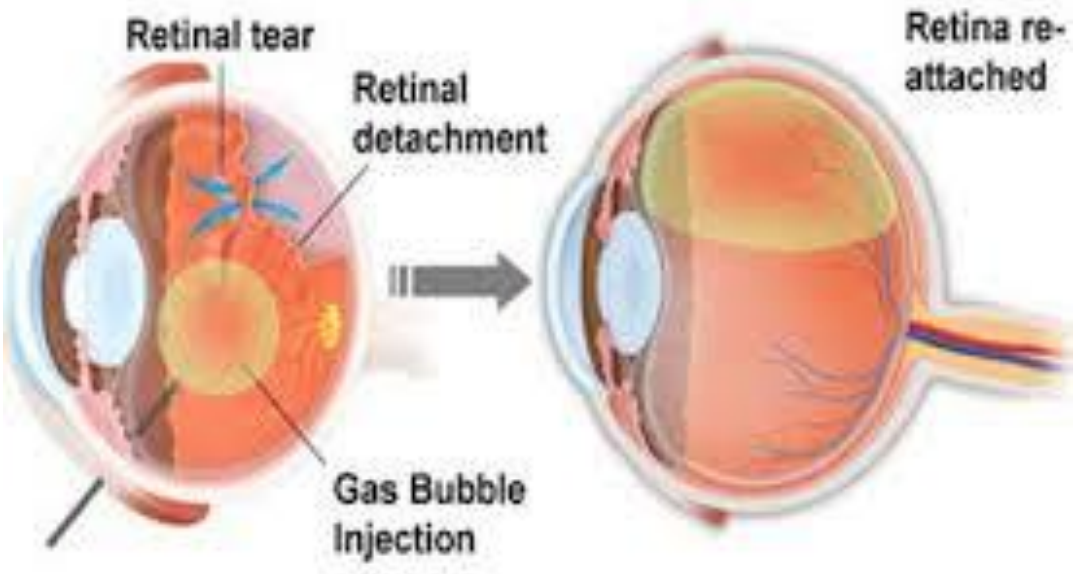


Silicone Oil

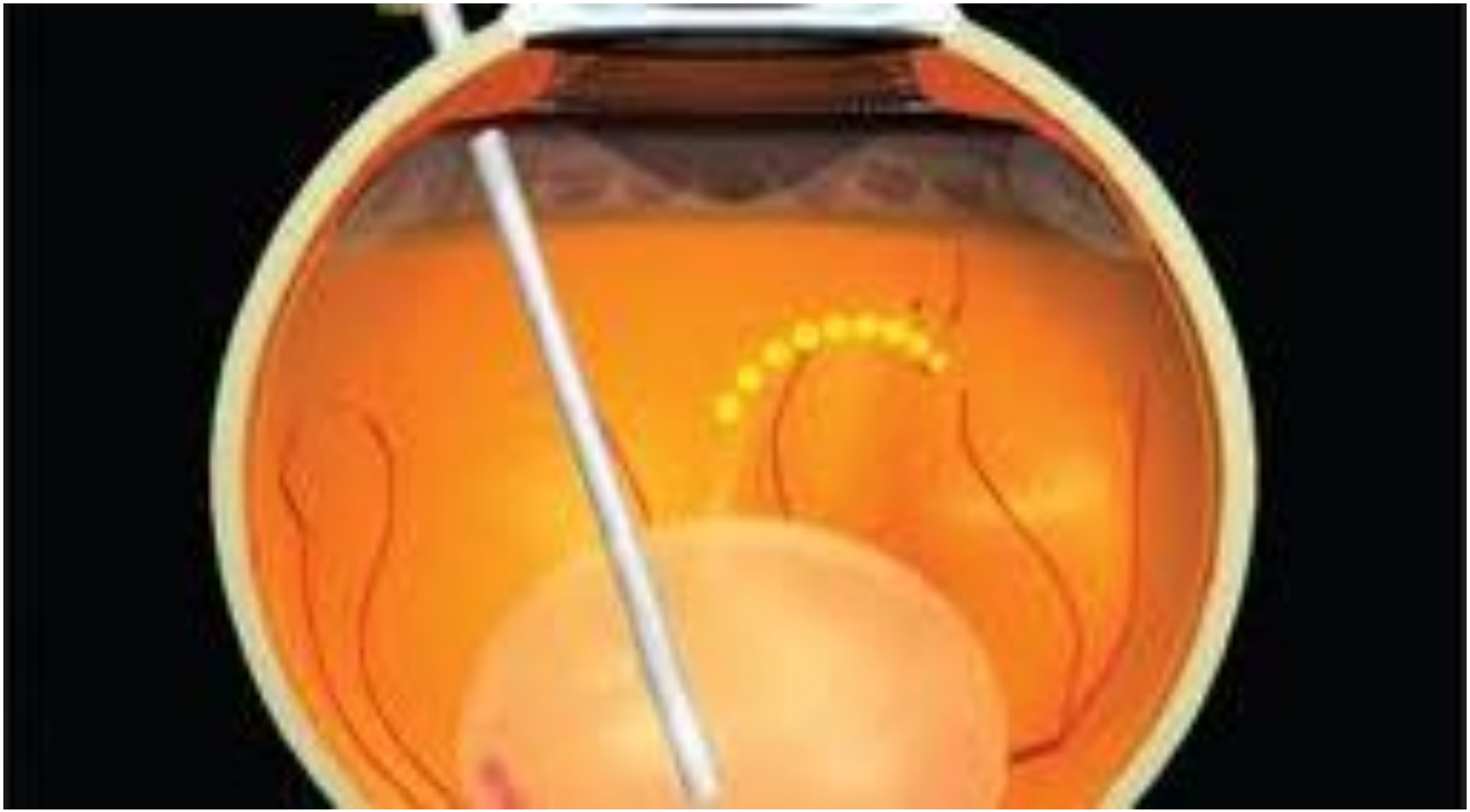


Silicone oil emulsification

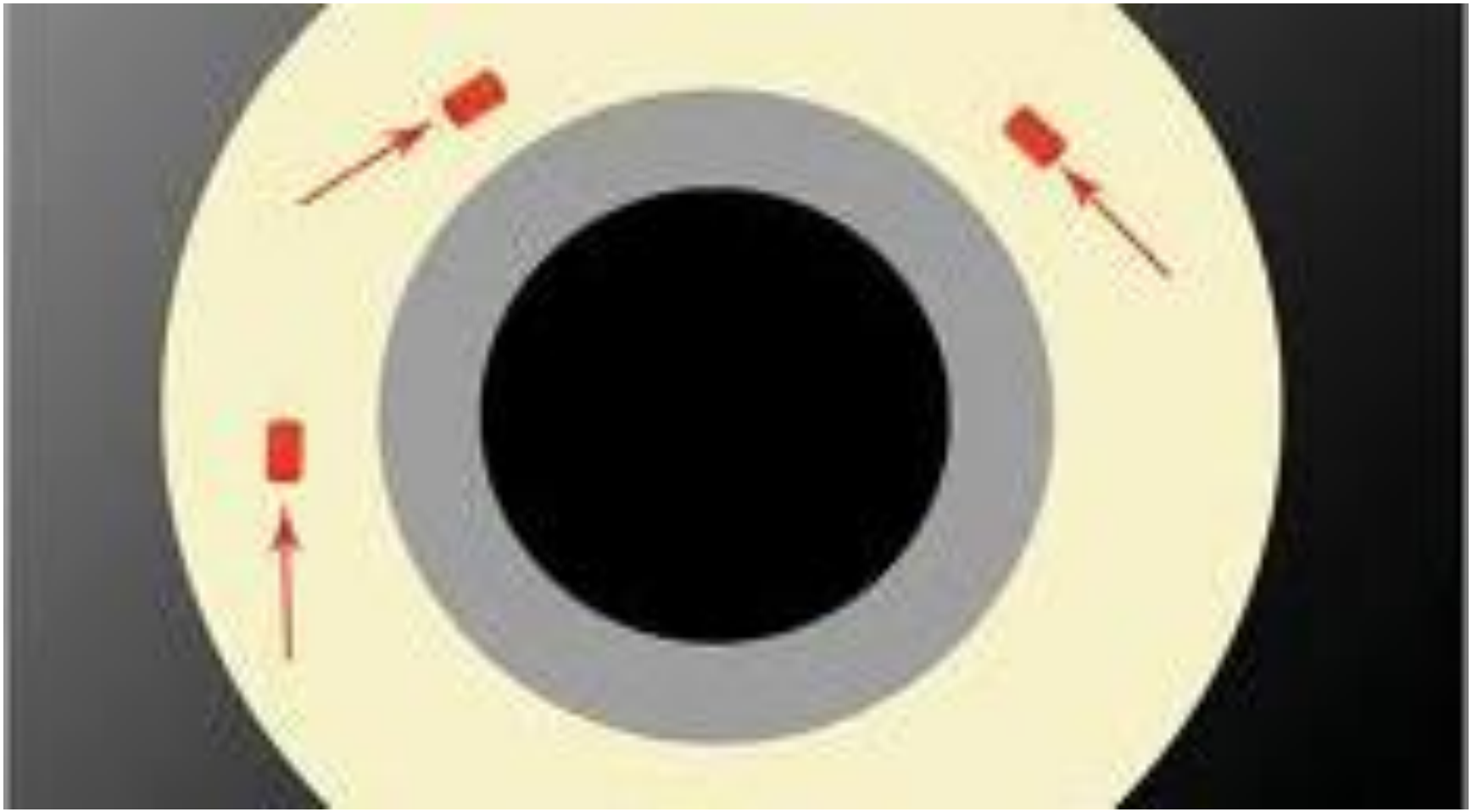
Pneumatic


Retinal tear
Retinal detachment
Gas Bubble Injection
Retina re-attached

Perfluron



Minimal incision – Suture-less



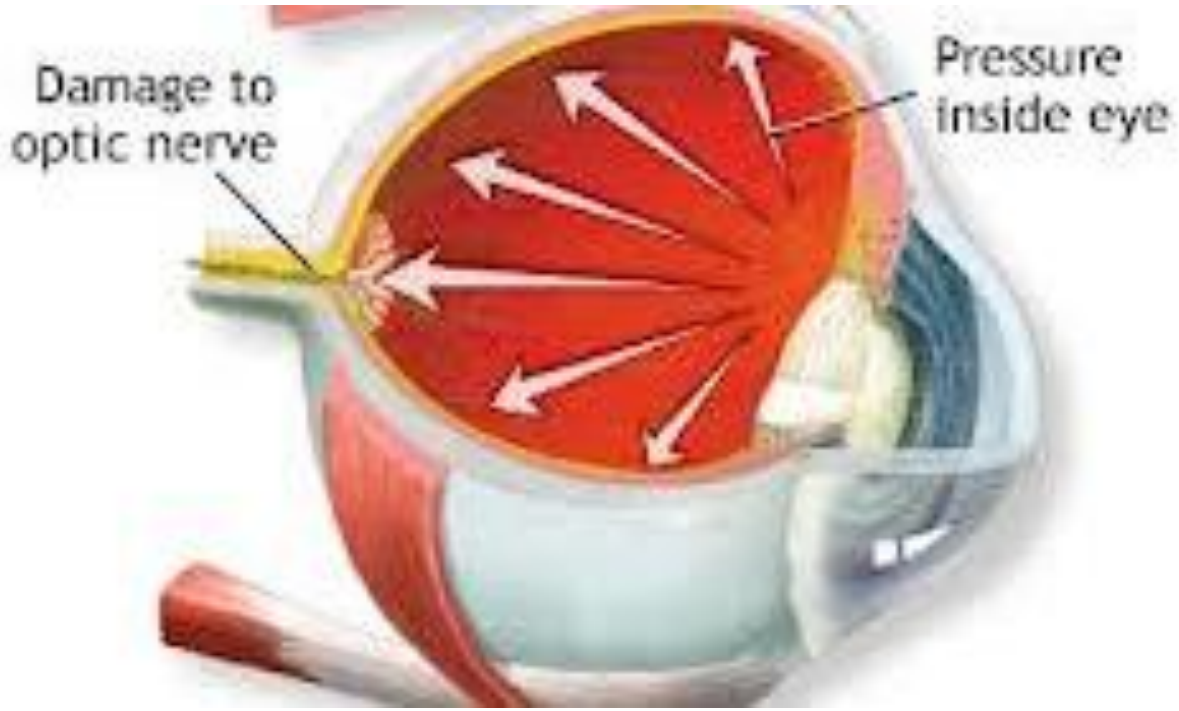
Other eye problems

Cataracts

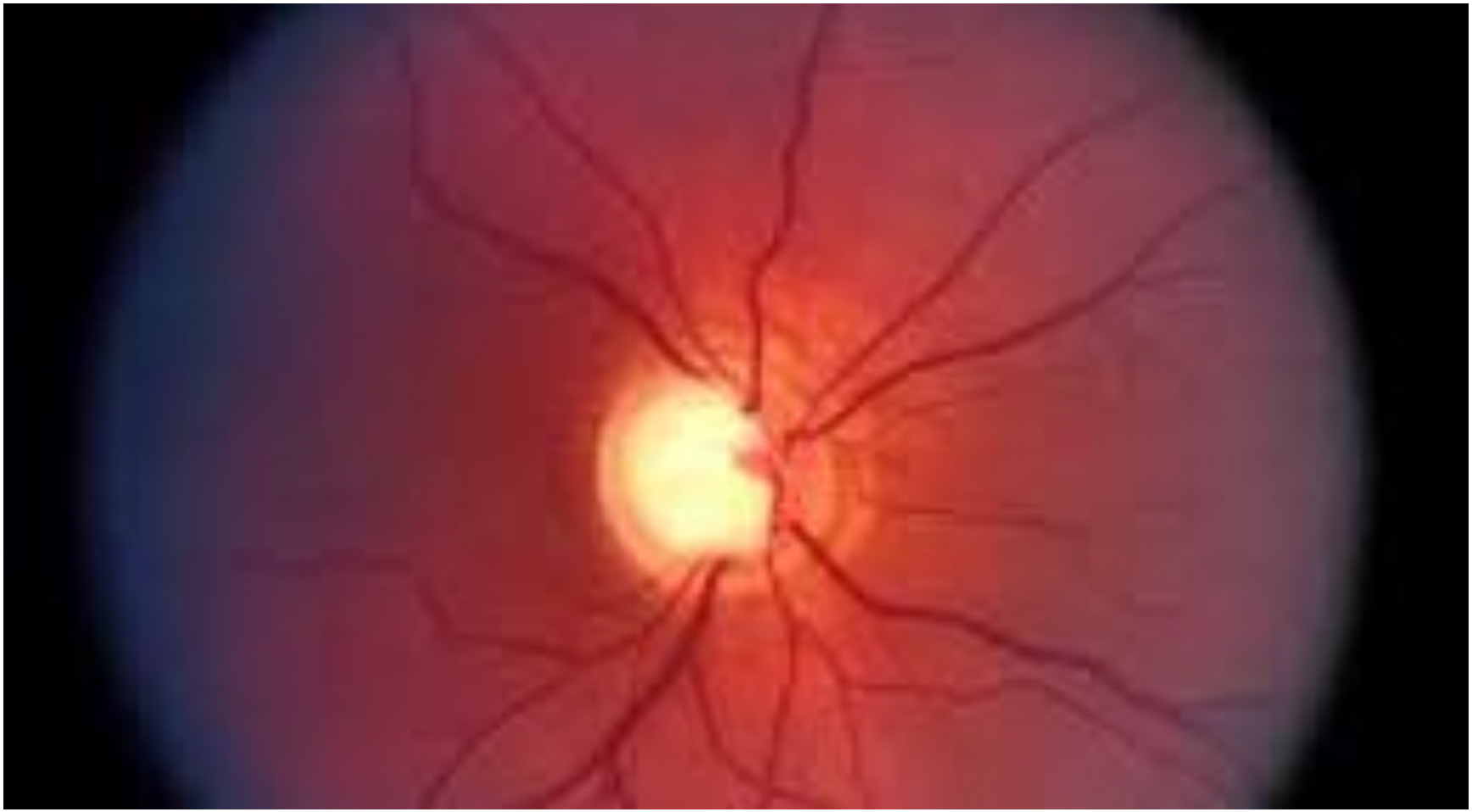
Cataract

Cataract effect on vision

Glaucoma



Glaucoma effect on optic nerve



Success rate

- Success is measured by patients vision and quality of life

Low vision


Low
Vision

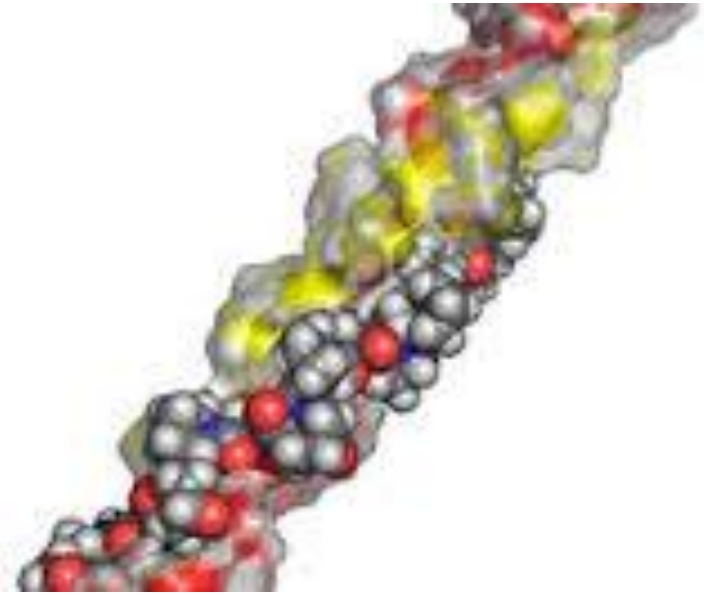

Chicken and Nooc
memade noodles make this the
for a family meal; a small sala
would complete it.
30 minutes Cook: 30 minutes Mai

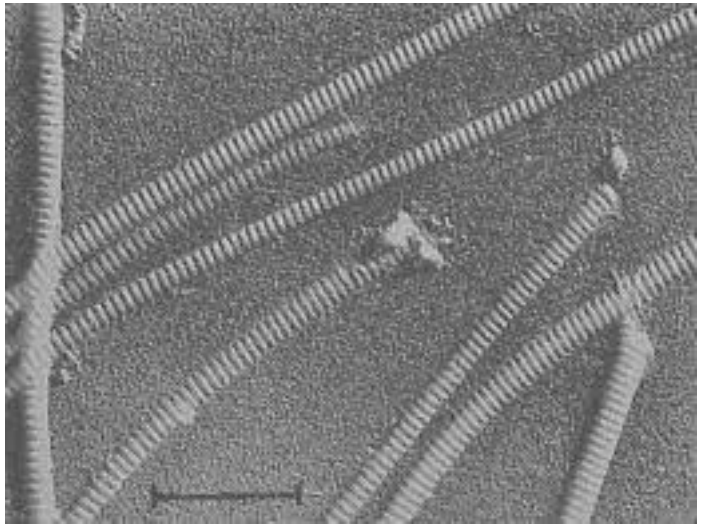

best fly fishing
angling equipme
river, that will p
lure, gives you t
standard bait ca

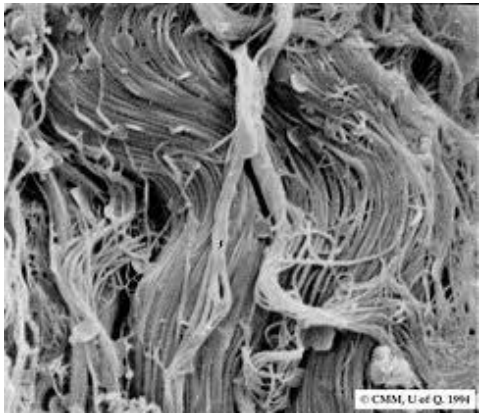
Stickler syndrome

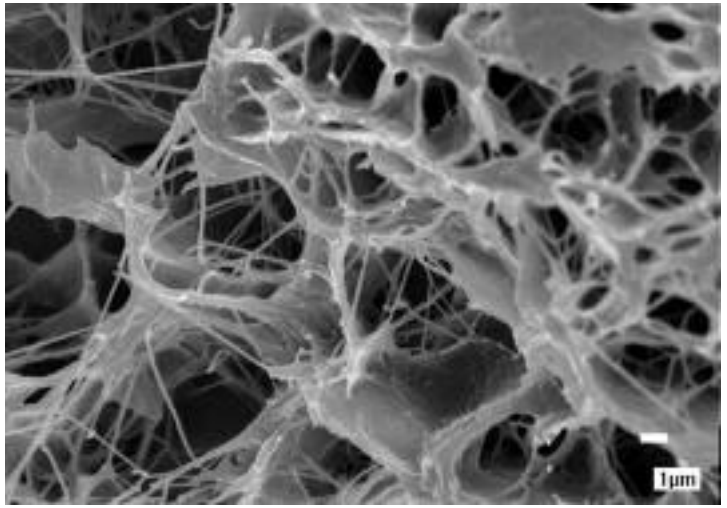
- Defect in Collagen gene
- Collagen is a main component of the vitreous
- Collagen is like rebar in cement

Collagen is a structural protein









Jelly fish jelly

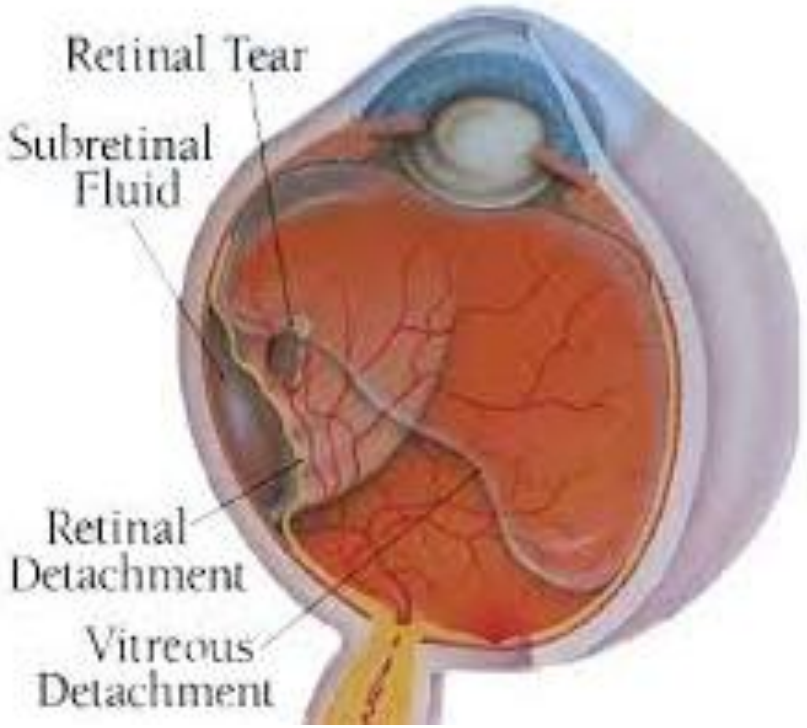
Vitreous jelly like the clear part of an egg

“vitreous” means “clear”

Lattice is vitreous attachment to retina

Stickler syndrome

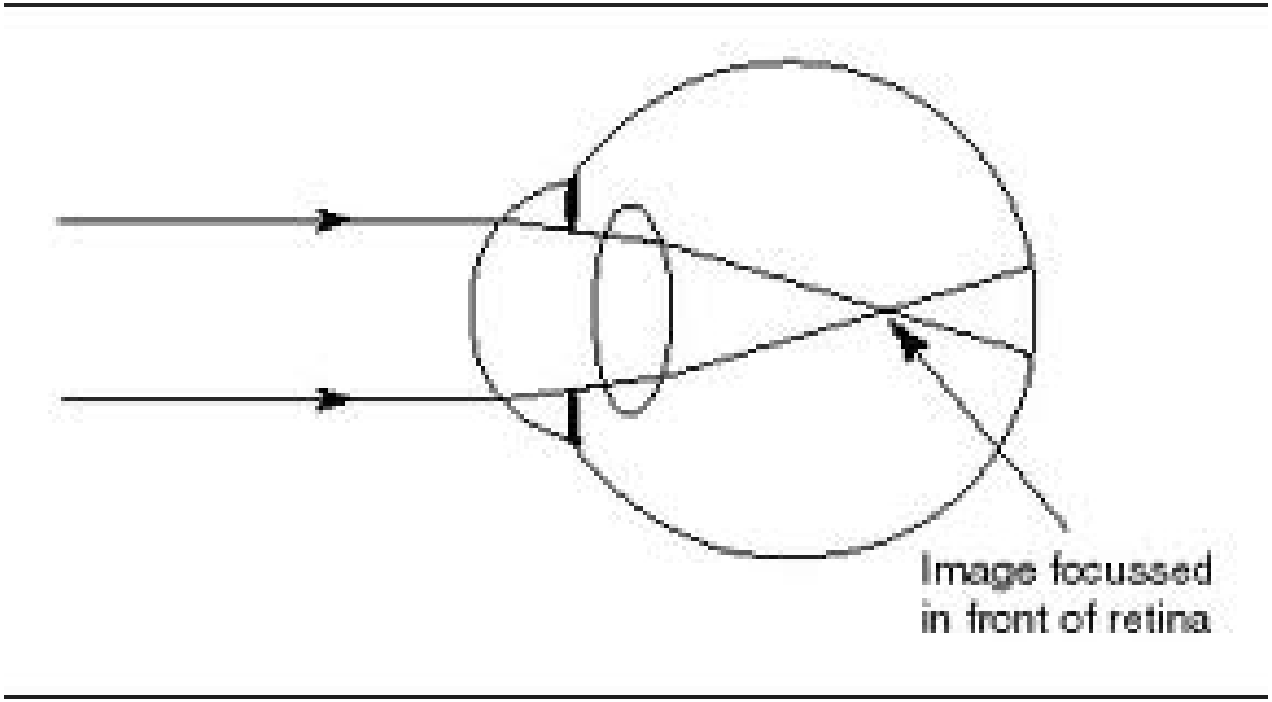
- Vitreous jelly not quite normal
- Prone to tear retina



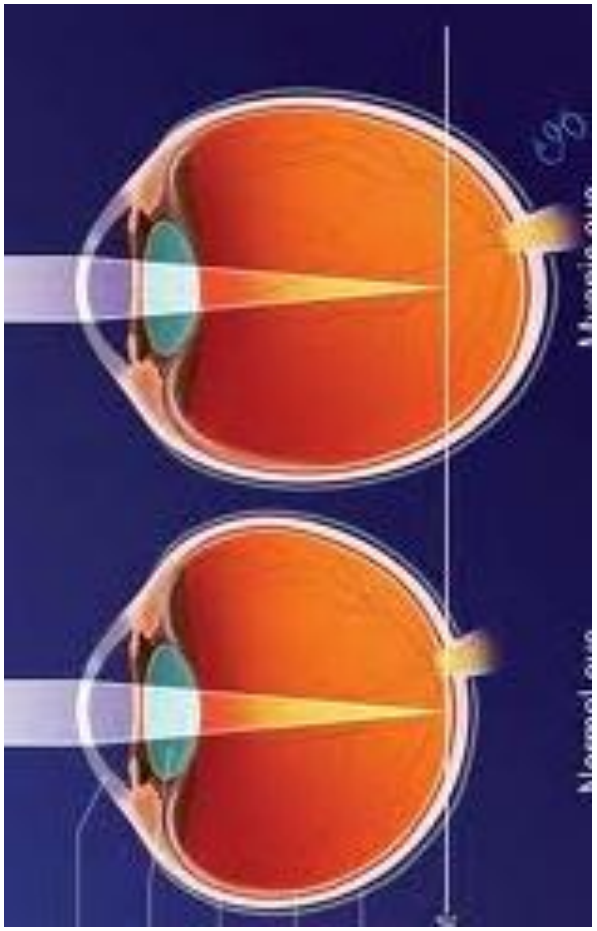
Myopia

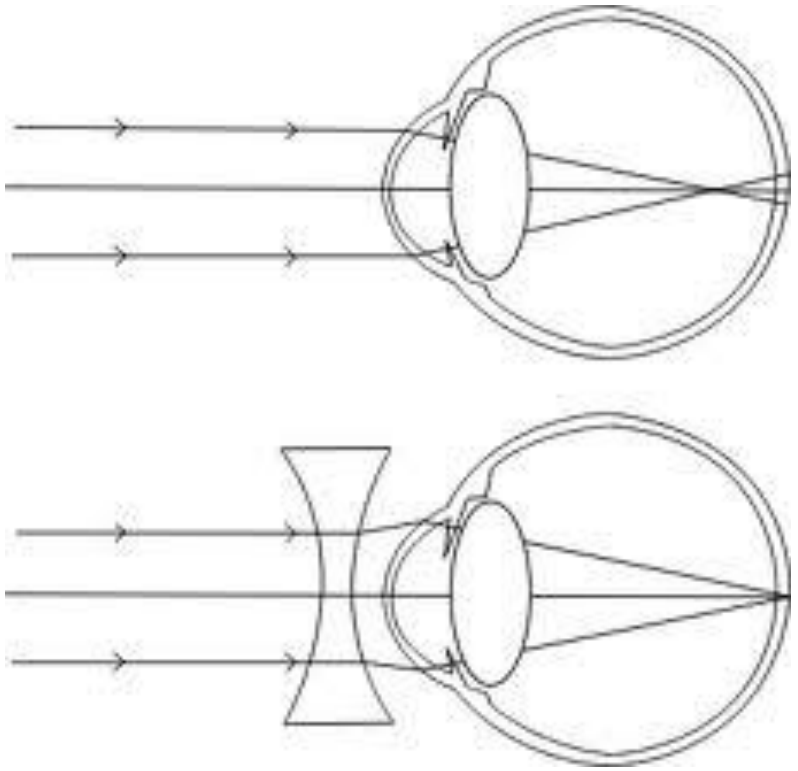
- Myopia is near-sightedness
- Myopia is common in Stickler syndrome
- Myopia is a risk factor for retinal detachment

Myopia = “near-sighted”



Myopic eyes are larger

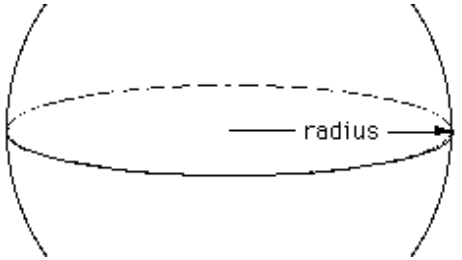




Myopic eyes are larger

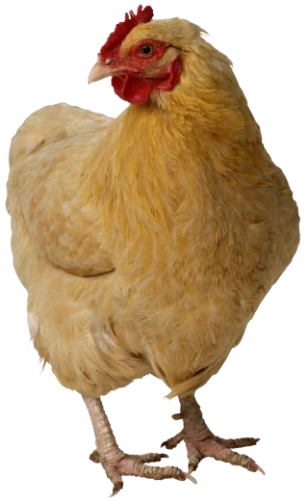
- Larger vitreous
- More leverage on retina

$$V = \frac{4}{3}\pi r^3$$



Heredity pattern a clue

- Advanced warning for kids
- If Stickler syndrome runs in the family, these kids need our help to monitor for eye trouble
- Can’t expect a kid to tell us if vision problems develop
- Need to monitor for
  - retinal detachment
  - cataract
  - glaucoma



Prevention

Challenges in Kids

- Don’t tell us when they have problems
- Don’t always wear their glasses
- Amblyopia

Ideas for the Future

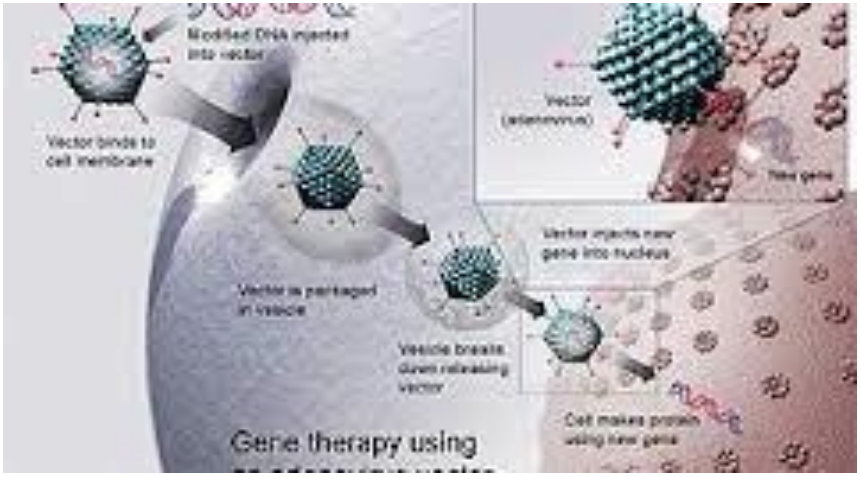
Artificial Eyes?

- Not yet

Gene therapy

- Stickler syndrome is caused by individual genetic defects
- Potentially a target
- Gene therapy is being used for other eye conditions
  - RPE65

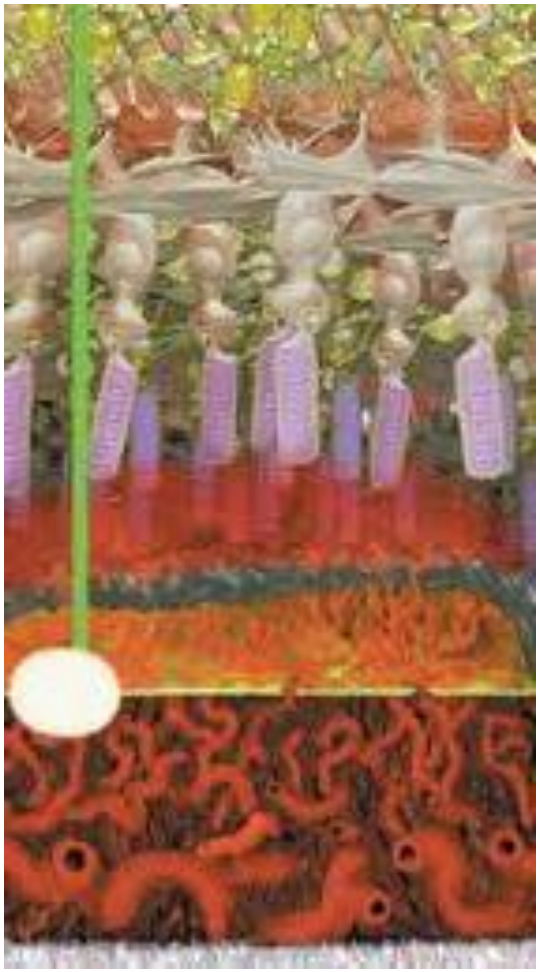
Use a virus to carry gene to cells



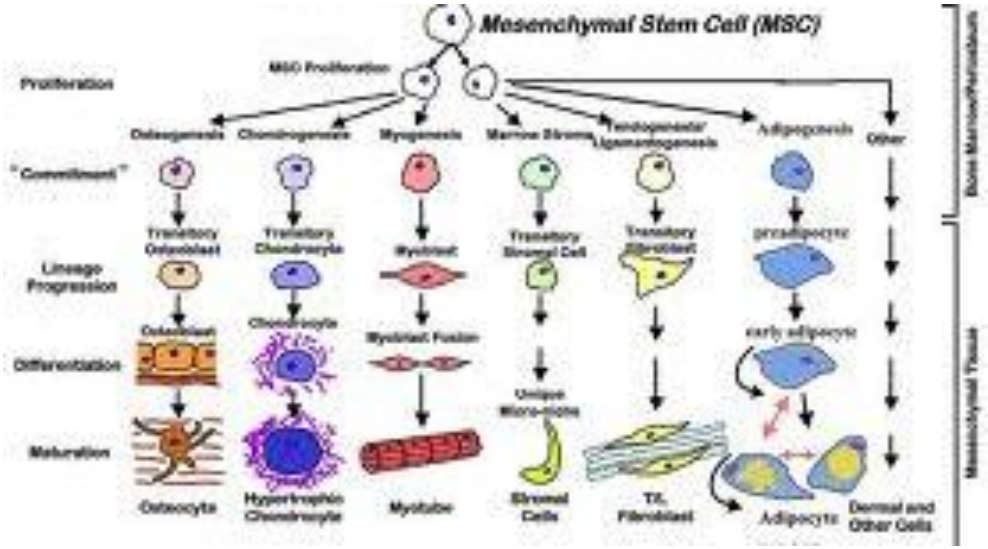
Genetic engineering ethics

Stem cells

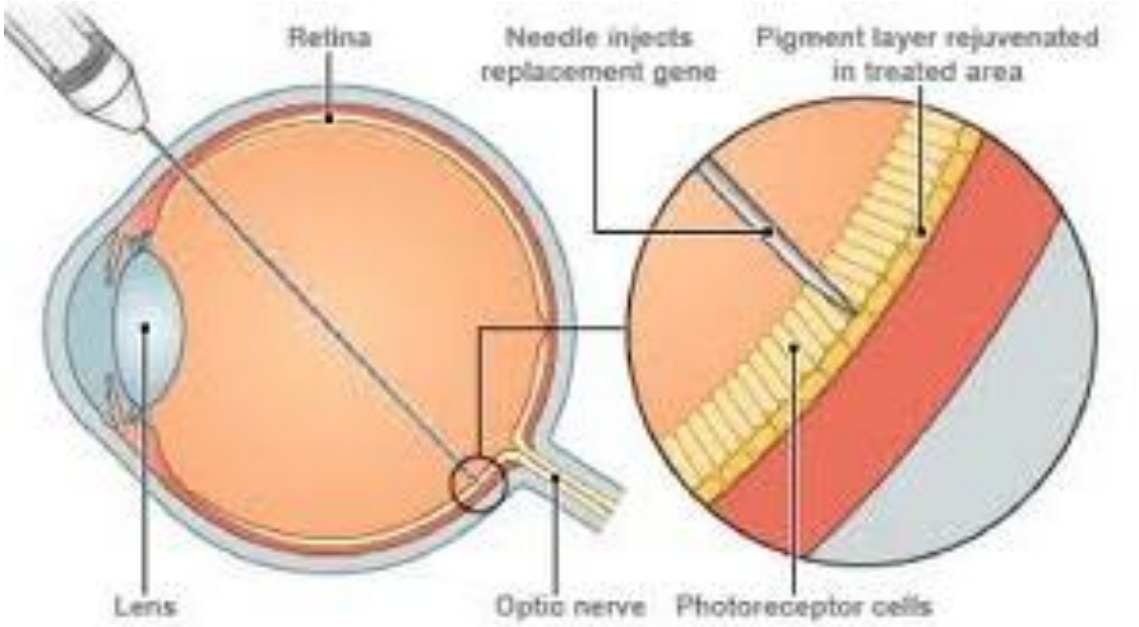
- Replace damaged tissue
- “Regenerative Medicine”
- Holy Grail
- Currently being tested at Harvard, other locations
- Caution needed

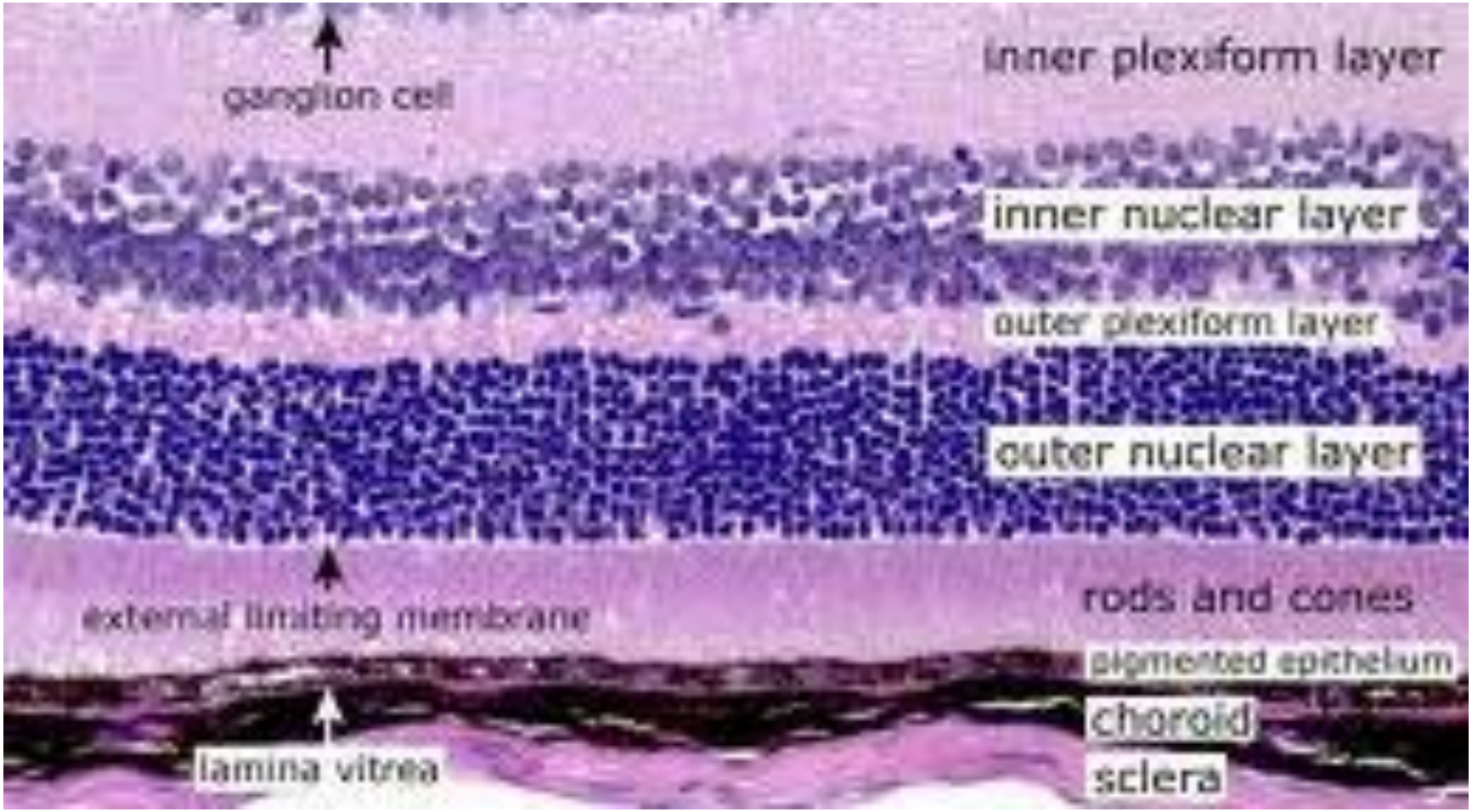


Retinal stem cells have been created



Subretinal surgery is now done




ganglion cell
inner plexiform layer
inner nuclear layer
outer plexiform layer
outer nuclear layer
external limiting membrane
rods and cones
pigmented epithelium
choroid
lamina vitrea
sclera


rods
cones
outer limiting membrane
Müller cells
horizontal cells
bipolar cells
amacrine cells
ganglion cells
nerve fiber layer

Dr. Stickler

Future is bright

Thank you!