

DeSanto-Shinawi syndrome

Ocular manifestations

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Desanto Shinawi Syndrome

Variable presentation

- +/- Developmental delay
- +/- Decreased muscle tone
- +/- Behavioral abnormalities
 - +/-GI abnormalities
 - +/- Eye abnormalities
 - +/- Facial differences

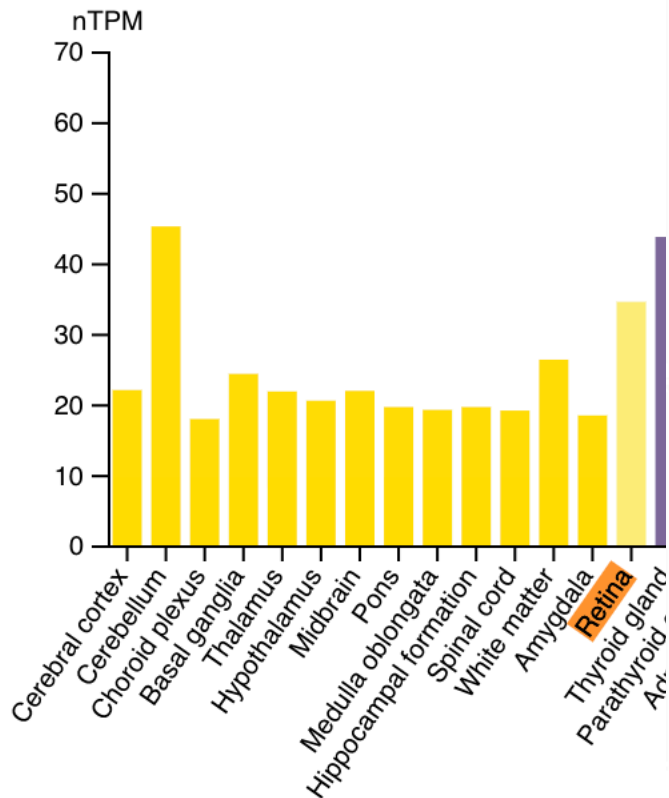
- **Secondary to a mutation in the WAC gene**

WAC gene

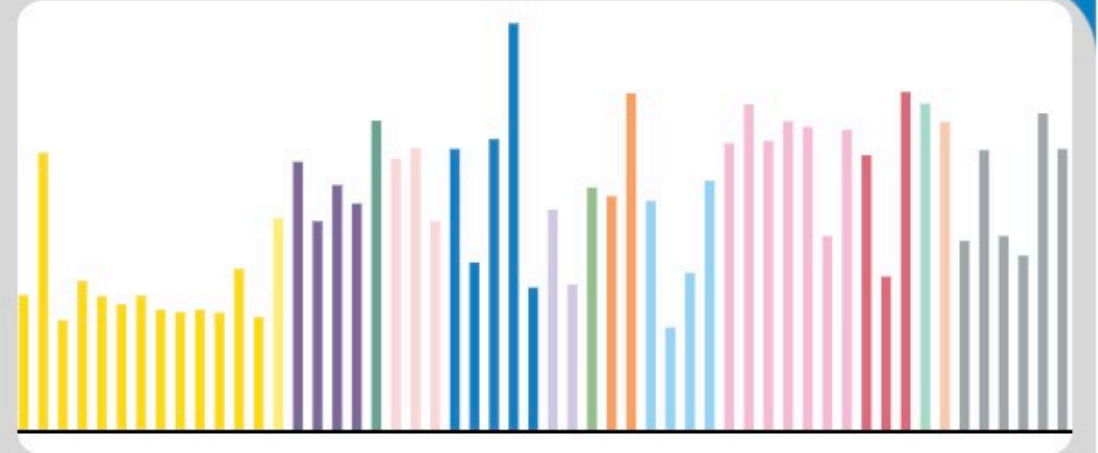
- This gene is expressed in all adult and fetal tissues. Including the eye

RNA EXPRESSION OVERVIEWⁱ

Consensus datasetⁱ



TISSUEⁱ



Tissue expression clusterⁱ

Non-specific - Transcription (mainly)

Tissue specificityⁱ

Low tissue specificity

Tissue distributionⁱ

Detected in all

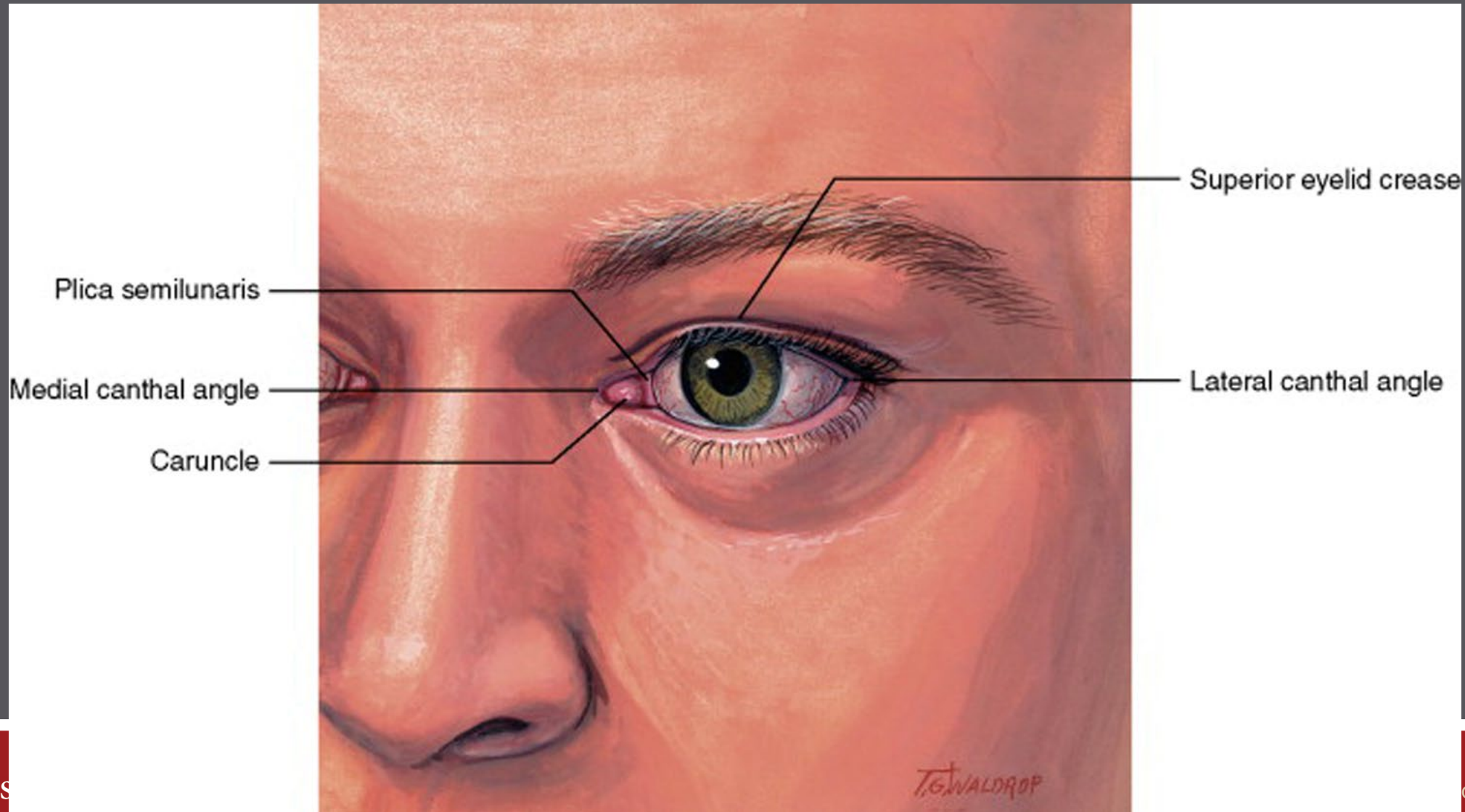
Protein expressionⁱ

Nuclear and cytoplasmic expression at variable levels in most tissues.

Commonly described eye findings

- Deep set eyes
- Hypertelorism
 - Ptosis
- Prominent brow
 - Strabismus
 - Myopia
 - Astigmatism
- Cortical visual impairment

Eyelids/orbit



Commonly described eye findings

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No implications
from an eye
health
standpoint.
Cosmetic

Commonly described eye findings

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Increased distance
between the orbits

No problems with health of
the eyes

Commonly described eye findings

- Deep set eyes
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 - **Ptosis**
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Ptosis

Can be caused by weakness in the muscles, nerves, or tendon that lifts the eyelid

If it is blocking the vision, can fix it with a surgery



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Prominent brow

Does not affect the health of the eye

Commonly described eye findings

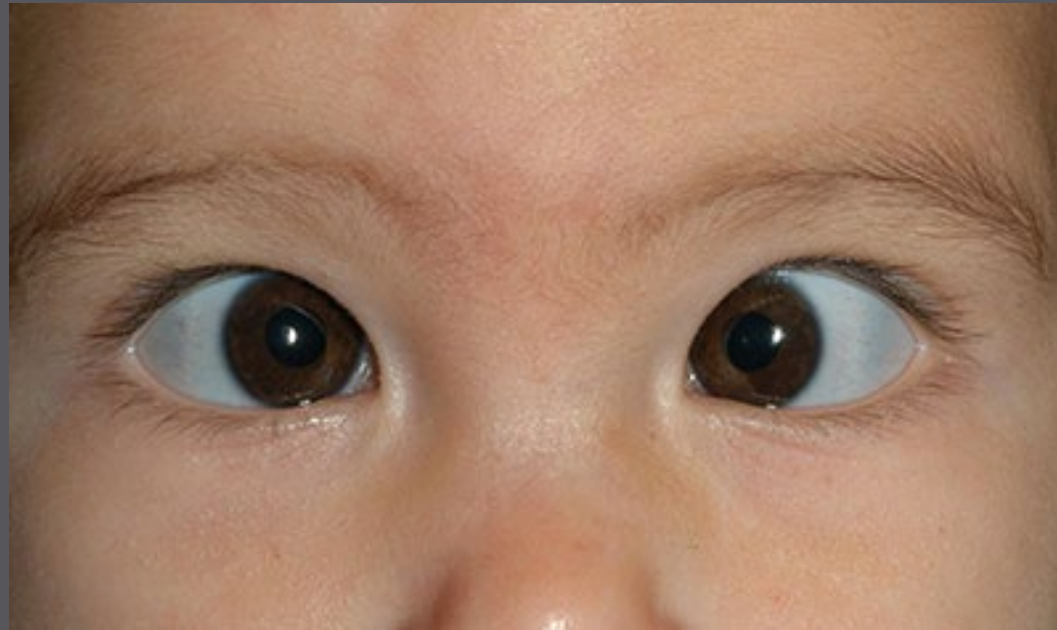
- Deep set eyes
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Strabismus

Any misalignment of the eyes

Common problem

Cause: unknown

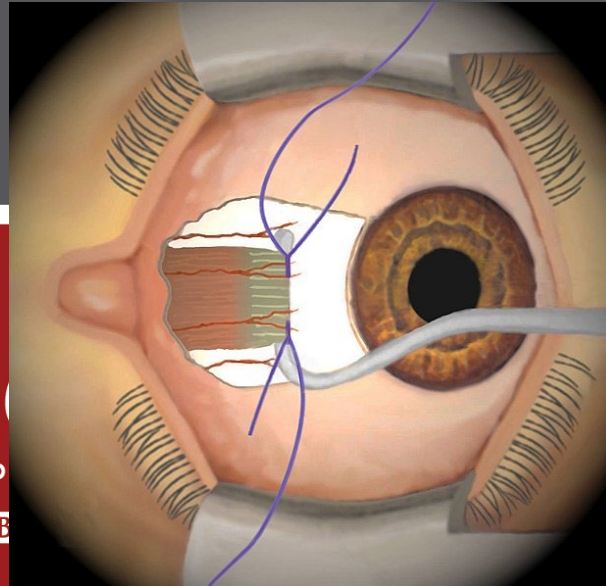


Strabismus: Eye Misalignment Terminology

- Horizontal strabismus
 - Esophoria / Esotropia (crossed inward)
 - Exophoria / Exotropia (drifted outward)
- Vertical Strabismus
 - Hypertropia (higher eye)
 - Hypotropia (lower eye)

Management of Strabismus

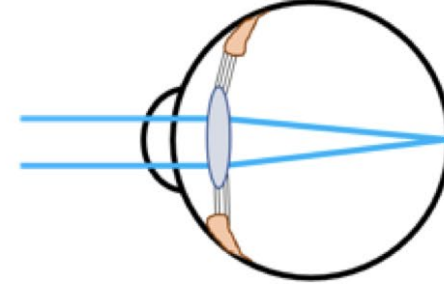
- Evaluation: full dilated examination, refraction
- Possible treatments:
 - Glasses
 - Patching (for amblyopia and strabismus control)
 - Surgery



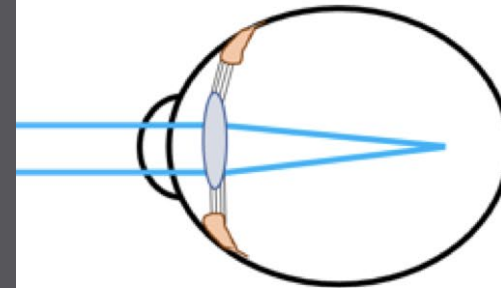
Commonly described eye findings

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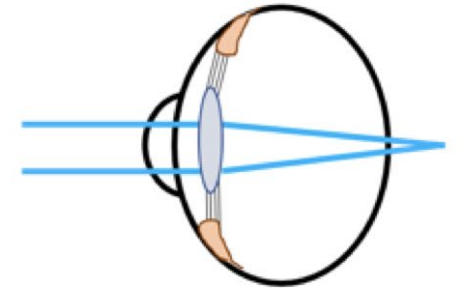
Can be corrected
with glasses



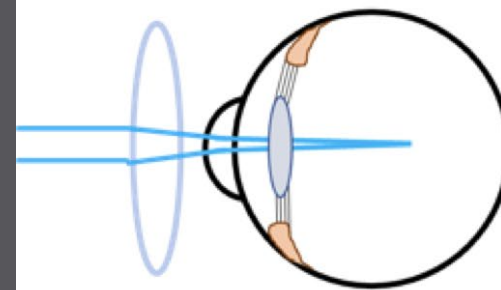
(a) Emmetropia



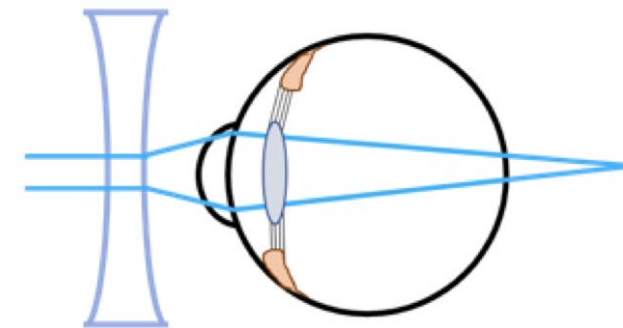
(b) Myopia



(c) Hyperopia



(d) Lens-Induced
Myopic Defocus



(e) Lens-Induced
Hyperopic Defocus

Myopia progression

Low Dose Atropine



Keep **Myopia** away,
go **outdoors** and **play!**

Our eyes are precious, we must take good care of them.

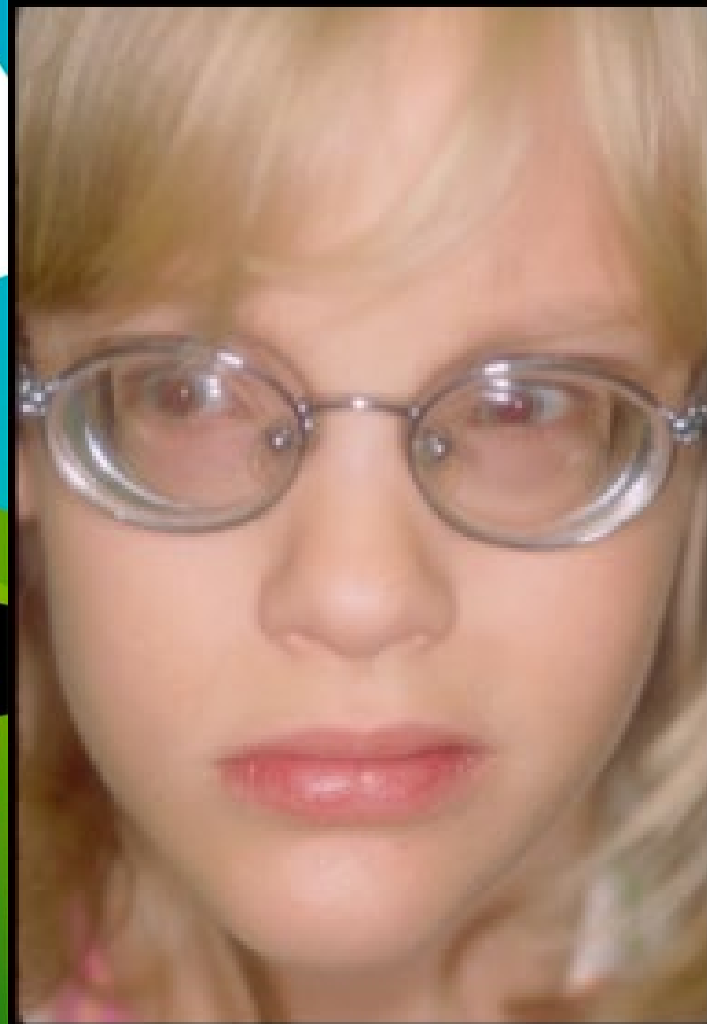
More time on

- Ball games
- Walks
- Fun at the playground and other outdoor activities every day!

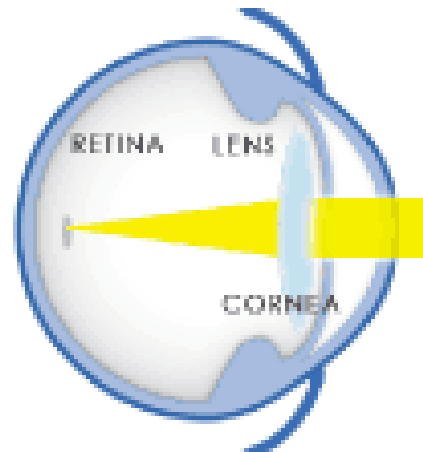
Less time on

- Computer games
- Hand-held games
- Handphone games

Health Promotion Board
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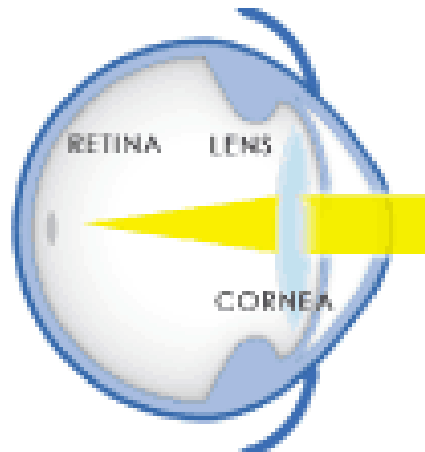


Astigmatism



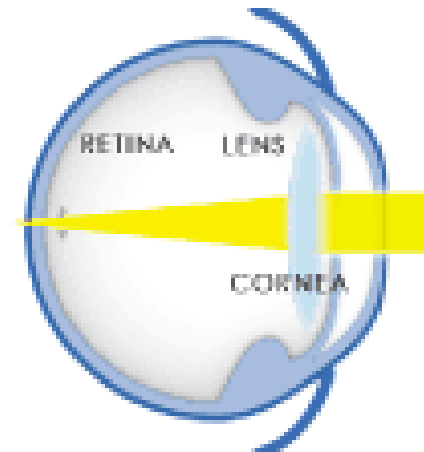
Emmetropia

Normal



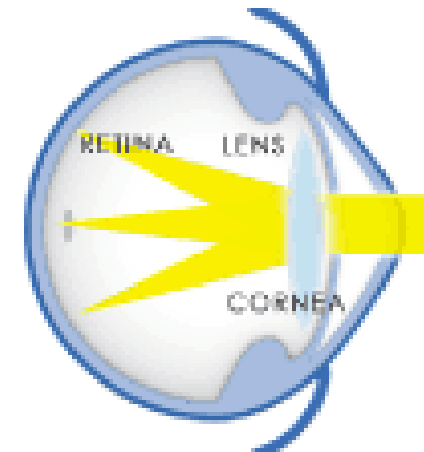
Myopia

Near-Sighted



Hyperopia

Far-Sighted



Astigmatism

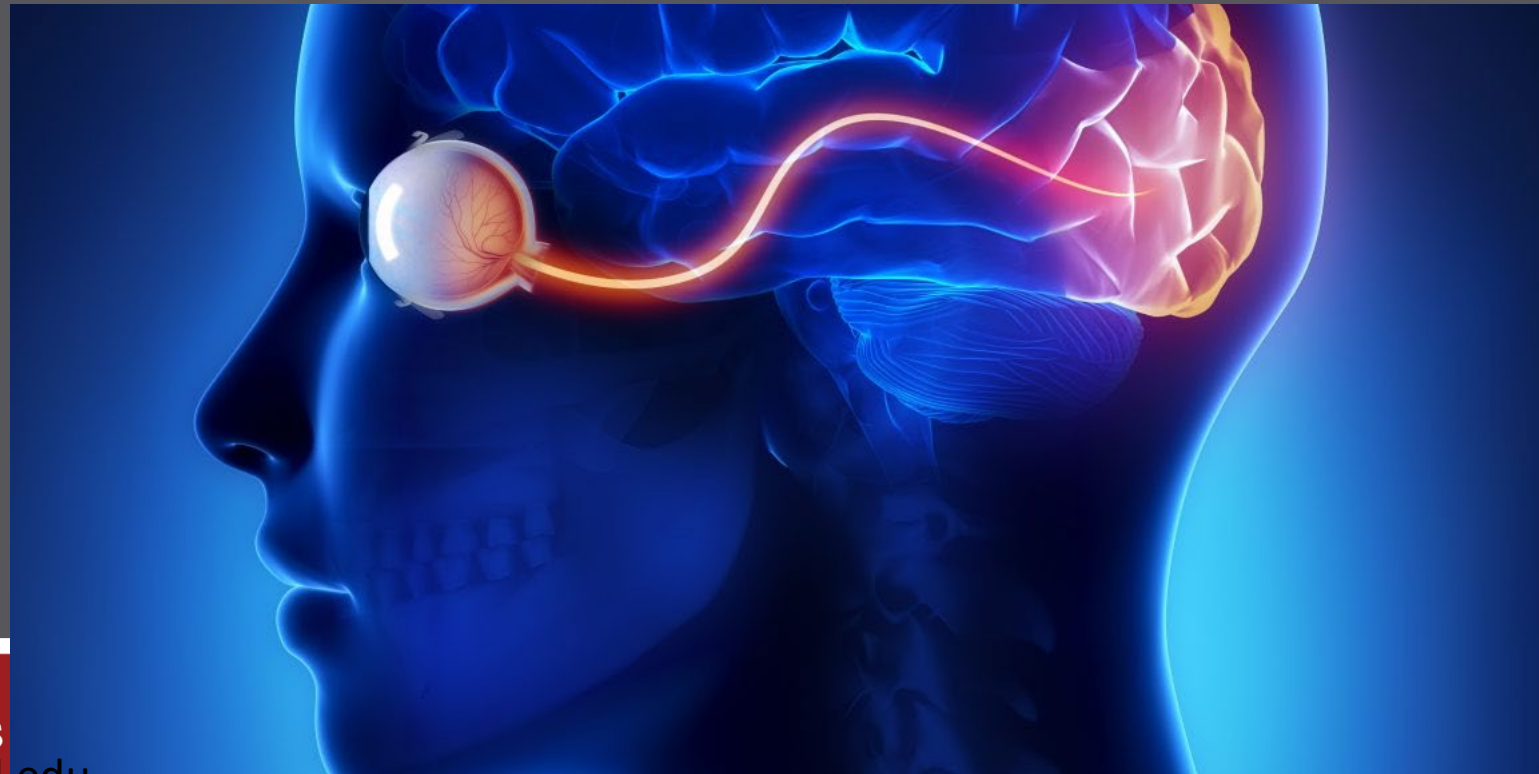
Irregular Vision

Commonly described eye findings

- Deep set eyes
- Hypertelorism
 - Ptosis
- Prominent brow
 - Strabismus
 - Myopia
 - Astigmatism
- **Cortical visual impairment**

Cortical blindness

- Cortical blindness
 - Stroke of the occipital lobe. Occipital lobe or pathology of the optic pathways. Depends on the extent of involvement, but patients can have no light perception vision.



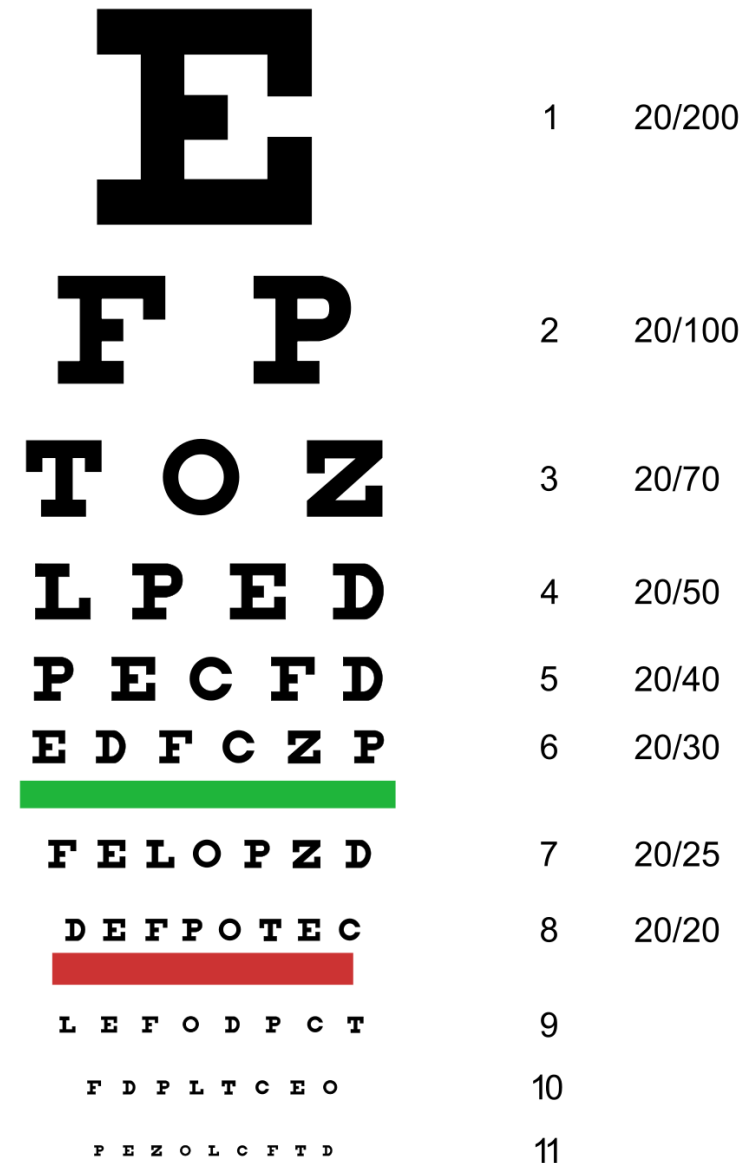
Visual acuity

- Visual acuity, i.e. 20/20
- Definition: how big an object has to be to see it.
- Cause: Any problem with the visual pathway.

E	1	20/200
F P	2	20/100
T O Z	3	20/70
L P E D	4	20/50
P E C F D	5	20/40
E D F C Z P	6	20/30
F E L O P Z D	7	20/25
D E F P O T E C	8	20/20
L E F O D P C T	9	
F D P L T C E O	10	
P E Z O L C F T D	11	

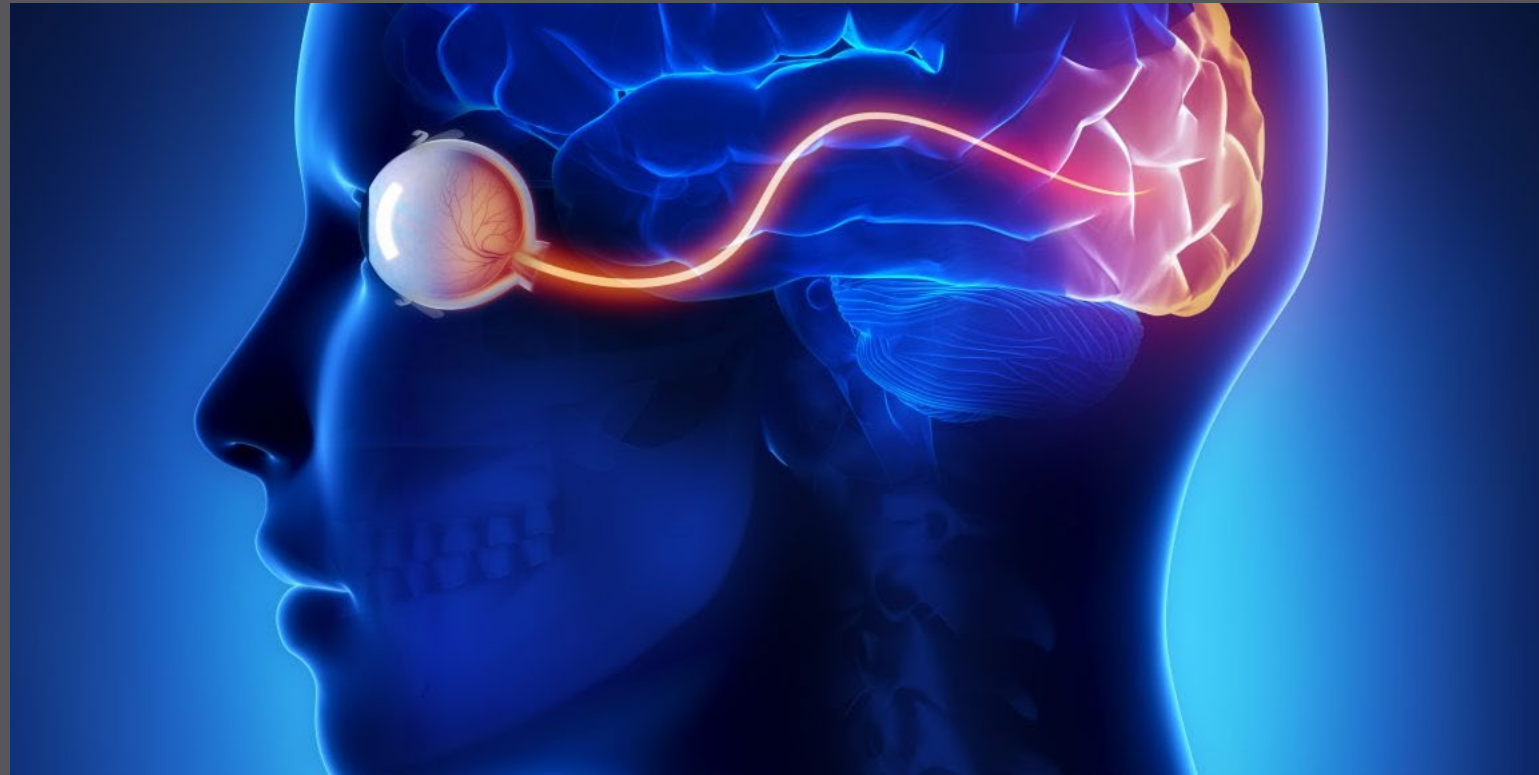
How does decreased visual acuity impact our patients?

- Clear images are important for learning, activities of daily living, acquisition of milestones.
- The brain isn't getting a clear image-makes it difficult to interact with the environment socially, difficult to interact with the environment-ADL's, difficult to see clearly in school-difficult to learn.
- The problem, as ophthalmologists, we most often "fix" or try to fix.



Visual Processing/Perceptual visual problems

- Visual processing (brain)
 - What you see
 - Is it important
 - Relating what you see to previously stored information.
 - Recognizing what you see.



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What can we offer to patients we cannot help as eye doctors?

- First, hope
 - Children often surprise us with how well they function and what they can do.
- Resources:
 - Delta Gamma-available for patients in the St. Louis area with low vision.
 - Occupational therapy –Vision service at WUSTL

ANNA PERLMUTTER, OTD

Pediatric Visual Disorders



Please call **(314) 286-1669** to schedule an appointment.

Therapist providing treatment: —

What can we do:

- Help teach children and their family strategies to improve their ability to be successful at home, in school, and out in the community.

- Visual perceptual strategies to improve the ability to safely mobilize throughout the community, read, and write
- Activities of daily living strategies including dressing, brushing teeth, bathing, feeding, toileting, etc.
- Sensory processing strategies to promote successful interaction with auditory, tactile, proprioceptive, and visual input
- Fine and gross motor strategies to increase the ability to successfully write, complete self-care tasks, play, etc.
- Executive function strategies to improve the ability to organize and plan out activities
- Social skill development to improve the ability to successfully interact with peers and family members

Email

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Questions?

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