



# Eye Care and Research Across the Lifespan: Keratoconus, Cataracts & More!

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uhealth

# Disclosures

- No financial disclosures
- Corneal collagen crosslinking and Photrexa are off label in children under 14 years old.
- Global Down Syndrome Foundation supported the purchase of the Pentacam topographer at our institution



Acknowledgement: Dr. Michael Puente



# Objectives

- Discuss the vision problems can affect development and quality of life in people with Down Syndrome from childhood to adulthood
- Review the latest research on vision and eye health in people with Down syndrome (and identify gaps that need more research)
- Recent advances in treatment for vision-threatening diseases strongly associated with Down Syndrome.

# Outline

- How can vision problems affect development and quality of life?
- Eye diseases in people with Down syndrome
- Unanswered research questions
  
- Who should examine me or my family member?
- How eye exams should be performed on patients with Down syndrome
- Recent advances in treatments for eye disease in people who have Down syndrome
- Recommended timelines for eye exams

# Outline

- **How can vision problems affect development and quality of life?**
- Eye diseases in people with Down syndrome
- Unanswered research questions

# Visual Development – General Population

## Forced-choice preferential looking

- 20/600 at birth
- 20/120 at 3 months of age
- 20/60 at 12 months of age
- 20/20 at 3 to 5 years of age

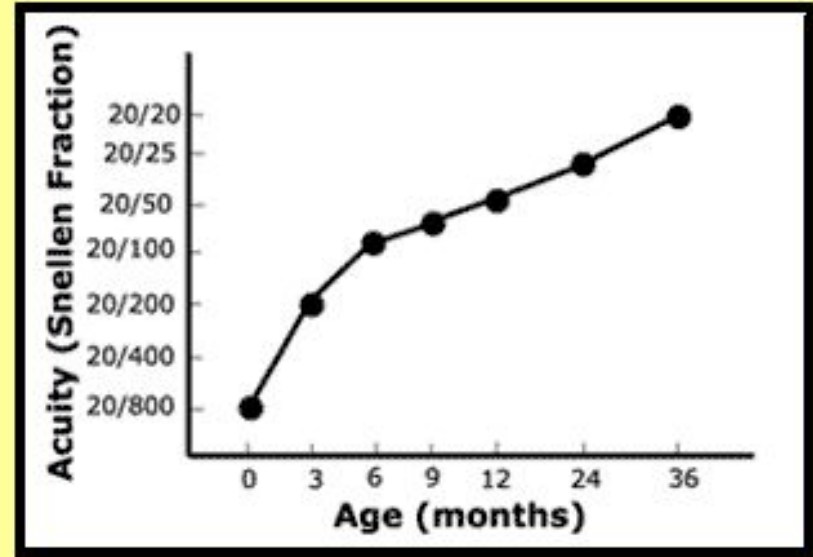
## Visual evoked potential

- 20/400 at birth
- 20/20 at 6 to 7 months of age

*Eustis & Guthrie, 2003*

*Visual Conditions 20*

Early Intervention Training Center for  
Infants and Toddlers With Visual Impairments  
FPG Child Development Institute  
University of North Carolina at Chapel Hill  
June 1, 2004



# Visual Milestones

Newborn – blinks to light

3 months – begins to make eye contact, tracks moving objects, social smiles

6 months – able to reach for objects, color vision develops

8-10 months – recognizes family and caregivers, looks at small objects like cereal, depth perception develops



# Visual Acuity in Children with Down Syndrome

- In one study of 35 children with Down syndrome, average visual acuity was 20/45
- This means that what someone with healthy eyes can see from 45 feet away, these kids could only see from 20 feet away

## Visual Psychophysics and Physiological Optics

### Static and Dynamic Measurements of Accommodation in Individuals with Down Syndrome

*Heather A. Anderson,<sup>1</sup> Ruth E. Manny,<sup>1</sup> Adrian Glasser,<sup>1</sup> and Karla K. Stuebing<sup>2</sup>*



# Visual Acuity in Children with Down Syndrome

- 94% of children with Down syndrome have below-average vision
- 79% of children with Down syndrome have visual acuity more than 2 standard deviations worse than average

**DMCN** Developmental Medicine & Child Neurology

## VISUAL ACUITY IN INFANTS AND CHILDREN WITH DOWN SYNDROME

Mary L. Courage, Russell J. Adams, Sandra Reyno, Poh-Gin Kwa

First published: July 1994 | <https://doi.org/10.1111/j.1469-8749.1994.tb11895.x> | Citations: 61

# Visual Acuity in Adults with Down Syndrome

- **46% of adults with DS between 50-59 years old have visual impairment (compared to 13% in adults with other causes of intellectual disability)**
- **85% of adults with DS older than 60 years old have visual impairment (compared to 20% in adults with other causes of intellectual disability)**



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Published online 2013 Jun 19. doi: [10.1111/jar.12062](#)

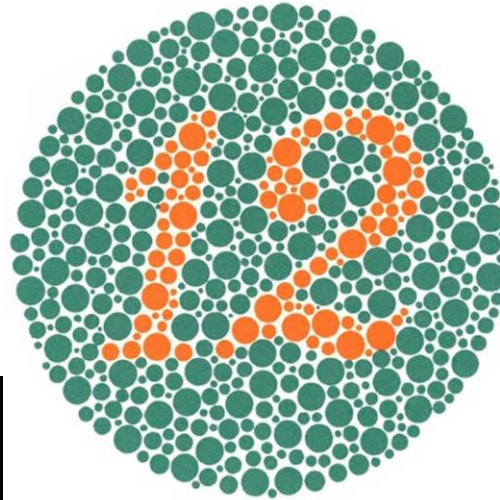
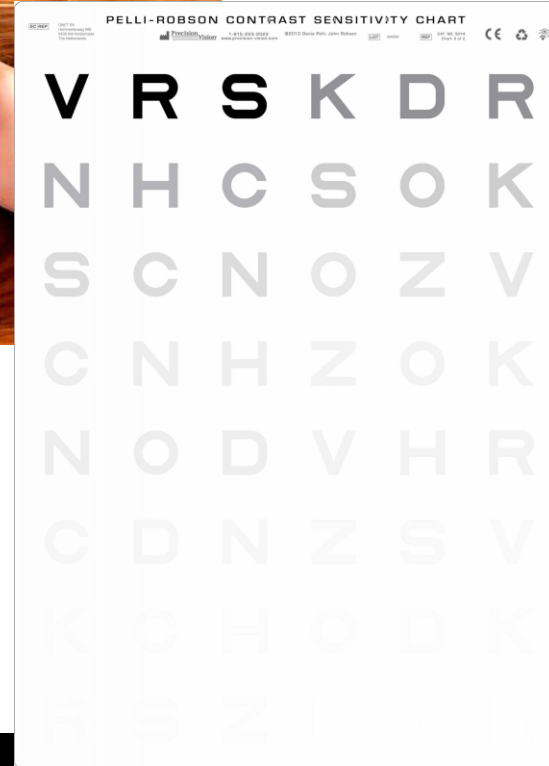
Vision Deficits in Adults with Down Syndrome

[Sharon J. Krinsky-McHale](#),<sup>1</sup> [Wayne Silverman](#),<sup>2</sup> [James Gordon](#),<sup>3,4</sup> [Darlynnne A. Devenny](#),<sup>1</sup> [Nancy Oley](#),<sup>5</sup> and [Israel Abramov](#)<sup>4,6</sup>

# Beyond Clarity

People with Down syndrome have been found to have decreased:

- *Depth perception*
- *Color vision*
- *Contrast sensitivity*



# Importance of good vision in childhood

- Vision affects all aspects of a child's development
  - *Social interaction*
  - *Language development*
  - *Motor skills*
  - *School*
  - *Independence*



# Impact of low vision in adults

1. Loss of the range and variety of experiences
2. Loss of the ability to move around
3. Loss of environmental control and self within it

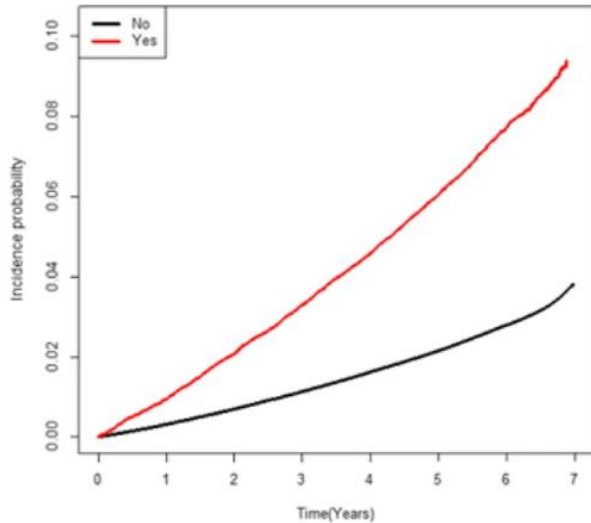


# Vision and dementia

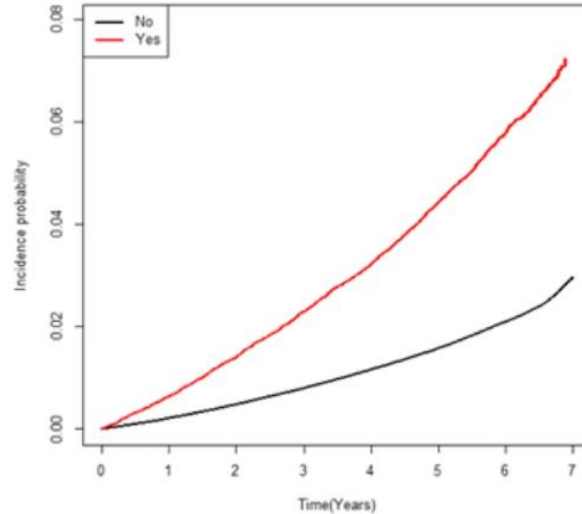
## *Risk of dementia in Koreans over 40 years old*

- **RED LINE** represents people with visual impairment
- **BLACK LINE** represents people without visual impairment

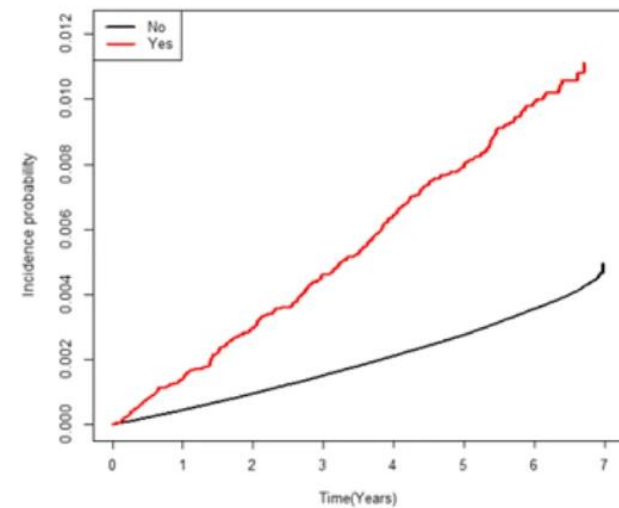
### Dementia



### Alzheimer's



### Vascular Dementia



# Vision and dementia

*Risk of dementia in Koreans over 40 years old*

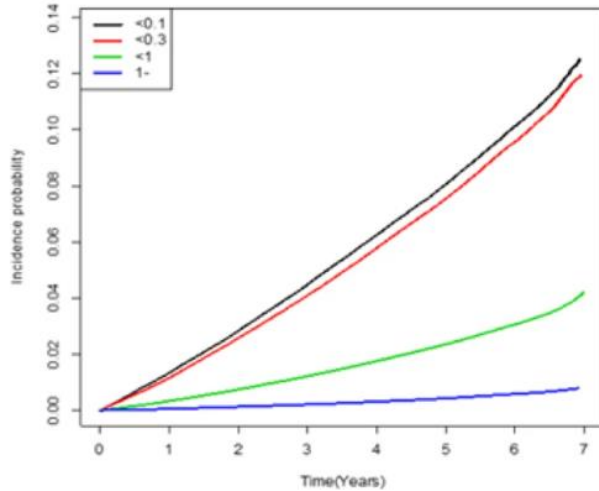
**BLUE LINE** = normal vision

**GREEN LINE** = mild visual impairment

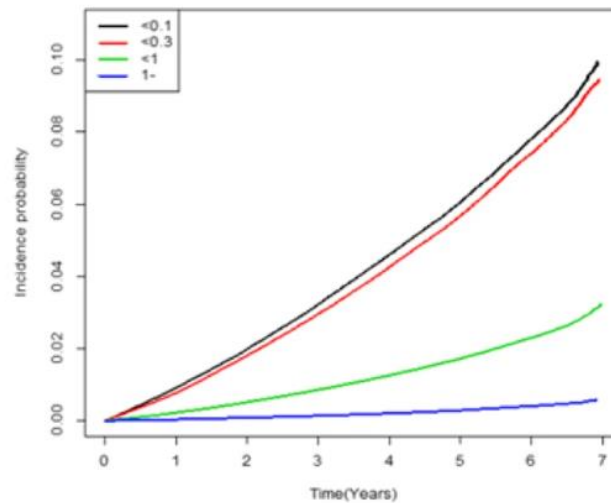
**RED LINE** = moderate visual impairment

**BLACK LINE** = severe visual impairment

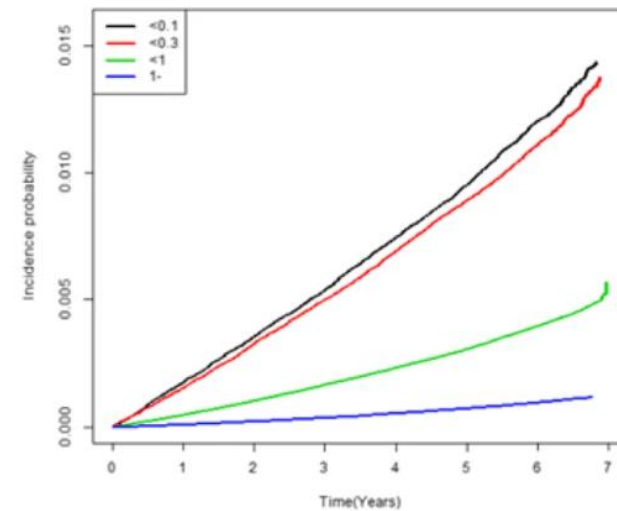
Dementia



Alzheimer's




Vascular Dementia



# Vision and dementia

- Vision problems may exacerbate cognitive decline
- Cataract surgery has been shown to reduce the risk of dementia and cognitive impairment
- Treating vision problems in adults may help preserve cognitive function
- **This specific issue has never been studied in Down syndrome**



Original Article |  Full Access

Cataract surgery is associated with a reduced risk of dementia: a nationwide population-based cohort study

W.-K. Yu, Y.-T. Chen, S.-J. Wang , S.-C. Kuo, B.-C. Shia, C. J.-L. Liu 

First published: 08 September 2014 | <https://doi.org/10.1111/ene.12561> | Citations: 17

## PLOS ONE

 OPEN ACCESS  PEER-REVIEWED

RESEARCH ARTICLE

Effect of cataract surgery on cognitive function in elderly: Results of Fujiwara-kyo Eye Study

Kimie Miyata, Tadanobu Yoshikawa, Masayuki Morikawa, Masashi Mine, Nozomi Okamoto, Norio Kurumatani, Nahoko Ogata 

Published: February 20, 2018 • <https://doi.org/10.1371/journal.pone.0192677>



American Journal of Ophthalmology

Volume 146, Issue 3, September 2008, Pages 404-409



Original article

The Impact of Cataract Surgery on Cognitive Impairment and Depressive Mental Status in Elderly Patients

Kotaro Ishii <sup>a, b</sup> , Takamichi Kabata <sup>a</sup>, Tetsuro Oshika <sup>a</sup>



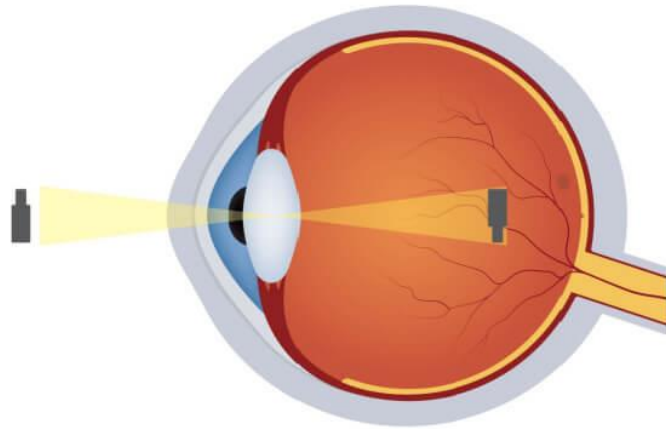
# Outline

- How can vision problems affect development and quality of life?
- **Eye diseases in people with Down syndrome**
- Unanswered research questions

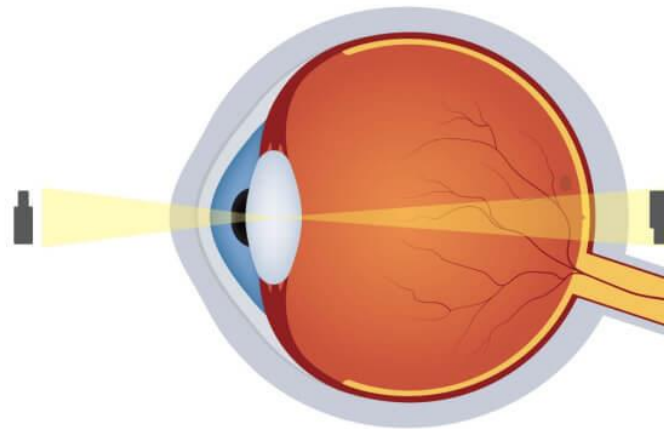
# Refractive error

- About 80% of children with Down syndrome need glasses
- Far-sightedness
- Near-sightedness
- Astigmatism

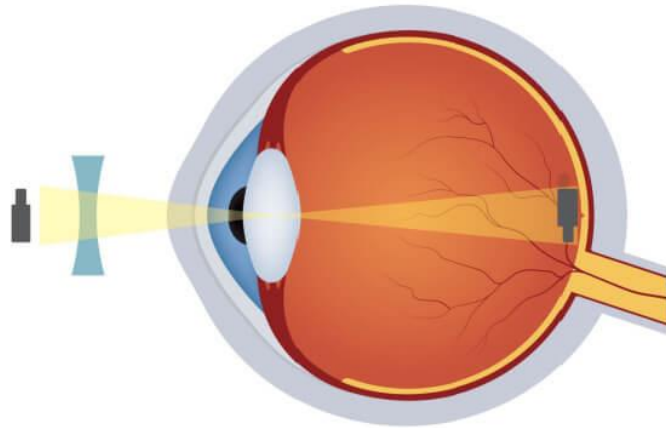




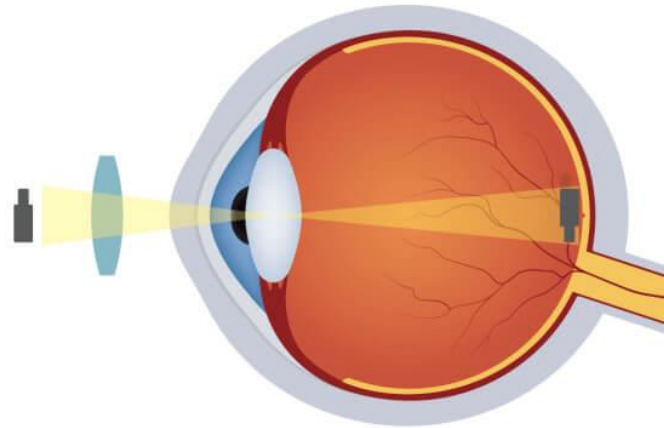
Myopia



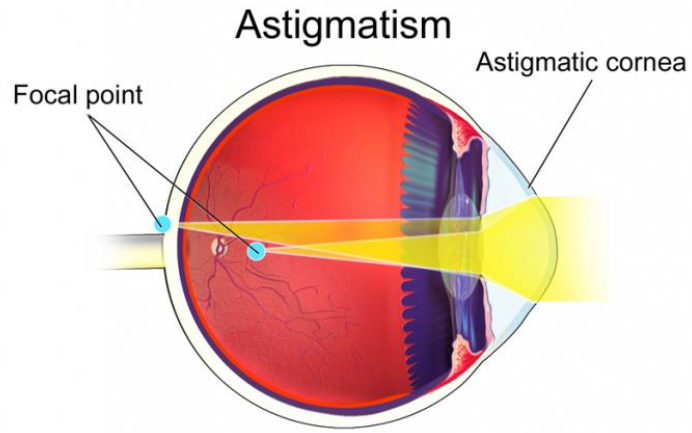
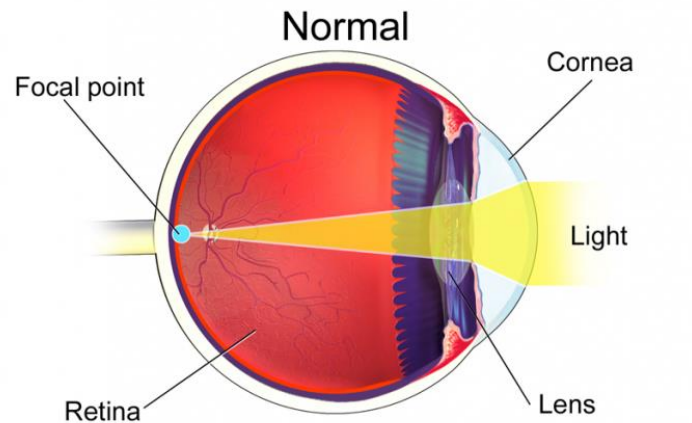
Hyperopia



Lenses for myopia



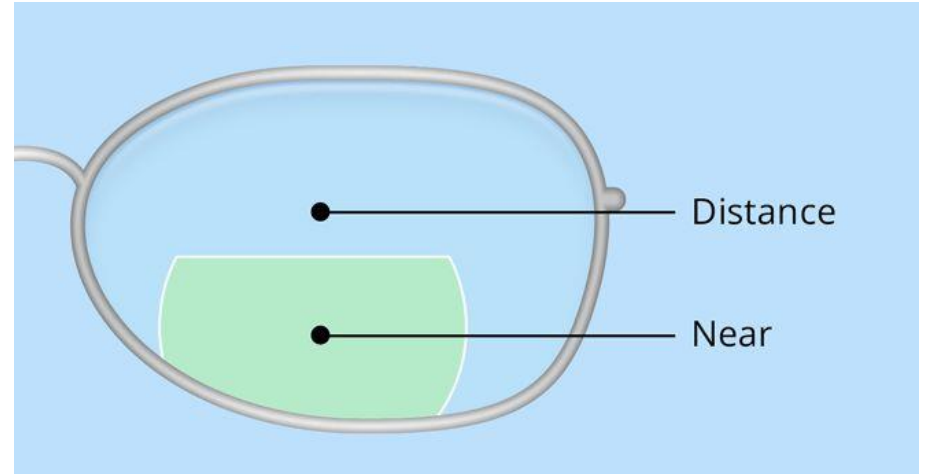
Lenses for hyperopia



*Astigmatic cornea distorts the focal point of light in front of and/or behind the retina*

# Hypoaccommodation

- “Accommodation” refers to the eye’s ability to focus on objects up close
- Up to 100% of people with Down syndrome have impaired accommodation
- Bifocal glasses have a stronger prescription at the bottom to help with near vision



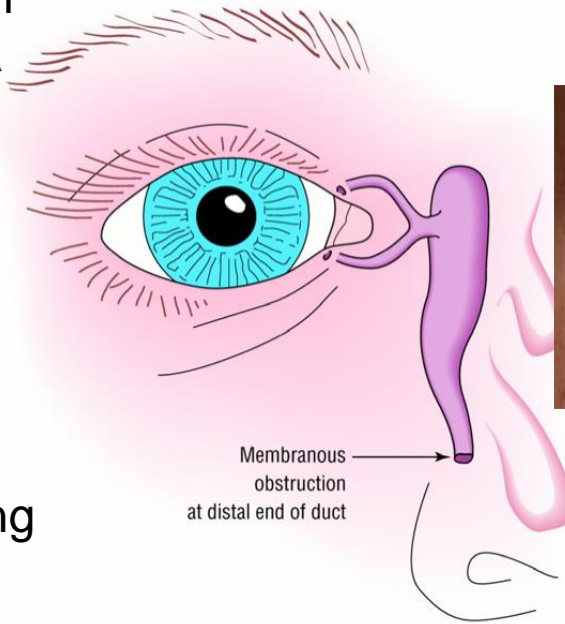
# Strabismus

- “Strabismus” refers to misalignment of the eyes
- Esotropia (or crossed eyes) is especially common
- Some people develop exotropia (or drifting eyes)
- Can affect depth perception
- *Treatment: glasses or surgery*



# Nasolacrimal duct obstruction

- Tears are supposed to drain to the inside of the nose via the nasolacrimal duct
- Up to 36% of children with Down syndrome have NLD obstruction
- Blocked tear ducts cause constant tearing and crusting
- *Treatment: surgery to open the blocked duct*



# Nystagmus

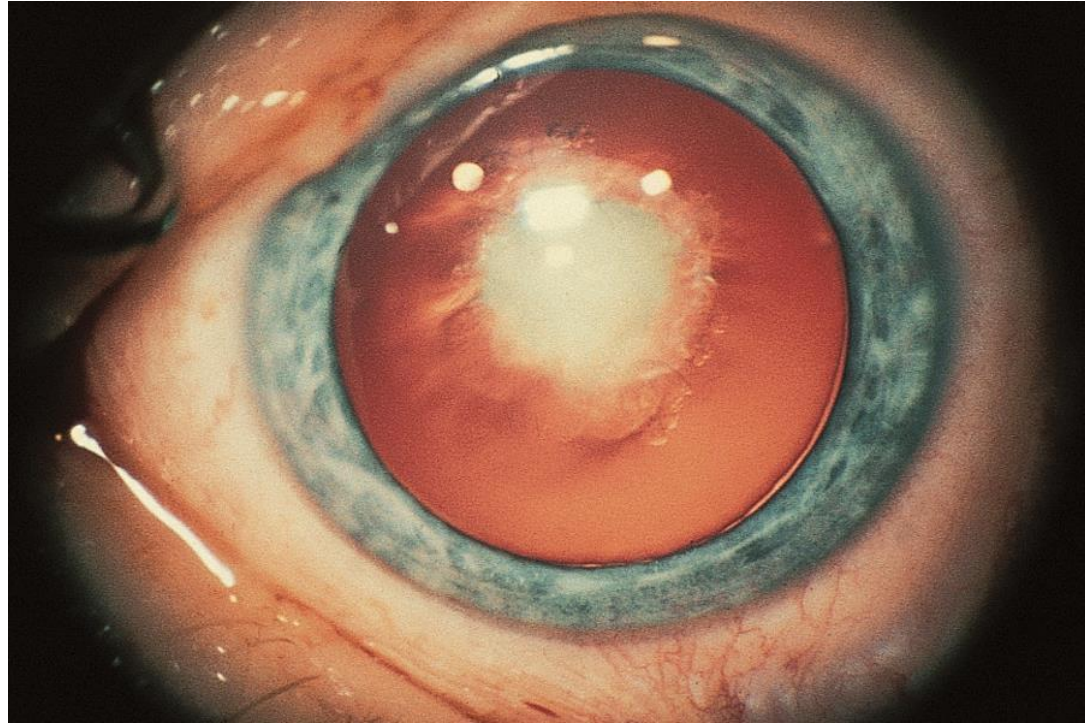
- “Nystagmus” refers to uncontrollable shaking movements of the eyes
- Up to 33% of people with Down syndrome have nystagmus
- Nystagmus is associated with low vision
- Treatment: no known treatment





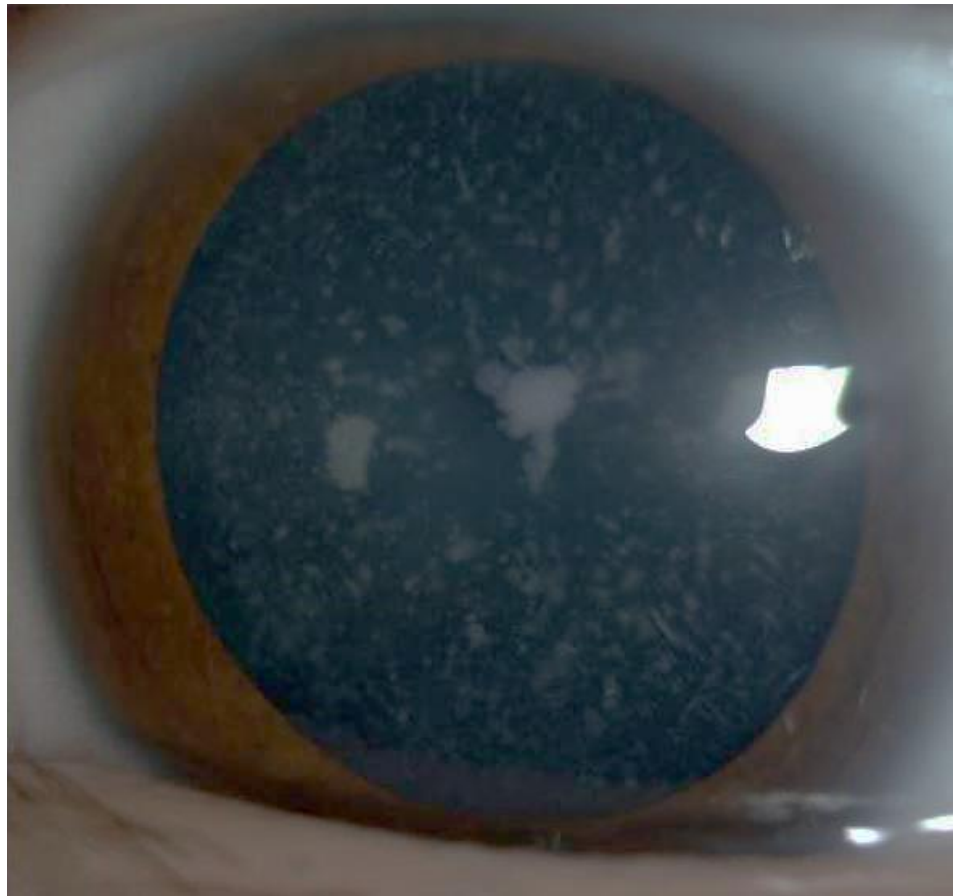
# Congenital cataracts

- “Cataract” refers to cloudiness of the lens inside the eye
- Babies with Down syndrome have about 300 times increased risk of being born with a cataract
- *Treatment: surgery to remove the cloudy lens*



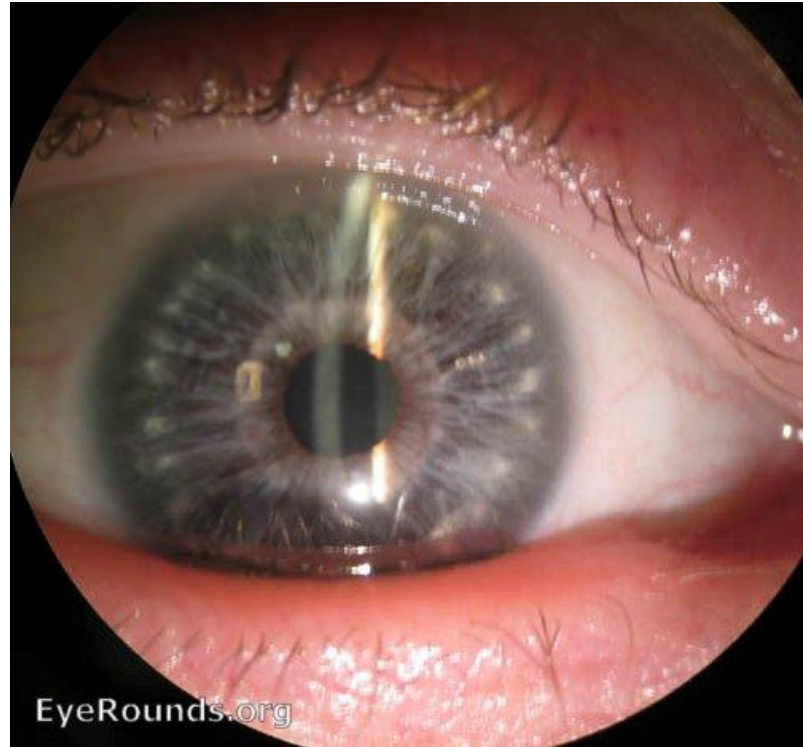
# Other cataracts

- People with Down syndrome develop cataracts at an earlier age than the general population
- Cataract surgery often needed as early as 40-50 years old
- About half of people with DS have “blue-dot cataracts,” which usually do not affect vision



# Brushfield spots

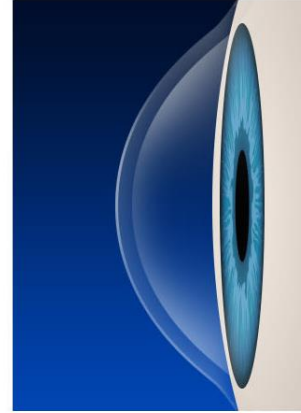
- White/gray/brown spots around the periphery of the iris
- No effect on vision
- Commonly seen in people with Down syndrome
- *Treatment: none needed*



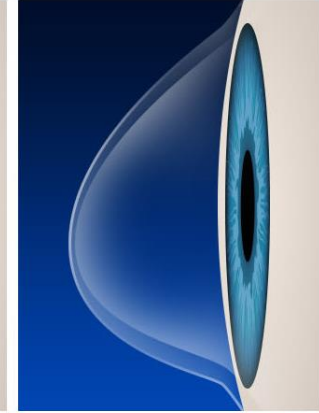
# Keratoconus

- The cornea is the clear front layer of the eye (like the windshield of a car)
- Keratoconus is a disease where the cornea changes shape from being round to being cone-shaped
- Causes progressively blurrier vision
- **Up to 32% of people with Down syndrome develop keratoconus**
- *Treatment: corneal cross-linking surgery, hard contact lenses, corneal transplant surgery*

Normal cornea

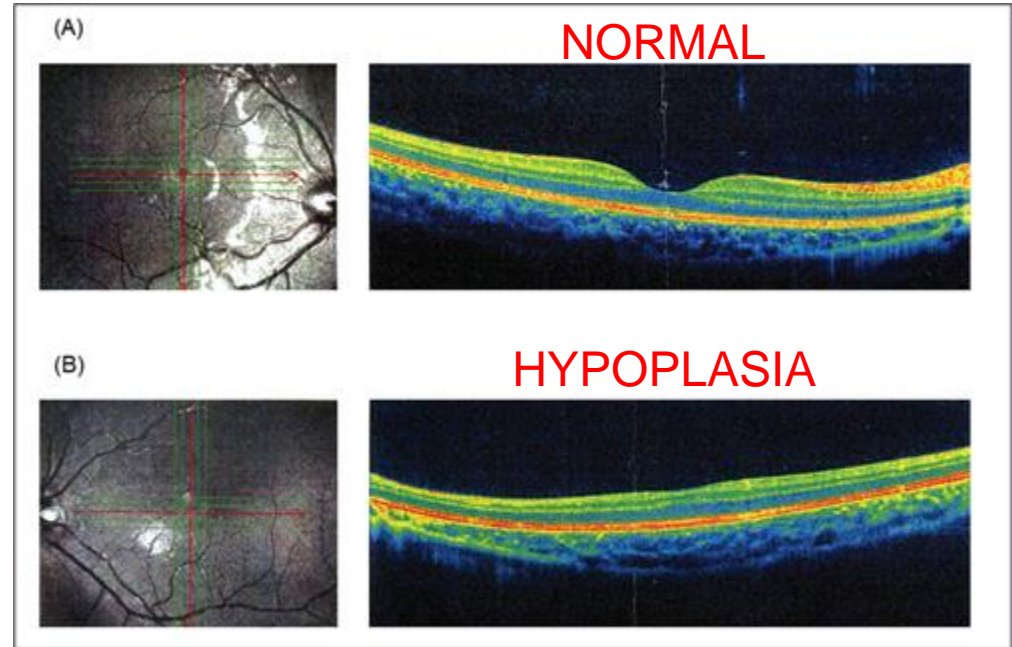


Keratoconus



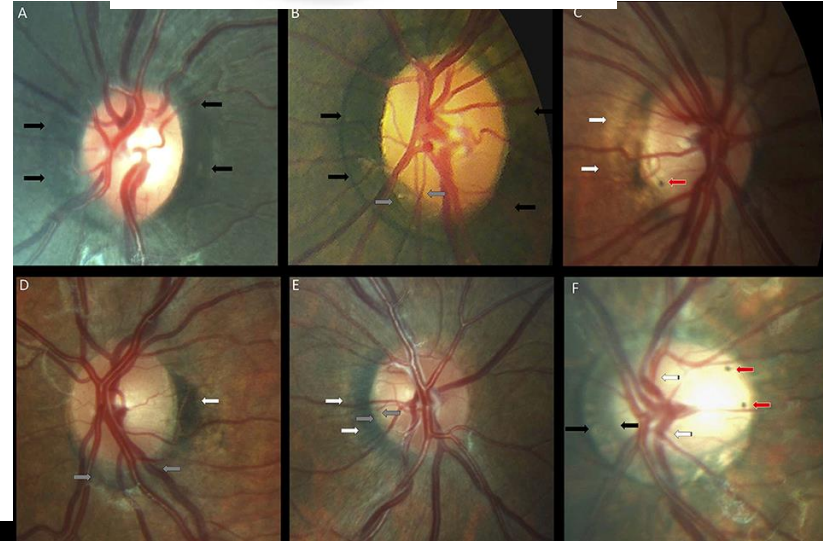
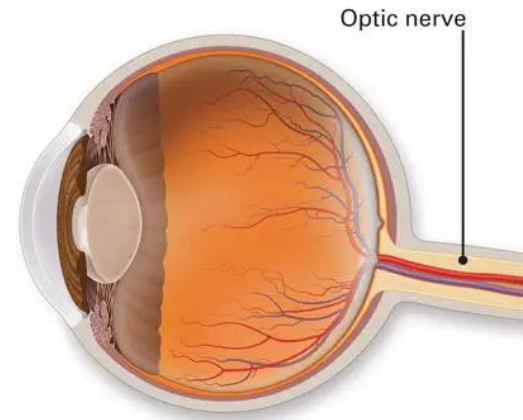
# Foveal hypoplasia

- The fovea is the part of the retina responsible for central vision
- Under-developed foveas lead to limited central vision
- Recent studies have shown that most children with Down syndrome have foveal hypoplasia
- *Treatment: no known treatment*



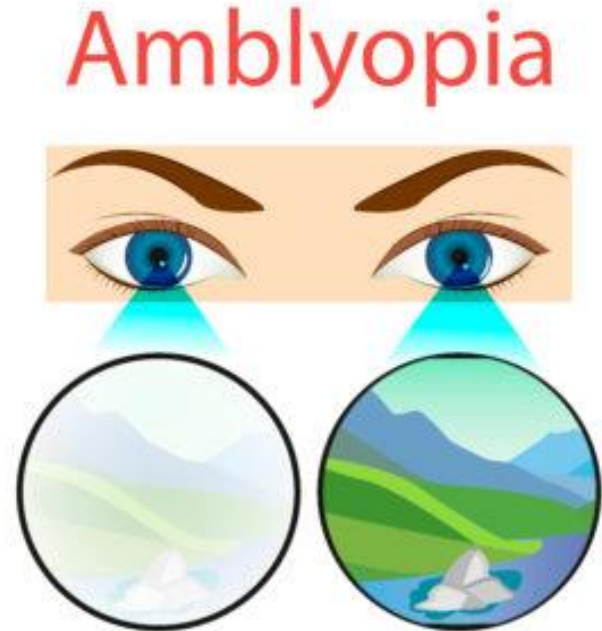
# Optic nerve anomalies

- The optic nerve is the cable that connects the eye and the brain
- Optic nerve abnormalities limit the eye's ability to send a clear signal to the brain
- People with Down syndrome commonly have abnormal optic nerves
- *Treatment: no known treatment*



# Amblyopia

- aka “lazy eye”
- Refers to blurrier vision in one eye due to the brain learning to ignore that eye
- Can be caused by any of the diseases discussed thus far
- Affects between 3-26% of people with Down syndrome
- *Treatment: glasses, patching, or eye drops*



# Cortical visual impairment

- The “visual cortex” is the part of the brain responsible for processing information from the eyes
- Problems with the visual cortex can cause decreased vision
- Down syndrome is associated with impaired cortical development, and cortical atrophy throughout life
- *Treatment: no known treatment*





# Outline

- How can vision problems affect development and quality of life?
- Eye diseases in people with Down syndrome
- **Unanswered research questions**

# Pediatric Research Questions

- How quickly does keratoconus worsen in children with Down syndrome?
- Why do children with Down syndrome develop nystagmus?
- How much do optic nerve abnormalities affect vision in children with Down syndrome?
- Why do babies with Down syndrome have such a high risk of cataracts?
- Should all kids with Down syndrome wear bifocal glasses?
- Why do people with Down syndrome develop keratoconus?

# Adult Research Questions

- What is the risk of common adult eye diseases in people with Down syndrome?
  - *Macular degeneration*
  - *Diabetic retinopathy*
  - *Glaucoma*
- Can retinal studies indicate risk of future dementia in people with Down syndrome?
- What type of lens implant works best for adults with Down syndrome after cataract surgery?
- Do visually impaired people with Down syndrome have higher risk of dementia?
- How frequently do adults with Down syndrome need an eye exam?

# Outline

- Who should examine me or my family member?
- How eye exams should be performed on patients with Down Syndrome
- Recent advances in treatments for eye disease in people who have Down Syndrome
- Recommended timelines for eye exams

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# Different types of eye doctors

## **Ophthalmologist – MD**

4 years undergraduate  
4 years medical school,  
1 year of internship  
3 years of ophthalmology residency

Pediatric Ophthalmologist – extra  
year of fellowship training (9 years)

## **Optometrist – OD**

4 years of undergraduate  
4 years optometry school

Pediatric Optometrist – extra  
year of training (5 years)

# Different types of eye doctors

## Ophthalmologist – MD

- Surgeon
- Specialist in medical and surgical diseases of the eye
- Extensive medical training
- Complex and medical / surgical care

## Optometrist – OD

- Glasses
- Contact lens experts
- Variable medical experience
- Great /appropriate for routine care

# Who should examine me or my family member?

.....It depends!

## Babies / Children

- Pediatric ophthalmology
- Pediatric optometrist with experience with DS

## Teens

- With years of stable exams optometry is a great option



# Who should examine me or my family member?

## Adults

- Known medical eye problem (cataracts, strabismus, keratoconus): Comprehensive ophthalmologist *at least* once a year
- Very stable eye exams as child: optometrist with experience in DS annually.
- At minimum, would check in with eye MD every 5 years

# Who should examine me or my family member?

## *Special situations:*

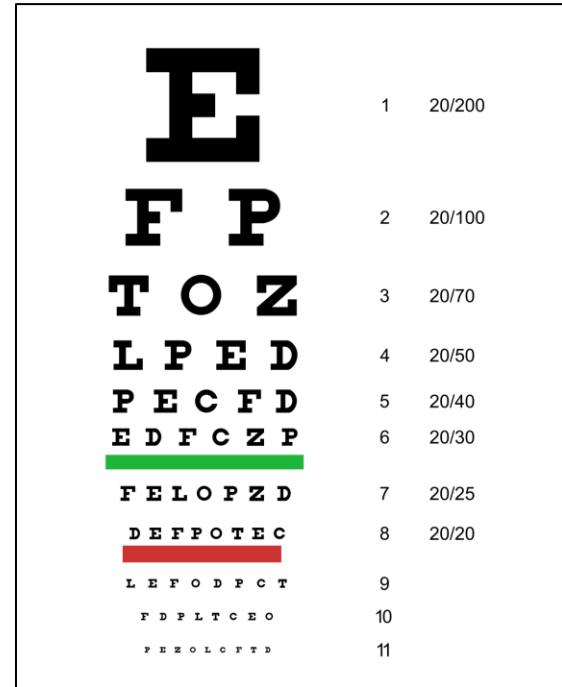
- ❖ Non verbal / difficult communication /behavior
  - Transition clinics?
  - MD with experience
- ❖ Cataracts
  - Needs surgeon
- ❖ Keratoconus
  - Pediatric or cornea specialist with experience in keratoconus in patients with DS
- ❖ Glaucoma
  - Pediatric or pediatric glaucoma specialist

# Outline

- Who should examine me or my family member?
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# What should a comprehensive eye exam look like in a person with Down Syndrome?

- Visual acuity testing
- Eye pressure
- Eye alignment and motility
- Dilation
- Refraction (glasses check)
- Special testing: topography, photos, etc when needed



# What should a comprehensive eye exam look like in a person with Down Syndrome?

- Visual acuity testing
  - Snellen chart (regular letters)
  - HOTV (4 letters to choose from)
  - Allen (pictures) – ok but unless young child, not desirable
  - Teller Acuity cards when needed
  - Don't forget about NEAR vision!



# What should a comprehensive eye exam look like in a person with Down Syndrome?



Checking eye pressure to evaluate for glaucoma

# What should a comprehensive eye exam look like in a person with Down Syndrome?



# What should a comprehensive eye exam look like in a person with Down Syndrome?

- Dilation +/- photos
- Should I pay extra for photos?

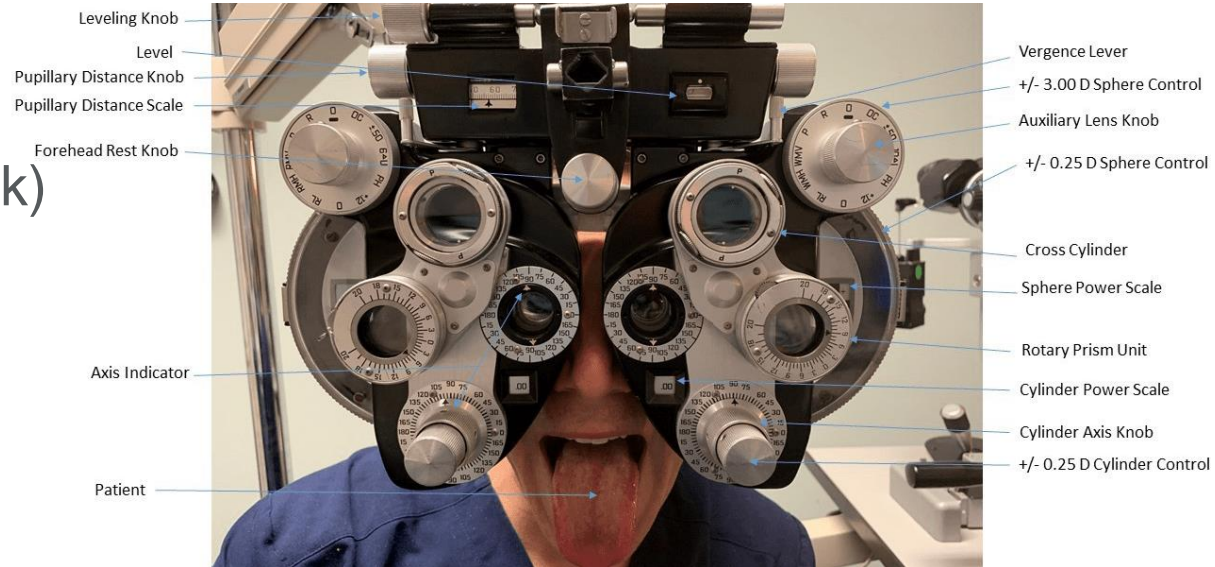




# What should a comprehensive eye exam look like in a person with Down Syndrome?

Refraction (glasses check)

PHOROPTER  
(lens 1 or lens 2)



# What should a comprehensive eye exam look like in a person with Down Syndrome?

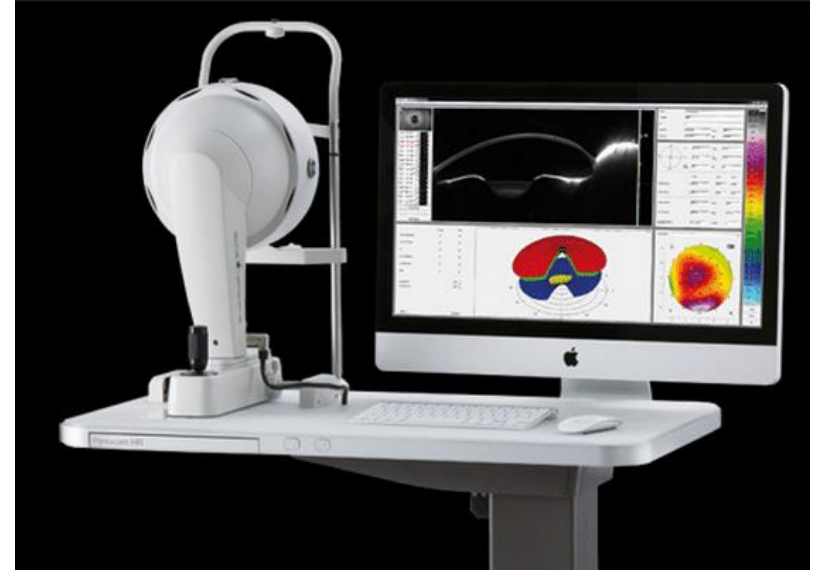
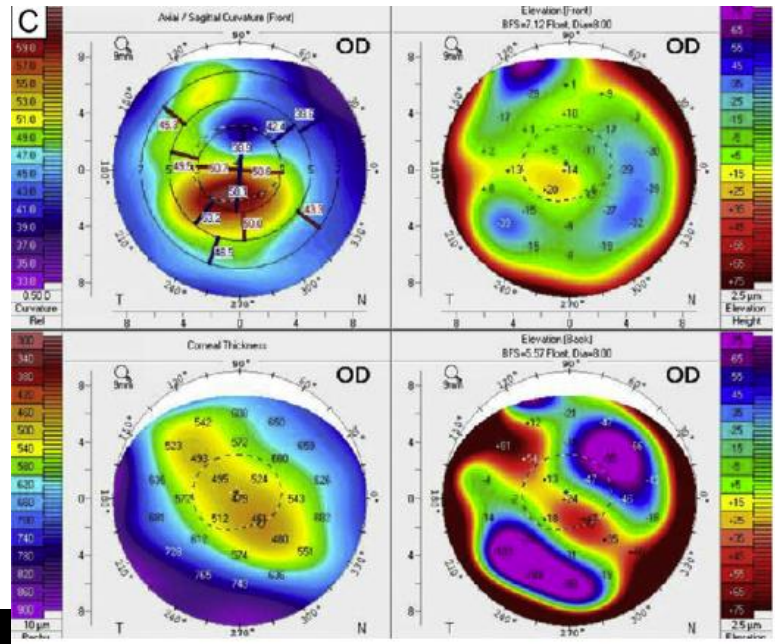
Refraction (glasses check)

**RETINOSCOPY**

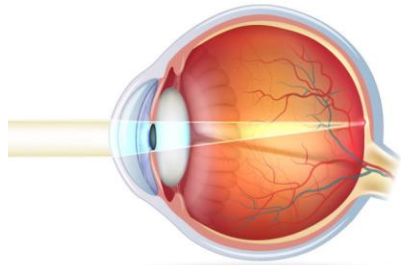


# What should a comprehensive eye exam look like in a person with Down Syndrome?

Topography!



# A few words about glasses



NORMAL VISION



# A few words about glasses

- ❖ People with Down Syndrome have much higher rates of needing glasses – higher astigmatism, nearsightedness (myopia), and farsightedness (hyperopia)
  - MUCH more likely to need a **bifocal** even as a child
  - Bifocals can help with acceptance of glasses in children



Specs4us.com

# A few words about glasses

Certain brands that fit better than others, however just like shoes some brands fit one person better than another

Glasses online? Generally would **avoid** for first pair or new fit.



<https://jonaspaleyewear.com/>

# Outline

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# Recent advances in treatments for eye disease in people who have Down Syndrome

- Crosslinking for Keratoconus
- Cataract surgery
- Strabismus (eye alignment)
- Blocked tear ducts



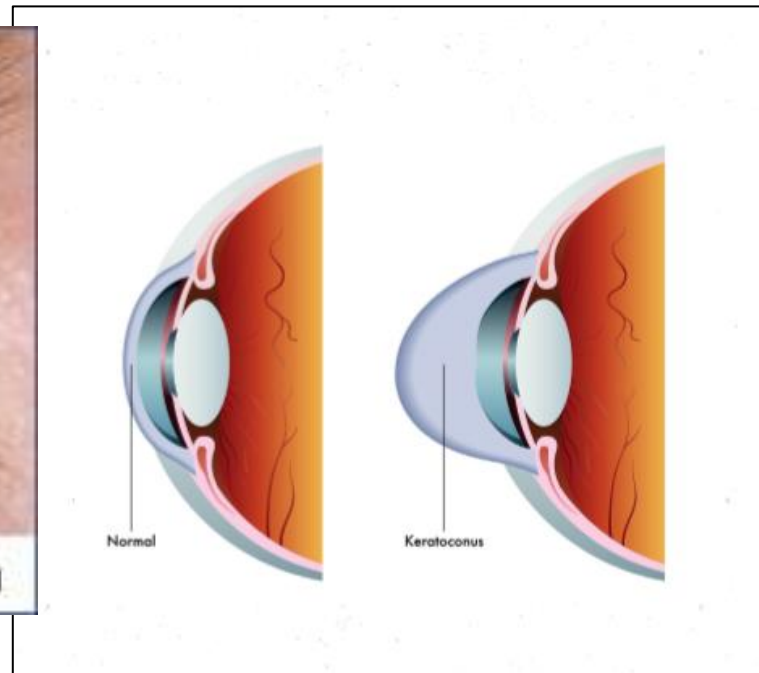




# A patient story



# Exam under anesthesia, April 2018



# November 2018 – severe complication of keratoconus called corneal hydrops





How could I prevent this?

Am I missing cases of keratoconus? How would I know?

Why can't I crosslink my patients?



# Keratoconus: focus on Down Syndrome

- **KCN affects people at a rate 10-300x more frequently in people with Down Syndrome**
- Alternations in chromosome 21 may affect collagen
- Eye rubbing/Allergy – very common in patients with DS
- Numbers are variable:
  - People with Down syndrome sometimes are hard to examine
  - Variable definition of keratoconus

# Corneal Morphologic Characteristics in Patients With Down Syndrome

Jorge L. Alio, MD, PhD, FEBO; Alfredo Vega-Estrada, MD, PhD; Pablo Sanz, OD, MSc; Amr A. Osman, MD, PhD; Ahmed M. Kamal, MD, PhD; Amr Mamoon, MSc; Hany Soliman, MSc

- 112 patients with Down Syndrome
- Included mostly adults, some children



It was observed that around 75% of the patients with DS diagnosis had corneal morphologic irregularities compatible with keratoconus. There are great variability and contradic-

# Keratoconus and corneal morphology in patients with Down syndrome at a pediatric hospital

Lauren M. Imbornoni, MD,<sup>a</sup> Ronald E. Wise, MD,<sup>a</sup> Michael J. Taravella, MD,<sup>a</sup> Francis Hickey, MD,<sup>b</sup> and Emily A. McCourt, MD<sup>a</sup>

- What about **children** with Down Syndrome?
- If anyone should be diagnosed early, shouldn't it be patients with Down Syndrome?

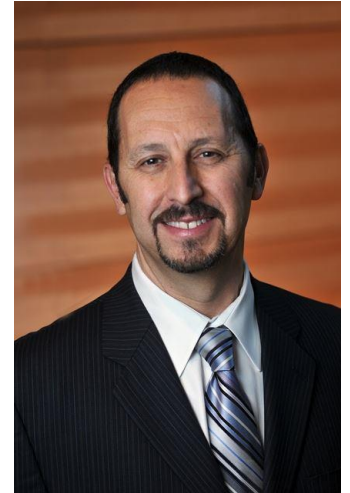
JAAPOS 2020 epub ahead of print



# Keratoconus and corneal morphology in patients with Down syndrome at a pediatric hospital

Lauren M. Imbornoni, MD,<sup>a</sup> Ronald E. Wise, MD,<sup>a</sup> Michael J. Taravella, MD,<sup>a</sup> Francis Hickey, MD,<sup>b</sup> and Emily A. McCourt, MD<sup>a</sup>

- We started to look at our patients
- 10 month period (August 2018 to May 2019)
- Pentacam  
**Generous gift from Global Down Syndrome Foundation**



JAAPOS 2020 epub ahead of print

# Results, continued

- Definite keratoconus: 4 eyes of 4 patients (13%)
- KCN suspect: 8 eyes of 8 patients (26%)
- 10 patients (32%) had KCN or KCN suspect in at least one eye
- Abnormal scan: 14 eyes of 10 patients (32%)
- 8 patients (26%) had normal scan of both eyes

**18 patients (58%) had at least one eye with abnormal cornea,  
KCN or KCN suspect**



# Conclusion



Keratoconus is often a much more aggressive in pediatric patients and needs to be treated.

We recommend screening patients with Down Syndrome for keratoconus with topography and tomography, regardless of age.

# Follow up on Lauren

- Right eye with significant scarring
- **Left eye crosslinked January 2019 – stable cornea**

Video on keratoconus



# Ongoing research

## Feasibility of topography in children with DS

- Of 219 patients, 58 children (26.5%) underwent corneal evaluation with Pentacam screening, and 161 did not have any screening attempted.
- Of the 58 children screened, **a quality scan was obtained in at least one eye of 44 of these patients (75.9%).**
- The children with successfully screening was significantly older than the other two groups and rate of keratoconus/keratoconus suspect was 34.9%

# Ongoing research

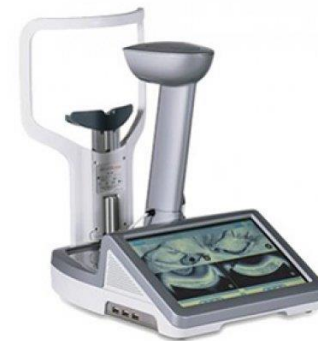
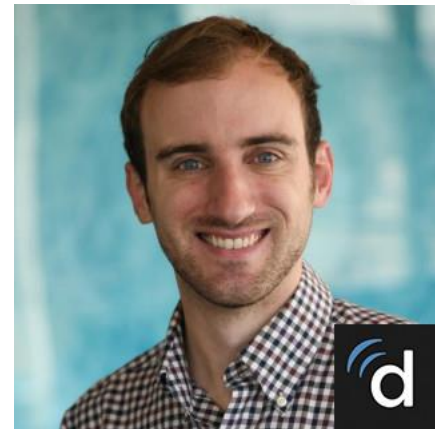
## Comparison of topography and refractive error in patients with and without DS

- **Children with DS diagnosed with keratoconus often have much lower amounts of astigmatism at the time of diagnosis.**
- This supports our hypothesis that screening tomography is a helpful adjunct in diagnosing keratoconus in patients with DS, as relying on high astigmatism as an indicator for disease may not be enough.
- Earlier detection and treatment is crucial to increasing favorable visual outcomes in children with DS and KCN.

# Ongoing Research

## Lipascan in patients with Down Syndrome

- Hypothesis is that decreased muscle tone -> weak blink -> blepharitis (eyelid inflammation) -> more rubbing -> more keratoconus
- Will image the eyelids of patients with and without DS to see if there's a difference in eyelid glands







# More to come on keratoconus

- **New grant!**



# More on recent advances in treatments....

- Crosslinking for Keratoconus
- Cataract surgery
- Strabismus (eye alignment)
- Blocked tear ducts

# Cataract Surgery

## Cataracts and Down Syndrome

- Lenses implant (IOL)
  - Monofocal
  - Multifocal
  - Toric



- NO studies guiding what type of lens to implant
- I would avoid multifocal lenses



# More on recent advances in treatments....

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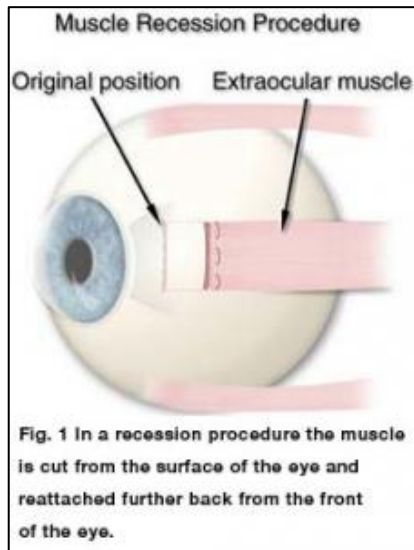


# Strabismus and Down Syndrome



video about strabismus and strabismus surgery

# Strabismus and Down Syndrome



- Prevalence is high!
- Surgical dose tables for DS
- Special kinds of strabismus in DS



video about strabismus and strabismus surgery



# More on recent advances in treatments....

- Crosslinking for Keratoconus
- Cataract surgery
- Strabismus (eye alignment)
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# Nasolacrimal Duct Obstruction (Blocked tear ducts)

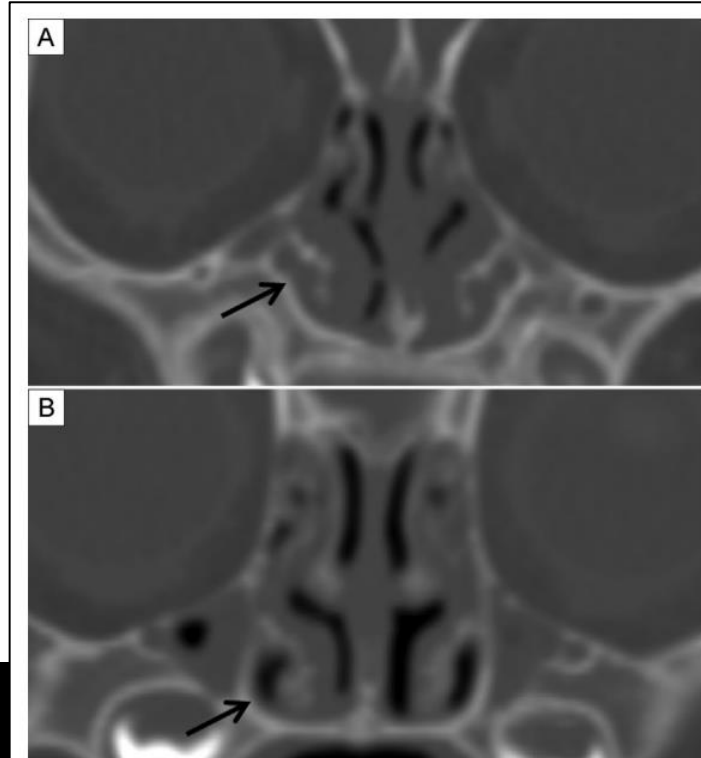
- High rates of surgical failure with probing and often need second surgery
- Can be very frustrating!





# Evaluation and treatment of failed nasolacrimal duct probing in Down syndrome

Francine Baran, MD,<sup>a,b</sup> John P. Kelly, PhD,<sup>a,b</sup> Laura S. Finn, MD,<sup>c</sup> Scott Manning, MD,<sup>d</sup>  
Erin Herlihy, MD,<sup>a,b</sup> and Avery H. Weiss, MD<sup>a,b</sup>



# Outline

- Who should examine me or my family member?
- How eye exams should be performed on patients with Down Syndrome
- Recent advances in treatments for eye disease in people who have Down Syndrome
- Recommended timelines for eye exams

# Recommended timelines for eye exams

- Children:
  - At 6 months of age.
  - Yearly until age of 8
  - After age of 8, could go to every other year if very stable



# Recommended timelines for eye exams

- Teens/Adults:
  - No formal recommendations exist!
  - Yearly in setting of known eye problems (cataract, etc)
  - Stable for year? Optometry yearly or every other year is appropriate
  - At minimum, would check in with eye MD every 5 years
  - Topography – ideally every 2 years between ages 13 and 30

# Our Team

- Michael Puente, MD
- Ronnie Wise, MD
- Casey Smith, MD
- Rich Davidson, MD
- Mike Taravella, MD
- Jennifer Jung, MD
- Francis Hickey, MD
- Lauren Imbornoni, MD
- Lauren Mehner, MD
- Eimi Rodriguez-Cruz
- Naresh Mandava, MD

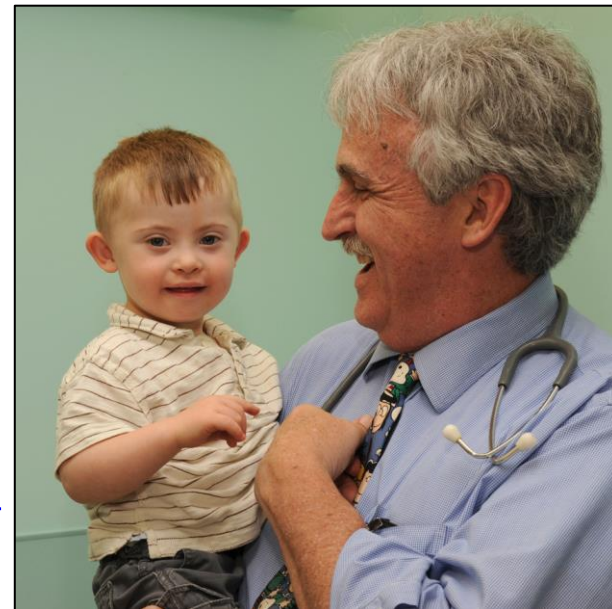


- **Our patients and their families**
- **Global Down Syndrome Foundation**
- **Sie Center for Down Syndrome**



# Links / resources

- [www.aapos.org](http://www.aapos.org)
- <https://www.childrenscolorado.org/doctors-and-departments/departments/down-syndrome/>
- <https://medschool.cuanschutz.edu/linda-crnice-institute>
- [https://eyewiki.aao.org/Trisomy\\_21/Down\\_Syndrome](https://eyewiki.aao.org/Trisomy_21/Down_Syndrome)
- <https://www.livingwithkeratoconus.com/>
- <https://publications.aap.org/pediatrics/article/128/2/393/30609/Health-Supervision-for-Children-With-Down-Syndrome>
- <https://www.uhealth.org/locations/uhealth-eye-center-anschutz-medical-campus/>
- <https://www.denverhealth.org/services/denver-health-and-global-down-syndrome-foundation-adult-down-syndrome-clinic>
- <https://www.globaldownsyndrome.org/>
- <https://www.childrenscolorado.org/doctors-and-departments/departments/eye/>



# Questions?

