Eye Research & Eye Care for Individuals with Down Syndrome Across the Lifespan – Part 1

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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

Outline – Part 1

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

Outline – Part 1

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

Visual Development – General Population

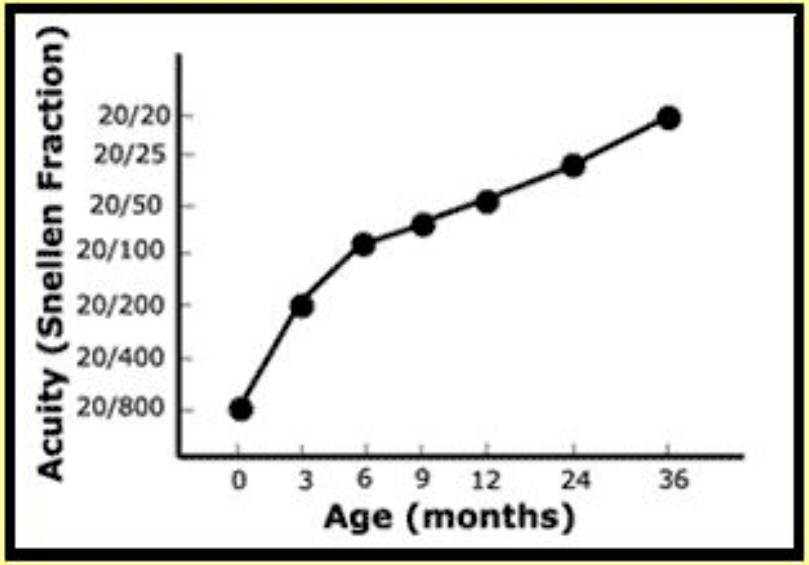
Forced-choice preferential looking

- 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 Milestones

Newborn – blinks to light

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

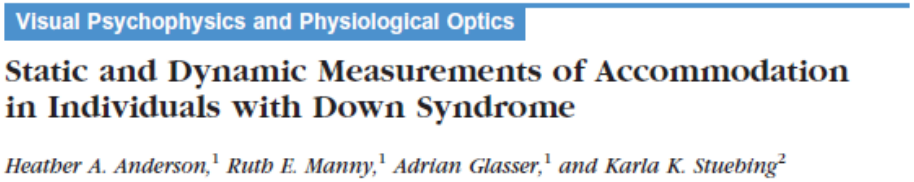
6 months – able to reach for objects, color vision develops

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 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,[1] Ruth E. Manny,[1] Adrian Glasser,[1] and Karla K. Stuebing[2]

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

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)

J Appl Res Intellect Disabil. Author manuscript; available in PMC 2015 May 1.
Published in final edited form as:
J Appl Res Intellect Disabil. 2014 May; 27(3): 247–263.
Published online 2013 Jun 19. doi: 10.1111/jar.12062
PMCID: PMC3841243
NIHMSID: NIHMS485753
PMID: 23784802

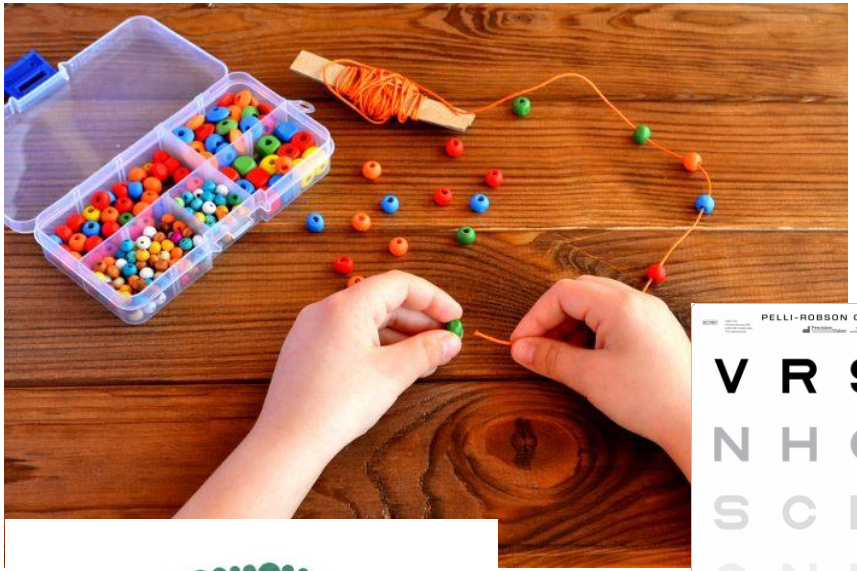
Vision Deficits in Adults with Down Syndrome

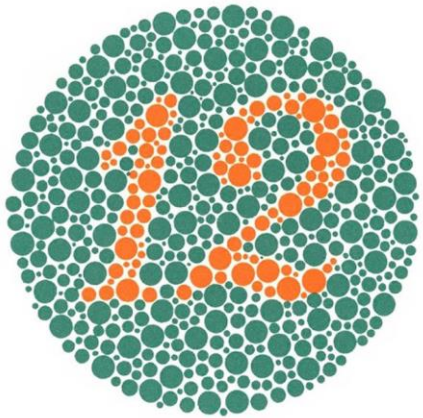
Sharon J. Krinsky-McHale,[1] Wayne Silverman,[2] James Gordon,[3,4] Darlynne A. Devenny,[1] Nancy Oley,[5] and Israel Abramov[4,6]

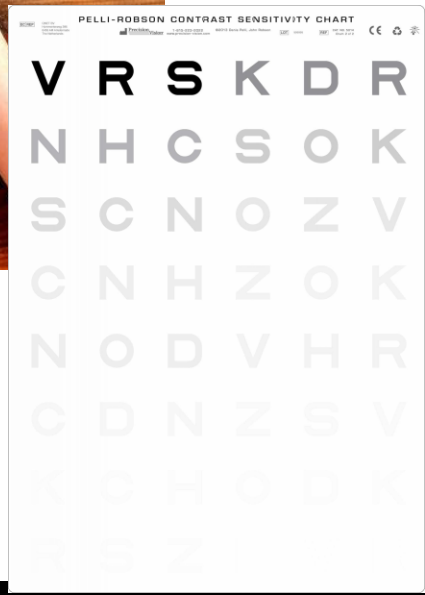
Beyond Clarity

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

"Lowenfeld Losses"

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

nature › scientific reports › articles › article

Article | Open Access | Published: 04 June 2020

Low vision and the risk of dementia: a nationwide population-based cohort study

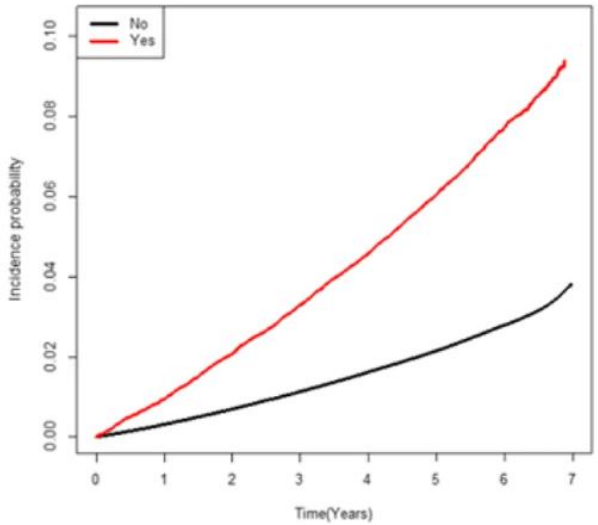
Ji-Sun Paik, Minji Ha, Youn Hea Jung, Gee-Hyun Kim, Kyung-Do Han, Hyun-Seung Kim, Dong Hui Lim & Kyung-Sun Na

Scientific Reports 10, Article number: 9109 (2020) | Cite this article

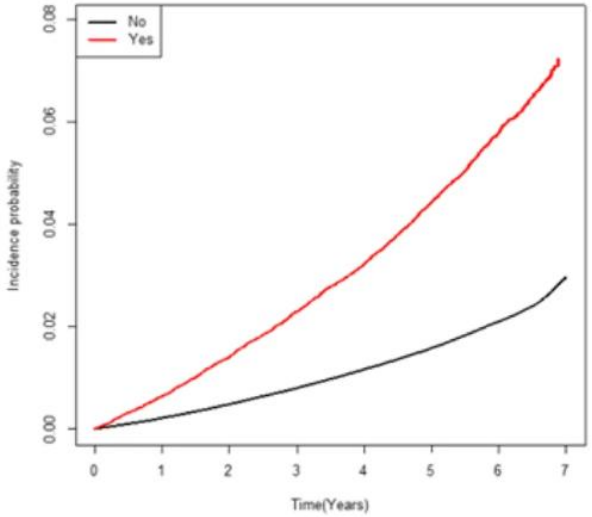
Risk of dementia in Koreans over 40 years old

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

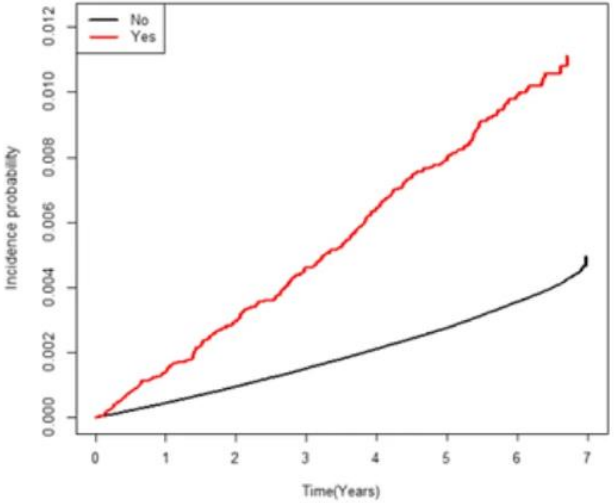
Dementia



Alzheimer's



Vascular Dementia



Vision and dementia

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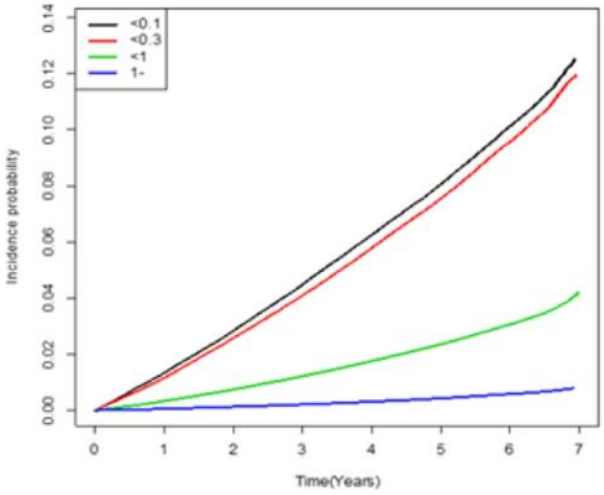
Risk of dementia in Koreans over 40 years old

BLUE LINE = normal vision
GREEN LINE = mild visual impairment
RED LINE = moderate visual impairment
BLACK LINE = severe visual impairment

Dementia



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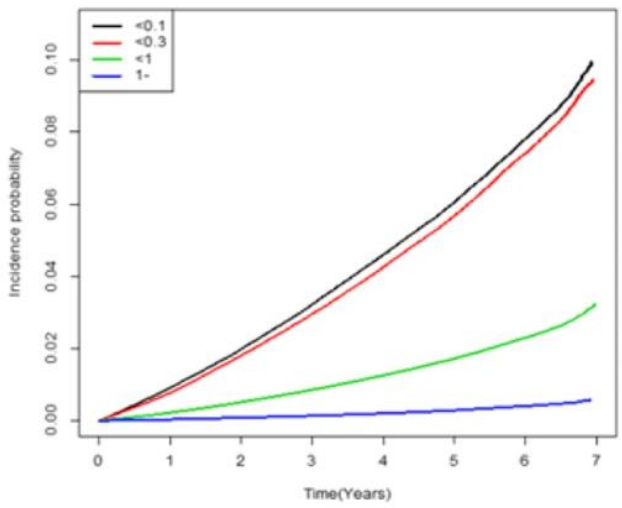
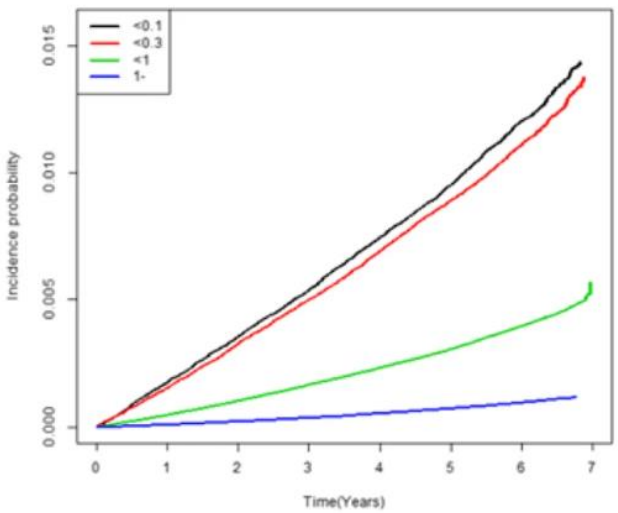


Vascular Dementia



Vision and dementia

- Cataract surgery has been shown to reduce the risk of dementia and cognitive impairment
- Vision problems may exacerbate cognitive decline
- Treating vision problems in adults may help preserve cognitive function

european journal of neurology
the official journal of the european academy of neurology

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

ELSEVIER

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 [a, b], Takamichi Kabata [b], Tetsuro Oshika [a]

Outline – Part 1

- 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

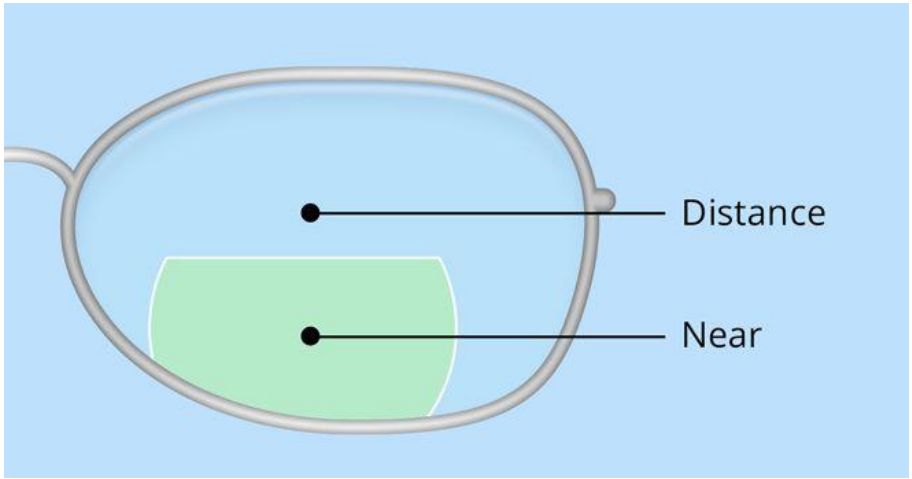
- Far-sightedness
- Near-sightedness
- Astigmatism

Hypoaccommodation

- “Accommodation” refers to the eye’s ability to focus on objects up close

- Up to 100% of people with Down’s syndrome have impaired accommodation

- Bifocal glasses have a stronger prescription at the bottom to help with near vision

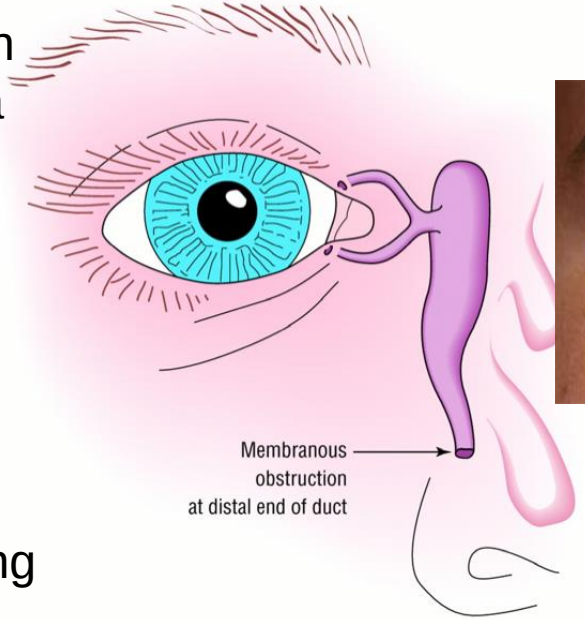


Strabismus

- “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

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

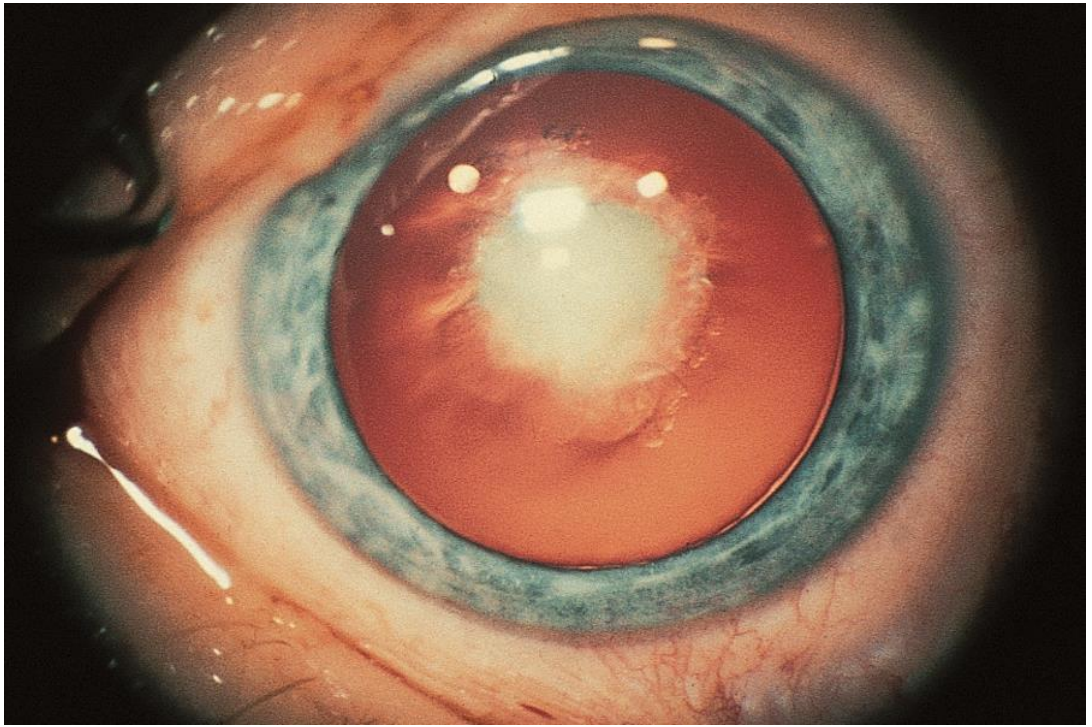


Nystagmus

- “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

Congenital cataracts

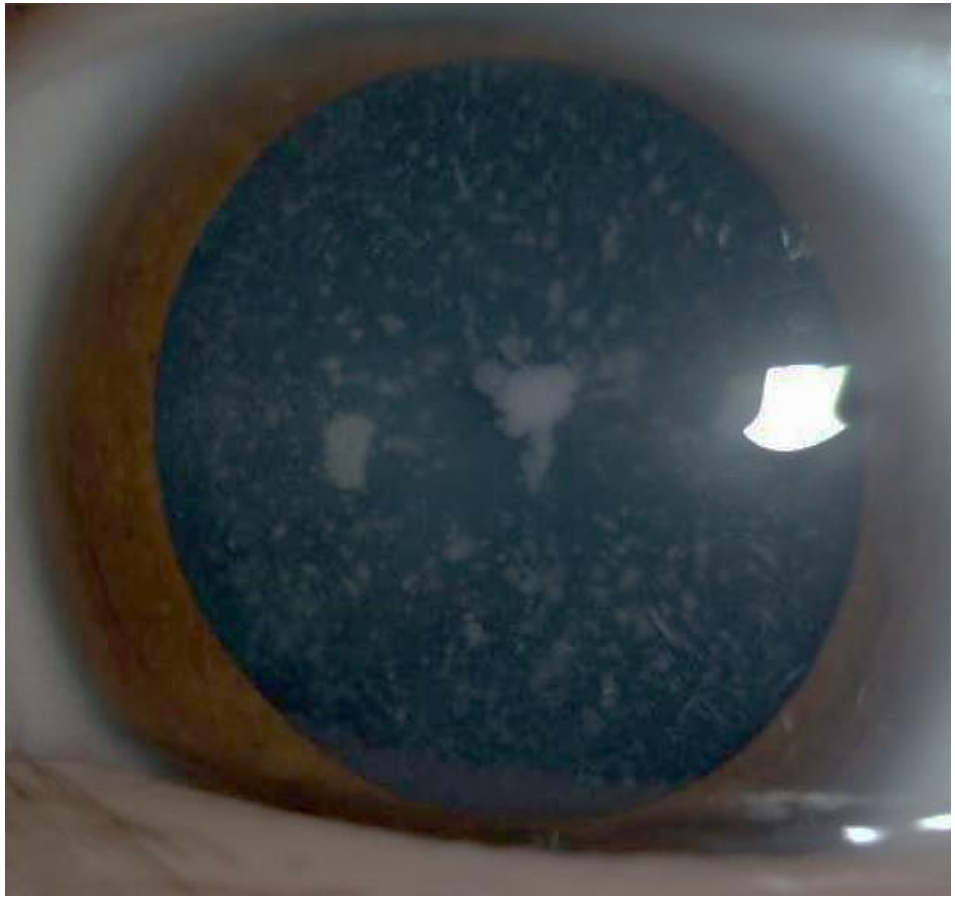
- “Cataract” refers to cloudiness of the lens inside the eye
- Babies with Down syndrome have about 300 times increased risk of being born with a cataract
- Treatment is surgery to remove the cloudiness



Other cataracts

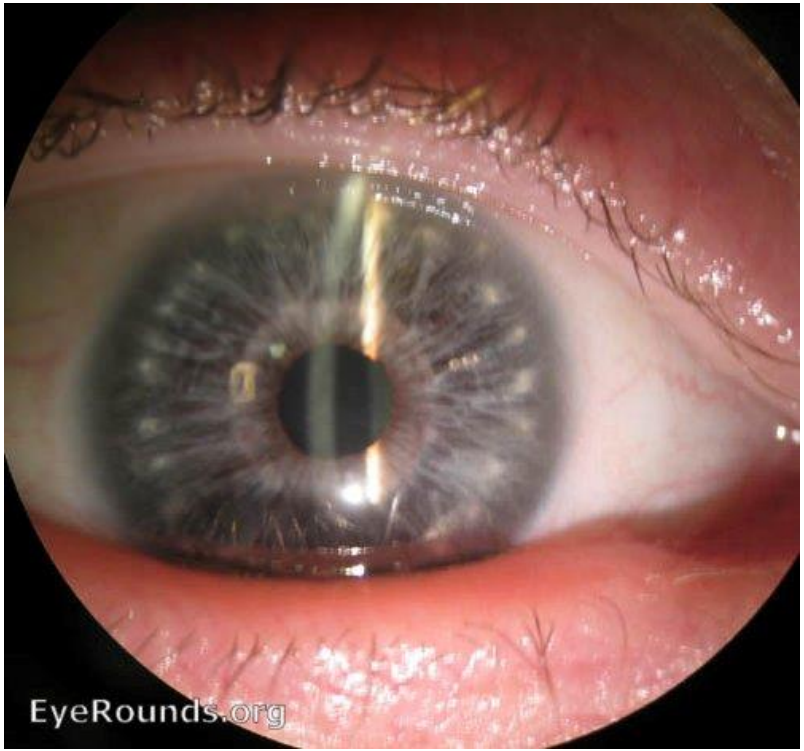
- People with Down syndrome develop cataracts at an earlier age than the general population, often needing cataract surgery in their 40's or 50's

- 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



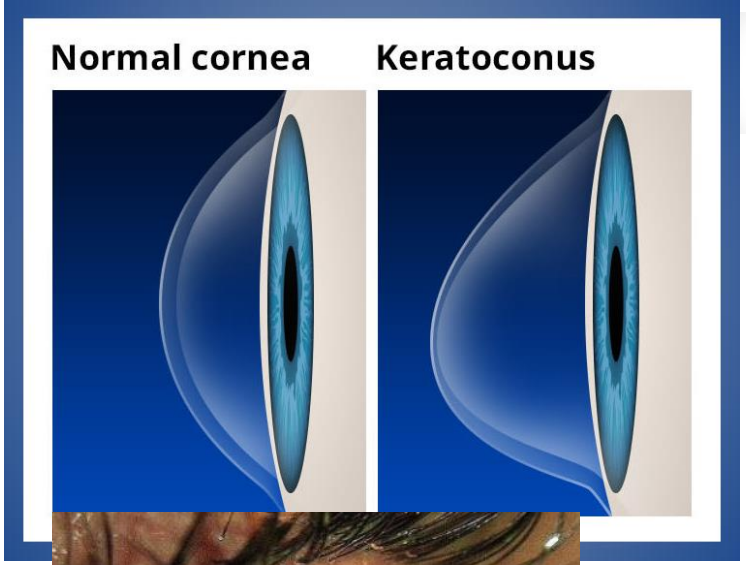
Keratoconus

- 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

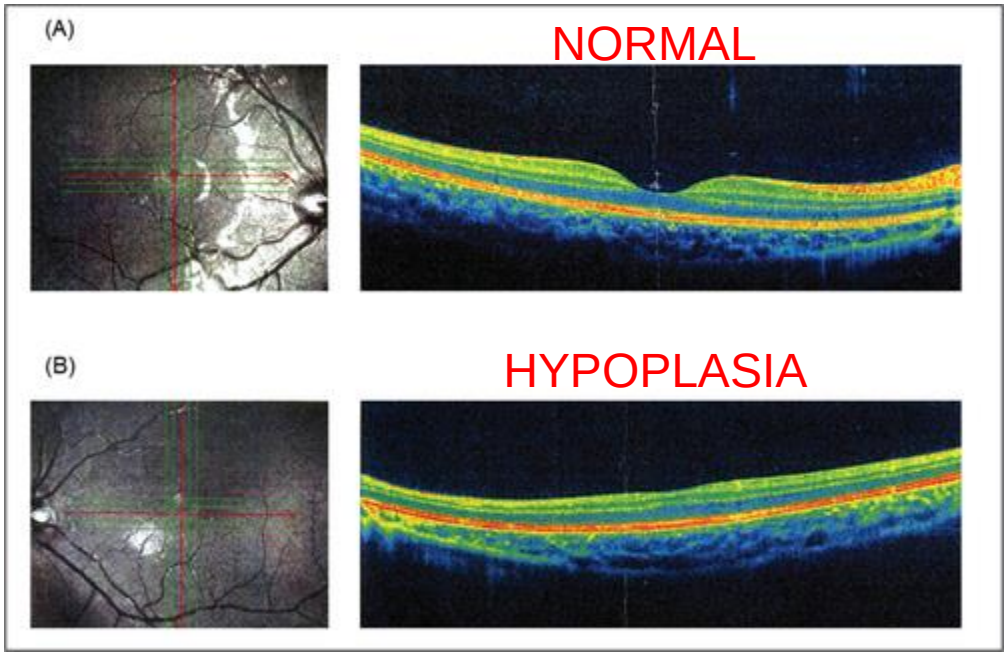
- Causes progressively blurrier vision

- Up to 32% of people with Down syndrome develop keratoconus



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

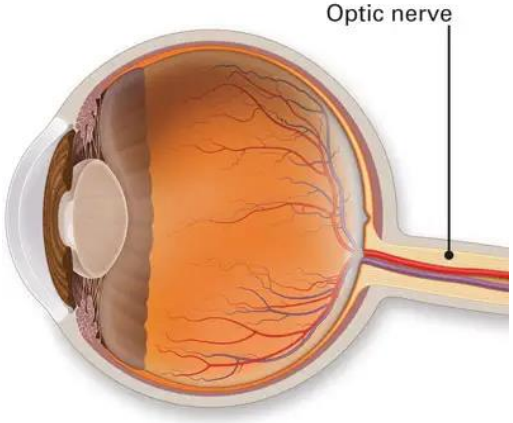


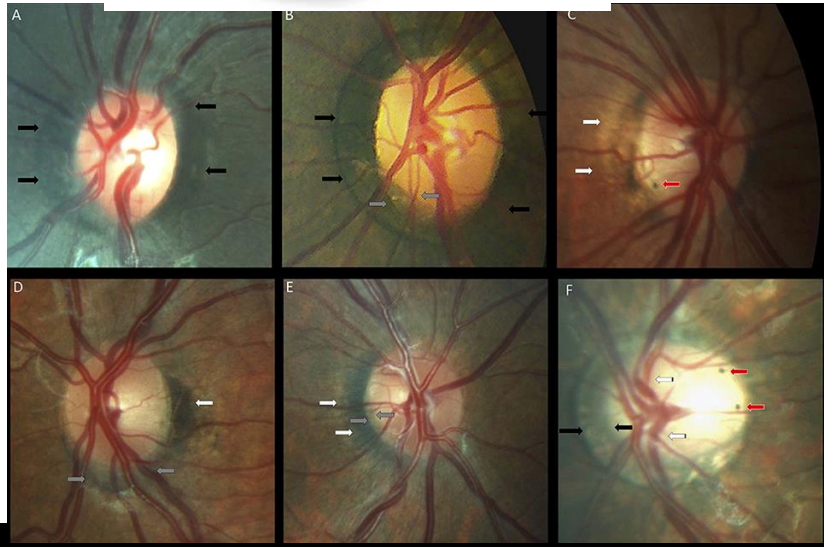
Optic nerve anomalies

- 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

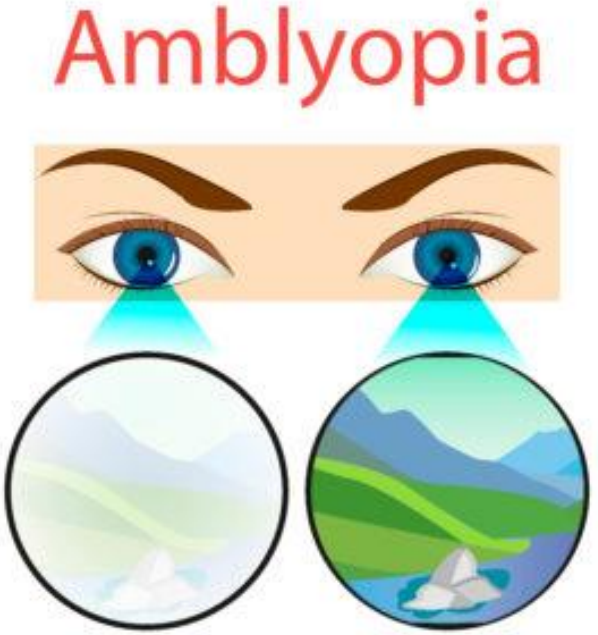
- People with Down syndrome commonly have abnormal optic nerves





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



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

Outline – Part 1

- 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?

Eye Research & Eye Care for Individuals with Down Syndrome Across the Lifespan – Part 2

Emily McCourt, MD

The Ponzio Family Chair for Pediatric Ophthalmology

Chief of Pediatric Ophthalmology, Children's Hospital Colorado
Associate Professor University of Colorado

Disclosures

- No financial disclosures

- **Global Down Syndrome Foundation** supported the purchase of the Pentacam topographer at our institution

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

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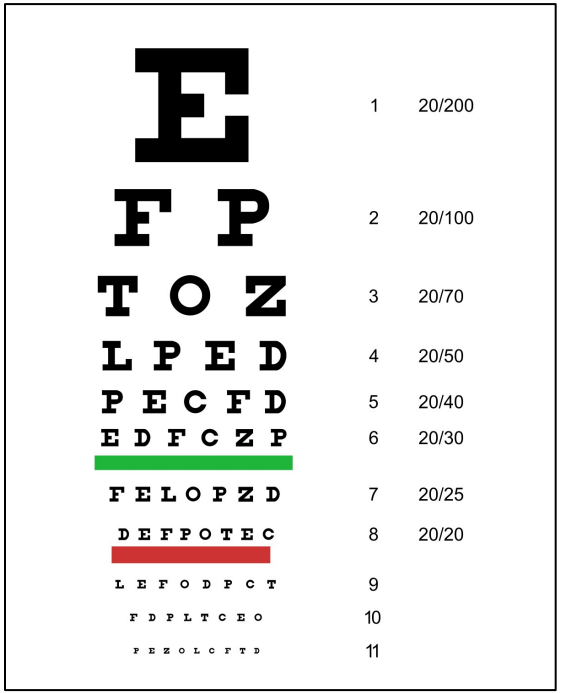
What should a comprehensive eye exam look like in a person with Down Syndrome?

- 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?

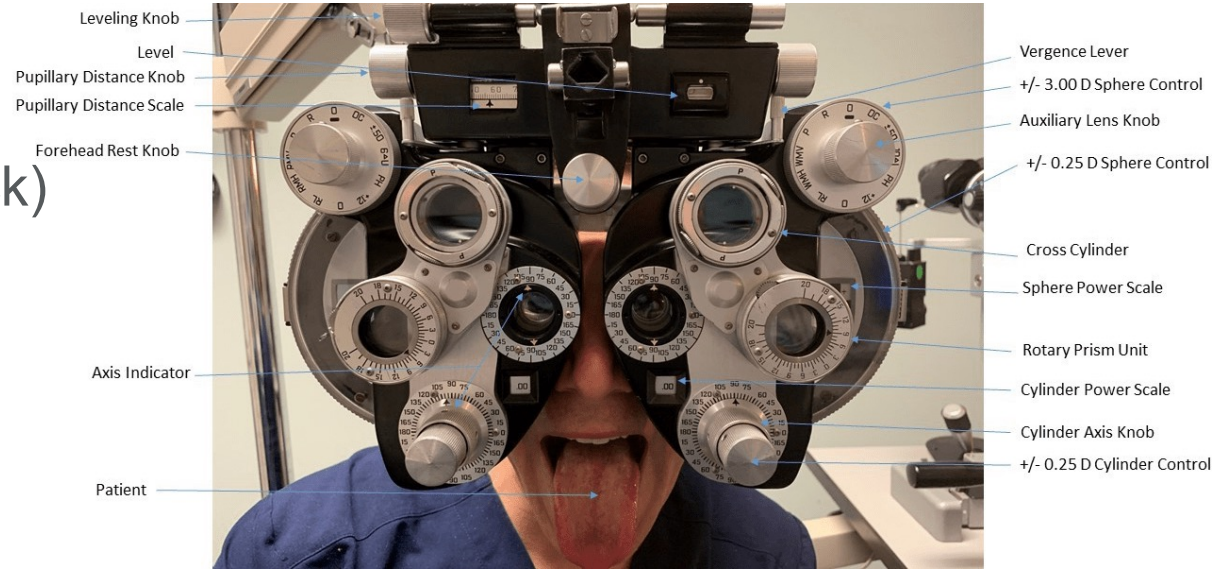
- Dilation +/- photos
- Should I pay extra for photos?

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

Refraction (glasses check)

PHOROPTER

(lens 1 or lens 2)



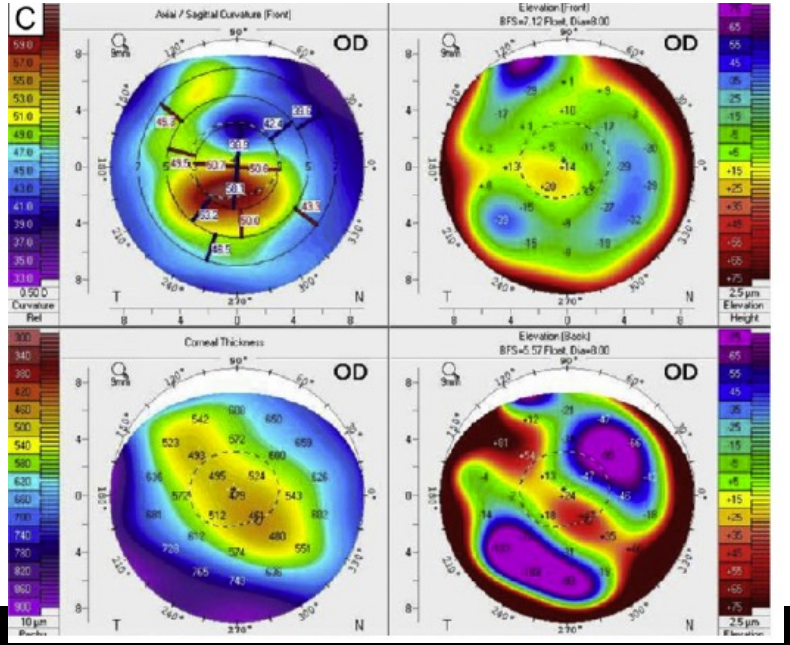
What should a comprehensive eye exam look like in a person with Down Syndrome?

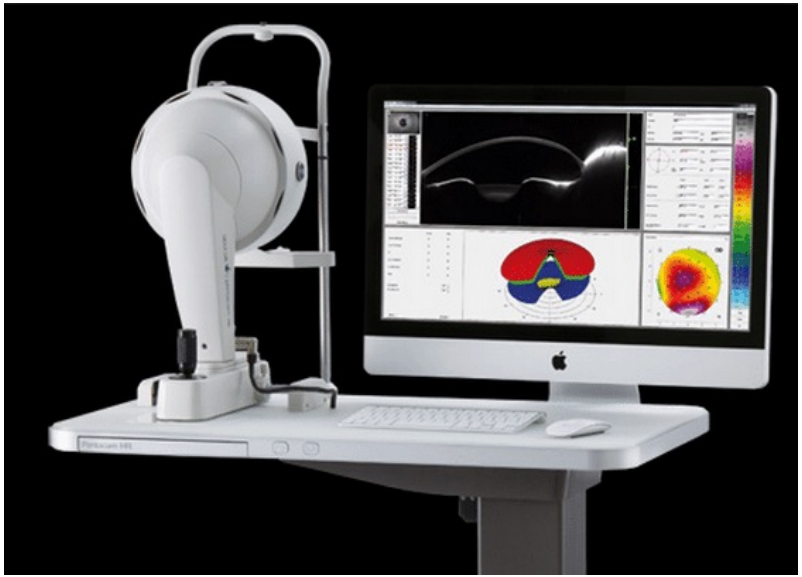
Refraction (glasses check)

RETINOSCOPY

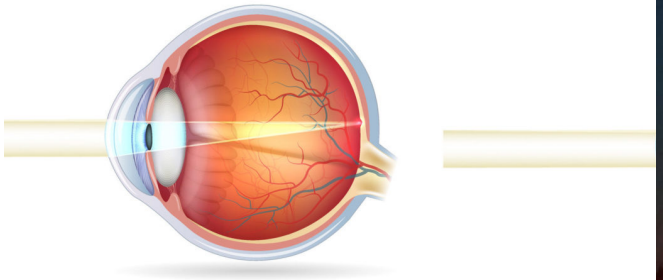
What should a comprehensive eye exam look like in a person with Down Syndrome?

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://jonaspauleyewear.com/

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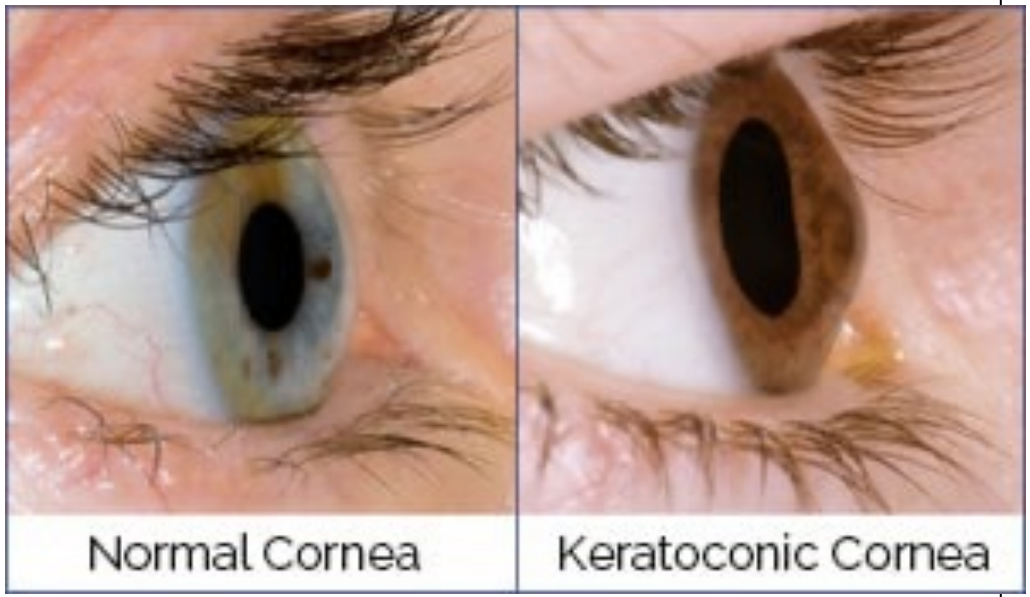
Recent advances in treatments for eye disease in people who have Down Syndrome

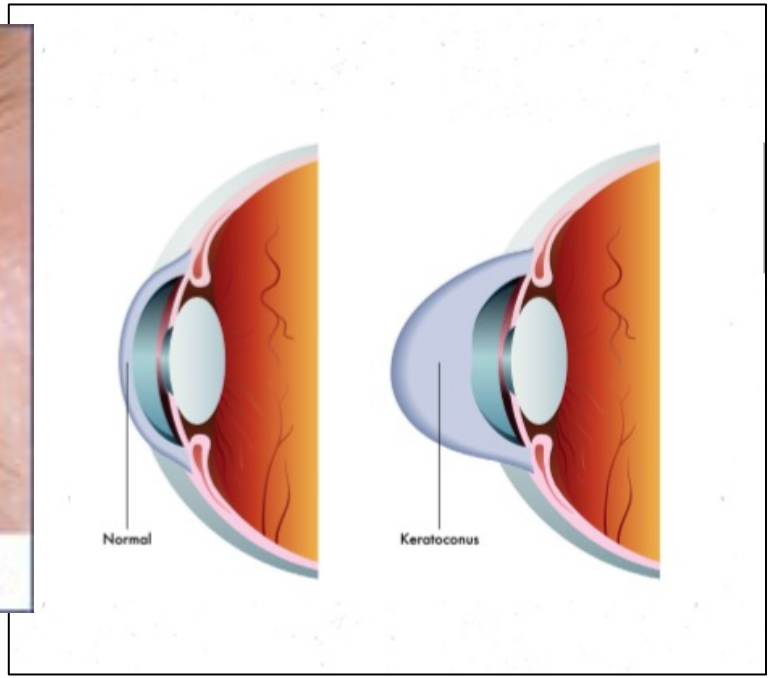
- Crosslinking for Keratoconus
- Cataract surgery
- Strabismus (eye alignment)
- Blocked tear ducts

A patient story

Lauren

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 – very common in patients with DS

- Numbers are variable:
  - Patients with Down syndrome sometimes are hard to examine
  - Variable definition of keratoconus

JAMA Ophthalmology | Original Investigation

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-

JAMA Ophthalmology September 2018 Volume 136, Number 9

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

Lauren M. Imbornoni, MD,[a] Ronald E. Wise, MD,[a] Michael J. Taravella, MD,[a] Francis Hickey, MD,[b] and Emily A. McCourt, MD[a]

- 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

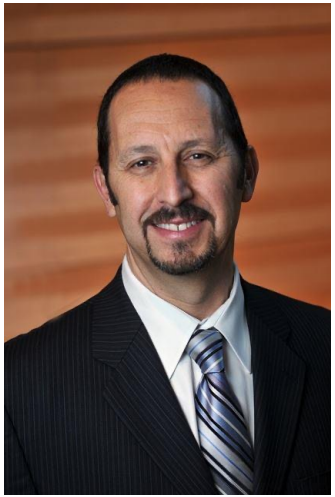
Keratoconus and corneal morphology in patients with Down syndrome at a pediatric hospital

Lauren M. Imbornoni, MD,[a] Ronald E. Wise, MD,[a] Michael J. Taravella, MD,[a] Francis Hickey, MD,[b] and Emily A. McCourt, MD[a]

- We started to look at our patients
- 10 month period (August 2018 to May 2019)

- Pentacam

  Generous gift from Global Down Syndrome Foundation



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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

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Conclusion

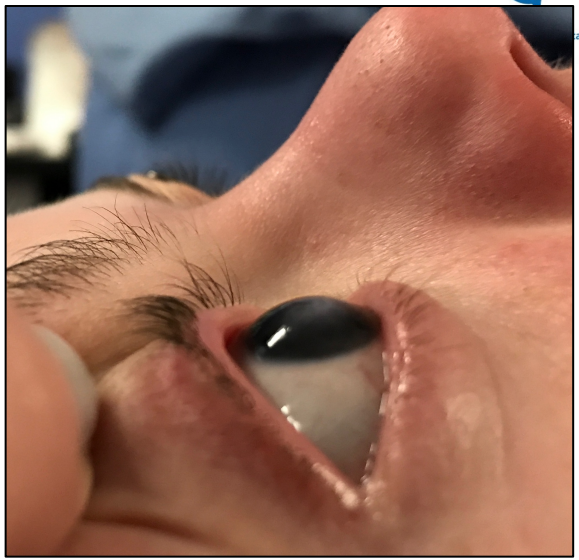
Keratoconus is often a much more aggressive in pediatric patients and needs to be treated.

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

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

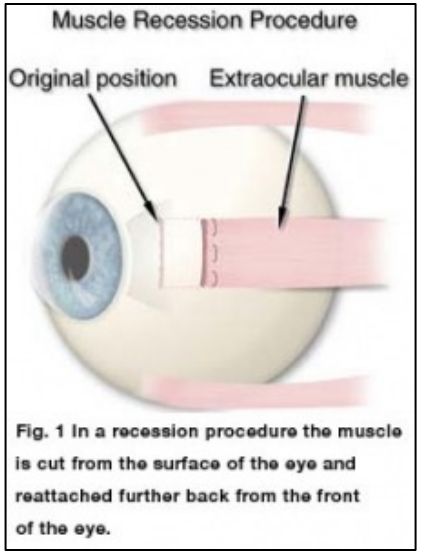

Fig. 1 In a recession procedure the muscle is cut from the surface of the eye and reattached further back from the front of the eye.

- 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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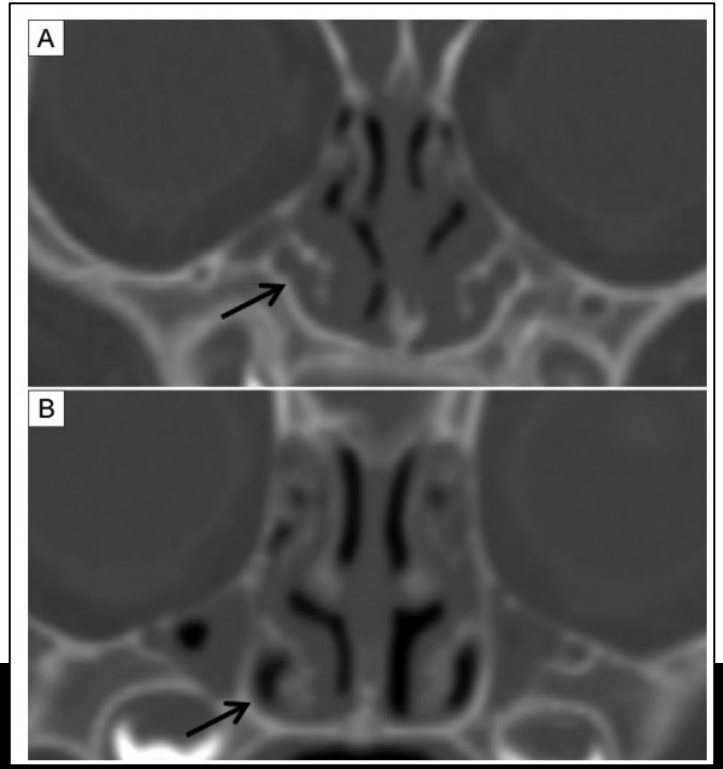
Nasolacrimal Duct Obstruction (Blocked tear ducts)

- 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,[a,b] John P. Kelly, PhD,[a,b] Laura S. Finn, MD,[c] Scott Manning, MD,[d] Erin Herlihy, MD,[a,b] and Avery H. Weiss, MD[a,b]



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

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

Links / resources

- www.aapos.org
- https://www.childrenscolorado.org/doctors-and-departments/departments/down-syndrome/
- https://medschool.cuanschutz.edu/linda-crnic-institute
- 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.uchealth.org/locations/uchealth-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?