Disclosure statement:
Nothing to disclose
What is Down Syndrome?

- First described by Langdon Down in 1866
- Occurs in 1 in 792 live births
- Affected children have medical conditions, cognitive impairment and multiple malformations due to extra chromosomal material
- Diagnosis confirmed by karyotype
- Phenotype is variable, but most children exhibit many similar features
Genetic etiology = Trisomy 21

- Majority (95%) affected have a sporadic, NONFAMILIAL condition = 47 chromosomes with an extra chromosome 21
- ~4% have 46 chromosomes with extra chromosomal material as result of unbalanced translocation between chromosome 21 and either chromosome 13, 14, 15 (Robertsonian translocation)
  - ¾ translocations are de novo, but ¼ are familial
  - Recurrence risk for families is variable / based on many factors, so genetic counseling recommended

- Mosaicism = ~1% of cases involve a mosaic pattern of cells, where there is a mixture of 2 types of cells: some with 46 chromosomes, some with 47 (may be more mildly affected)
Growth failure
Mental retardation
Flat back of head
Abnormal ears
Many "loops" on finger tips
Palm crease
Special skin ridge patterns
Unilateral or bilateral absence of one rib
Intestinal blockage
Umbilical hernia
Abnormal pelvis
Diminished muscle tone

Broad flat face
Slanting eyes
Epicranthic eyelid
Short nose
Short and broad hands
Small and arched palate
Big, wrinkled tongue
Dental anomalies

Congenital heart disease
Enlarged colon
Big toes widely spaced
Common physical features

- Hypotonia
- Small, broad (brachycephalic) head
- Large protruding tongue
- Small nose with low, flat nasal bridge
- Upward (temporal) slanting palpebral fissures
- Small, poorly defined ears
- Short, thick neck
- Stubby hands with a single palmar crease
- Short, stubby feet with wide gap b/t 1st and 2nd toes
Clinical findings

- **Cognitive impairment** is variable – mild (IQ 50-70) to severe (IQ 20-35)
- **Developmental Delay**
- **Social skills** can be improved through early intervention techniques
  - Often function more effectively in social situations than what would be predicted by cognitive level
- **Significant risk for**...
  - Hearing loss (75%)
  - Otitis media (50-70%)
  - Obstructive sleep apnea (50-79%)
  - Congenital heart defects (50%)
  - Delayed dental eruption and hypodontia (23%)
Common eye findings

- Why are WE an important part of the care team?

  - 60% of children with Down Syndrome have general VISION PROBLEMS
Common eye findings

- 50% are found to have significant **REFRACTIVE ERROR**
- 15% are found to have **CATARACTS**
- Up to 47% have **STRABISMUS**
  - also **PSEUDOSTRABISMUS / EPICANTHUS**
- 33% have **NYSTAGMUS**
- Up to 75% have **NASOLACRIMAL DUCT OBSTRUCTION**
- 35-78% have **BRUSHFIELD SPOTS**
- Also commonly seen...
  - Amblyopia
  - Keratoconus
  - Ptosis and corneal ectasia to a lesser extent
Reduced visual acuity

- Seen in many children with Down Syndrome (in absence of significant refractive error or ocular pathology)
- Studies have shown reduced low contrast and Vernier acuity
- Etiology of this poor visual performance is not fully understood

**TAKE HOME MESSAGE** = it is not necessarily “abnormal” for your patient to have 20/30 to 20/60 best-corrected acuities
High refractive error

- Most commonly high astigmatism and hyperopia

- Prescribe to prevent amblyopia in preschool children
  - What is enough to cause amblyopia?
    - Greater than 1.5 D anisometropia of hyperopia or astigmatism
    - Greater than 2.5 D cylinder both eyes
    - Greater than 4.50 D hyperopia in both eyes

- May see more behavioral challenges to wear glasses
  - So many kids will refuse to wear glasses at home!
High refractive error

- Need frames to fit flat nasal bridge
- Regular nose pads often don’t fit well
  - **Specs 4 Us**
    - SPECS = Superior Precision Eyewear for Children who are Special
    - Company founded by mother of child with Down Syndrome
      - Maria Dellapina, one of 2015 Toyota Mothers of Invention
    - Frames have lower-set bridge and nosepads
Accommodative Insufficiency

- Seen often in children with Down Syndrome – related to generalized hypotonia
- Could be an additional barrier to literacy and early learning in these children
- Many school age children, especially those who are avidly reading, may benefit from a BIFOCAL or reading glasses – don’t hesitate to prescribe!
- Attempt simple accommodative tests to look for adequate, age-appropriate accommodation
Accommodative Insufficiency

- **Monocular Estimation Method (MEM near point retinoscopy)**
  - Most OBJECTIVE method to evaluate accommodative response
  - Attach MEM card to retinoscope that is appropriate for age and reading level
  - Use normal room illumination
  - As patient reads the words or looks at pictures on the card, perform retinoscopy along horizontal axis
  - QUICKLY place lens before the eye to evaluate
  - Norm = plano to +0.75
    - i.e. MEM of +1.25 would represent underaccommodation
Epicanthus

- Pseudostrabismus / pseudoesotropia
- Commonly misdiagnosed as true strabismus by pediatricians or family physicians
- Note corneal reflex centered in each eye (no true strabismus)
Nasolacrimal duct obstruction

- Chronic, unilateral or bilateral tearing caused by...
  - *Redundant skin tissue* more often in this population
  - To a lesser extent, the typical causes: persistence of an embryonic membrane at the valve of Hasner or stenosis of partial or entire nasolacrimal duct
- Signs include increased tear prism, overflow of tears down cheek, red/swollen eyelids with yellow discharge when bacteria not properly flushed down the duct
- May continue past the age of 12 months (when 90% cases will spontaneously resolve) = indication for surgery
Nasolacrimal duct obstruction

**Treatment**
- Higher failure rate even with treatment
  - Mainly when redundant skin is the cause
- Common to need multiple procedures
- Referral to pediatric ophthalmologist or oculoplastic specialist

**Techniques**
- NLD probing
- Silicone (Crawford) tube placement
- Balloon catheter dilation (dacrocystoplasty)
  - Indicated when probing and irrigation has failed
  - Advantage: no silicone tube is left in the lacrimal system, eliminating any concern about the child removing it or dislocation
Blepharitis

- The prevalence of blepharitis in children with Down syndrome ranges from 3% to 34.5%

- The high rate of blepharitis has been speculated to be due to impaired immune system or due to skin abnormalities in Down syndrome individuals

- Treatment important to prevent any cause of irritation that could lead to eye rubbing
Brushfield spots

- Multiple, round, focal areas on anterior surface of iris that appear beige or light brown / gray
- Represent areas of iris stromal hyperplasia surrounded by relative hypoplasia
- More commonly seen in Down Syndrome kids with...
  - Lightly pigmented (blue, green, hazel) irises
  - European descent
- *NOT* pathognomonic – can be seen in normal patients as well...
Cataracts

- Higher risk
  - Congenital cataracts
  - **Cerulean cataracts**
    - “Blue” dot opacities in anterior and posterior capsule
    - Typically bilateral, but can be asymmetric
    - Often do not affect overall visual acuity, but can
    - Can be stable or progress very slowly over time
Nystagmus

- Nystagmus is present in 3 to 33% of children with Down Syndrome
- Typically seen form is *rapid horizontal nystagmus*
  - To a lesser extent see latent nystagmus
- Decreased visual acuity can certainly be associated with any type of congenital nystagmus
Strabismus

- Exotropia
- Vertical strabismus less common
- **Esotropia** - most common!!
  - Congenital or infantile ET
  - Accommodative ET
  - Do NOT confuse with spasm of over-accommodation
    - Common phenomena in Down Syndrome
    - Mainly seen when focusing on near target
    - Due to poor accommodative skills (as is often the case with hypotonic kids)
Strabismus

TREATMENT

- Glasses considerations
  - Always full plus with presence of esotropia, especially accommodative esotropia
  - Since poor accommodation skills seen more frequently, would consider Rx low plus more often

- Strabismus surgery
  - Required for correction of infantile ET
Accommodative esotropia

- Presents between 1½ to 3 years of age
- Caused by the over-convergence associated with increased accommodation
  - Uncorrected hyperopia causes accommodation!
- Associated with significant hyperopia
  - Above +2.00...typically +4-6 D
- Prescribing full refractive correction improves (partially accommodative) or totally corrects the ET
  - Cosmetic and functional cure!
Amblyopia

- **Refractive**
  - *Isometropic*
    - VERY common with high bilateral astigmatism or hyperopia
    - Treatment involves full-time wear of glasses
  - *Anisometropic*
    - Treatment – occlusion or optical penalization
    - May be more difficult due to behavioral challenges
- **Strabismic**
  - Commonly seen with unilateral esotropia
- **Important Message:**
  - AVOID ATROPINE PENALIZATION therapy due to so many children with cardiac abnormalities
Keratoconus

- Onset more commonly presents in early adulthood
- Increased rubbing of eyes in Down Syndrome puts these individuals at higher risk
- Will see more in eye on the same side as their dominant hand!
- Increased prevalence could be due to...
  - Cornea is steeper with higher rates of astigmatism
  - ? Genetic link between genes on chromosome 21 and keratoconus
Keratoconus

- **Treatment**
  - More challenging
  - Poor candidate for contact lenses
  - Poor candidate for corneal transplant... **but... new transplant technique**
    - Deep lamellar keratoplasty using femtosecond laser, often referred to as “Top Hat Keratoaplasty”
    - Precisely shaped and customized incisions fit together like Legos and heal faster and stronger
    - Less concern about eye rubbing behavior
Increased macular thickness?

- Study published in *Graefe’s Archives for Clinical and Experimental Ophthalmology* July 2015
- Dr. Jingyn Wang, now research faculty at Salus
- Investigated macular structural characteristics in children with Down Syndrome compared to those in healthy children
- Also investigated the correlation between the reduced visual acuity seen in many Down Syndrome patients to increased macular thickness
Increased macular thickness

Conclusions

- On average, central subfield thickness (CST) in the Down Syndrome group was greater than that in the control group, suggesting abnormal macular development in Down Syndrome.

- Depth of foveal pit was shallower in the Down Syndrome group.

- Thicker macula was not found necessarily to correlate with poorer visual acuity.
Eye Exam Recommendations

- When should children with Down Syndrome have eye exams?
  - First exam at age 6 months
  - Annual exams up to age 5
    - evaluate for refractive error, strabismus, or other conditions which could result in amblyopia
  - Every 2 years thereafter
    - evaluate for the onset of new ophthalmic disorders
Eye exam recommendations

- Best techniques depend on cognitive developmental level, cooperation and co-morbidities...quite a range in Down Syndrome!
- Pediatric tests will often be ideal for also working with adults with Down Syndrome (or other developmental delays)
- As in any pediatric exam, you should have the proper “tools of the trade” at your reach...
Visual acuity

- Light detection
  - Wince to light

- Object detection
  - Fix, follow, maintain

- Preferential looking
  - Teller acuity
  - Lea Gratings
Visual acuity

- Recognition test
  - Lea Symbols
    - Matching cards and blocks
    - Near acuity card

- Snellen acuity
  - Try for older children/teens, especially for those with higher cognitive levels
Other helpful exam tools:

- Hand-held slit lamp
- Interesting fixation targets
- Pediatric color vision test
- Loose prisms
Resources for families

- National Association for Down Syndrome
  - www.nads.org
- Down Syndrome Diagnosis Network
  - www.dsdiagnosisnetwork.org
- National Down Syndrome Society:
  - www.ndss.org/Resources/Local-Support
- Global Down Syndrome Foundation:
  - www.globaldownsyndrome.org/about-down-syndrome/resources/local-organizations/
- American Academy of Pediatrics Healthcare Guidelines
- GiGi’s Playhouse – Down Syndrome Achievement Centers
  - www.gigisplayhouse.org
Advice from parents of a child with Down Syndrome...

- “Talk to my child as if he understands everything you say – just talk to him like you would anyone else.”

- “Don’t be afraid to use the term ‘down syndrome’ – there’s no shame or embarrassment in it.”

- “Be ready for hugs – many kids with down syndrome like to show physical affection.”
Questions?

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