

# Down Syndrome Update

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Sanford Children's

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## Down Syndrome

- Goals
  - Review most significant changes
  - Share experiences
  - Encourage following guidelines
  - Discuss local and rural resources

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## Coordinated Treatment Center

- Sanford Broadway Medical Center – downtown Fargo
- Multidisciplinary clinic
  - General pediatrician
  - RN
  - Social worker
  - Nutritionist
  - PT, OT, Speech
  - PM and R
  - Family Voices
- Coordination of Specific Care
  - Audiology, Vision, Sleep ,Labs
  - Cardiology, GI, Endocrine, Heme/Onc, etc

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## Down Syndrome Organizations

- Typical origins
  - Lack of knowledge
  - Insensitivity
  - Poorly researched – outcomes, therapies, interventions, screening
  - Poorly organized

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## Down Syndrome Organizations

- National Association of Down Syndrome
  - 1961, parents who wanted to care for child at home
- Global Down Syndrome Foundation 2009
  - Physician's first grandchild
  - "least funded genetic condition" - decreased 2000 by NIH
- GiGi's Playhouse
  - Early 2000's
  - Nearly 60 sites, 200 applications

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## 2022 Update

- Health Supervision For Children and Adolescents with Down Syndrome
  - Pediatrics Volume 149, number 5, May 2022
  - Last update 2011, recommendations "expire after 5 years unless reaffirmed"
  - No fundamental changes, but enough updates to rewrite and improve

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## Down Syndrome

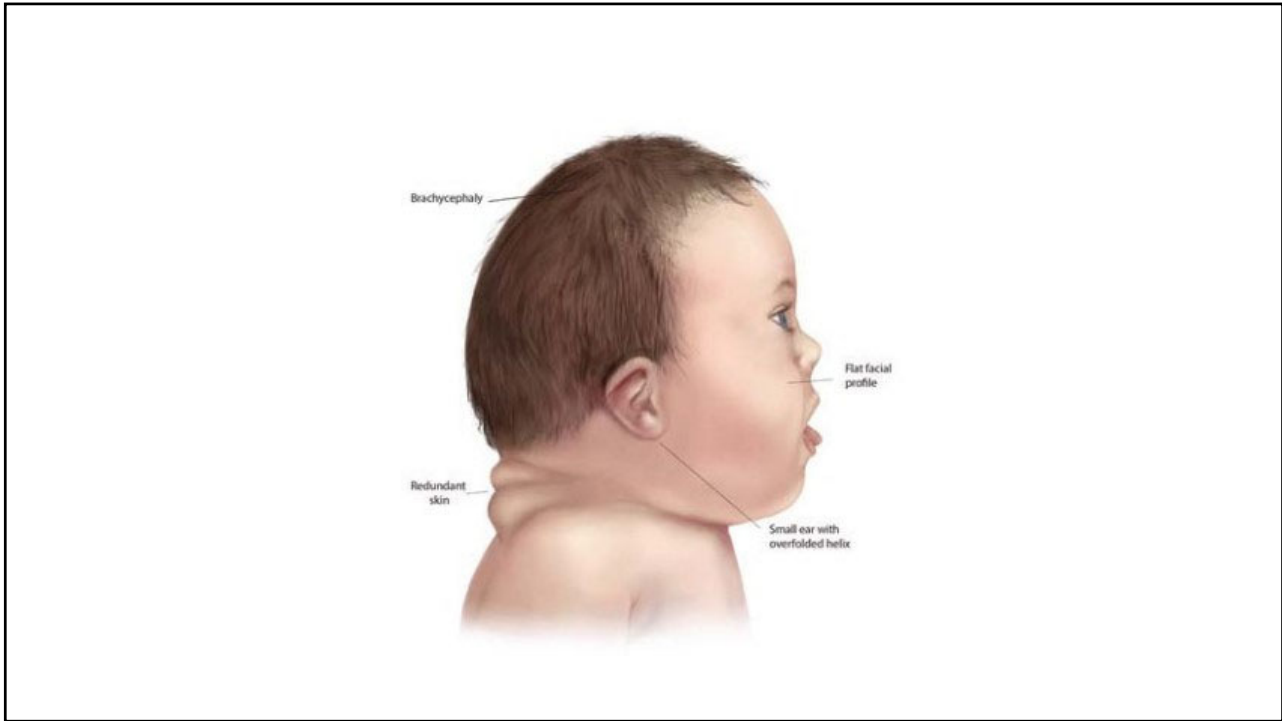
- Most common chromosomal anomaly
- Most common known cause of developmental delay
- 1 of 691 live births
- Likely 350,000 in the United States

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## Physical findings

- Hypotonia
- Small brachycephalic head
- Epicanthal folds
- Flat nasal bridge
- Upward-slanting palpebral fissures
- Brushfield spots
- Small mouth
- Small ears
- Excessive skin at the nape of the neck
- Single transverse palmar crease
- Short fifth finger with clinodactyly
- Wide spacing between the first and second toes often with a deep plantar groove

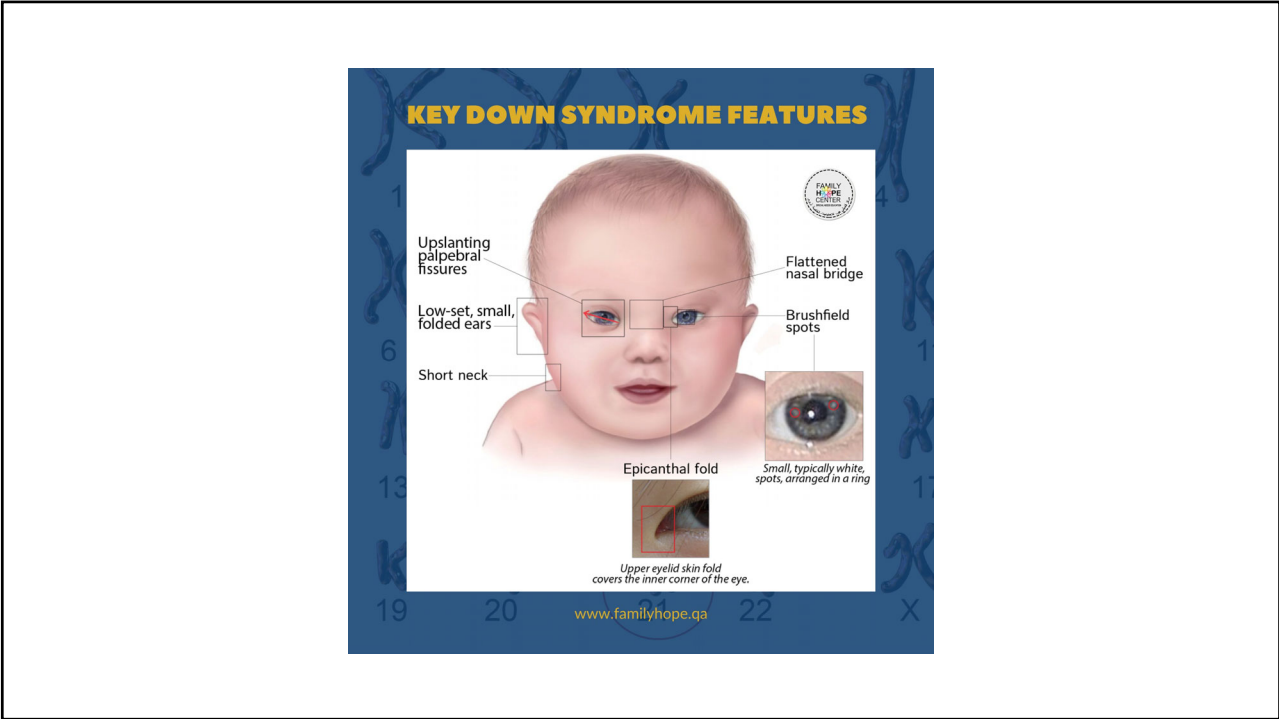
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## Genetics - "offer referral"

- cfDNA – cell free DNA
  - 99.7% detection rate, .04 false positive
- Confirmed by CV sampling or amnio prenatal
  - Because it only screens for 21, 13, 18, X and Y
- Fish postnatally is most efficient
- Still recommend formal Chromosomes for translocation

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## Strengths and positives

- First communication should be congratulations
- Lifespan increased from 30yrs in 1973 to 60 yrs in 2002
- Function more effectively in social situations
- Nearly 99% report being happy with their lives
- 79% of parents life outlook was more positive
- 88% of siblings felt they were better people

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## Genetics

- Subtypes
  - ~96% sporadic
  - ~4% translocation
    - 75% de novo, 25% parent
  - ~1% mosaic
- Recurrence – from 1-100%
  - Sporadic: 1% and increasing after 40 yrs
  - 14:21 : 2-15 % depending if maternal or paternal
  - 21:21 : 100% if parent carrier (1 in 14)

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## Eyes (60-80%)

- Photoscreening is foundation
  - Well child **OR** eye provider yearly before 5yo and every 2 yrs after 5yo
  - Can be used at any age or development
- Formal eye exam by ophthalmologist experienced with children with developmental delay
  - Rural difficulty
  - Local experience
  - Strictly only by 6 months and an “or” recommendation after that

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## Eyes

- Nystagmus up to 33%
- Glaucoma up to 7%
- Nasolacrimal duct obstruction up to 36%
- Cataracts 3%
- Strabismus 36%
- Refraction errors up to 80%
- Keratoconus up to 13% - after school age increase
  
- My opinion: See an eye doctor yearly

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## Ears – hearing problem 75%

- Little confusing and perhaps most difficult
  - Birth "objective testing"
    - OAE or BAER
    - Referral to ENT if failed
      - Unable to view TM - humility
    - Tympanometry if unable to view TM
  - Rescreen at 6 months
  - Attempt behavioral audiogram at 1 year
    - If unable BAER

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## Ears

- Behavioral audiogram and tympanometry every 6 months (6m-4yr)
  - BAER if fail
- Behavioral hearing tests annually:
  - 4 yrs – adult (No more standard tympanometry)
  - If fail then OAE or BAER
- Testing after treatment with BAER
- 50-70% OME in the first year
  - Same percent age 3-5 yrs

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## Thyroid

- 2-7% congenital
- 50% by late childhood
  - Many have mild TSH elevation with normal T4
  - Hyperthyroid in 3%
  - Antithyroid antibodies are common and increase risk of later hypothyroid
- Management should be discussed with endocrinology
  - Thyroid doesn't always behave as "expected"

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## Thyroid

- 6 and 12 months and then annually or sooner if symptoms
- Every 6 months if previous antibodies detected
- If mild TSH abnormal would check antithyroid antibodies
- Risk increases with age

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## Atlanto-axial instability (10%)

- No study links radiographic atlanto-axial instability with injury
- 41 "well documented AAI" ever
- 50,000 special olympics participants with no spinal cord injuries
  - Screen specific sports
  - Gymnastics, pentathlon, butterfly stroke, diving, diving start, high jump, soccer, equestrian, squat lift, judo, snowboarding, alpine skiing

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## Cervical Spine

- Education more than screening (every other year after 1)
  - Neutral position
    - Anesthesia, surgery, radiography
  - Signs of myelopathy
    - Asymmetry, weakness, increased DTR - pursue
    - Change in gait, use of arms or hands, bowel or bladder, neck pain or stiffness, head tilt, torticollis, change in general function, weakness - discuss
- Radiographs after 3 can be done but....
  - Do not predict or reassure of risk, not recommended screening

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## Cervical spine

- Symptomatic
  - Xray in neutral position
  - If neutral normal - flexion and extension "in collaboration with subspecialist"
  - Pediatric neurosurgeon or orthopedic surgeon with expertise
  - ASAP referral if any abnormality
- No trampolines for anybody and particularly with Down Syndrome
  - Football, soccer, gymnastics risk for spinal cord injury

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## Heart 50%

- Echo at birth regardless of prenatal echo
  - Referral if abnormal
- If left to right shunt monitor for heart failure
  - Tachypnea, feeding struggles, poor weight gain
- Pulmonary hypertension risk without cardiac defect
- Acquired mitral and aortic valvular disease – risk begins in teens
  - Fatigue, SOB, exertional dyspnea, new murmur

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## Gastrointestinal

- Duodenal or anal atresia (12%), Hirschsprungs, GERD
- Celiac (5%)
  - Diarrhea, constipation, slow growth, FTT, anemia, abdomen pain or bloating, difficult behavior or development concerns
  - TTG IgA and total IgA
  - Don't start diet prior to GI referral
  - Retesting at a later time may still be necessary if negative

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## Hematologic

- CBC by three days of age
  - Transient abnormal myelopoiesis (TAM) (9%)
    - Many immature blood cells, particularly megakaryocytes
    - "Blasts"
  - Polycythemia (33%)
  - Leukocytosis

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## Hematologic

- TAM (>10% blasts)
  - Usually regresses by 3 months
  - May require chemo
  - Increases risk of death up to 20% before 6 mo
  - Increase AML (30%) by age 4yrs

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## Hematologic

- Neutrophilia (80%), Thrombocytopenia (60%), thrombocytosis
  - Generally resolve after 1 week, refer if persist
  - Macrocytosis often lifelong
- Leukemia more common (1%)
  - Typically better outcomes

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## Anemia (1.2%)/iron deficiency(6.7%)

- CBC yearly **and**
  - Ferritin and CRP (preferred) **or**
  - Serum iron and TIBC
- Iron deficiency – subtle, perhaps justifies screening
  - Macrocytosis can confuse diagnosis
  - Precedes anemia
  - Neurological
  - Sleep

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## Sleep – OSA up to 79%

- At birth begin discussing OSA
  - Heavy breathing, snoring, uncommon sleep positions, frequent awakenings, daytime sleepiness, apnea, behavior problems
  - Refer if concerns
- Sleep study between 3 and 4 years
  - Poor correlation between parent report and sleep study results

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## Sleep

- Repeat sleep study after T and A
  - Minimally symptomatic sleep apnea can persist despite
- Low ferritin
  - Restless sleep and ferritin <50 prescribe iron supplement
  - <75 if adults
- Car seat test before discharge

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## Solid tumors

- Overall not increased
- Decreased risk
  - Breast cancer, neuroblastoma, medulloblastoma
- Increased risk
  - Testicular cancer
    - Palpation by provider during all well child
    - Maybe trusted adult continues with routine screening

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## Autism

- Often delayed diagnosis
  - Avoid overshadowing
- **Better** imitation, relating, and receptive skills than without Down Syndrome
- **Worse** stereotypies, repetitive language, overactivity, social withdrawal, anxiety, self-injury, language skills, cognitive skills
- Screen with same tools at well child visits

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## Neurologic

- Seizures up to 13%
  - Infantile spasms
  - Benign movement disorders
- Increased incidence of Moya Moya
  - Down Syndrome is 26 times greater with Moya Moya
- Acute regression
  - "Loss of skills, marked mood changes, or catatonia, or who develop repetitive thoughts or behaviors that interfere with usual life activity"
  - No known cause, look for someone who specializes in treatment

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## Resources

- Very important though "non-medical"
  - Largest struggle for CTC is rural living
- Support groups and organizations
  - Family voices, Gigi's playhouse
- Respite care
- Financial resources
- Early intervention
- Preschool at 36 months
- [www.dsdiagnosisnetwork.org](http://www.dsdiagnosisnetwork.org) - cohorted, moderated Facebook

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## More

- Cognitive delay
  - Mild can occur
  - Moderate for most
  - Severe occasionally
  - Goal of prevention/treatment is maximize cognition and function
- JIA
- Type 1 DM

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## More

- Skin – alopecia, vitiligo, keratosis pilaris, folliculitis
- Medication sensitivity
- Obesity
- Feeding/eating disorders 30-80%
- Complementary or alternative medication (38%)
  - 20% are asked
  - High dose fat soluble vitamins, cell therapy
- Renal anomalies

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## More

- Synagis, PSV 23 if otherwise qualify
- Hypodontia
- Vulnerability
- Pubertal changes – similar to general population
- Obesity – standard BMI chart after 10 years
- Wandering
- 50% of pregnancy will have Down syndrome
- Majority of men are infertile

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## More

- Contraception and menstruation
- Work independence
- Transition of medical care
- Guardianship
- Alzheimers
- Newly developed Health Supervision for adults
- Need for rigorous research about atlanto-axial instability

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# References

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