Quarterly Webinar Series

What Families Need to Know: Utilizing the Pediatric Medical Care Guidelines for Down Syndrome

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Dr. Fran Hickey, Medical Director

Wednesday, September 11, 2019
The Global Down Syndrome Foundation is part of a network of affiliate organizations that work closely together on a daily basis to deliver on our mission, vision, values, and goals:

**Global & Affiliates**

- **Global**: was established as a 501(c)3 in 2009 and is “Dedicated to significantly improving the lives of people with Down syndrome through Research, Medical Care, Education, and Advocacy”

- **Affiliates are**:
  - Established with a lead gift from Anna & John J. Sie Foundation
  - Must work closely together to benefit people with Down syndrome
  - Must be self-sustaining financially
Incidence/Prevalence of Down Syndrome

❖ Incidence
  • Approximately 100-120 births/year in Colorado
  • 5,000 to 6,000 births in the US

❖ Prevalence
  • Prevalence is rising due to improved medical care assisted by DS Guidelines
  • Improved understanding of potential that a good quality of life is possible, also community inclusion
  • Improved life expectancy into 60’s
Average Life Expectancy of Individuals with Down Syndrome
Improved Life Expectancy Over Past 30-40 Years: Why?

- Improved OUTCOME of repair of cardiology defects
- Leukemia Treatment
- Improved Medical Care
  Down Syndrome Guidelines
- Deinstitutionalization
- Integration of Schools and Community
History of Guidelines for Individuals with Down Syndrome

- 1981 Healthcare Guidelines for Individuals with Down Syndrome published in *Down Syndrome Papers and Abstracts for Professionals* by Dr. Mary Coleman
- 1992 authored by Ohio/Western Penn Down Syndrome Network
- 1996 authored by Down Syndrome Medical Interest Group
- 1999 authored by Down Syndrome Medical Interest Group
- 2011 Last update “Health Guidelines for Children with Down syndrome” published in *Pediatrics*
References for Healthcare Guidelines for Individuals with Down Syndrome

Health Guidelines for Children with Down syndrome 2011
http://pediatrics.aappublications.org/content/128/2/393.full

Health Care Information for Families of Children with Down Syndrome
American Academy of Pediatrics- A Summary Document for Families
# AAP Guidelines Chart

## Down Syndrome Healthcare Guidelines (2011 Revision) Record Sheet

| Genetic Counseling¹, Karyotype | Birth | 6 mo | 1 | 2 | 3 | 4 | 5 | 6 | 7 | 8 | 9 | 10 | 11 | 12 | 13 | 14 | 15 | 16 | 17 | 18 | 19 | 20 |
| Cardiology | Echo² | | | | | | | | | | | | | | | | | | | | | |
| CBC to R/O transient myeloproliferative disorder, polycythemia | | | | | | | | | | | | | | | | | | | | | | |
| Swallowing assessment if feeding problems or aspiration | | | | | | | | | | | | | | | | | | | | | | |
| Parent Group Info and Support | Parent-to-parent contact, support groups, current books and pamphlets | | | | | | | | | | | | | | | | | | | | | |
| Hemoglobin | Hemoglobin annually beginning at 1 year old. If Hgb≤11, do (a) CRP and ferritin, or (b) reticulocyte Hemoglobin Content (CHr). If possible risk for iron deficiency, do (a) or (b) regardless. | | | | | | | | | | | | | | | | | | | | | |
| 23-valent pneumococcal vaccine² | | | | | | | | | | | | | | | | | | | | | | |
| Audiological Evaluation | ABR or OAE | Every 6 months until 3 years of age | | | | | | | | | | | | | | | | | | | | |
| Ophthalmologic Evaluation | Nstd Reflex | Ophtho Appt | Annual ophthalmology appt | Q1 Ophthalmology appointment | Q2 Ophthalmology appointment | | | | | | | | | | | | | | |
| Celiac Disease Screening | | | | | | | | | | | | | | | | | | | | | | |
| Thyroid – TSH, T4 | State Screen | Lab | Labs 12mo & 18mo | Test TSH and T4 annually | | | | | | | | | | | | | | |
| Neck X-ray (AAI)³ | | | | | | | | | | | | | | | | | | | | | | |
| Dental Exam | Annual Dental Exam: Reassure parents that delayed or irregular eruption, hypodontia are common. | | | | | | | | | | | | | | | | | | | | |
| Sleep Study by age 4 years | Done prior to 4 years of age | | | | | | | | | | | | | | | | | | | | |
| Early Intervention | | | | | | | | | | | | | | | | | | | | |
| Childhood | Discuss self-help, ADHD, OCD, wandering off, transition to middle school | | | | | | | | | | | | | | | | | | | | |
| Puberty | Discuss physical and psychosocial changes through puberty, need for gynecologic care (pelvic exams) in pubescent females | | | | | | | | | | | | | | | | | | | | |
| Facilitate transition | Guardianship, financial planning, behavioral problems, school placement, vocational training, independence with hygiene and self-care, group home, work settings | | | | | | | | | | | | | | | | | | | | |
| Sexual development and behaviors | Discuss Contraception, STDs, recurrence risk for offspring | | | | | | | | | | | | | | | | | | | | |
| Preventive care | Annually monitor for signs and symptoms of constipation, OSA, and aspiration. | | | | | | | | | | | | | | | | | | | | |

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1. Discuss Recurrence rate of future pregnancies with parents
2. 23-valent pneumococcal vaccine if chronic or pulmonary disease.
3. AAI: See AAP Guidelines page 391 - X-rays only if myopathic signs or symptoms
4. Follow up to be determined by Cardiologist

* Peds 2011;128:393-406 Chart by Sie Center for Down Syndrome

Updated 09/2019
Health Supervision for Children with Down Syndrome: AAP 2011

- Pediatric Journal, 2011
  - Initially published in 1981

- These guidelines are designed to assist the pediatrician and subspecialties in caring for the child with a diagnosis of Down syndrome
Timing
- Allow mother to recover from immediate delivery
- Wait until husband/partner/support person and infant are present in the room
- Private setting

Information
- Ideal to have delivering MD and PCP for infant both present for discussion in order to coordinate message
- Initially offer congratulations on birth of baby
- Inform parents of suspicion of DS diagnosis immediately
- Recommend offering private hospital room pending diagnosis

Also offer:
- Balanced approach to information
- Up-to-date printed info on Down syndrome
- Access to other families with children with DS and/or DS support organizations

Clinician should be aware “of realities and possibilities for healthy, productive lives of individuals with DS in society”

Cooley 1992; Skotko 2012; AAP Guidelines
Early Intervention (EI) Services: County-based usually

- Provides in-home therapy services (PT, OT-feeding, S/L), case management, and transition to preschool

- Early Intervention referral should be made prior to discharge from the nursery; need to confirm this has happened at 1st visit

- Speech therapy recommended by 9 months of age - strongly encourage the use of sign language
  - Signing videos provide good introduction, often available at public libraries

- Continue follow up with the family at office visits to ensure they have EI

- Access to other families with children with DS and referral to DS parent group
Review parental concerns and chromosomal karyotype
  • Discuss specific findings/potential clinical manifestations

Refer for genetic counseling - especially if need recurrence rate discussion (1%) or Translocation

If vomiting or absence of stools, check for gastrointestinal tract blockage (duodenal web or atresia, Hirschsprung disease)

Echocardiogram with follow up evaluation by a pediatric cardiologist

Exam for plethora, saturation for hypoxia

Review feeding history to ensure adequate caloric intake and consider swallow study (this guideline was added by AAP ‘11)
Newborn: Obtain CBC

More specific info provided regarding
- Transient Myeloproliferate Disorder (TMD)
- Management of TMD or polycythemia per subspecialty guidance
- Counsel parents of infants with TMD regarding increased risk (20%) for leukemia
- Need close follow up with Pediatric Oncology
High Rate of Neonatal Complications

- 73% requiring NICU stay as newborns
  - indicating the importance of appropriate medical readiness and intervention from birth to discharge

- NICU admission rate = 46%
- Require oxygen = 31% (n=725)
- PAH = 13%
- Feeding problem required NG tube = 48%

- Oxygen in over 56% of admits

- 60% required phototherapy.

- These numbers are higher in comparison to the largest NICU dataset for DS published from the United Kingdom
40 - 50% of all children with Down syndrome have CHD

20% will need surgery by 4 months

Every newborn needs an echo, evaluate for Pulmonary Hypertension
  - Echo is an evidence based guideline

As increasing blood crosses the septal defect:
  - More blood needs to be pumped to maintain cardiac output (flow to the body)
  - Burns extra calories and causes poor growth
  - The increased pulmonary blood flow may ultimately lead to irreversible lung disease = PULMONARY HYPERTENSION
Cardiology: Improved Treatment of Cardiology Defects

- Improved life expectancy in patients with CHDs over the past 20 years

- Earlier:
  - Identification with echo and fetal echo
  - Initial surgery

- Improved Areas of Care

- Pre-op care
  - Surgery techniques
  - CICU post-op care
  - PAH management
  - Cardiology follow-up
Increased incidence of PH in individuals with DS when compared to the general population
• Due to the cardiac and pulmonary co-morbidities and undefined predisposition of this population to PH

Etiologies of PH include CHD, hypoxia, polycythemia, OSA, feeding/aspiration, DS and abnormal pulmonary vasculature
Cua 2010

Keeping these children appropriately oxygenated is integral for prevention and treatment of PH

Sie Center incidence of PH in children with DS is 28% (n=346) (Bush 2018)
Hematologic Challenge - Anemia Assessment

❖ Challenge to identify anemia because RBC, Hemoglobin and Hematocrit, and MCV indices elevated

❖ Obtain CBC

❖ Serum ferritin

❖ CRP

❖ Iron studies
Guideline

- Newborn hearing screen approximately 20% initially fail
- Rescreen age 6 months with Behavioral Audiogram (BA)
- Hearing test every 6 months until 3 years of age
  - Then may need sedated ABR if unable to obtain accurate result
- After 3 years of age then yearly hearing test

Research

- Hearing Loss = 25%
- A recent published paper from our clinic and audiology dept at CHCO, indicates a high rate of hearing loss by 6 years of age (Nightengale 2017)
  - 25% of children with DS with permanent hearing loss at age 6 years
Issues in Down syndrome

- Stenotic ear canals (narrow ear canals) are common in up to 30 - 40% of infants with DS
- Narrow ear canals make it difficult to see the tympanic membrane and evaluate hearing
- If a child with Down syndrome has stenotic ear canals, he should see an ENT specialist for an otoscope with a microscope in order to avoid undiagnosed serous otitis media and subsequent hearing loss
Guideline provides indications for swallowing assessment:

- Marked hypotonia
- Slow feeding
- Choking with feeds
- Recurrent pneumonia - especially RUL or other recurrent or persistent respiratory symptoms
- Unexplained Failure to Thrive (FTT)
Morbidity and Mortality

- Respiratory illnesses are the cause of 80% of admissions to the hospital for children with DS
- After surgery, children with DS contract respiratory illnesses at a rate of 3x higher
- Leading cause of mortality in recent published studies
- Aspiration issues: Recurrent Pneumonias, Laryngeal clefts

Anatomically, children with DS have an increased risk of airway anomalies such as laryngeal clefts, laryngomalacia, tracheal ring, and tracheal bronchus, as well as lung anomalies including hypoplastic lungs and decreased alveoli in lungs

McDowell 2011; Yang 2002
Provided preliminary evidence that swallow study findings may be more stable after five months of age.

Children with DS and laryngomalacia, PAH, recurrent pneumonias, and/or congenital cardiac defects are at an increased risk for dysphagia.

- For these patients, early multidisciplinary care with the appropriate medical subspecialists and feeding and swallowing therapists trained to work with these populations is critical.

Providers should consider timing of instrumental swallowing studies and have a low threshold for ordering in children with DS and medical comorbidities.
Associations between Age, Respiratory Comorbidities, and Dysphagia in Infants with Down Syndrome (in review)

Timing and Predictive Value of Instrumental Swallow Assessments

Children with initial swallow study performed at >5 months of age were more likely (80.0%) to have consistent findings on repeat studies compared to children imaged at a younger age (35.3%).

Comorbidities Associated with Dysphagia

Children with laryngomalacia, pulmonary hypertension, pneumonia, or congenital cardiac disease were at a statistically significant increased risk for dysphagia (p > 0.05).

Comparison group: Patients with DS receiving care at the Sie Center for DS who were not referred for a swallow study matching the same age and date inclusion parameters (n=525)
Sleep Study Recommendations

❖ 2011 Guidelines
  • Initial Sleep Study recommended at 4 years old, unless earlier clinical indication

❖ Sie Center for Down Syndrome patients
  • Sleep studies N=678 (61.2% of patients)
    • Abnormal sleep studies (71.1%)
    • Obstructive sleep apnea (66.1%)
• Birth: Evaluate for cataracts by looking for red reflux

• AAP Guidelines: Refer all infants with DS for ophthalmology evaluation within first 6 months

• Refer to ophthalmologist with expertise managing children with DS

• Nystagmus common (20%): Needs follow up with experienced Pediatric Ophthalmologist
**Why do we see the Ophthalmologist annually?**

<table>
<thead>
<tr>
<th>Abnormality</th>
<th>Prevalence Range</th>
<th>Sie Center</th>
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<tr>
<td>Overall Ophthalmic Disorders</td>
<td>46 - 61%</td>
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<td>Astigmatism</td>
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<td>Optic Nerve Abnormality</td>
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<td>Issue Compliance with Glasses</td>
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Vision Challenges
Newborn Test

- Obtain TSH if Newborn State Screen (NBSS) measures only FT4
- Continue to screen at:
  - 6 and 12 months and 18 months
  - Annually from 2 years on with TSH, T4 (sooner is symptoms of concern for thyroid dysfunction)
Autoimmune

- Thyroid Abnormal = 247 31.3%
- Hypothyroidism = 232 29.4%
- Hyperthyroidism = 7 0.9%
- Diabetes Mellitus = 7 0.9%
- Alopecia Areata = 12 1.5%
Autism and other behavioral problems occur at increased frequency in children with Down children and may manifest as early as 2 or 3 years of age.

Refer children who may have autism for appropriate evaluation and intervention as soon as suspected.
Compliance to AAP Down syndrome Guidelines at Sie Center

- 77% referred to Audiology
- 35% referred to Cardiology
- 67% referred to get labs (TSH, T4)
- 60% referred to Ophthalmology
- 39% referred to get a Sleep Study
Average Life Expectancy of Individuals with Down Syndrome
Enjoy your children!!
## NIH Historic Increases in Down Syndrome Research

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<th>Year</th>
<th>NIH Actual Total Obligations by Budget Mechanism (In Millions &amp; Rounded)</th>
<th>CF Research Funding (Dollars in Millions)</th>
<th>Fragile X Research Funding (Dollars in Millions)</th>
<th>MS Research Funding (Dollars in Millions)</th>
<th>Autism Research Funding (Dollars in Millions)</th>
<th>DS Research Funding (Dollars in Millions)</th>
<th>DS Research Funding to NIH Budget (Rounded)</th>
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