Medical Management: Children with Down Syndrome

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Objectives

- Briefly discuss the genetics of DS
- Detail the developmental & behavioral differences observed in children with DS
- Review the medical management
- Highlight updates in basic science research
- Mention resources available to families of children with DS
Introduction

• Most common chromosome abnormality among live-born infants
• Most frequent cause of intellectual disability due to genetic abnormality
• The Committee on Genetics of the AAP has provided recommendations to assist providers in the care of children with DS
  • Evaluation and monitoring for associated abnormalities & prevention of common disorders
Genetics

- 95% trisomy 21 (nondisjunction)
- 3-4% unbalanced translocation
- 1% mosaic
Development & Behavior

- Range of cognitive abilities
  - Usually moderate ID
- Comorbid diagnosis of ASD
- Behavior problems
  - Inattention
  - Obsessions & compulsions
  - Defiance
  - Elopement

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The average age for walking independently in a child with DS is which of the following:

A. 1-1.5 yo
B. 2-2.5 yo
C. 3-3.5 yo
# Development and Behavior

<table>
<thead>
<tr>
<th>Milestone</th>
<th>Range for Children with Down Syndrome</th>
<th>Typical Range</th>
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<tr>
<td><strong>GROSS MOTOR</strong></td>
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<tr>
<td>Sits Alone</td>
<td>6 – 30 Months</td>
<td>5 – 9 Months</td>
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<tr>
<td>Crawls</td>
<td>8 – 22 Months</td>
<td>6 – 12 Months</td>
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<tr>
<td>Stands</td>
<td>1 – 3.25 Years</td>
<td>8 – 17 Months</td>
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<td>Walks Alone</td>
<td>1 – 4 Years</td>
<td>9 – 18 Months</td>
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<td><strong>LANGUAGE</strong></td>
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<td>First Word</td>
<td>1 – 4 Years</td>
<td>1 – 3 Years</td>
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<tr>
<td>Two-Word Phrases</td>
<td>2 – 7.5 Years</td>
<td>15 – 32 Months</td>
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<tr>
<td><strong>SOCIAL/SELF-HELP</strong></td>
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<tr>
<td>Responsive Smile</td>
<td>1.5 – 5 Months</td>
<td>1 – 3 Months</td>
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<tr>
<td>Finger Feeds</td>
<td>10 – 24 Months</td>
<td>7 – 14 Months</td>
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<tr>
<td>Drinks From Cup Unassisted</td>
<td>12 – 32 Months</td>
<td>9 – 17 Months</td>
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<tr>
<td>Uses Spoon</td>
<td>13 – 39 Months</td>
<td>12 – 20 Months</td>
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<tr>
<td>Bowel Control</td>
<td>2 – 7 Years</td>
<td>16 – 42 Months</td>
</tr>
<tr>
<td>Dresses Self Unassisted</td>
<td>3.5 – 8.5 Years</td>
<td>3.25 – 5 Years</td>
</tr>
</tbody>
</table>

"You have to forget the timetable you reserve for your other kids. This child will succeed at his own pace."
Hearing Conditions

- Children with DS require an annual hearing screen from birth to 3 years of age:
  - True
  - False
Hearing Screening

Screening Recommendations

- All newborns
- Repeat at 6 mos
- Every 6 mos until 5 yo
- Once yearly thereafter
Ophthalmic Screening

Screening Recommendations

- Initially in the first 6 mos of life
- Annual exams until 5 yo
- Every two years from 5-13 yo
- Every 3 years thereafter
Ophthalmic Disorders

- Strabismus is important to detect early because if left untreated it can lead to:
  A. Double vision
  B. Amblyopia (permanent vision loss)
  C. Headaches
  D. All of the above
Growth & Nutrition

- Higher risk of medical conditions leading to growth disturbances in children with DS
- Generally have **short stature**
  - Infants may be underweight
  - But older children tend to be overweight
Growth & Nutrition

- Are there growth charts for children with Down Syndrome?
  A. Yes
  B. No

- Should we use these growth charts?
  A. Only DS specific growth charts
  B. Use both CDC and DS growth charts
Growth & Nutrition

• Goal of obesity prevention
• Promotion of *physical activity*
• Attention to *diet*
Length for Age Curve for children with DS
https://www.cdc.gov/nchdddbirthdefects/downdysndrome/growth-charts.html
Growth & Nutrition

- Calcium & vitamin D intake should be monitored closely to minimize bone loss
Immune System

- Increased risk of infections
  - Respiratory primarily
- Increased risk for autoimmunity
  - Thyroid dysfunction
  - Celiac disease
  - Diabetes mellitus
  - Vitiligo
  - Alopecia
Thyroid Function

• Thyroid disease occurs in greater than 50% of individuals with DS
  • True
  • False
Thyroid Function

• Screening with TSH & free T4
  • Birth
  • 6 months
  • 1 yo
  • Annually thereafter

- Measurement of height & weight yearly
GI Problems

- Constipation
  - Very common, can be quite problematic
- Celiac disease (gluten-sensitive enteropathy)
  - Screening recommended beginning at 1 yo if signs/symptoms develop
- Hirschsprung disease
Hematological Disease

- Increased risk of transient myeloproliferative disorder (TMD) and polycythemia >> CBC-diff as newborn

- TMD is “pre-leukemia”
  - Usually self-resolves
  - If TMD, higher risk of leukemia >> CBC-diff regularly until 6 yo

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Hematological Disease

• For other children, screen for anemia
  >> Hgb annually from 1-13 yo
  • Anemia usually due to iron deficiency
  ▪ Also evaluate for signs/symptoms of leukemia
    ▪ Pallor/fatigue, increased infections, excessive bruising
Cardiac Disease

What is the percent of children with DS with congenital heart disease?

A. 20-40%
B. 40-60%
C. 60-80%
Cardiac Disease

• All newborns with DS should be evaluated
• Continued cardiac evaluation is necessary
Hypotonia & Gait

• Almost universally have hypotonia
• Pes planovalgus creates problems with stable ambulation
• “Chaplinesque gait”
Arthropathy

- Juvenile rheumatoid-like arthropathy occurs in 1-2% (RF neg)
  - Polyarticular
  - Small joints (hands/wrists)
  - Associated joint subluxations
Atlanto-Axial Instability

- Based on the most current guidelines, the best way to screen for atlanto-axial instability is performing a cervical spine radiograph in extension/flexion
  - True
  - False
Atlanto-Axial Instability

- Signs & symptoms consistent with SCI is the best predictor for AAI or dislocation
  - Loss of motor skills/change in gait
  - Loss of bowel/bladder control
  - Neck pain or stiffness
  - Head tilt/torticollis
- **Caution** regarding contact sports & trampoline use should be discussed
Atlanto-Axial Instability

- Positive screen >> lateral cervical spine radiograph (neutral)
- Anesthesia/Surgical procedure >> lateral cervical spine radiographs (neutral, flexion/extension)
Atlanto-Axial Instability

- Despite these recommendations, the Special Olympics requires neck films for participation.
Atlanto-Axial Instability

- Symptomatic AAI occurs in what percent of the population in individuals with DS?
  
  A. 3%
  B. 8%
  C. 20%
Neurologic Disease

- Seizures 10%
  - Infantile spasms
  - https://www.youtube.com/watch?v=9iRs-uyHA8
- Moya moya disease
- Alzheimers disease
Sleep Apnea

• Symptoms related to sleep apnea include
  • Snoring
  • Restless sleep
  • Atypical sleep position
Sleep Apnea

- Screening with polysomnography or pulse oximetry monitoring is only recommended for children with DS who are symptomatic:
  - True
  - False
Skin Problems

- Cutis Marmorata
- Fissured Tongue
- Folliculitis
- Palmar Hyperkeratosis
Dental Issues

- Delayed & irregular dental eruption patterns
  - Hypodontia 23%
- Periodontal disease is common
Life Expectancy

• Life expectancy is shorter
• However, survival rates have improved
  • Increased placement of infants in homes rather than institutions
  • Changes in the treatment of common causes of death, especially CHD
Basic Research Update

- The overexpression of many genes found on chromosome 21 contributes to learning deficits.
- The development of a DS mouse model has provided an opportunity to study emerging pharmacotherapies.
Basic Research Update

• Oxidative stress may contribute to some features of DS
  • Decreased immune function
  • Premature aging
  • Impaired mental function
  • Malignancy

• In particular, the activity of superoxide dismutase is increased
Alternative Treatments

- Supplementation with antioxidant nutrients has been proposed as potential therapy for individuals with DS
  - Zinc
  - Selenium
  - Megavitamins & minerals
  - Vitamin A
  - Vitamin B6
  - 5-hydorxytryptamine
  - Coenzyme Q10
Counseling

• Prenatal diagnosis of DS
• Confirmed diagnosis after birth and during childhood
• Young adolescence and transition to adulthood
“A promising future together: A guide for new & expectant parents” available in ENGLISH & SPANISH can be downloaded from the NDSS website.
Summary

• DS is the most common chromosome abnormality
• These children have increased medical and developmental needs
  • The Committee on Genetics of the American Academy of Pediatrics has provided recommendations to assist in the care of children with DS
  ▪ Caring for these children requires a multi-disciplinary effort
Questions/Comments

The Battle Building
UVA Health System