Making Sense of the Updated American Academy of Pediatrics Guidelines for Individuals with Down Syndrome

In July of 2011, the American Academy of Pediatrics (AAP) Committee on Genetics published revised guidelines to assist physicians in providing care to individuals with Down syndrome (Ds). Previous guidelines were published in 1999. Eight years later, the most welcomed and updated guidelines provide clinicians assistance in the key areas including prenatal testing and counseling; feeding in the infant with Down syndrome; airway, cardiac , neurologic and gastrointestinal assessment; growth in children with focus on BMI or weight for length (no longer using Ds growth curves); evaluation for atlantoaxial instability (AAI), clarification about screening for thyroid disorders and celiac screening; and vision and hearing screening. See below for a reorganized outline and summary of the recommendations based on organ system and age of the patient.

Cardiac

- Echocardiogram (~50% risk) with appropriate referral to cardiologist
- If symptoms or signs of congenital heart disease, monitor for congestive heart failure (CHF) at all visits
- Perform a car seat safety challenge prior to discharge from nursery, especially if the infant has significant hypotonia or cardiopulmonary disease.

Feeding and Gastrointestinal

- Check of symptoms/signs of celiac: if present then, obtain TTG IgA and quantitative IgA.

Evidence does not show that routine screening for celiac is beneficial. Know symptoms that might indicate celiac disease. Screening and appropriate referral when symptoms/signs present.

Growth

- Assess growth using NCHS or CDC standard growth charts (not former Down syndrome growth charts); follow also weight for length and / or BMI.

Airway and Breathing

- Assess of obstructive sleep apnea (*Sleep study by 4 years of age).*

Evaluation for Atlantoaxial Instability (AAI)

- C-spine positioning precautions are needed for all procedures.
- In the Asymptomatic Child

Routine cervical spine imaging (note, the Special Olympics has their own participation requirements so a C-spine film may be necessary).

- In the Symptomatic Child

Obtain C-spine imaging in neutral position first. If this is normal, then obtain Flexion and Extension views. Then, refer to a neurosurgeon with experience in children with Ds.

Hematologic

- Evaluate for anemia, polycythemia and TMD (transient myeloproliferative disorder): obtain CBC with differential at birth.
- Then hemoglobin annually (evaluate for Fe deficiency anemia).
- Obtain CRP and iron studies (ferritin) if there is a risk for iron deficiency anemia (note that children with Ds may have higher MCV).

Thyroid

- Evaluate for hypothyroidism: obtain TSH and T4 at birth, *if not already done by state screening*.
- Obtain TSH at 6 months of age then annually.

Understand the importance of child's communication skills, including need for regular evaluation of hearing and vision to maximize ability to communicate.

Ears

- Pay vigilant attention to risk of middle ear effusion.
- Perform a newborn hearing screen, such as ABR or OAE.
- Audiology evaluation at 6 months of age.
- If normal hearing established, behavioral audiogram and tympanometry every 6 months until bilateral ear specific testing possible.
- If normal ear specific hearing established (usually after age 4y), behavioral audiogram annually.
- Referral to otolaryngologist with experience in children with Ds if tympanic membrane cannot be visualized, if abnormal hearing.

Eyes

- Examine the newborn's eyes with particular attention to red reflex. If the exam is abnormal (i.e. absence of red reflex) then promptly refer to ophthalmologist who has experience in children with Ds.
- Ophthalmology referral by 6 months of age.
    - *Annual eye exam age 1 to 5 years.*
    - *Eye exam every two years ages 6-13 years.*
    - *Eye exam every three years age 13+ .*

References

Health Supervision for Children with Down Syndrome

Marilyn J. Bull and the Committee on Genetics

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