Down syndrome: Update

Dianne M. McBrien, MD
Clinical Associate Professor of Pediatrics
University of Iowa Children’s Hospital
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In 2011, the American Academy of Pediatrics (www.aap.org) last updated clinical practice guidelines for management of patients with Down syndrome.
Differences

- Evaluate for iron deficiency anemia
- Which growth charts to use
- Some routine studies excluded
Most common referral reasons

- Rule out dementia
- ADHD
- Behavior
Car seat evaluation prior to nursery discharge

Why?
- Hypotonia
- More likely to have congenital CV lesion, pulmonary hypertension
- Controversial
Monitoring for iron deficiency anemia

- Incidence probably same as in typical population
- Disproportionate impact on motor and cognitive outcomes
- Certain risk factors: Dietary intake, feeding problems
- Diagnosis can be obscured by macrocytosis
• For most patients, obtain an annual Hgb concentration
• For those patients with risk factors (poor growth, feeding problems, history of past anemia) also assess iron stores with ferritin and CRP
What about those cervical spine films?
Routine cervical spine films are no longer recommended!
Monitor for symptoms and signs of cord compression

- Head, neck, back pain
- Head tilt
- Change in gait
- Change in bowel/bladder continence
- Weakness
Questions about activity restriction

- Encourage lots of free play
- Keep Special Olympics restrictions in mind
- Activities with high risk of head trauma and neck hyperflexion should not be encouraged
The screening guidelines for celiac disease in this population have changed.
Routine screening for celiac disease is no longer recommended!
Monitor for symptoms and signs

- Insufficient growth or weight gain
- Unexplained anemia
- Unexplained micronutrient deficiencies
- Chronic diarrhea
- Refractory behavioral problems
ADHD

- Examine expectations
- Processing speed and learning style
- Look at comorbidities (hearing, sleep)
- Modest expectations
- Moderate dosage
Alzheimer’s dementia
People with Down syndrome present questions about genetics of aging:

- ALL autopsy studies in adults over 35 show characteristic plaques and tangles.

However, NOT all of those individuals were demented in life.
Many things mimic dementia in people with Down syndrome

- Hearing/vision loss
- Depression
- Acquired valvular lesion
- Obstructive sleep apnea
Some may find it useful to consider adults with Down syndrome as having a biological or medical age roughly 20 years older than chronological age.
Down Syndrome Clinic
Center for Disabilities and Development
University of Iowa Children’s Hospital
(319)-353-6900