Practice Guidelines

Medical Care for Adults With Down Syndrome: Guidelines From the Global Down Syndrome Foundation

Key Points for Practice

- Assess adults with Down syndrome annually for dementia starting at 40 years of age.
- Screen adults with Down syndrome and obesity for type 2 diabetes mellitus with an A1C level every two to three years starting at 21 years of age and earlier if other diabetes risk factors are present; in adults with Down syndrome at a healthy weight, start screening at 30 years of age.
- Avoid routine cervical radiography in adults with Down syndrome unless suggested by neurologic symptoms.
- Screen adults with Down syndrome for hypothyroidism with a thyroid-stimulating hormone test every one to two years starting at 21 years of age.

From the *AFP* Editors

Down syndrome occurs in one of every 691 live births in the United States, making it the most common chromosomal condition and cause of developmental delay. Screening for comorbid conditions, early intervention programs, and subsequent medical management have helped increase life expectancy from 25 years in 1983 to 60 years today. In addition to mild to moderate intellectual disability, people with Down syndrome have increased risks of congenital cardiac and gastrointestinal anomalies, autoimmune disorders, leukemias, respiratory infections, sleep disorders, hearing and vision loss, and early development of dementia. People with Down syndrome often have limited expressive language skills, delayed motor skill development, and reduced executive functioning capacity. Inequitable access to care, issues of guardianship, and social stigma also contribute to the vulnerability of adults with Down syndrome.

The Global Down Syndrome Foundation (GLOBAL) has published the first medical care guidelines for adults with Down syndrome.

Behavior and Mental Health

Behavior and mental health conditions are common in adults with Down syndrome, and common medical conditions such as sleep apnea, hypothyroidism, and celiac disease can worsen behavioral concerns. Challenges in assessing an adult with Down syndrome may lead to misdiagnosis of a medical issue, such as hearing loss, as behavioral. The *Diagnostic and Statistical Manual of Mental Disorders*, 5th ed., and the *Diagnostic Manual-Intellectual Disability 2* are moderately helpful in the diagnosis of mental health conditions in these patients.

Early dementia is common in adults with Down syndrome, affecting nearly 9% of adults between 45 and 50 years of age and nearly one-third of those between 55 and 59 years of age. Given the benefits of early diagnosis and treatment, annual screening for dementia is recommended starting at 40 years of age using the National Task Group—Early Detection Screen for Dementia assessment (https://www.the-ntg.org/_files/ugd/c53c74_6b1f8d20f969401b87a6328c679d71c4.pdf), which involves interviewing patients and caregivers. This assessment looks for signs of dementia across the six domains of cognition and memory, behavior and personality, communication, adaptive functioning, motor skills, and established skill decline. Dementia is uncommon in adults with Down syndrome before 40 years of age.

Diabetes Mellitus

Obesity and diabetes are more common in adults with Down syndrome. Diabetes rates in adults with Down syndrome are four times those of unaffected adults younger than 30 years and

Coverage of guidelines from other organizations does not imply endorsement by *AFP* or the AAFP.

This series is coordinated by Michael J. Arnold, MD, contributing editor.

A collection of Practice Guidelines published in *AFP* is available at https://www.aafp.org/afp/practguide.

CME This clinical content conforms to AAFP criteria for CME. See CME Quiz on page 358.

Author disclosure: No relevant financial relationships.

twice those of unaffected adults 30 years and older. Down syndrome is associated with premature aging, earlier onset of cataracts, and other organ dysfunction. Despite limited evidence, GLOBAL recommends diabetes screening with an A1C level every three years beginning at 30 years of age in those without obesity, at 21 years of age in those with obesity, and earlier if other risk factors are present. Adults with Down syndrome and obesity should be encouraged to follow the U.S. Preventive Services Task Force behavioral intervention recommendations to prevent obesity-related morbidity and mortality in adults (https://www.uspreventiveservicestaskforce.org/uspstf/recommendation/obesity-in-adults-interventions), although common physiologic differences such as lower heart rate and blood pressure, joint hyperflexibility, hypotonia, and balance issues can limit physical activity in adults with Down syndrome.

Cardiovascular Disease

Because congenital heart disease increases the risk of cardioembolic stroke, consider periodic cardiac monitoring in patients with congenital heart disease. In patients without congenital heart disease, cardiovascular prevention recommendations are the same for adults without Down syndrome.

Atlantoaxial Instability

Although one in 10 adults with Down syndrome has atlantoaxial instability, these patients do not have a higher risk of spinal cord injury. Cervical spine radiography should be avoided in adults with Down syndrome unless symptoms of cervical myelopathy are present.

Osteoporosis

Because prevention of osteoporosis has not been studied in adults with Down syndrome, evidence-based recommendations cannot be made. Secondary causes of osteoporosis, including hyperparathyroidism, hyperthyroidism, vitamin D deficiency, celiac disease, and medications, are common and should be considered after a fragility fracture.

Thyroid

Hypothyroidism is common in adults with Down syndrome, occurring 10 to 30 times more than in the overall population. Because symptoms such as fatigue, weight gain, and constipation are common in people with Down syndrome, screening with serum thyroid-stimulating hormone testing every one to two years starting at 21 years of age is recommended.

Celiac Disease

Celiac disease is common in children with Down syndrome, but the prevalence in adults is unknown. Although symptoms can be challenging to detect and differentiate from other conditions, annual clinical assessment for celiac disease is recommended, with laboratory evaluation if indicated.

Editor's Note: Dr. Christopher Bunt is an assistant medical editor with *AFP*.

These guidelines are the first attempt at taking an evidence-based approach to the management of adults with Down syndrome. Although evidence is limited, there are some important recommendations. The screening recommendations for diabetes mellitus, dementia, and hypothyroidism provide an initial template for routine care. Because we all learned about atlantoaxial instability and Down syndrome in medical school, it is important to know that this finding is not clinically significant.—Michael J. Arnold, MD, Contributing Editor

Guideline source: Global Down Syndrome Foundation (GLOBAL)

Evidence rating system used? Yes

Systematic literature search described? Yes

Guideline developed by participants without relevant financial ties to industry? Yes

Recommendations based on patient-oriented outcomes? Yes

Published source: *JAMA*. October 20, 2020;324(15):1543-1556

PDF available at: https://www.globaldownsyndrome.org/wp-content/uploads/2020/10/Global-Down-Syndrome-Foundation-Medical-Care-Guidelines-for-Adults-with-Down-Syndrome-v.1-10-20-2020.pdf (globaldownsyndrome.org)

Christopher W. Bunt, MD, FAAFP
Medical University of South Carolina
Charleston, S.C.
Email: cwbunt@gmail.com

Stephanie K. Bunt, PhD
University of Iowa
Iowa City, IA ■