

# Understanding the Co-Occurrence of Disease in People with Down Syndrome & the GLOBAL Guideline

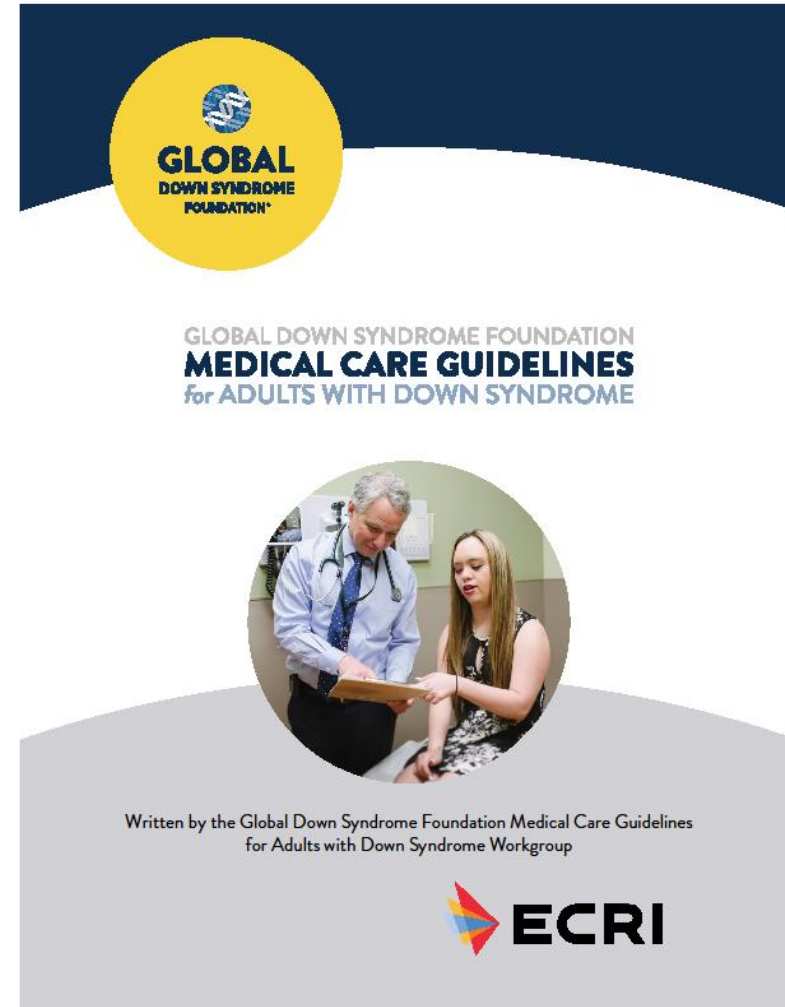
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**Objective:** Review care for adults with Down syndrome throughout the lifespan related to our findings from the *GLOBAL Medical Care Guidelines for Adults with Down Syndrome*.

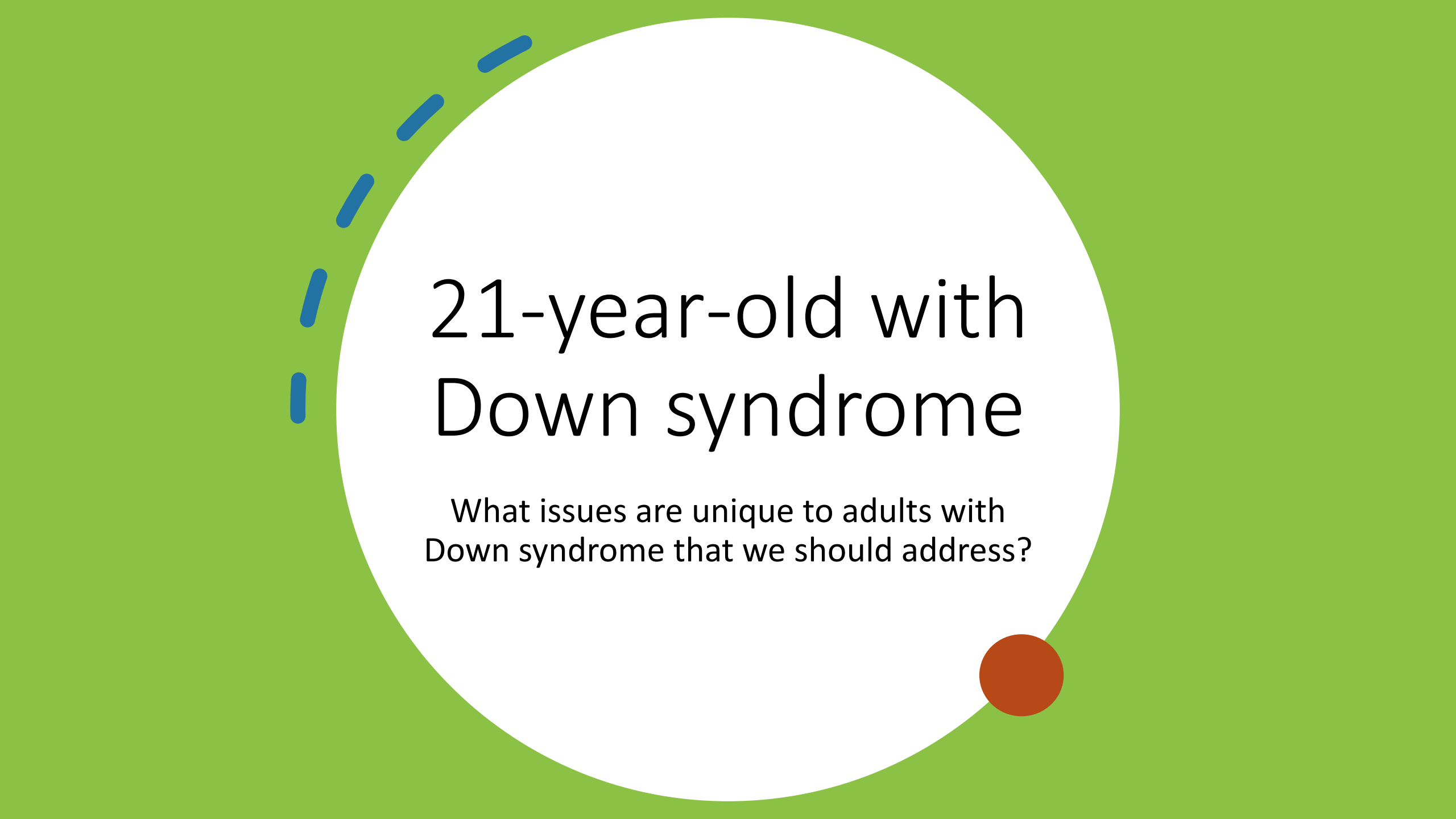


**JAMA | Special Communication**

# Medical Care of Adults With Down Syndrome

## A Clinical Guideline

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for the Global Down Syndrome Foundation Medical Care Guidelines for Adults with Down Syndrome Workgroup



# 21-year-old with Down syndrome

What issues are unique to adults with  
Down syndrome that we should address?

21-year-old

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Thyroid

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Diabetes

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Celiac

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Is a Cardiologist needed

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Exercise

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Screen for Atlanto-Axial Instability

21-year-old

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Thyroid? Yes!

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Diabetes? Only if BMI greater than 30

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Celiac? Only if symptomatic

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Is a Cardiologist needed? If history of congenital heart disease

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Exercise? Yes!

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Screen for Atlanto-Axial Instability? Only if symptomatic



# 30-year-old with Down syndrome

Mood seems changed from the last visit-  
Less interested in friendships, activities

30-year-old

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Thyroid

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Diabetes

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Behavior change

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Screen for high cholesterol?

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Screen for Dementia?



30-year-old

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Thyroid? Yes! Every 1-2 years

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Diabetes? Yes! Every 1-3 years

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Behavior change? Screen for medical issues, treat any mental health issues

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Screen for high cholesterol? Not routinely at age 30

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Screen for Dementia? Not at age 30



# 40-year-old with Down syndrome

With some new gastrointestinal issues

40-year-old

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Screen for high cholesterol?

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Screen for dementia?

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How about celiac testing now? Is it too late?

40-year-old

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Screen for high cholesterol?

Yes

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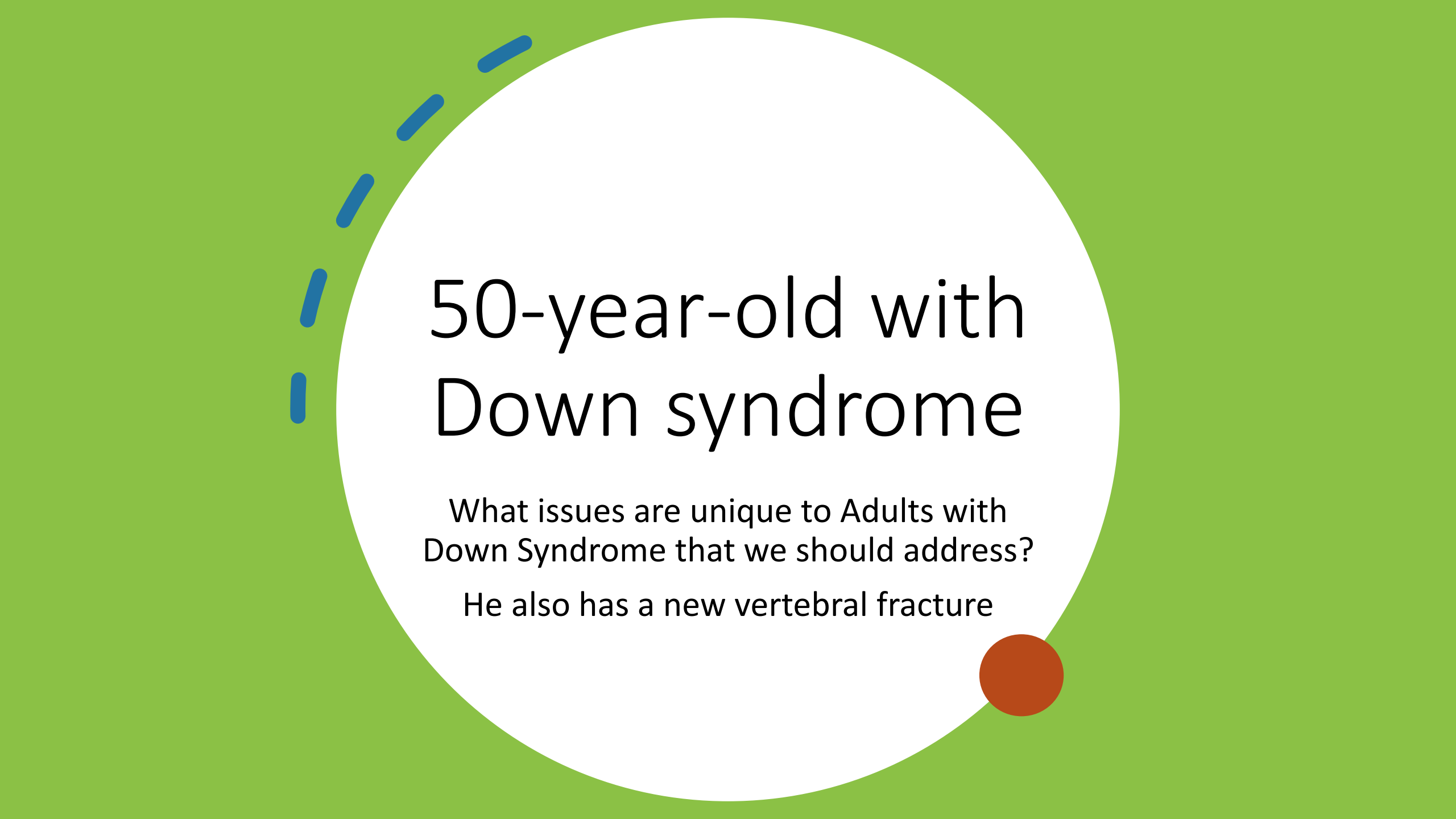
Screen for dementia?

Yes

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How about celiac testing now? Is it too late?

Not too late – do screen



# 50-year-old with Down syndrome

What issues are unique to Adults with  
Down Syndrome that we should address?

He also has a new vertebral fracture

50-year-old

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Diabetes screening now?  
Thyroid?

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What to do about the  
fracture?

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More screening for  
dementia?

50-year-old

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Diabetes screening now? Thyroid?

Yes no age limit

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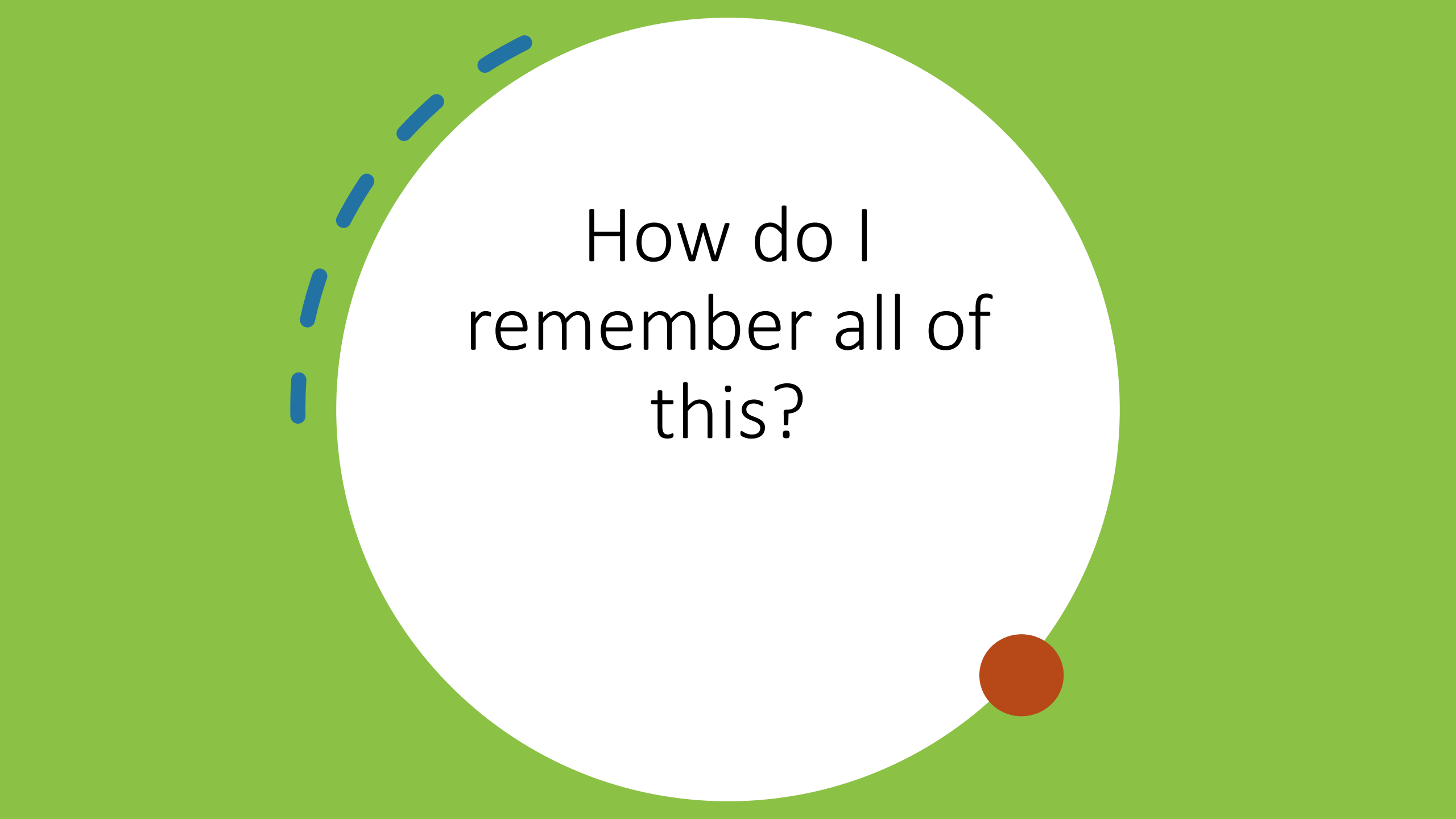
What to do about the fracture?

Refer to endocrinologist experienced  
with osteoporosis

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More screening for dementia?

Yes – in definitely



How do I  
remember all of  
this?



# GLOBAL MEDICAL CARE GUIDELINES

## for Adults with Down Syndrome Checklist



This checklist is intended to support the health of adults with Down syndrome directly or through their caregivers. We encourage this checklist to be shared with your medical professionals. Statements in blue represent our recommended, periodic health screenings/assessments that should begin at a specific age. Below each blue screening/assessment recommendation, there are blank boxes. Caregivers or individuals with Down syndrome can check off, date, or initial each blank box when the screening/assessment is completed. For screening/assessment recommendations with a time range (e.g. 1-2 years), the box size represents the longer possible time frame, such as 2 years versus 1. Statements in gray represent advisory recommendations that individuals with Down syndrome and caregivers should follow throughout adulthood.

Screening/Assessment  
  Advisory  
  Checkbox  
  No Recommendations

	21-29 Years	30-39 Years	40-49 Years	50-59 Years	60+ Years
<b>Behavior</b>	A review of behavioral, functional, adaptive, and psychosocial factors should be performed as part of an annual history that clinicians obtain from all adults with Down syndrome, their families, and caregivers. (Boxes below represent 1 year increments)				
	When concern for a mental health disorder in adults with Down syndrome is present medical professionals should: a) Evaluate for medical conditions that may present with psychiatric and behavioral symptoms and b) Refer to a clinician knowledgeable about the medical, mental health disorders, and common behavioral characteristics of adults with Down syndrome.				
	When concern for a mental health disorder in adults with Down syndrome is present, medical professionals should follow guidelines for diagnosis in the Diagnostic and Statistical Manual of Mental Disorders (DSM 5). The Diagnostic Manual-Intellectual Disability 2 (DM-ID-2) also may be used to adapt diagnostic criteria from the DSM-5.				
<b>Dementia</b>	Caution is needed when diagnosing age-related, Alzheimer's Type Dementia in adults with Down syndrome less than age 40.		Medical professionals should assess adults with Down syndrome and interview their primary caregivers about changes from baseline function annually beginning at age 40. Decline in the six domains as per the National Task Group - Early Detection Screen for Dementia (NTG-EDSD) should be used to identify early-stage age-related Alzheimer's-type dementia and/or a potentially reversible medical condition. (Boxes below represent 1 year increments)		
<b>Diabetes</b>			For asymptomatic adults with Down syndrome, screening for type 2 diabetes using HbA1c or fasting plasma glucose should be performed every 3 years beginning at age 30. (Boxes below represent 3 year increments)		
	For any adult with Down syndrome and comorbid obesity, screening for type 2 diabetes using HbA1c or fasting plasma glucose should be performed every 2-3 years beginning at age 21. (Boxes below represent 3 year increments)				
<b>Cardiac</b>			For adults with Down syndrome without a history of atherosclerotic cardiovascular disease, the appropriateness of statin therapy should be assessed every 5 years starting at age 40 and using a 10-year risk calculator as recommended for adults without Down syndrome by the U.S. Preventive Services Task Force. (Boxes below represent 5 year increments)		
	For adults with Down syndrome, risk factors for stroke should be managed as specified by the American Heart Association/American Stroke Association's Guidelines for the Primary Prevention of Stroke.				
	In adults with Down syndrome with a history of congenital heart disease, given the elevated risk of cardioembolic stroke, a periodic cardiac evaluation and a corresponding monitoring plan should be reviewed by a cardiologist.				
<b>Obesity</b>	Healthy diet, regular exercise, and calorie management should be followed by all adults with Down syndrome as part of a comprehensive approach to weight management, appetite control, and enhancement of quality of life.				
	Monitoring for weight change and obesity should be performed annually by calculating Body Mass Index in adults with Down syndrome. The U.S. Preventive Services Task Force Behavioral Weight Loss Interventions to Prevent Obesity-Related Morbidity and Mortality in Adults should be followed. (Boxes below represent 1 year increments)				
<b>Atlantoaxial Instability</b>	In adults with Down syndrome, routine cervical spine x-rays should not be used to screen for risk of spinal cord injury in asymptomatic individuals.				
	Annual screening for adults with Down syndrome should be based on a review of signs and symptoms of cervical myelopathy using targeted history and physical exam. (Boxes below represent 1 year increments)				
<b>Osteoporosis</b>	For primary prevention of osteoporotic fractures in adults with Down syndrome, there is insufficient evidence to recommend for or against applying established osteoporosis screening guidelines, including fracture risk estimation; thus, good clinical practice would support a shared decision-making approach to this issue would support a shared decision-making approach to this issue.				
	All adults with Down syndrome who sustain a fragility fracture should be evaluated for secondary causes of osteoporosis, including screening for hyperthyroidism, celiac disease, vitamin D deficiency, hyperparathyroidism and medications associated with adverse effects on bone health.				
<b>Thyroid</b>	Screening adults with Down syndrome for hypothyroidism should be performed every 1-2 years using a serum thyroid-stimulating hormone (TSH) test beginning at age 21. (Boxes below represent 2 year increments)				
<b>Celiac Disease</b>	Adults with Down syndrome should receive an annual assessment for gastrointestinal and non-gastrointestinal signs and symptoms of celiac disease using targeted history, physical examination and clinical judgement of good practice. (Boxes below represent 1 year increments)				

This checklist is not intended to be diagnostic. Presentation of medical and mental health conditions for people with Down syndrome may be atypical. Similar signs and symptoms may be a consequence of multiple reasons, including different disease processes. Thus, the patient evaluation should include considerations of additional causes for any detected sign or symptom. The development of new and/or changes in signs or symptoms should prompt a comprehensive evaluation with your clinician.

# What's Next for the GLOBAL Guideline?

Family-Friendly Version **NOW AVAILABLE!**

Translations:

- Spanish & Japanese (August '22)

New topics to be covered in the next version:

- Sleep apnea
- Cancer screening
- Ophthalmology and more...

