

Addressing Common Health Conditions in Adults with Down Syndrome

Adult Down Syndrome Center Health Education Series

February 10, 2021 | Brian Chicoine, MD

 Advocate Health Care

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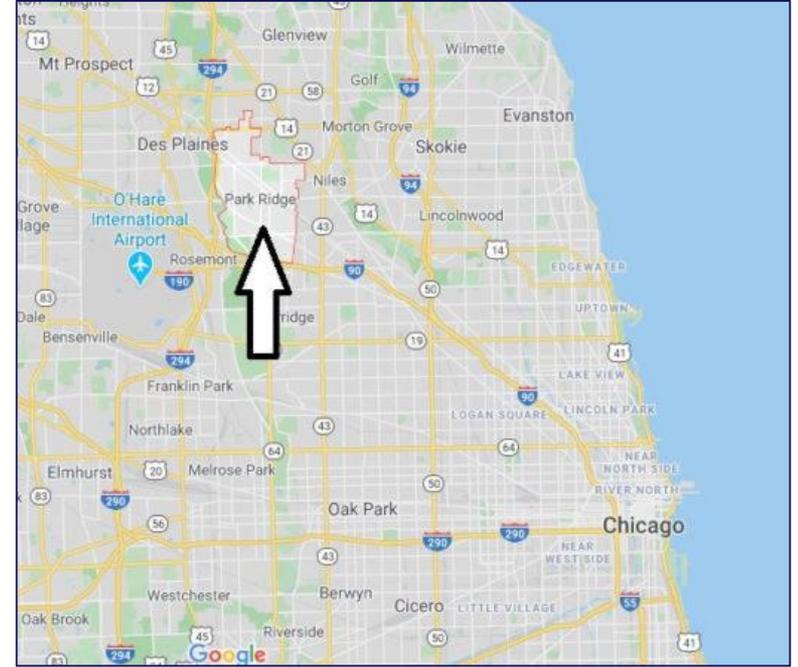
This information is provided for educational purposes only and is not intended to serve as a substitute for a medical, psychiatric, mental health, or behavioral evaluation, diagnosis, or treatment plan by a qualified professional.

Adult Down Syndrome Center



Our mission is to enhance the well-being of people with Down syndrome who are 12 and older by using a team approach to provide comprehensive, holistic, community-based health care services.

Adult Down Syndrome Center



We are **AdvocateAuroraHealth**

Online Resource Library

- Contains videos, articles, and booklets on a variety of topics, including aging, Alzheimer's disease, and mental health.
- Resources are continually being updated.

The screenshot shows the website for the Advocate Medical Group Adult Down Syndrome Center. At the top, there is a navigation bar with the center's name and a search bar. Below this is a yellow banner for "COVID-19 Resources" with a sub-header and a brief description. The main content area is titled "Adult Down Syndrome Center | Resource Library" and features three large image-based buttons: "For People with Down Syndrome" (with a photo of a young woman), "For Families & Caregivers" (with a photo of a family), and "For Health Care Professionals" (with a photo of a healthcare professional). Below these are five smaller buttons for "Events, Classes & Programs", "Video Gallery", "Related Organizations", "Projects", and "News". At the bottom, a yellow banner displays the URL adsresources.advocatehealth.com.

Objectives

- Provide an overview of common health conditions in adults with Down syndrome, including presentation and prevention.
- Describe guidelines and best practices for addressing common health conditions in adults with Down syndrome.
- Identify resources to learn more about common health conditions in adults with Down syndrome.

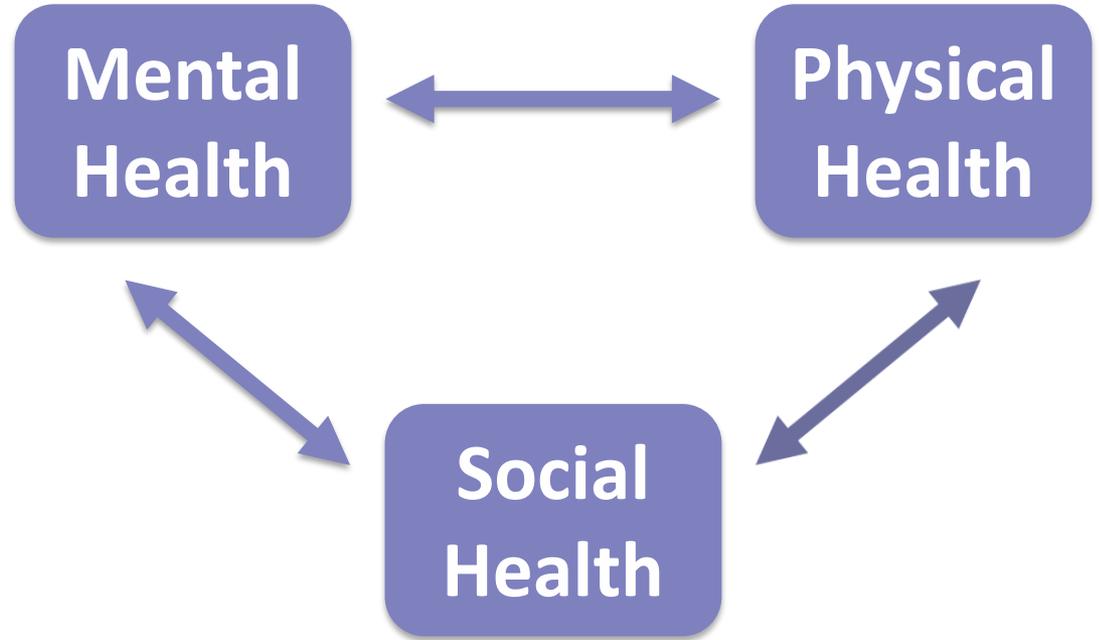
What is health?

We are **AdvocateAuroraHealth**[™]

Health and well-being

“Health is a state of complete physical, mental, and social well-being and not merely the absence of disease or infirmity.”

(World Health Organization)



Health conditions

We are **AdvocateAuroraHealth™**

Common health conditions

- Definition
- Presentation
- Prevention
- Treatment

GLOBAL Medical Care Guidelines

- Evidence-based recommendations intended to offer guidance to healthcare providers who see adults with DS
- Newest set published in October 2020



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 screening/treatments that should begin at a specific age. Below each blue screening/treatment recommendation, there are black boxes. Caregivers or individuals with Down syndrome can check off, date, or initial each black box when the screening/treatment is completed. For screening/treatment recommendations with a time range (e.g. 12 years), the box size represents the longest 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/treatment
 Advisory
 Done
 No recommendation

	21-29 Years	30-39 Years	40-49 Years	50-59 Years	60+ Years
Behavior					
Dementia					
Diabetes					
Cardiac					
Obesity					
Adolescent Incontinence					
Osteoporosis					
Thyroid					
Celiac Disease					

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<https://www.globaldownsyndrome.org/global-adult-guidelines/>

ADSC Health Questionnaire



NEW PATIENT HEALTH QUESTIONNAIRE
ADULT DOWN SYNDROME CENTER || ADVOCATE LUTHERAN GENERAL HOSPITAL

Date of Appointment _____

ALL QUESTIONS REFER TO THE PERSON WITH DOWN SYNDROME

Name _____ Date of Birth _____

Person Filling Out the Form: _____

Do you have any specific concerns regarding new or ongoing health/behavioral issues about the Down syndrome? (Please write in the space below. Use another sheet of paper if necessary).

SAFETY ISSUES:

Are there safety issues that have affected or impacted his or her daily routine (i.e. sex, physical) If so, please describe:

REVIEW OF SYSTEMS:

SAFETY ISSUES:

Are there safety issues that have affected or impacted his or her daily routine (i.e. sex, physical abuse, etc.)? If so, please describe:

REVIEW OF SYSTEMS:

BEHAVIOR, FEELINGS AND MEMORY

Has the patient had trouble remembering things or been forgetful? Yes _____ No _____

Can the patient learn to do new things? Yes _____ No _____

Stopped being able to do things he or she used to be able to do? Yes _____ No _____

Had any change in his or her usual behavior or outlook on life? Yes _____ No _____

Had any change in his or her interest in life or activities? Yes _____ No _____

Seemed sad or withdrawn? Yes _____ No _____

Are there concerns about how the patient is acting or feeling? Yes _____ No _____

If yes, please describe:

ENDOCRINE

Has a medical person ever said that the patient has a thyroid problem? Yes _____ No _____

If yes, has the medical person said the thyroid is underactive (working too little) or overactive (working too much)?

Underactive _____ Overactive _____

Ever been diagnosed with diabetes mellitus? Yes _____ No _____

Hasn't been drinking more liquids recently? Yes _____ No _____

Drinking more? Yes _____ No _____

Ever had seizures (spasms, convulsions)? Yes _____ No _____

Has he or she ever had any other neurological issues (epilepsy, multiple sclerosis, stroke, etc)? Yes _____ No _____

Reason: _____

Does he or she seem more tired? Yes _____ No _____

Has he or she ever been diagnosed with a sleep disorder? Yes _____ No _____

Reason: _____

Does the patient go to bed? _____

Does the patient get up? _____

https://www.advocatehealth.com/assets/documents/subsites/luth/downsyndrome/new_patient/new-patient-health-questionnaire.pdf

Overweight and obesity

- Body mass index (BMI)
- More common
 - Estimates vary
 - Review of literature combined results from several studies:
 - 38% of study participants were obese, 34% were overweight
- Associated with other physical, mental, and social health issues

Capone et al. 2018 Tsou et al. 2020

Overweight and obesity



- GLOBAL Guidelines
 - Monitor for weight change and obesity annually
 - Follow a healthy diet, regular exercise, and calorie management

Tsou et al. 2020

Prevention and treatment

Healthy Lifestyle

- Healthy eating
 - Hydration
 - Balanced meals (all the food groups)
 - Portion size
 - Limit processed foods and pop/soda
 - Small changes add up
- Exercise / physical activity
 - Find what works for the individual
 - Make it fun and social
 - Incorporate it throughout the day
 - Get outdoors

Prevention and treatment

- Medications
- Surgery

Vitamin deficiencies

- Vitamin B12
 - Seems to be more common in people with Down syndrome
 - More common in people with digestive disorders such as celiac disease
 - Autoimmune?
- Vitamin D
 - Bone health
 - COVID?

Prevention and treatment

- Healthy diet
- Regular exercise
- Outdoor activities

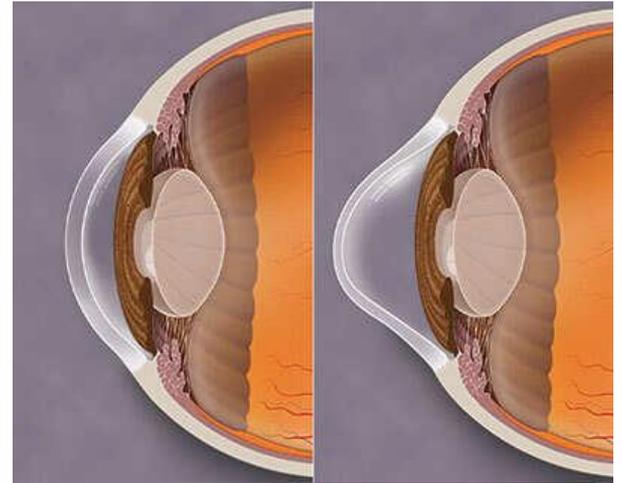
Nutrition & Exercise Resources



- [Hunger and Satiety in People with Down Syndrome](#)
- [Low Carb Diets](#)
- [Vitamin B12 Deficiency](#)
- [Exercise in People with Down Syndrome](#)

Vision and hearing

- Cataracts
- Keratoconus
- Hearing impairment
- Cerumen impaction (ear wax)



A healthy cornea

A cornea with
keratoconus

Source: [American Academy of Ophthalmology](#)

Prevention and treatment

- Consider screening at least every 2 years

Vision and Hearing Resources



- [Keratoconus in People with Down Syndrome](#)
- [Vision in Adults with Down Syndrome](#)
- [Cerumen Impaction](#)

Atlantoaxial instability

- Condition of the vertebrae in the neck

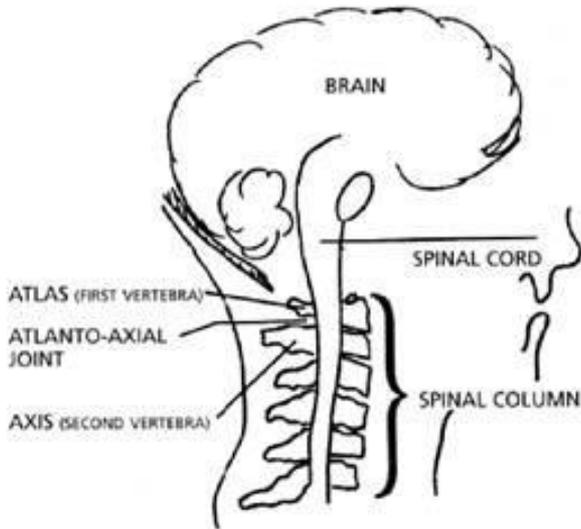
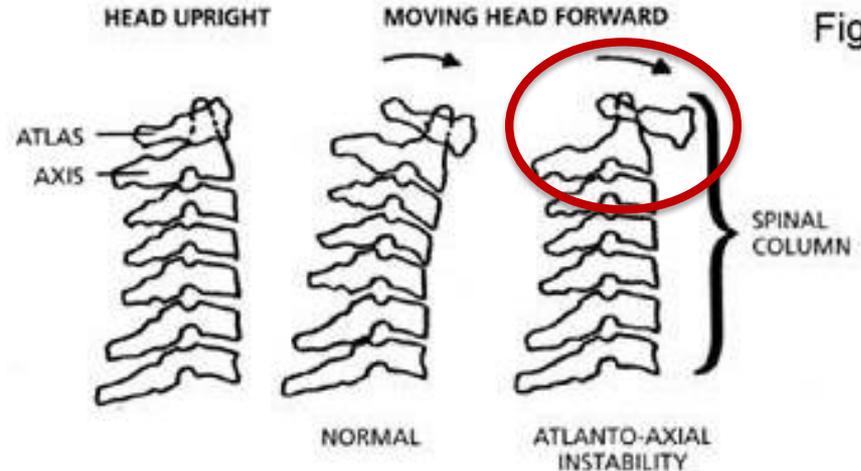


Fig 1



Source: [Down's Syndrome Association](#)

Atlantoaxial instability



- GLOBAL Guidelines

- For adults with Down syndrome *without* symptoms:

- Routine cervical spine X-rays should not be used to screen for risk of spinal cord injury.
 - Review of signs and symptoms of cervical myelopathy annually. Do an annual screening using targeted history and physical exam.

Tsou et al. 2020

Seizures

- Types
- Alzheimer's disease
- Treatment



AGE

The onset of seizures is most common in children and older adults.



Neurology Resources

- [Atlantoaxial Instability Screening](#)
- [Seizures](#)
- [Psychogenic Non-Epileptic Seizures \(“Fake” Seizures\)](#)
- [Seizures in People with DS and Alzheimer’s Disease](#)

Mental health disorders

- Behavioral and mental health disorders are common in people with Down syndrome.
 - Depression, anxiety, OCD
- Studies have shown that 25-40% of people with Down syndrome experience a behavioral or mental health disorder during their life.

Dykens 2007 Mantry et al. 2008 Alexander 2015 Palumbo & McDougle 2018

Common characteristics

- The Groove
- Self-talk
- Strong memory and the concept of time

Mental health disorders



- GLOBAL Guidelines
 - When concern for a mental health disorder in adults with Down syndrome is present:
 - Refer to a clinician knowledgeable about the medical, mental health disorders, and common behavioral characteristics of adults with Down syndrome.
 - Follow guidelines in the:
 - Diagnostic and Statistical Manual of Mental Disorders (DSM-5) and/or
 - Diagnostic Manual – Intellectual Disability (DM-ID-2)

Tsou et al. 2020

Mental health disorders

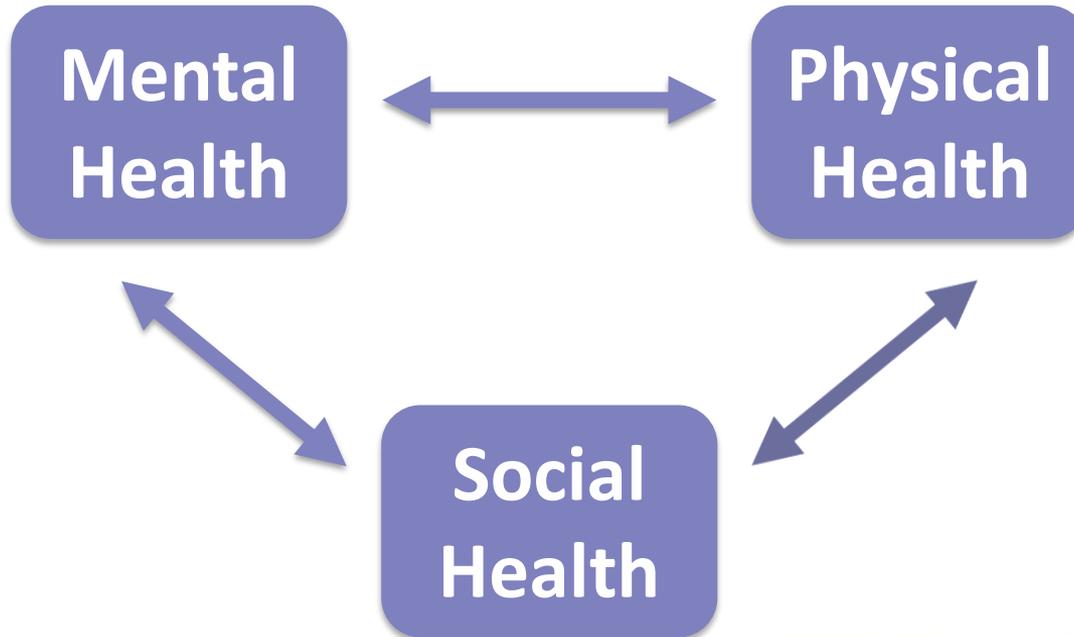


- GLOBAL Guidelines
 - Clinicians should perform a review of behavioral, functional, adaptive, and psychosocial factors on an annual basis.
 - When concern for a mental health disorder in adults with Down syndrome is present:
 - Medical professionals should evaluate for medical conditions that may present with psychiatric and behavioral symptoms.

Tsou et al. 2020

Prevention and treatment

- Connection between physical, mental, and social health



Prevention and treatment

- Importance of sleep
- Be aware of “empathy radar” or “social sensitivity”
- Social skills
- Work with a mental health provider
 - Consider therapies (e.g., music, art)
- Medications



Mental Health Resources

- [Behavior Changes in Adolescents & Adults with DS Webinar](#)
- [Down Syndrome and Mental Health](#)
- [Mental Wellness and Aging in Adults with DS Webinar](#)
- [Social Sensitivity](#)
- [“Self-Talk” in Adults with Down Syndrome](#)

Alzheimer's disease and dementia

- More common in people with Down syndrome
 - Uncommon before age 40.
 - Incidence estimated to be 55% in those between ages 50-59.
 - Incidence estimated to be greater than 75% in those 60 years of age and older.
- Why?
 - One of the genes associated with Alzheimer's disease is on the 21st chromosome.
 - Amyloid precursor protein (APP)

Coppus et al. 2006 McCarron et al. 2017 Zis & Strydom 2018

Alzheimer's disease and dementia

- Symptoms
 - Behavioral/psychological/personality changes
 - Memory
 - Loss of skills
 - Incontinence
 - Gait changes
 - Swallowing problems

Alzheimer's disease and dementia



- GLOBAL Guidelines
 - For adults with Down syndrome younger than age 40, be cautious when diagnosing age-related, Alzheimer's type dementia.
 - Assess baseline function each year beginning at age 40.
 - NTG-EDSD: National Task Group – Early Detection Screen for Dementia

ntg
National Task Group
on Early Detection of Dementia

NTG-EDSD

4/2020.1

The NTG-EDSD is a screening tool for dementia, adapted from the DSQIID[®], can be used for the early detection screening of those adults with an intellectual disability who are suspected of or may be showing early signs of mild cognitive impairment or dementia. The NTG-EDSD is not an assessment or diagnostic instrument, but an administrative screen that can be used by staff and family caregivers to note functional decline and health problems and record information useful for further assessment, as well as to serve as part of the mandatory cognitive assessment review that is part of the Affordable Care Act's annual wellness visit for Medicare recipients. This instrument complies with Action 2.B of the US National Plan to Address Alzheimer's Disease.

It is recommended that this instrument be used on an annual or as indicated basis with adults with Down syndrome beginning with age 40 and with other at-risk persons with intellectual or developmental disabilities when suspected of experiencing cognitive change. The form can be completed by anyone who is familiar with the adult (that is, has known him or her for over six months), such as a family member, agency support worker, or a behavioral or health specialist using information derived by observation or from the adult's personal record.

The estimated time necessary to complete this form is between 15 and 60 minutes. Some information can be drawn from the individual's medical/health record. Consult the NTG-EDSD Manual for additional instructions (www.aadmd.org/ntg/screening).

(1) File #: _____ (2) Date: _____
Name of person: (3) First _____ (4) Last: _____
(5) Date of birth: _____ (6) Age: _____
(7) Sex:

Female
Male

(8) Best description of level of intellectual disability

No discernible intellectual disability
Borderline (IQ 70-75)
Mild ID (IQ 55-69)
Moderate ID (IQ 40-54)
Severe ID (IQ 25-39)
Profound ID (IQ 24 and below)
Unknown

(9) Diagnosed condition (check all that apply)

Autism
Cerebral palsy
Down syndrome
Fragile X syndrome
Intellectual disability
Prader-Willi syndrome
Other: _____

Instructions:
For each question block, check the item that best applies to the individual or situation.

Current living arrangement of person:

<input type="checkbox"/> Lives alone
<input type="checkbox"/> Lives with spouse or friends
<input type="checkbox"/> Lives with parents or other family members
<input type="checkbox"/> Lives with paid caregiver
<input type="checkbox"/> Lives in community group home, apartment, supervised housing, etc.
<input type="checkbox"/> Lives in senior housing
<input type="checkbox"/> Lives in congregate residential setting
<input type="checkbox"/> Lives in long term care facility
<input type="checkbox"/> Lives in other: _____

Tsou et al. 2020

Prevention and treatment

- Prevention – possibly exercise and brain stimulation
- Medications
 - Cholinesterase inhibitors (e.g., donepezil / Aricept)
 - NMDA receptor antagonist (memantine / Namenda)

Treat associated symptoms

- Depression
- Anxiety
- Agitation
- Sleep challenges
 - E.g., day/night reversals
- Medication choices are influenced by a patient's particular symptoms and the particular effects and side effects of the medication.
- Observation and report of symptoms are key to assisting with medication selection.

Supporting an individual with AD

- Quality of life
 - Doing what can still be done
 - “Bingo Pace”
- Communication
 - Simple instructions, do not argue, avoid asking them if they remember, get down on their level
- Home safety and adaptive equipment

Alzheimer's Disease Resources



- [List of resources on Alzheimer's disease and Down syndrome](#)
 - [Aging and Alzheimer's Disease in Adults with Down Syndrome Webinar](#)
 - [Alzheimer's Disease & Down Syndrome: A Practical Guidebook for Caregivers](#)
 - [Living with Dementia](#)
 - [Alzheimer's Disease Video Journal](#)
 - [NTG-EDSD - Early Detection and Screen for Dementia](#)

Thyroid conditions

- Hypothyroidism
 - Underactive thyroid
 - Symptoms: fatigue, dry skin, constipation, cognitive impairment, depression, weight gain
- Hyperthyroidism
 - Overactive thyroid
 - Symptoms: fatigue, anxiety, agitation, sweating, tremor, weight loss

Thyroid conditions



- GLOBAL Guidelines

- Screen for hypothyroidism every 1-2 years beginning at age 21.

- Serum thyroid-stimulating hormone (TSH) test

Tsou et al. 2020

Treatment

- Hypothyroidism
 - Thyroid replacement medication (levothyroxine, Armour thyroid)
- Hyperthyroidism
 - Medications
 - Radioactive iodine
 - Surgery

Diabetes

- Type 1 diabetes is more common
- Type 2 diabetes is more common (?)
 - Estimates vary

Capone et al. 2018 Tsou et al. 2020

Diabetes



- GLOBAL Guidelines
 - For adults without symptoms:
 - Screen for type 2 diabetes every 3 years beginning at age 30.
 - For adults with comorbid obesity:
 - Screen for type 2 diabetes every 2-3 years beginning at age 21.
 - Hemoglobin A1c (HbA1c) or fasting plasma glucose

Tsou et al. 2020

Prevention and treatment

- Type 1 diabetes
 - No known prevention
 - Treatment: insulin
- Type 2 diabetes
 - Diet, exercise
 - Metformin?
 - Treatment: oral or injectable medications and sometimes insulin



Endocrine Resources

- [Diabetes in Adults with Down Syndrome](#)
- [Hypothyroidism](#)
- [Thyroid, Weight, and Metabolism](#)

Cardiovascular disease

- Hypertension
- Atherosclerotic disease
- Vasovagal syncope

Cardiovascular disease



- GLOBAL Guidelines
 - For adults with Down syndrome without a history of atherosclerotic cardiovascular disease (ASCVD):
 - Assess the appropriateness of statin therapy every 5 years starting at age 40
 - Use a 10-year risk calculator
 - This is the same recommendation for adults without Down syndrome (U.S. Preventive Services Task Force)

Tsou et al. 2020

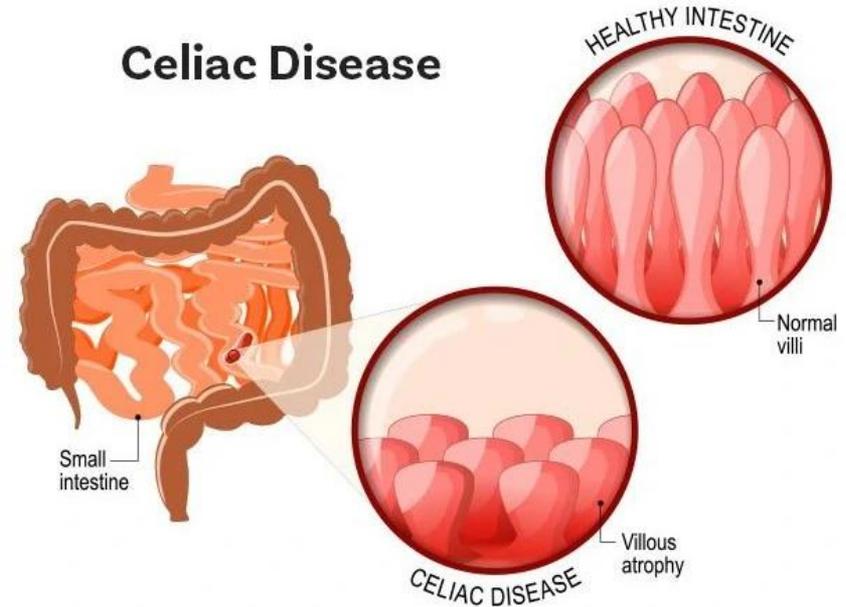
Cardiology Resources



- [Heart Disease in Adults with DS Between 1996 and 2016](#)
- [Vasovagal Syncope](#)

Celiac disease

- More common
- Symptoms
 - Direct symptoms
 - Symptoms secondary to vitamin/mineral deficiencies



Source: [Celiac Disease Foundation](https://www.celiac.org/)

Celiac disease



- GLOBAL Guidelines

- Do an annual assessment for gastrointestinal and non-gastrointestinal signs and symptoms of celiac disease

- Use targeted history, physical examination, and clinical judgement of good practice.

Tsou et al. 2020



Gastroenterology Resources

- [Celiac Disease](#)
- [Constipation in Down Syndrome](#)
- [Gastroesophageal Reflux Disease](#)
- [Treating Gastroesophageal Reflux Disease with Surgery](#)

Skin, hair, and nail conditions

- Dry skin (xerosis)



Source: [American Academy of Dermatology](#)

Skin, hair, and nail conditions

- Folliculitis
- Boils
- Athlete's foot
- Fungal infections of the nails



Source: [DermNet NZ](#)



Source: [NHS](#)



Source: [Nemours](#)



Source: [Mayo Clinic](#)



Skin, Hair, & Nail Resources

- [Skin, Hair, and Nail Concerns in People with DS Presentation](#)
- [Folliculitis and Boils in People with Down Syndrome](#)
- [Lotion Options](#)
- [Seborrheic Dermatitis](#)
- [Ringworm, Athlete's Foot, and Jock Itch](#)
- [Onychomycosis – Toenail Fungal Infection](#)

Cancer

- MORE common – leukemia, testicular cancer
- LESS common – most solid tumor cancers (breast, colon, etc.)

Published: 31 March 2016

Low risk of solid tumors in persons with Down syndrome

Henrik Hasle MD, PhD , Jan M. Friedman MD, PhD, Jørgen H. Olsen MD, DMSc & Sonja A. Rasmussen MD, MS

Genetics in Medicine 18, 1151–1157(2016)

JIDR Journal of Intellectual Disability Research
Published on behalf of mencap and in association with IASSIDD

The profile and incidence of cancer in Down syndrome

S. G. Sullivan , R. Hussain, E. J. Glasson, A. H. Bittles

First published: 08 December 2006 | <https://doi.org/10.1111/j.1365-2788.2006.00862.x>



European Journal of Medical Genetics

Volume 63, Issue 4, April 2020, 103783



Cancer screening in adults with down syndrome, a proposal

Marie-Odile Rethoré ^{a, b}, Jacques Rouëssé ^b, Da

American Journal of Medical Genetics 78:207–216 (1998)

A Tumor Profile in Down Syndrome

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Cancer

- Implications for future guidelines
- How does this affect screening?
- Should my loved one be screened?
 - Discuss with healthcare provider.

Screening

Consider the potential risks and benefits.

Questions to ask

- What does the procedure involve?
- What do follow-up procedures involve?
- Is anesthesia required?
- What is your family history?



Cancer Resources

- [Testicular Cancer](#)
- [Colon Cancer Screening for People with Down Syndrome](#)
- [Prostate Cancer Screening in Men with Down Syndrome](#)
- [Breast Cancer and Breast Cancer Screening for Women with DS](#)
- [Low Risk of Solid Tumors in Persons with Down Syndrome](#)

Respiratory infections

- Influenza (flu)
- Pneumonia
- Presentation
 - Confusion or change in mental status
- Prevention
 - Consider earlier vaccination for pneumococcal pneumonia



Respiratory Resources

- [Recurrent Pneumonia](#)
- [Pneumococcal Vaccine](#)
- [Flu Vaccine Information for People with Down Syndrome](#)

COVID-19

- In people with Down syndrome age 40 and older, the risk for severe complications from COVID-19 is increased.
- In people with Down syndrome younger than 40 *with* co-occurring conditions, the risk is also increased.



Resources

- [List of COVID-19 Resources](#)
 - [COVID and Down Syndrome Webinar](#)
 - [T21RS COVID-19 and Down Syndrome Survey](#)
 - [DSMIG-USA IDD COVID-19 Vaccination Position Statement](#)
 - [Routines during the Pandemic](#)

Questions?

Resource Library

adsresources.advocatehealth.com

Facebook

www.facebook.com/adultdownsyndromecenter

E-mail Newsletter

<http://eepurl.com/c7uV1v>

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