

Physical and Mental Health Conditions of Adults with Down Syndrome

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Down Syndrome Connection of the Bay Area Conference






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Medical
Group


Adult Down
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1610 Luther Lane




Resource Library





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<https://adsresources.advocatehealth.com/>

Please note:

- This presentation is intended for families, caregivers, health care professionals, and service providers of individuals with Down syndrome.
- The information in this presentation 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.
- We are unable to provide diagnosis or treatment recommendations specific to an individual. We recommend that you bring specific questions about an individual with Down syndrome to their medical and/or therapy professionals.

Objectives

- Identify common and uncommon physical and mental health conditions of adults with Down syndrome.
- Describe unique aspects of diagnosing and treating health conditions of individuals with Down syndrome.
- Share resources on health of individuals with Down syndrome.

Reminders

**Each person with
Down syndrome
is unique.**

**Many people with
Down syndrome
share common
characteristics.**

Cognitive and behavioral

- Better receptive language than expressive language
- Concrete thinking
- Visual memory
- Self-talk and imaginary friends
 - Change in quality or frequency may be a sign of stress or mental illness
- The "Groove"
- Empathy radar

THE "GROOVE"

The "groove" is a preference for **sameness, repetition, and routine.**

Possible Advantages

The groove can...

- Give structure and order to daily life
- Support successful completion of tasks
- Increase independence
- Help manage stress

Possible Disadvantages

The groove can make it difficult to...

- Be flexible
- Transition from one task to the next
- Deal with changes
- Apply skills across different settings



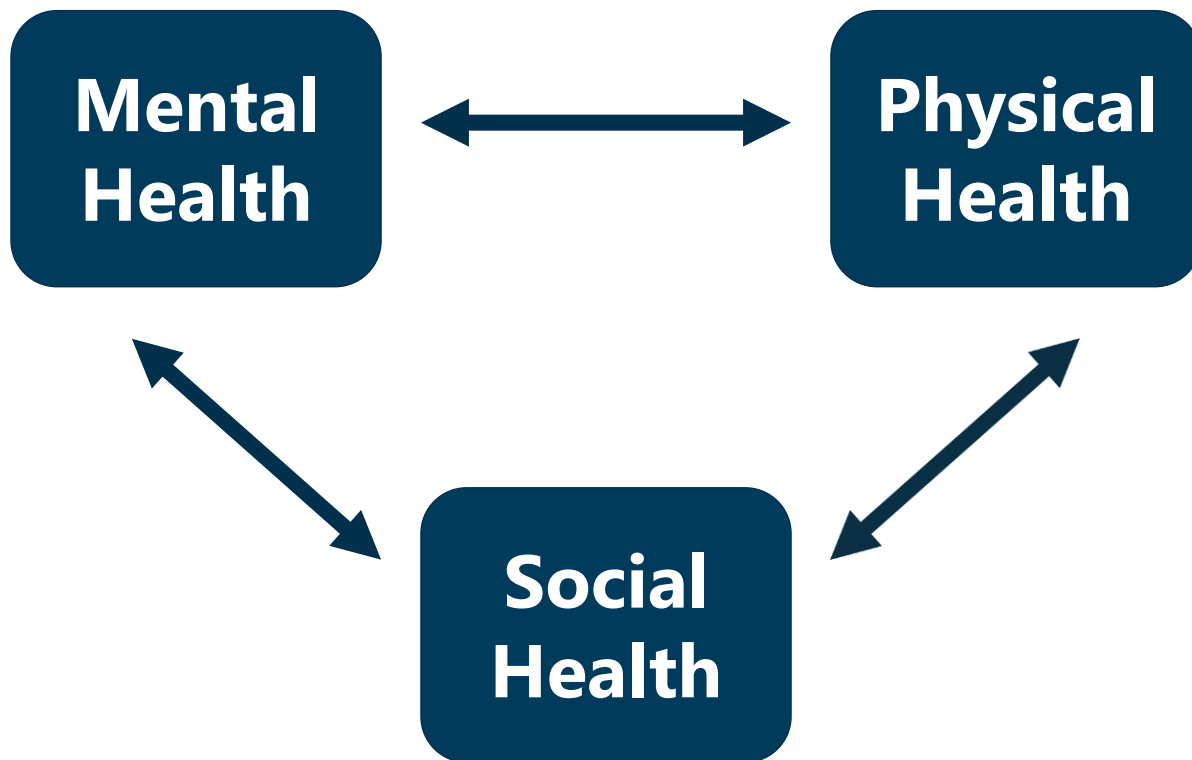
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**Any and all behavior
change should be
viewed as a possible
communication tool.**



Rosa

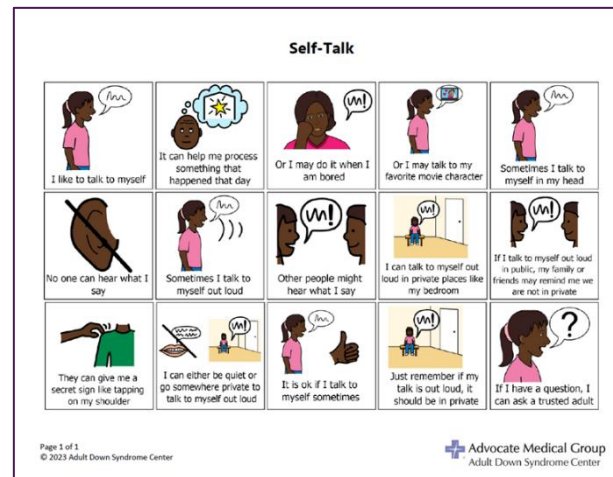
Age: 43

- Had become mildly aggressive
- Lived in a group home with several other women
- Behavior occurred after dinner during chore time
 - Chores rotated among housemates
 - If Rosa wasn't assigned to sweep, she took the broom from the assigned housemate
- New chore schedule
 - Housemates didn't mind that she wanted to sweep



Resources

- [Behavior Change](#) (article)
- [Diagnostic Overshadowing](#) (article)
- [Misunderstandings Caused by Different Perceptions](#) (article)
- [Self-Talk](#) (article)
- [Self-Talk](#) (webinar)
- [The Groove](#) (article)
- [The Groove](#) (webinar)
- [Two Syndromes](#) (article)



[Self-Talk Visual](#)

Health conditions

**Each person with
Down syndrome
has a unique
health history.**

**Many people with
Down syndrome
have 1 or more
common health
conditions.**

Data

CME Accredited

ORIGINAL RESEARCH

Prevalence of Common Disease Conditions in a Large Cohort of Individuals With Down Syndrome in the United States

Brian Chicoine, MD,^{1,3} Anne Rivelli, MPH, MA,^{1,2} Veronica Fitzpatrick, DrPH,^{1,2} Laura Chicoine, BA,^{1,3} Gengjie Jia, PhD,^{4,5} Andrey Rzhetsky, PhD^{4,5}

¹Advocate Aurora Health, Downers Grove, IL; ²Advocate Aurora Research Institute, Downers Grove, IL; ³Advocate Medical Group Adult Down Syndrome Center, Park Ridge, IL; ⁴Department of Medicine, University of Chicago, Chicago, IL; ⁵Institute of Genomics and Systems Biology, University of Chicago, Chicago, IL

BRIEF REPORT

Prevalence of Endocrine Disorders Among 6078 Individuals With Down Syndrome in the United States

Anne Rivelli, MA, MPH,^{1,2} Veronica Fitzpatrick, DrPH,^{1,2} Danielle Wales, BS,³ Laura Chicoine, BA,^{1,4} Gengjie Jia, PhD,⁵ Andrey Rzhetsky, PhD,⁴ Brian Chicoine, MD^{1,4}

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BRIEF REPORT

Prevalence of Infectious Diseases Among 6078 Individuals With Down Syndrome in the United States

Veronica Fitzpatrick, DrPH,^{1,2} Anne Rivelli, MA, MPH,^{1,2} Sagar Chaudhari, MA,³ Laura Chicoine, BA,^{1,4} Gengjie Jia, PhD,⁵ Andrey Rzhetsky, PhD,⁴ Brian Chicoine, MD^{1,4}

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BRIEF REPORT

Prevalence of Mental Health Conditions Among 6078 Individuals With Down Syndrome in the United States

Anne Rivelli, MA, MPH,^{1,2} Veronica Fitzpatrick, DrPH,^{1,2} Sagar Chaudhari, MA,³ Laura Chicoine, BA,^{1,4} Gengjie Jia, PhD,⁵ Andrey Rzhetsky, PhD,⁴ Brian Chicoine, MD^{1,4}

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JAMA[®]
Journal of the
American Medical Association

<p>Research</p> <p>1. Effect of a Novel Intervention on the Prevalence and Incidence of Mental Health Conditions in Children With Down Syndrome: A Systematic Review 2. Prevalence of Common Disease Conditions in a Large Cohort of Individuals With Down Syndrome in the United States 3. Prevalence of Endocrine Disorders Among 6078 Individuals With Down Syndrome in the United States 4. Prevalence of Infectious Diseases Among 6078 Individuals With Down Syndrome in the United States 5. Prevalence of Mental Health Conditions Among 6078 Individuals With Down Syndrome in the United States</p>	<p>Original Research</p> <p>1. Effect of a Novel Intervention on the Prevalence and Incidence of Mental Health Conditions in Children With Down Syndrome: A Systematic Review 2. Prevalence of Common Disease Conditions in a Large Cohort of Individuals With Down Syndrome in the United States 3. Prevalence of Endocrine Disorders Among 6078 Individuals With Down Syndrome in the United States 4. Prevalence of Infectious Diseases Among 6078 Individuals With Down Syndrome in the United States 5. Prevalence of Mental Health Conditions Among 6078 Individuals With Down Syndrome in the United States</p>	<p>Clinical Research & Education</p> <p>1. Effect of a Novel Intervention on the Prevalence and Incidence of Mental Health Conditions in Children With Down Syndrome: A Systematic Review 2. Prevalence of Common Disease Conditions in a Large Cohort of Individuals With Down Syndrome in the United States 3. Prevalence of Endocrine Disorders Among 6078 Individuals With Down Syndrome in the United States 4. Prevalence of Infectious Diseases Among 6078 Individuals With Down Syndrome in the United States 5. Prevalence of Mental Health Conditions Among 6078 Individuals With Down Syndrome in the United States</p>
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AMERICAN MEDICAL ASSOCIATION



Chicoine et al. 2021, Fitzpatrick et al. 2022, Rivelli et al. 2022, Tsou et al. 2020

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Common health conditions

- Thyroid disorders
- Obsessive-compulsive disorder
- Depression
- Obesity
- Pneumonia
- Swallowing dysfunction
- Skin conditions (e.g., folliculitis)
- Gum disease
- Gastroesophageal reflux (GERD)
- Celiac disease
- Constipation
- Flat feet and overpronation
- Seizures
- Atlantoaxial instability
- Vision and hearing problems
- Sleep apnea
- Congenital heart disease
- Alzheimer's disease

Less common health conditions

- Many solid tumor cancers
- Atherosclerotic disease ("hardening of the arteries," plaques in the arteries)
- Myocardial infarctions (heart attacks)
- Hypertension (high blood pressure)



Resources

- [At-Home Treatments for Common Health Conditions of People with Down Syndrome](#) (webinar)
- [Common and Uncommon Health Conditions in Adolescents and Adults with Down Syndrome](#) (webinar)
- [Global Medical Care Guidelines for Adults with Down Syndrome](#) (Global Down Syndrome Foundation)
- [Health Supervision for Children and Adolescents with Down Syndrome](#) (American Academy of Pediatrics)

GENERAL

Vital signs

Differences

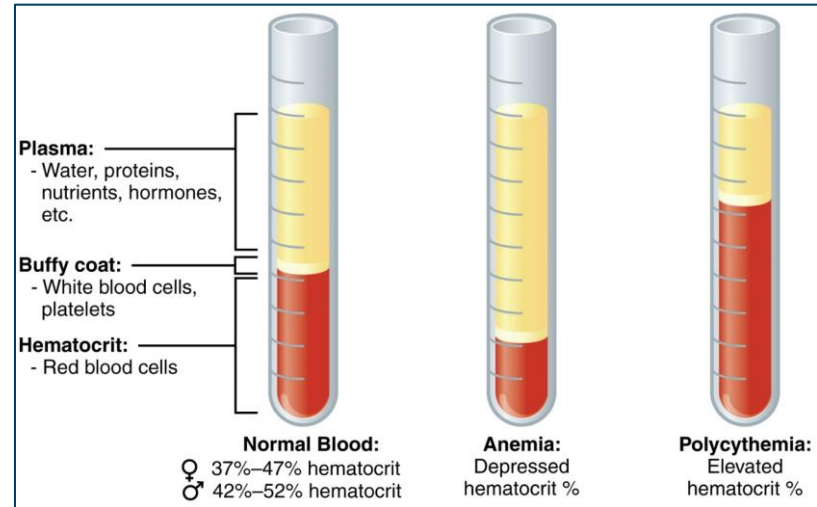
- Lower heart rate (pulse)
- Lower blood pressure
- Body temperature



Blood

More common

- Polycythemia (high red blood cell count)
- Macrocytosis (large red blood cells)
- Neutropenia (low white blood cell count)
- Abnormal blood clotting?



Composition of blood

Anatomy and Physiology, [OpenStax](#)

Nutrition

More common

- Vitamin deficiencies (e.g., vitamin B12)
- Folate metabolism

Pain

- People with Down syndrome experience pain.
 - Some people with Down syndrome report it less often than people without Down syndrome.
 - May have difficulty localizing pain.
 - May have delayed response to pain.
- Some people with Down syndrome appear to experience greater pain than people without Down syndrome.

Joshua

Age: 28

- Intermittently getting upset
- Can last for a few hours
- Sometimes seems to occur after eating
- Developed episodes of vomiting
- Ultrasound demonstrated gallstones
- Vomiting and behavioral changes resolved after removal of gall bladder



Resources

- [Differences in Red Blood Cells](#) (article)
- [Fainting](#) (article)
- [Heart Rates and Exercise](#) (article)
- [High and Low Blood Pressure](#) (article)
- [Normal Abnormal Lab Values](#) (article)
- [Pain](#) (article)
- [Polycythemia](#) (article)
- [Temperature Regulation](#) (article)
- [The Recipe for Health](#) (webinar)
- [Vitamin B12 and Folate](#) (article)

MENTAL HEALTH

Mental health

More common

- Depression
- Obsessive-compulsive disorder
- Impulse control disorder
- Attention deficit disorder

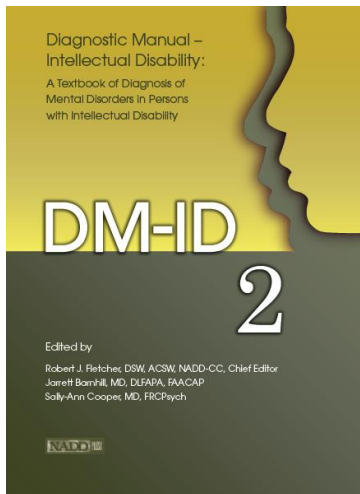
Less common

- Substance use/abuse
- Anxiety disorder?
- Posttraumatic stress disorder?

Mental health

Presentation

- Communication
- Behavior
- Self-report
- Introspection
- Impulse control



[Diagnostic Manual – Intellectual Disability \(DM-ID-2\)](#)

Screening/diagnosis

- Behavioral changes
- Changes in routines
- Underlying physical health conditions

Symptoms of depression

- Persistent feelings of sadness and/or decreased interest in things the individual previously enjoyed
- Depressed or irritable mood
- Loss of interest or pleasure in activities
- Weight loss or gain
- Change in sleep patterns
- Slowing down of physical movement or restlessness
- Fatigue
- Feelings of worthlessness
- Decreased concentration
- Recurrent thoughts of death
- Psychotic features (extreme withdrawal, hallucinatory self-talk, etc.)
- Inappropriate fears or avoidances of people/things
- Strong refusal to leave the home

Symptoms of anxiety disorder

- Anxiety that interferes with day-to-day life on a long-term basis
- Feeling worried or anxious most days
- Mild self-injurious behavior
- Being easily fatigued
- Difficulty sleeping
- Restlessness
- Difficulty concentrating
- Muscle tension
- Irritability

Obsessive-compulsive disorder

- Obsessions – persistent thoughts or thoughts that preoccupy the mind
- Compulsions – repetitive behaviors or acts that one feels compelled to perform
- Individual with Down syndrome often is not upset by the thoughts/behaviors
- May get upset if the behaviors are blocked or prevented
- Impairs function
- Cannot participate in usual activities due to being “stuck”

Mental health

Treatment

- Counseling
- Art therapy
- Occupational therapy
- Animal-assisted therapy
- Music therapy
- Medications



Lexi

Age: 22

- Aged out of school, started to gain weight
- Had a disagreeable mood and difficulty sleeping
- History, physical exam, labs, sleep study (negative)
- Initial diagnosis: adjustment reaction with depressed mood and sleep issues
 - Established new routine, counseling, sleep hygiene
- Didn't respond; diagnosed with depression
 - Bupropion (Wellbutrin) for depression
 - Melatonin for sleep



Mental Health

- [Behavior Changes](#) (webinar)
- [Depression](#) (article)
- [Generalized Anxiety Disorder](#) (article)
- [How to Cope with Stress](#) (article)
- [Post-Traumatic Stress Disorder](#) (article)
- [Promoting Mental Health Across the Lifespan](#) (webinar)
- [Psychotherapy](#) (article)



[Mental Health Visuals and Videos](#)

Topics

- **Endocrine & weight**
 - Thyroid conditions, diabetes, obesity
- **Gastrointestinal**
 - Reflux, celiac disease, constipation
- **Musculoskeletal**
 - Joint laxity, flat feet, atlantoaxial instability
- **Nose, mouth & upper airway**
 - Swallowing problems
- **Sensory**
 - Vision and hearing, sensory processing
- **Respiratory**
 - Pneumonia, sleep apnea
- **Down syndrome regression disorder**
- **Neurology**
 - Seizures, Alzheimer's disease
- **Urinary**
 - Urinary retention, UTIs
- **Gynecology**
 - Menstruation, menopause
- **Heart & vascular system**
 - Heart disease, embolic strokes
- **Skin & nail conditions**
 - Alopecia, dry skin, folliculitis/boils
- **Cancer**
 - Solid tumor cancers
- **Wrap-up**

ENDOCRINE & WEIGHT

Endocrine and weight

More common

- Hypothyroidism – underactive thyroid
- Hyperthyroidism – overactive thyroid
- Type 1 diabetes mellitus
- Obesity and overweight
- Adrenal insufficiency
- Type 2 diabetes mellitus
 - More common age 5 to 34 years of age; less common overall

Hypothyroidism

- Underactive thyroid
- Symptoms
 - Weakness, fatigue
 - Dry, pale, and/or cold skin
 - Mood changes, depression
 - Forgetfulness, difficulty with thinking process
 - Puffy eyelids and/or face
 - Constipation
 - Weight gain
 - And others
- Screening/diagnosis
 - Thyroid blood tests of TSH and possibly T4 (less commonly T3)
 - Screen adults with Down syndrome every 1 to 2 years using a TSH test
- Treatment
 - Medication (levothyroxine, Synthroid, Levoxyl)
 - Check TSH level 6-8 weeks after starting medication or changing dose

Overweight and obesity

- Weight of adults with Down syndrome in 2020 study
 - 27.3% - BMI in overweight range
 - 53.8% - BMI in obesity range
- Lower resting metabolic rate
- Hormonal differences
- Co-occurring conditions
- Factors that affect weight
 - Genetics
 - Hormones
 - Sleep
 - Physical activity
 - Diet
 - Stress
 - Some medications
 - Life changes

Overweight and obesity

- Screening/diagnosis
 - Monitor for weight change and obesity annually by calculating BMI
- Treatment
 - Lifestyle (e.g., diet, physical activity, hydration, stress, sleep)
 - Medications
 - Surgery



[Healthy Lifestyle Visuals and Videos](#)

Lisa

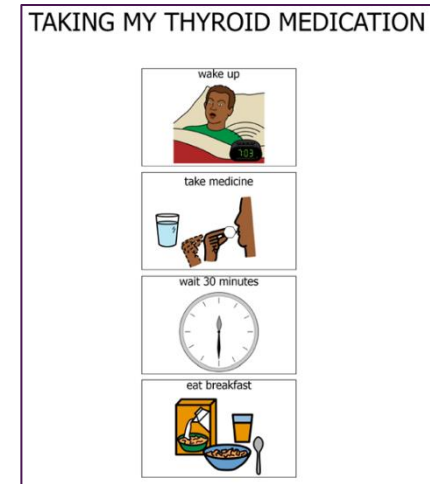
Age: 31

- "Nervousness" and increasing compulsive behavior - repeatedly straightening objects in her bedroom making it difficult to leave the house
- Labs showed TSH low and T4 high
- Diagnosis: hyperthyroidism
- Improved with treatment
- Six months later
 - Depressed mood
 - High TSH and low T4
 - Diagnosis: hypothyroidism
 - Fluctuation back and forth
 - Radioactive ablation of thyroid and thyroid replacement medication (levothyroxine)



Resources

- [Diabetes](#) (article)
- [Gout](#) (article)
- [Hypoglycemia](#) (article)
- [Hypothyroidism and Hyperthyroidism](#) (article)
- [Thyroid, Weight, and Metabolism](#) (article)
- [Weight Management](#) (article)



[Thyroid Visuals](#)

GASTROINTESTINAL

Gastrointestinal

More common

- Gastroesophageal reflux disease (GERD, heartburn)
- Celiac disease
- Constipation
- Motility problems
- Achalasia
- Gall bladder disease
- Hernias
- Fatty liver

Gastroesophageal reflux disease

- GERD, heartburn, reflux
- Backward flow of stomach acid into the esophagus
- Risk factors
 - Obesity
 - Sleep apnea
 - Overeating
 - Consuming caffeine, spicy foods, carbonated beverages
 - Lying down after eating

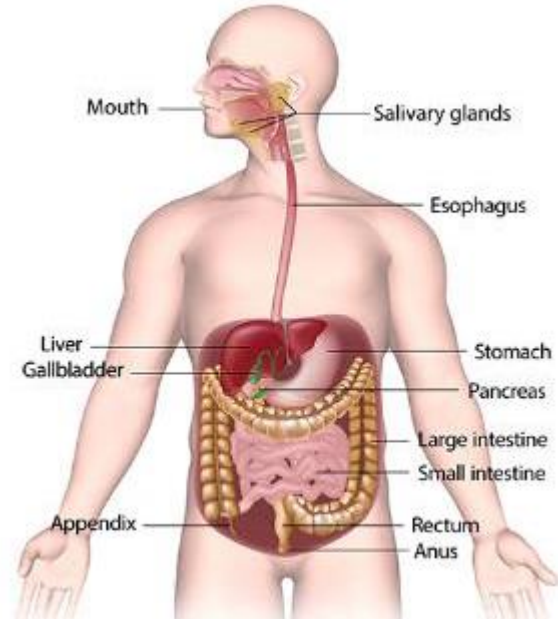


Image from [National Institute of Diabetes and Digestive and Kidney Diseases, National Institutes of Health](#)

Gastroesophageal reflux disease

- Symptoms

- Chest pain, abdominal pain
- Sore throat
- Regurgitation of digested food
- Difficulty swallowing
- Aspiration (breathing food into the lungs)
- Chronic cough
- Bronchospasm (asthma)
- Loss of enamel on the teeth
- Change in behavior

- Possible complications

- Barrett's esophagitis
- Scarring of the esophagus

- Diagnosis

- Clinical diagnosis
- Upper endoscopy (EGD)
- Upper GI (barium)

Gastroesophageal reflux disease

- Treatment

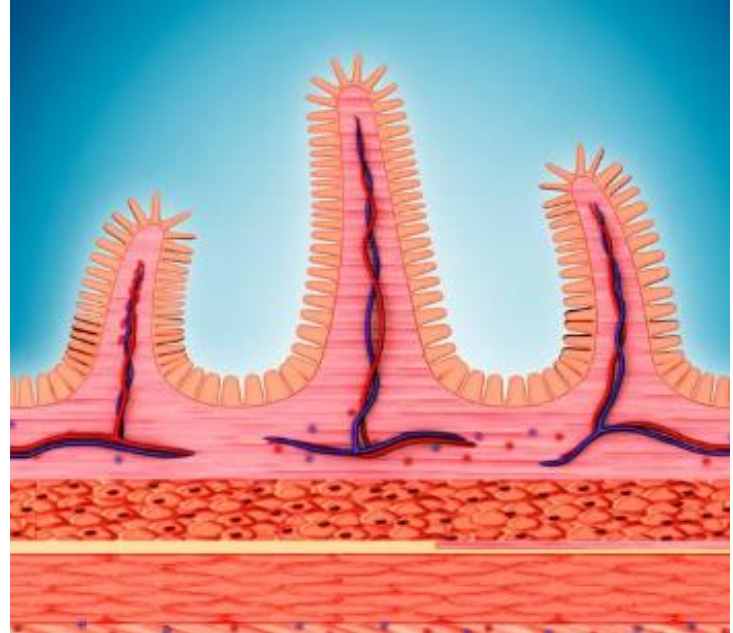
- Avoid overeating
- Reach/maintain ideal body weight
- Limit caffeine and carbonated drinks
- Avoid lying down after a meal
- Avoid clothing that constricts the abdomen
- Elevate the top of the bed
- Treat sleep apnea

- Medications

- Antacids (e.g., Tums)
- H2 blockers (e.g., ranitidine)
- Combination (e.g., Pepcid Complete)
- Proton pump inhibitors (PPI) (e.g., omeprazole)
- Metoclopramide

Celiac disease

- Sensitivity to gluten that causes an inflammatory response
- Damages villi →
- Affects absorption of nutrients
- Symptoms
 - Diarrhea
 - Bloating
 - Weight loss
 - Mood and behavior change



Celiac disease

- Diagnosis
 - History and physical exam
 - Blood test
 - Anti-tissue transglutaminase IgA
 - Total IgA
 - Genetic testing
 - Endoscopy (EGD) with biopsy
- Treatment
 - Gluten-free diet
 - Supplements
 - Dietician consult
- Screening
 - Guidelines for adults with Down syndrome: annual assessment for GI and non-GI signs and symptoms of celiac disease
 - Guidelines for children with Down syndrome: review for symptoms at each health supervision visit

Constipation

- Change in the frequency, size, consistency, and/or ease of bowel movements
- Symptoms
 - Infrequent, hard, or difficult to pass bowel movements
 - Diarrhea
 - Bloating
 - Abdominal discomfort
 - Pain with having a bowel movement
 - Fecal impaction
- Diagnosis
 - History and physical
 - X-ray
 - Colonoscopy
 - Celiac testing
- Treatment and prevention
 - Fluids
 - Exercise
 - Fiber
 - Medications

Jim

Age: 35

- Depressed mood, reduced verbal abilities, change in his ability to complete ADLs
- History and physical exam, lab testing, OT assessment
- "Does he have Alzheimer's disease?"
- Diagnosed with celiac disease, vitamin B12 deficiency, and depression
- Treatment
 - Gluten-free diet
 - Occupational therapy
 - Medications



Resources

- [Celiac Disease](#) (article)
- [Constipation](#) (article)
- [Gallstones](#) (article)
- [Gastroesophageal Reflux Disease](#) (article)
- [Gastrointestinal Health](#) (webinar)
- [Hernias](#) (article)



[Celiac Disease Visuals](#)

MUSCULOSKELETAL

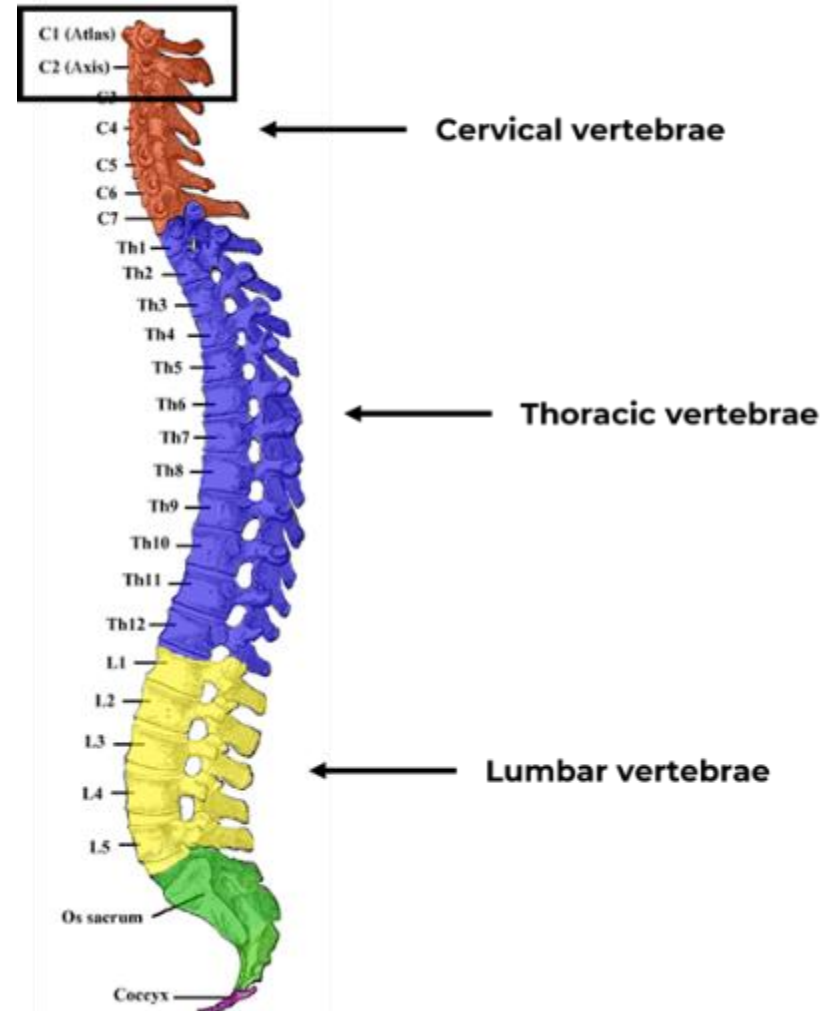
Musculoskeletal

More common

- Atlantoaxial instability (AAI), cervical myelopathy
- Autoimmune arthritis
- Joint laxity and subluxation
- Flat feet and overpronation
- Gout
- Osteoarthritis?

AAI

- Atlantoaxial instability
- Abnormal movement between the 1st and 2nd vertebrae in the neck
- Symptoms
 - Pain in the neck or head
 - Holding the head in unusual positions
 - Difficulty holding the head up
 - Weakness of the arms and/or legs
 - Incontinence (urine or stool)
 - Not wanting to participate in activities



AAI

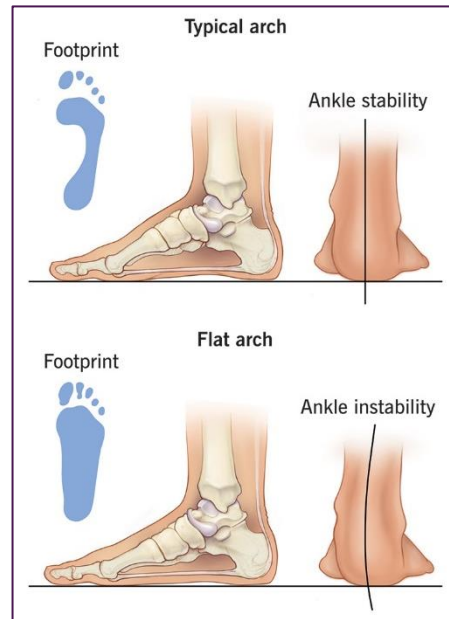
- Screening
 - Annual assessment for symptoms and physical findings for spine and nerve compression
 - Increased deep tendon reflexes
 - Clonus
 - Abnormal plantar reflex
 - Muscle weakness
 - Period neck x-rays are NOT recommended unless a person has symptoms

AAI

- Diagnosis
 - Neck x-rays
 - MRI
 - CT scan
 - Electromyogram (EMG)
- Prevention
 - Avoid trampolines
 - Teach methods to reduce injury when playing contact sports
 - Anesthesia – do not use posterior extension for placement of breathing tube
- Treatment
 - Observation through regular exams and possibly imaging
 - Surgery

Overpronation & flat feet

- Overpronation: feet/ankle roll inward too far
- Flat feet: fallen arches of the feet
- Symptoms
 - Foot/ankle pain
 - Bunions
 - Plantar fasciitis
 - Shin splints
 - Knee, hip, and/or back pain



[Image from Cleveland Clinic](#)

Overpronation & flat feet

- Diagnosis
 - Physical exam
- Treatment
 - Treat associated symptoms – icing, nonsteroidal anti-inflammatory medications (e.g., ibuprofen, naproxen, aspirin)
 - Supportive shoes
 - Orthotics/inserts
 - Superfeet, Sole, Spenco, etc.
 - Custom
 - Physical therapy



Image from [Superfeet](#)

John

Age: 17

- Teachers and parents concerned that he might be depressed
- Would put his head on the desk and not participate in class (more as the day progressed)
- Physical exam found increased reflexes
- Imaging of neck demonstrated AAI
- Surgery to correct
- With physical therapy, his strength improved, and symptoms thought to be depression resolved



Resources

- [Anesthesia](#) (article)
- [Atlantoaxial Instability](#) (article)
- [Changes in Gait](#) (article)
- [Connective Tissue](#) (article)
- [Ligamentous Laxity](#) (article)
- [Overpronation](#) (article)

SENSORY

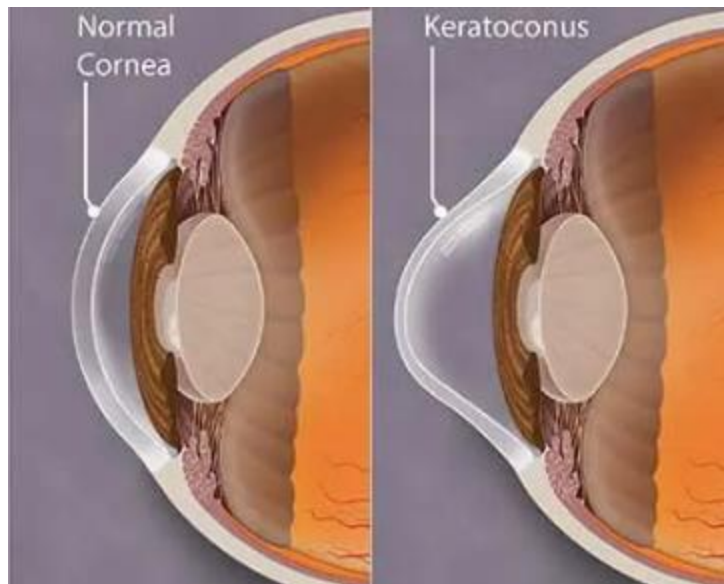
Sensory

More common

- Vision impairment
- Cerebral vision impairment
- Depth perception differences
- Hearing impairment
- Sensory processing differences

Vision

- Myopia – near-sightedness
- Hyperopia – far-sightedness
- Keratoconus
- Cataracts
- Blepharitis
- Conjunctivitis
- Cerebral vision impairment
 - Depth perception differences



[Image from American Academy of Ophthalmology](#)

Hearing

- Hearing impairment
- Cerumen impaction
- Recurrent infections
- Middle ear complications
 - Cholesteatomas
 - Otosclerosis (abnormal bone growth)
 - Persistent fluid

Sensory processing

- “Problems with the ability to process information received through the senses which impact the ability for a person to function in their daily life” (Katie Frank, PhD, OTR/L)
- 8 senses
 - Touch, sight, smell, sound, taste
 - Vestibular, proprioception, interoception

Sensory Activities & Accommodations



Examples

- Headphones
- Air purifiers
- Weighted objects
- Vibration
- Joint compression
- Physical activity
- TheraPutty
- Massage

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Adult Down Syndrome Center

[Sensory Resources](#)

Lisa

Age: 27

- Needs blood drawing and is afraid
- Gets hot and sweats when just discussing blood drawing
- Discussed calming strategies but still having difficulty allowing blood to be drawn
- A few appointments with OT to learn sensory strategies
- Doing better with blood drawing

Mohammed

Age: 41

- Concern about cognitive decline
 - "I think he has developed Alzheimer's disease."
- Not following directions, seems confused
- On exam, ear canals were completely occluded with cerumen (ear wax)
- Ear wax removed
- Referred to audiology for hearing test, hearing was only slight decreased
- Noted to be following directions again



Resources

- [Balance](#) (article)
- [Cerumen Impaction](#) (article)
- [Down Syndrome EyeWiki](#) (American Academy of Ophthalmology)
- [Hearing Loss](#) (article)
- [Proprioceptive Input](#) (article)
- [Sensory Processing](#) (article)
- [Sensory Processing](#) (webinar)



[Wearing My Hearing Aids Story](#)

NOSE, MOUTH, AND UPPER AIRWAY

Nose, mouth, & upper airway

More common

- Chronic rhinitis
- Chronic sinusitis
- Gum disease
- Malocclusions
- Dysphagia (swallowing problems)

Dysphagia (swallowing problems)

- Why is it more common?
 - Differences in anatomy and physiology
 - Eating too fast, not chewing well
 - GERD, esophageal conditions
- Symptoms
 - Choking
 - Gagging
 - Eating refusal or eating selectivity
 - Aspiration pneumonia
- Diagnosis
 - Speech therapy
 - Video swallow study
 - Esophagram
 - Upper endoscopy (EGD)
- Treatment
 - Eat slower, chew thoroughly
 - Swallowing therapy
 - Diet change
 - Feeding tube (e.g., PEG)

Elisha

Age: 38

- Concern about his eating becoming “picky”
- Eating less solid food
- Coughing after eating sometimes
- Video swallow study normal
- Esophagram demonstrated abnormal contractions in the esophagus
- Further evaluation by GI with EGD
- Diagnosis: Achalasia
- Treated with Peroral Endoscopic Myotomy (POEM)



Resources

- [Brushing Teeth](#) (visuals and video)
- [Healthy Pace for Eating](#) (visuals and video)
- [Swallowing Problems](#) (article)

RESPIRATORY

Respiratory

More common

- Smaller airways
- Obstructive sleep apnea
- Pneumonia
- Infections
- Pulmonary hypertension (high blood pressure in the lungs)
- COVID-19

Less common (more severe?)

- Influenza?
- Upper respiratory infection?

Obstructive sleep apnea

- Temporary stoppage of breathing while sleeping
- Symptoms
 - Daytime sleepiness
 - Snoring
 - Pauses in breathing
 - Nighttime arousal and/or awakening
 - Headaches
 - Changes in behavior and mood

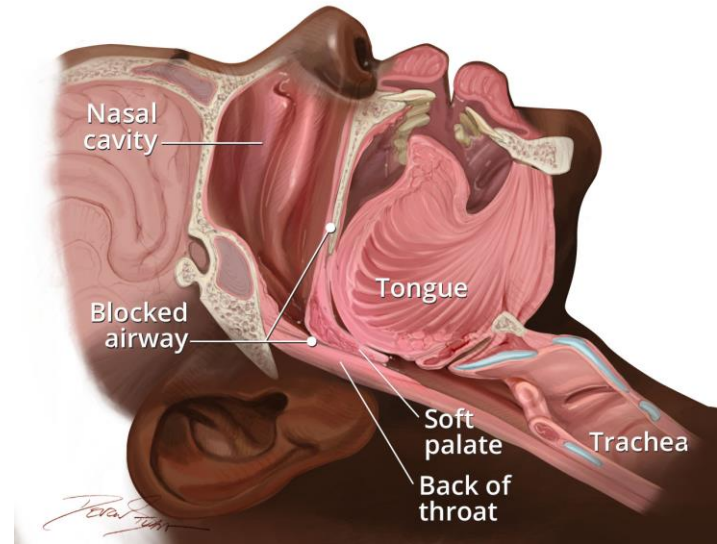


Image from the [National Heart, Lung, and Blood Institute, National Institutes of Health](https://www.nhlbi.nih.gov/health/obstructive-sleep-apnea)

Obstructive sleep apnea

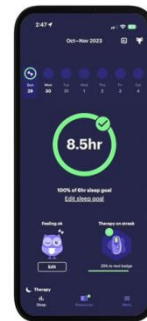
- Screening/diagnosis
 - Sleep study in a sleep lab
 - Home sleep study
 - Hand or wrist devices
- Treatment
 - CPAP or BiPAP
 - Positional treatment
 - Oral appliances
 - Surgery
 - Medications



The Inspire implant keeps your airway open while you sleep, so you can breathe regularly and sleep soundly.



The handheld Inspire™ remote simply turns on your therapy when you're ready to sleep. No mask or hose required.



The Inspire® app tracks your sleep goals and allows you to share data directly with your doctor.

Images from [Inspire Therapy](#)

Pneumonia

- Infection of the lungs
- Symptoms
 - Cough
 - Fever, sweating, and/or chills
 - Fatigue
 - Confusion
 - Shortness of breath, low oxygen level
 - Lower than normal body temperature
 - Low blood pressure
- Increased risk
 - Abnormal swallowing
 - GERD
 - Sleep apnea
 - Immune dysfunction
 - Tooth and gum disease
 - AAI
 - Alzheimer's disease
- Prevention
 - Pneumococcal vaccines

Omar

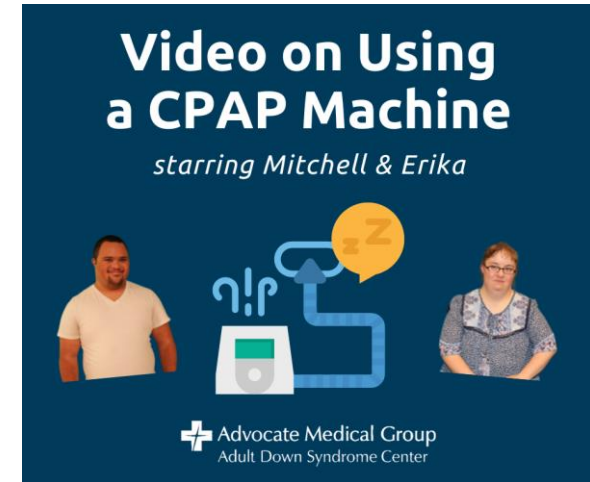
Age: 27

- Psychoses and more rigid adherence to “grooves”
- History and physical, lab tests
- Diagnosed with sleep apnea and obsessive-compulsive disorder
- Treatment
 - Continuous positive airway pressure (CPAP)
 - Anti-depressant
 - Did not require anti-psychotic medication



Resources

- [Chronic Cough](#) (article)
- [Pneumococcal Vaccines](#) (article)
- [Pneumonia](#) (article)
- [Respiratory Syncytial Virus \(RSV\) Vaccination](#) (article)
- [Sleep apnea](#) (article)



[How to Use a CPAP Machine Video](#)

DOWN SYNDROME REGRESSION DISORDER

DSRD

- Has also been referred to as:
 - Down syndrome disintegrative disorder
 - Regression
 - Adult regression syndrome
 - Catatonia
- Limited data on prevalence
- Continues to be studied and discussed

Expert consensus

- Paper published in July 2022
- 27 panelists who previously published on regression in Down syndrome or were involved in national or international working groups
- Name, diagnostic work up, diagnostic criteria



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Assessment and Diagnosis of Down Syndrome Regression Disorder: International Expert Consensus

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Objective: To develop standardization for nomenclature, diagnostic work up and diagnostic criteria for cases of neurocognitive regression in Down syndrome.

Background: There are no consensus criteria for the evaluation or diagnosis of neurocognitive regression in persons with Down syndrome. As such, previously published data on this condition is relegated to smaller case series with heterogeneous data sets. Lack of standardized assessment tools has slowed research in this clinical area.

Methods: The authors performed a two-round traditional Delphi method survey of an international group of clinicians with experience in treating Down syndrome to develop a standardized approach to clinical care and research in this area. Thirty-eight

Diagnostic criteria

- **Symptom onset:** New neurologic, psychiatric, or mixed symptoms over a period of <12 weeks in previously healthy individual with Down syndrome
- **Exclusion of other causes**

Symptoms

- Altered mental status or behavioral dysregulation
- Cognitive decline
- Developmental regression with/without new autistic features
- New focal neurologic deficits on examination and/or seizure
- Insomnia or circadian rhythm disruption
- Language deficits
- Movement disorder (excluding tics)
- Psychiatric symptoms

Catatonia

- Abnormality of movement and behavior
- Can (but may not) be associated with a mental illness
- Various presentations
 - Repetitive or purposeless overactivity
 - Resistance to movement

Health Condition	Prevalence (6,078)	Prevalence %	Prevalence (30,326)	Prevalence %	More or less common?	Odds Ratio
	Down Syndrome		Controls			
Catatonia	73	1.20%	70	0.23%	More	5.25*

Naya

Age: 16

- Developed rash and treated with oral steroids for poison ivy
- Over next few weeks, severe decline in ADLs, eating refusal, exceedingly slow moving
- Initially diagnosed with depression
- Limited response to anti-depressant
- Diagnosed with catatonia
- Treated very successfully with ECT



Resources

- [Decline in Skills and Regression](#) (webinar)
- [Eating Refusal](#) (article)
- [Regression & Down Syndrome](#) (National Down Syndrome Society)
- [Selective Mutism and Other Causes of Loss of Speech](#) (article)

NEUROLOGY

Neurology

Common

- Epilepsy/seizures
- Alzheimer's disease
- Autism

Uncommon

- Atherosclerotic strokes
- Multiple sclerosis??
- Parkinson's??

Seizures

- Wide variety of causes and types
 - Seizure – usually not recurrent
 - Due to infection, electrolyte abnormalities, etc.
 - Epileptic seizures – recurrent
 - Variety of types – e.g., tonic-clonic (grand mal), absence (petit-mal), etc.
 - Two peaks of onset – childhood and later adulthood (Alzheimer's)
 - Psychogenic non-epileptic seizures (PNES)
- Diagnosis
 - EEG (electroencephalogram)
- Treatment
 - Depends on the cause and type

Alzheimer's disease

- By age 40, nearly all people with Down syndrome have the brain pathology (plaques and tangles) of Alzheimer's disease.
- HOWEVER, symptoms of Alzheimer's disease are uncommon before age 40.
- Average age at diagnosis: 54 to 55 years old
- Symptoms
 - Changes in self-talk, routines
 - Psychological changes
 - Memory impairment
 - Loss of previously mastered skills
 - Incontinence
 - Weight loss
 - Seizures
 - Unsteady gait
 - Swallowing difficulties

McCarron et al. 2017, Sinai et al. 2018, Fortea et al. 2021, Tsou et al. 2020, Rubenstein et al. 2024., Ballard et al. 2016, Altuna et al. 2021, Menendez 2005

Alzheimer's disease

Global Medical Care 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.
 - [National Task Group Early Detection Screen for Dementia \(NTG-EDSD\)](#)

The NTG-EDSD form is a screening tool for dementia in adults with Down syndrome. It includes a header with the NTG logo and the title 'NTG-EDSD'. A detailed instruction box explains the purpose of the form and its use. The form contains several sections for data entry: 1) Identification (Name, Date of birth, Sex, Date), 2) Intellectual disability level (a table with categories: No discernible intellectual disability, Borderline (IQ 70-75), Mild (IQ 55-69), Moderate (IQ 40-54), Severe (IQ 25-39), Profound (IQ 24 and below), Unknown), 3) Diagnosed condition (a table with categories: Autism, Cerebral palsy, Down syndrome, Fragile X syndrome, Intellectual disability, Prader-Willi syndrome, Other), 4) Current living arrangement (a list of options: Lives alone, Lives with spouse or friends, Lives with parents or other family members, Lives with paid caregiver, Lives in community group home, apartment, supervised housing, etc., Lives in senior housing, Lives in congregate residential setting, Lives in long term care facility, Lives in other:). A final instruction box states: 'For each question block, check the item that best applies to the individual or situation.'

Alzheimer's disease

- Diagnosis

- Rule out other causes
 - History and physical exam
 - Tests – CBC, metabolic panel, TSH/T4, vitamin B12 and folic acid, celiac, sleep study
- Neuropsychological testing
 - Limitations
- Imaging

- Treatment

- Treat associated symptoms
 - Depression, anxiety
 - Behavior change
 - Seizures
 - Sleep challenges
 - Pain
- Cholinesterase inhibitor
- NMDA receptor antagonist
- Amyloid-beta therapy

Research



[Understanding key differences among the current and emerging Alzheimer's clinical trials in Down syndrome](#) (LuMind IDSC Foundation)

Seamus

Age: 51

- Sister noted he was no longer sending birthday cards to the family members
- Upon further evaluation at home, discovered their parents had been doing more for Seamus –he was much less independent with his ADLs
- Physical exam and labs did not find an alternative cause
- Diagnosed with Alzheimer's disease
- Had his first ever seizure



Resources

- [Aging in Adults with Down Syndrome](#) (webinar)
- [Change in Behavior in a Person with DS and AD](#) (article)
- [How is Alzheimer's Disease Diagnosed in a Person with Down Syndrome?](#) (article)
- [Loss of Skills and Alzheimer's Disease](#) (article)
- [Psychogenic Non-Epileptic Seizure](#) (article)
- [Reducing Risk of Getting Alzheimer's Disease](#) (article)
- [Seizures in People with Down Syndrome and Alzheimer's Disease](#) (article)

URINARY

Urinary

More common

- Urinary retention
- Impairment of kidney function
- Urinary incontinence
- Urinary tract infections

Urinary retention

- Build-up of excessive urine in the bladder
- Possible complications
 - Urinary tract infections
 - Urinary incontinence
 - Abdominal pain/discomfort
 - Kidney damage
- Diagnosis
 - Urine sample
 - Ultrasound, catheter, voiding study
 - Cystoscope
 - CT scan
- Treatment
 - Urinating regularly (2-3 hours)
 - Catheters
 - Medications
 - Surgery

Impairment of kidney function

- Decreased kidney function
- Contributing factors
 - Genetic differences
 - Dehydration
 - Urinary retention
 - Gout
- Labs
 - Blood urea nitrogen (BUN)
 - Creatinine

José

Age: 37

- He is non-verbal, has impaired vision
- Intermittent yelling and pushing on his abdomen
- No loss of skills
- Participating in activities
- Wakes at night sometimes yelling
- Bladder scan revealed 600 ml in his bladder before he voided and about 400 ml after he voided
- Diagnosed with urinary retention; referred to Urology



Resources

- [Kidney Function](#) (article)
- [Urinary Incontinence](#) (article)
- [Urinary Retention](#) (article)
- [Urinary Tract Infections](#) (article)

GYNECOLOGY

Gynecology

More common

- Earlier start of menstruation and menopause?

Less common

- Sexually transmitted infections (STIs)
- Breast cancer
- Cervical cancer

Menstruation

- Period hygiene
- Birth control
 - Oral hormonal contraceptive pills
 - Injections
 - IUDs

Menstrual Hygiene Strategies



Try out different types of pads (length, with/without wings, etc.)



Wear period panties instead of or in addition to pads/tampons.



Pack a period bag with all necessary supplies (pads, wipes, underwear, etc.).



Use visual supports. Check out our Resource Library!



Advocate Medical Group
Adult Down Syndrome Center



Advocate Health Care

Now part of  **ADVOCATEHEALTH**

Li

Age: 15

- Having difficulty managing her period hygiene
 - Missing out on opportunities, impeding her independence
- “Can we make her periods stop?”
- Reviewed options
 - Supporting her in using pads and tampons
 - Period underwear
 - Medication



Resources

- [Birth Control](#) (article)
- [Enhancing Women's Health](#) (webinar)
- [Menopause](#) (article)
- [Menopause](#) (visuals)
- [Menstrual Hygiene](#) (article)
- [Menstruation \(Period\)](#) (visuals)
- [PMS and PMDD](#) (article)



[National Down Syndrome Society](#)

HEART & VASCULAR SYSTEM

Heart and vascular system

More common

- Congenital heart disease
- Valvular heart disease
- Embolic strokes
- Moyamoya
- Varicose veins

Less common

- Hypertension (high blood pressure)
- Coronary artery disease/
myocardial infarctions

History of congenital heart disease

- “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.” ([Global Medical Care Guidelines for Adults with Down Syndrome](#))

Joan

Age: 45

- Went to the emergency room because of chest pain
- History, physical exam
- Normal EKG, blood work negative for myocardial infarction (heart attack)
- Treatment
 - Antacids resolved the chest pain
 - Is there need for additional evaluation and treatment?



Resources

- [High and Low Blood Pressure](#) (article)
- [Strokes](#) (article)
- [When Veins Don't Work](#) (article)

SKIN & NAIL CONDITIONS

Skin and nail conditions

More common

- Alopecia
- Folliculitis and boils
- Hidradenitis suppurativa
- Seborrhea
- Psoriasis
- Infections
- Dry skin
- Onychomycosis (fungal infection of the nails)

Folliculitis and boils

- Folliculitis – infection or inflammation of the hair follicles
- Boils – painful, pus-filled bumps that can form under the skin if infections around the hair follicles spread deeper
- Prevention and treatment
 - Antibacterial soaps such as Lever 2000 or Dial Soap
 - Antibacterial and antimicrobial skin cleanser such as Hibiclens
 - Dry well after bathing, baby powder
 - Zinc supplement?
 - Antibiotic
 - Boils – heat, triple antibiotic cream, topical cream (e.g., Boil-Ease)

Alopecia areata

- Sudden loss of hair
- Autoimmune condition
- Treatment
 - Steroids
 - Topical minoxidil (Rogaine)
 - JAK inhibitors

Veronica

Age: 19

- Has become very distraught due to hair loss
- Multiple bald patches
- Labs unremarkable
- Referred to Dermatology
- Treatment options discussed
- Started on baricitinib (JAK inhibitor)
- Marked improvement in hair loss
- Mood improved



Resources

- [Alopecia Areata](#) (article)
- [Down Syndrome and Hidradenitis Suppurativa](#) (handout from the Society for Pediatric Dermatology)
- [Dry Skin](#) (article)
- [Folliculitis and Boils](#) (article)
- [Onychomycosis](#) (article)
- [Skin Conditions in Down Syndrome](#) (podcast episodes)

DRY SKIN TIPS



- Creams and ointments are usually better than lotions because they are thicker.
- A good time to apply creams and ointments is after taking a shower or bath.

Learn more tips in our dry skin video!

 Advocate Medical Group
Adult Down Syndrome Center

[Tips for Dealing with Dry Skin Video](#)

CANCER

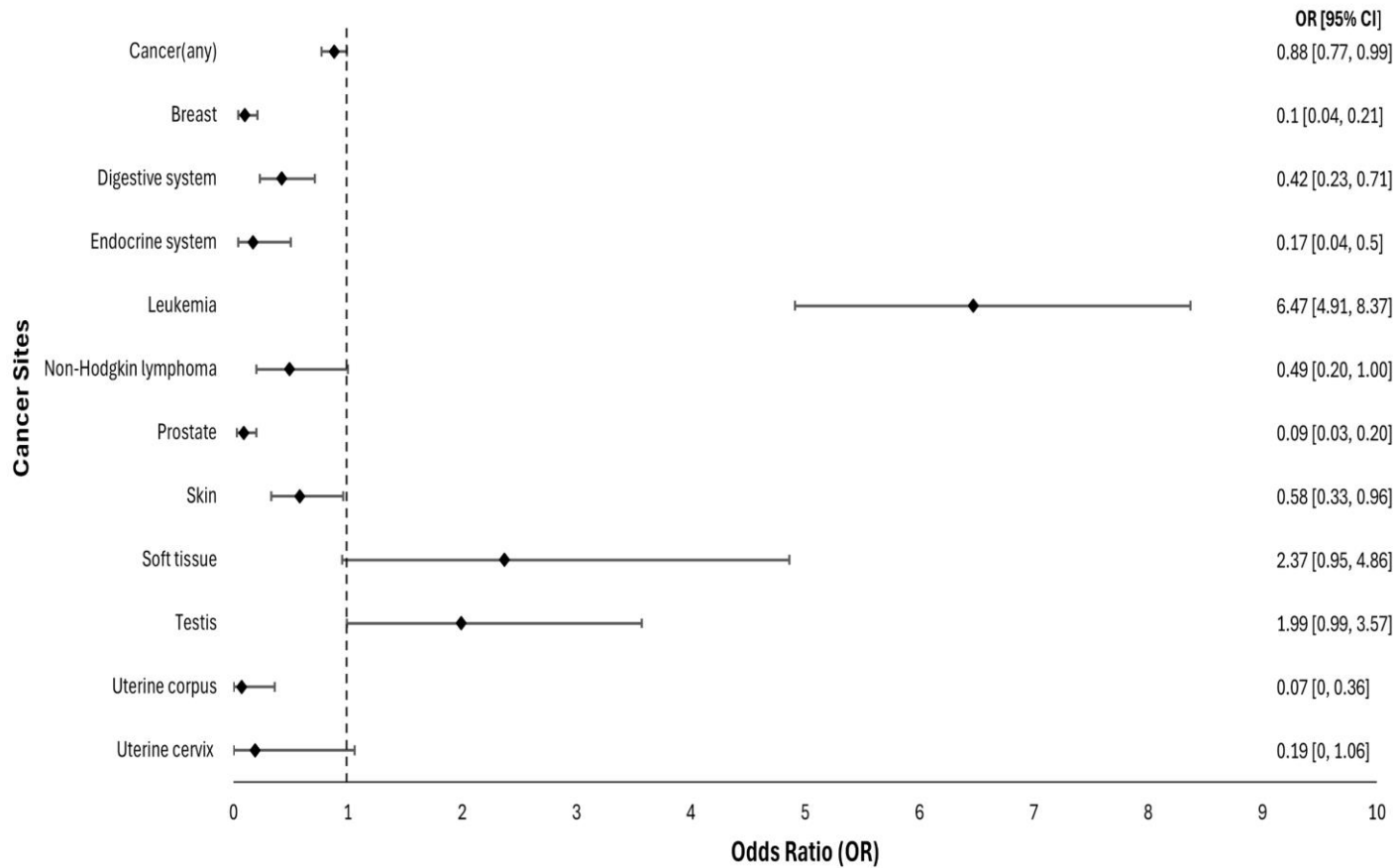
Cancer

More common

- Leukemia (childhood)
- Testicular cancer
- Lymphoma?

Less common

- Many solid tumor cancers
 - Breast cancer
 - Cervical cancer
 - Colorectal cancer



*Chicoine et al. 2025, unpublished data

Cancer

Presentation

- Lack of self-report
- May present later due to lack of report of sense of ill health
- Behavioral change
- Mostly similar
- Limited experience

Screening/treatment

- Treatment recommendations generally not different
 - AML (leukemia) – lower doses
- Screening
 - Mammograms
 - Colonoscopies
 - Pap smears
 - Etc.

Vivian

Age: 45

- Came in for an annual appointment
- No concerns
- History, physical exam, labs
- Asked about mammography



Resources

- [Breast Cancer and Breast Cancer Screening](#) (article)
- [Colon Cancer Screening](#) (article)
- [Dysgerminomas](#) (article)
- [Primary Care Provider's Guide to Women's Health and Down Syndrome](#) (guidebook from National Down Syndrome Society)
 - Includes information about breast, cervical, endometrial, and ovarian cancer
- [Prostate Cancer Screening](#) (article)
- [Testicular Cancer](#) (article)

Wrap-up



Takeaways

- People with Down syndrome are living longer and healthier than ever before.
- Some health conditions are more common in people with Down syndrome, and some are less common.
- Thorough evaluations are recommended to assess for conditions that may present as behavioral change.
- Ongoing research and review of the literature will help us decide on appropriate screening, diagnosis, and treatment of co-occurring conditions in people with Down syndrome.

Resources

DSMIG-USA

Down Syndrome Medical Interest Group-USA

- Speaker Series
- Project ECHO

A promotional graphic for DSMIG-USA. It features a dark blue background with white and orange text. At the top right is the DSMIG-USA logo with the tagline 'Down Syndrome Medical Interest Group'. On the left, a green and orange banner reads 'FREE RESOURCES'. To the right of this, the text 'SHARE WITH YOUR HEALTH CARE PROVIDER' is displayed. Below this, a bulleted list describes three offerings: Project ECHO, the Speaker Series, and vetted resources. A megaphone icon is on the right side. At the bottom, it says 'find out more at: DSMIG-USA.ORG'.

DSMIG-USA®
Down Syndrome Medical Interest Group

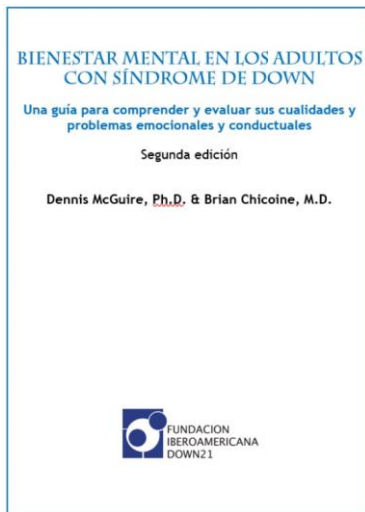
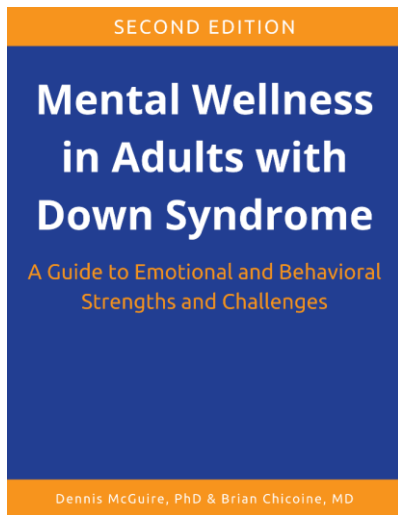
FREE RESOURCES

SHARE WITH YOUR HEALTH CARE PROVIDER

- Down Syndrome Project ECHO is a monthly virtual meeting for health care providers to learn and seek input from expert providers.
- The DSMIG Speaker Series consists of webinars and enduring materials designed to share knowledge and experience related to the care of people with Down syndrome and clinical research related to Down syndrome.
- DSMIG vetted resources including articles and important guidelines related to child and adult health issues, and health utilization by people with Down syndrome.

find out more at:
DSMIG-USA.ORG

Mental Wellness book



**Available as a free
PDF in English
and Spanish**

<https://adsresources.advocatehealth.com/mental-wellness-in-adults-with-down-syndrome-2nd-edition/>

Adult Down Syndrome Center

[Resource Library](#)

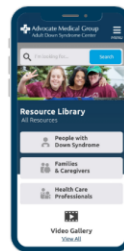
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FREE HEALTH RESOURCES

for people with Down syndrome, families and caregivers, and professionals



Resource Library



Find information on aging, puberty, mental health, self-talk, weight management, Alzheimer's disease, social skills, and more.

adscresources.advocatehealth.com

Facebook & Instagram



[@adultdownsyndromecenter](https://www.facebook.com/adultdownsyndromecenter)



www.eepurl.com/c7uV1v

Email List



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Questions?

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