Common and Uncommon Health Conditions in Adolescents and Adults with Down Syndrome

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Reminders

- This webinar will be recorded.
 - The link will be emailed to attendees and posted in our Resource Library within 1 week.
- Q&A
 - Please submit questions using the Q&A option.



Please note:

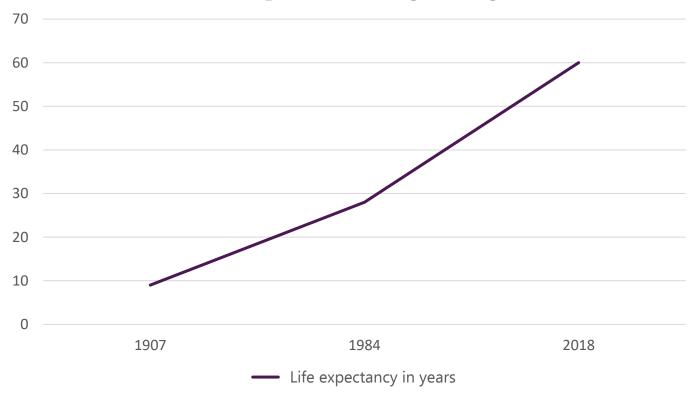
- This webinar is intended for families, caregivers, health care professionals, and service providers of individuals with Down syndrome.
- The information in this webinar is provided for educational purposes only and is not intended to serve as a substitute for a medical, psychiatric, mental health, or behavioral evaluation, diagnosis, or treatment plan by a qualified professional.
- We recommend that you bring specific questions about an individual with Down syndrome to their medical and/or therapy providers.

Objectives

- Provide an overview of common and uncommon health conditions in adolescents and adults with Down syndrome.
- Describe differences in prevalence of health conditions in people with and without Down syndrome.
- Discuss comprehensive and individualized health care for adolescents and adults with Down syndrome.

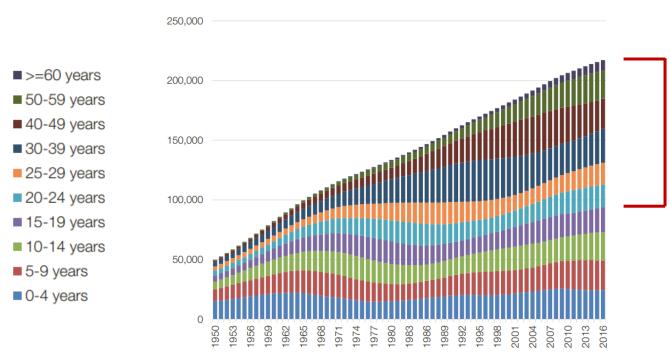
Today, people with Down syndrome are living *longer* and *healthier* than any other time in the past.

Life expectancy in years



There are more adults with Down syndrome living now than ever before.

People with DS in the United States



Estimated # of people with DS 20 years of age or older

Figure 5. Population of people with Down syndrome in the USA, 1950-2016

Health conditions

Prevalence of common diseases

Follow-up reports

- Mental health conditions
- Infectious diseases
- Endocrine disorders

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CME Accredited

ORIGINAL RESEARCH

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

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Purpo

Given the current life expectancy and number of individuals living with Down syndrome (DS), it is important to learn common occurrences of disease conditions across the developmental lifespan. This study analyzed data from a large cohort of individuals with DS in an effort to better understand these disease conditions, inform future screening practices, tallor medical care guidelines, and improve utilization of health care resources.

Mothods

This retrospective, descriptive study incorporated up to 28 years of data, compiled from 6078 individuals with DS and 30,326 controls matched on age and sex. Data were abstracted from electronic medical records within a large Midwestern health system.

Results

In general, individuals with DS experienced higher prevalence of testicular cancer, leukemias, moyamoya disease, mental health conditions, bronchitis and pneumonia, gastrointestinal conditions, thyroid disorder, neurological conditions, atlantoaxial subluxation, osteoporosis, dysphagia, diseases of the eyes/adnexa and of the ears/mastoid process, and sleep apnea, relative to matched controls. Individuals with DS experienced lower prevalence of solid tumors, heart disease conditions, sexually transmitted diseases, HIV, influenza, sinusitis, urinary tract infections, and diabetes. Similar rates of prevalence were seen for lymphomas, skin melanomas, stroke, acute myocardial infarction, hepatitis, cellulitis, and osteoarthritis.

Conclusion

While it is challenging to draw a widespread conclusion about comorbidities in individuals with Down syndrome, it is safe to conclude that care for individuals with DS should not automatically mirror screening, prevention, or treatment guidelines for the general U.S. population. Rather, care for those with DS should reflect the unique needs and common comorbidities of this population. (J Patient Cent Res Rev 2021:8:88-97.)

Keywords

Down syndrome; epidemiology; comorbidities; family medicine; population health

own syndrome (DS) is the most frequent chromosomal abnormality among live-born infants and the most common genetic cause of intellectual disability in the world. ¹² Due to improved screening and advances in medical technology and care, the lifespan for an individual with DS has more than doubled to age 60 in the past few decades. ¹² Recent U.S.-based population estimates of major birth defects

concluded that, while prevalence of most birth defects has remained stable over the past 15 years, prevalence of DS is increasing. 8 With increasing prevalence and longer lifespans of individuals with DS, it is important to understand common occurrences of co-occurring conditions, or comorbidities, across the developmental lifespan for more appropriate screening practices, tailored medical care guidelines, improved utilization of health care resources, and up-to-date planning and management of care for individuals with DS.

Corresponding author: Veronica Fitzpatrick, DrPH, Advocate Lutheran General Hospital, 1775 Dempster Street, W-939, Park Ridge, IL 60068 (veronica.fitzpatrick@aah.org) Individuals with DS represent a unique population in need of health care guidelines that address their specific medical care.⁹⁻¹³ A high prevalence of comorbidity has been documented among adults with DS, with types

Link to article

GLOBAL Medical Care Guidelines

- Evidence-based recommendations intended to offer guidance to health care providers who see adults with Down syndrome
- Article published in JAMA, guidelines document, familyfriendly guidelines



MEDICAL CARE GUIDELINES

for ADULTS WITH DOWN SYNDROME



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



Common

- Leukemia (childhood)
- Testicular cancer

Uncommon

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

Health Condition	Prevalence (6,078)	Prevalence %	Prevalence (30,326)	Prevalence %	More or less common?	Odds Ratio
	Down Sy	yndrome	Con	trols		
Breast	10	0.16%	246	0.81%	Less	0.20*
Cervix	38	0.63%	653	2.15%	Less	0.29*
Colorectal	3	0.05%	115	0.38%	Less	0.13*^
Testis	10	0.16%	19	0.00%	More	2.63**
Leukemia	50	0.82%	59	0.19%	More	4.23*

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

Screening

- The risks and benefits may be different for individuals with Down syndrome.
- Shared decision-making with the person and their family or caregiver.
 - What is the prevalence of the condition?
 - How old is the person with Down syndrome?
 - What does the procedure involve?
 - What do follow-up procedures involve?
 - Is anesthesia required?
 - What is the family history?

Breast cancer

Biennial screening (50-74)

Women without Down syndrome

- **2,240** # of mammograms / averted breast cancer death
- **27** # of benign biopsies / averted breast cancer death

Women with Down syndrome

- 16,735 # of mammograms / averted breast cancer death
- **209** # of benign biopsies / averted breast cancer death



Case study

- 42-year-old woman who came in for an annual physical
- History, physical exam, labs
- Asked about mammography

Common

- Depression
- Obsessive-compulsive disorder
- Impulse control disorder

Uncommon

- Substance use/abuse
- Generalized anxiety disorder?
- Posttraumatic stress disorder?

Health Condition	Prevalence (6,078)	Prevalence %	Prevalence (30,326)	Prevalence %	More or less common?	Odds Ratio
	Down Syndrome		Controls			
Depression	571	9.39%	2297	7.57%	More	1.27*
Anxiety disorders	1029	16.92%	4773	15.74%	More	1.09**
Generalized anxiety disorder	24	0.39%	474	1.56\$	Less	0.24*
Posttraumatic stress disorder	25	0.41%	208	0.69%	Less	0.60***
Obsessive- compulsive disorder	447	7.35%	119	0.39%	More	20.15*
Substance use/abuse	10	0.16%	4095	13.5%	Less	0.08*

Health Condition	Prevalence (6,078)	Prevalence %	Prevalence (30,326)	Prevalence %	More or less common?	Odds Ratio
	Down Sy	/ndrome	Con	trols		
Mental disorders due to physiologic cause	20	0.33%	34	0.11%	More	2.94*
Psychosis	24	0.39%	31	0.10%	More	3.87*
Impulse control disorder	73	1.2%	16	0.053%	More	23.03*

Presentation

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

Screening/treatment

- Screening/diagnosing
 - Behavioral changes, changes in routines
 - Underlying physical health conditions
- Treatment
 - Multidisciplinary
 - Medications



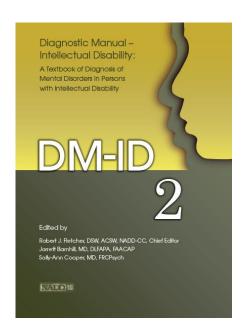
Mental health/behavior

- When concern for a mental health disorder is present:
 - Refer to a clinician knowledgeable about the medical, mental health disorders, and common behavioral characteristics of adults with Down syndrome.
 - Medical professionals should evaluate for medical conditions that may present with psychiatric and behavioral symptoms.



Mental health/behavior

- When diagnosing mental health disorders, follow guidelines in the:
 - Diagnostic and Statistical Manual of Mental Disorders (DSM-V) and/or
 - Diagnostic Manual Intellectual Disability (DM-ID-2)





Case study

- 22-year-old man who aged out of school, started to gain weight, and had a disagreeable mood. Also had difficulty sleeping.
- History, physical exam, labs, sleep study.
- Diagnosed with depression and sleep issues.
- Treatment
 - Established new routine
 - Counseling
 - Bupropion (Wellbutrin) for depression
 - Sleep hygiene and trazadone for sleep



Common

- Hypothyroidism
- Hyperthyroidism
- Type 1 diabetes mellitus
- Gout

Less common

Type 2 diabetes mellitus

Health Condition	Prevalence (6,078)	Prevalence %	Prevalence (30,326)	Prevalence %	More or less common?	Odds Ratio
	Down Syndrome		Controls			
Adrenal insufficiency / Addison's	41	0.67%	122	0.40%	More	1.68***
Hypothyroidism	2212	36.39%	1507	4.97%	More	10.94*
Hyperthyroidism	155	2.55%	319	1.05%	More	2.46*
Hashimoto's disease	69	1.14%	144	0.47%	More	2.41*
Graves' disease	28	0.46%	74	0.24%	More	1.89^
Gout	261	4.29%	502	1.66%	More	2.67*

Health Condition	Prevalence (6,078)	Prevalence %	Prevalence (30,326)	Prevalence %	More or less common?	Odds Ratio
	Down Syndrome		Controls			
Diabetes insipidus	7	0.12%	12	0.04%	More	2.91^^
Type 1 diabetes mellitus	66	1.01%	203	0.67%	More	1.63**
Type 2 diabetes mellitus	240	3.95%	2066	6.81%	Less	0.56*

Presentation

- Behavioral
- Psychological
- Physical symptoms

Screening/treatment

- Screen
 - Thyroid every 1-2 years
 - DM every 2-3 years
- Treatment
 - Similar
 - Thyroid more than the lab value



Diabetes

- For asymptomatic adults with Down syndrome beginning at age 30:
 - Screen for type 2 diabetes mellitus using hemoglobin A1c or fasting plasma glucose every 3 years.
- For any adult with Down syndrome and comorbid obesity beginning at age 21:
 - Screen for type 2 diabetes mellitus using hemoglobin A1c or fasting plasma glucose every 2-3 years.



Thyroid

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



Obesity

- Monitor for weight change and obesity annually by calculating BMI.
 - Follow the U.S. Preventive Services Task Force (USPSTF) Behavioral Weight Loss Intervention to Prevent Obesity-Related Morbidity and Mortality in Adults.
- Use a comprehensive approach to weight management, appetite control, and enhancement of quality of life.
 - Healthy diet, regular exercise, calorie management

Infections

Common

- Pneumonia
- Skin infections (dry skin, boils, etc.)

Less common

- Influenza?
- Upper respiratory infection?
- Sexually transmitted infections

Health Condition	Prevalence (6,078)	Prevalence %	Prevalence (30,326)	Prevalence %	More or less common?	Odds Ratio
	Down Syndrome		Controls			
Influenza	128	2.11%	1010	3.33%	Less	0.62*
Upper respiratory infection	756	12.44%	8384	27.65%	Less	0.37*
Bronchitis	236	3.88%	2334	7.70%	Less	0.48*
Otitis externa	193	3.18%	582	1.92%	More	1.68*
Otitis media	564	9.28%	3541	11.68%	Less	0.77*

Health Condition	Prevalence (6,078)	Prevalence %	Prevalence (30,326)	Prevalence %	More or less common?	Odds Ratio
	Down Sy	yndrome	Controls			
Pneumonia	746	12.27%	993	3.27%	More	4.13*
Skin infections	1035	17.03%	3201	10.56%	More	1.74*
Sexually transmitted infections	12	0.20%	304	1.00%	Less	0.20*

Presentation

- Lack of fever
- Lack of elevated WBC
- Hypotension (low blood pressure) sooner
- Sepsis and adrenal insufficiency

Screening/treatment

- Similar to people without DS
 - Sexually transmitted infections

Common

- Gastroesophageal reflux disease
- Celiac disease

Uncommon

 Difficult to know due to incomplete history/complaint of pain

Health Condition	Prevalence (6,078)	Prevalence %	Prevalence (30,326)	Prevalence %	More or less common?	Odds Ratio
	Down Syndrome		Controls			
Intestinal obstruction w/o hernia	82	1.35%	167	5.51%	More	2.47*
Esophageal disorders	1054	17.34%	3679	12.13%	More	1.52*
Gastroduodenal ulcer except hemorrhage	71	1.17%	249	0.82%	More	1.43**
Celiac disease	328	5.40%	93	0.31%	More	18.54*

Presentation

- Possible reduced pain
- Chest pain
- Change in function
- Change in mood and/or behavior

Screening/treatment

- Screening colon cancer
- Diagnosis abdominal evaluations
- Treatment similar
 - Function



Celiac disease

- Do an annual assessment for gastrointestinal and nongastrointestinal signs and symptoms of celiac disease.
 - Use targeted history, physical examination, and clinical judgment of good practice.



Case study

- 21-year-old man with depressed mood, reduced verbal abilities, and a change in his ability to complete ADLs
- History and physical exam, lab testing, occupational therapy assessment
- Diagnosed with celiac disease, vitamin B12 deficiency, catatonia, regression, and depression
- Treatment
 - Gluten-free diet
 - Occupational therapy
 - Medications



Common

- Epilepsy/seizures
- Alzheimer's disease
- Cervical myelopathy
- Atlantoaxial instability
- Osteoarthritis

Uncommon

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

Health Condition	Prevalence (6,078)	Prevalence %	Prevalence (30,326)	Prevalence %	More or less common?	Odds Ratio
	Down Syndrome		Controls			
Epilepsy	804	13.23%	878	2.90%	More	5.11*
Alzheimer's disease	739	12.06%	114	0.38%	More	36.68*
Myelopathy, cervical	51	0.84%	51	0.84%	More	2.51*
Atlantoaxial subluxation	137	2.25%	9	0.00%	More	77.68*
Osteoarthritis	380	6.25%	1841	6.07%	More	1.03**

Presentation

- Osteoarthritis change in function more common than pain
- AAI depression
- AD psychological, behavioral

Screening/treatment

- Screening
 - AAI
 - Alzheimer's disease
- Treatment
 - Medications for AD
 - Seizures



Atlantoaxial instability

- 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.



Osteoporosis

- Use a shared decision-making approach.
- Insufficient evidence to recommend for or against applying established osteoporosis screening guidelines for primary prevention of osteoporotic fractures in adults with Down syndrome.
- Evaluate all adults with Down syndrome who sustain a fragility fracture for secondary causes of osteoporosis.



Alzheimer's disease/dementia

- 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)

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Heart disease

Heart disease/cardiovascular disease

More common

- Embolic strokes
- Moyamoya

Uncommon

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

Heart disease

Health Condition	Prevalence (6,078)	Prevalence %	Prevalence (30,326)	Prevalence %	More or less common?	Odds Ratio
	Down Sy	yndrome	Con	trols		
High blood pressure / hypertension	345	5.68%	18,139	59.81%	Less	0.04*
Late effects of cerebrovascular disease (stroke)	36	0.59%	138	0.46%	More	1.30**
Coronary atherosclerosis and other heart disease	109	1.80%	1035	3.41%	Less	0.52*
Acute myocardial infarction	40	0.66%	213	0.70%	Less	0.94***
Moyamoya	14	0.23%	4	0.00%	More	17.50*

Chicoine et al. 2021 | *p<0.0001, **p<0.1568 (not statistically significant), ***p<0.7046 (not statistically significant)

Heart disease and risk factors

Health Condition	Prevalence % DS	Prevalence % Controls	More or less common?	Odds Ratio
Coronary heart disease	0.38%	6.70%	Less	0.05*
Myocardial infarction	0.04%	3.00%	Less	0.01*
Stroke	2.95%	2.50%	More	1.18***
Hypertension	2.69%	46.0%	Less	0.03*
Overweight	73.43%	69.40%	More	1.22**
Obesity	43.00%	36.30%	More	1.32*

Heart disease

Presentation

- Limited experience
- Function

Screening/treatment

- Screening
 - Cholesterol
- Treatment
 - High blood pressure



Cardiovascular disease

- 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).



Stroke

- Manage risk factors for stroke.
 - Follow the American Heart Association/American Stroke Association's *Guidelines for the Primary Prevention of Stroke.*
- In adults with a history of congenital heart disease:
 - There is an elevated risk of cardioembolic stroke.
 - See a cardiologist for a periodic cardiac evaluation and a corresponding monitoring plan.



Case study

- 45-year-old man presents to the emergency room with 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?

Common

- Dysphagia swallowing dysfunction
- Vision problems
- Hearing problems
- Sleep apnea

Uncommon

- Chronic pain??
- Headaches??
- Back pain??

Health Condition	Prevalence (6,078)	Prevalence %	Prevalence (30,326)	Prevalence %	More or less common?	Odds Ratio
	Down Syndrome		Controls			
Dysphagia	824	13.56%	743	2.45%	More	6.24*
Diseases of the eyes and adnexa	1710	28.13%	6177	20.37%	More	1.53*
Diseases of the ears and mastoid process	2484	40.87%	8117	26.77%	More	1.89*
Sleep apnea	581	9.56%	1713	5.65%	More	1.77*

Presentation

- Behavioral change
- Swallowing
 - Symptoms
 - Pneumonia
 - Consider esophagus
- Decline in skills
- Sleep apnea

Screening/treatment

Sleep apnea



Case study

- 29-year-old woman with psychoses and more rigid adherence to grooves
- History and physical, lab tests
- Diagnosed with sleep apnea and OCD
- Treatment
 - Continuous positive airway pressure (CPAP)
 - Anti-depressant
 - Did not require anti-psychotic medication

Down syndrome regression disorder

DSRD

- Also referred to as
 - Down syndrome disintegrative disorder
 - Regression
 - Adult regression syndrome
- 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 DS or were involved in national or international working groups
- Name, diagnostic work up, diagnostic criteria



ORIGINAL RESEARCH published: 15 July 2022 doi: 10 3380/fneur 2022 040174



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 heterogenous data sets. Lack of standardized assessment tools has slowed research in this clinical series.

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

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Diagnostic criteria

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

Co-occurring co-occurring conditions



Case study

- 51-year-old woman with decreased attention to grooves
- History and physical, lab tests
- Diagnosed with hypothyroidism
- Developed memory impairment, challenges with depth perception, and difficulty completing multi-step tasks
- Diagnosed with Alzheimer's disease



Treatment

- Medications
 - Levothyroxine
 - Should we use medications for Alzheimer's disease?
- Physical therapy
- Occupational therapy
- Adaptive equipment

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 DS.

Resources

DSMIG-USA

- <u>Down Syndrome</u>
 <u>Medical Interest Group</u>
 - Membership
 - Speaker Series
 - Project ECHO



Adult Down Syndrome Center

- Resource Library
- **Email List**
- Facebook
- **Instagram**

FREE HEALTH RESOURCES

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



Resource Library



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

adscresources.advocatehealth.com

Facebook & Instagram









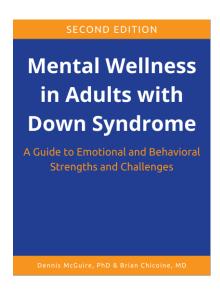
@adultdownsyndromecenter

www.eepurl.com/c7uV1v

Email List



Mental Wellness book





Available as a free PDF in English and Spanish

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



Anesthesia

Celiac Disease

Constipation

Depression in People with DS

Generalized Anxiety Disorder in People with DS

Global Medical Care Guidelines for Adults with Down Syndrome

Iron Deficiency and Down Syndrome

Pain in People with Down Syndrome

Post-Traumatic Stress Disorder

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

Psychotherapy for Individuals with Down Syndrome

Sensory Processing and Down Syndrome



Webinars and podcasts

At-Home Treatments for Common Health Conditions of People with Down Syndrome Webinar

Aging and Alzheimer's Disease in Adults with DS Webinar

Behavior Changes in Individuals with DS Webinar

Decline in Skills and Regression in Adolescents & Adults with DS Webinar

Skin Conditions in Down Syndrome Podcast

Sensory Processing Strategies to Promote Mental Wellness Webinar

Sleep in Adolescents and Adults with DS Webinar





Resource lists by topic

Aging and Alzheimer's Disease Neurology

<u>Cancer</u> <u>Nutrition and Weight</u>

<u>Decline in Skills and Regression</u>
<u>Self-Care and Hygiene</u>

Endocrinology Sensory

Grief and Loss Sleep

Healthy Lifestyle Social Skills

Mental Health Symptoms

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