Decline in Skills & Regression in Adolescents and Adults with Down Syndrome

Adult Down Syndrome Center

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AdvocateAuroraHealth

Adult Down Syndrome Center

Park Ridge, IL

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.

Disclaimer

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.

Outline

1. Define decline in skills.
2. Describe causes of decline in skills, including regression and Alzheimer's disease, in adolescents and adults with Down syndrome.
3. Identify strategies to assist with diagnosis and treatment of decline in skills in individuals with Down syndrome.

What is decline in skills?

Decreased ability to perform or use previously mastered skills or abilities

- Cognition
- Behavior / psychological changes
- Ability to perform activities of daily living
- Motor function
- Speech

Why might a person with Down syndrome decline?

Causes

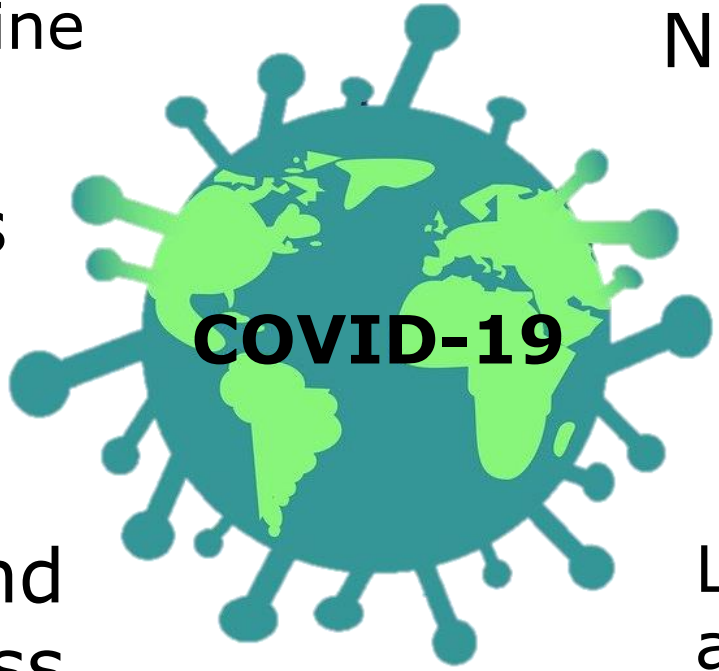
- Autism
- Down syndrome regression disorder (DSRD)
- Alzheimer’s disease (AD)
- Other

Causes during & after COVID-19

Changes in routine

News overdose

Grief and loss



Changes in diet

Social isolation

Family stress

Family and personal illness

Lack of physical activity

Medical conditions

- Medication side effects
- Sleep apnea
- Seizures
- Vitamin B12 deficiency
- Endocrine disorders
  - Hypothyroidism or hyperthyroidism
  - Adrenal insufficiency
  - Diabetes mellitus
  - Puberty-related
- Cervical myelopathy (subluxation, spinal stenosis)
- Chronic pain
  - Dental
  - Sinus
  - Cervical spine
  - Menstrual
  - Gastrointestinal, severe constipation

Medical conditions (cont.)

- Cardiovascular disease
  - Uncorrected congenital heart disease with pulmonary hypertension, congestive heart failure
  - Eisenmenger's syndrome
  - Stroke: thrombotic or hemorrhagic
- Neuropsychiatric disorders
  - Catatonia
  - Mood disorder
  - Obsessive compulsive disorder
  - Psychotic disorder
  - Complex tic disorder
  - Post-traumatic stress disorder
  - Parkinsonism, dystonia

Medical conditions (cont.)

- Infectious disease
  - Urinary tract infections
  - Pneumonia
  - Sepsis
  - Viral/bacterial meningitis/encephalitis
  - Lyme’s disease
- Toxic-metabolic
  - Numerous etiologies
- Sensory
  - Visual impairment
    - Glaucoma
    - Retinal detachment
    - Cataracts
    - Keratoconus
  - Hearing impairment
    - Hypo- or hyperacusis
    - Tinnitus
    - Vertigo

Adjustment to life events

- Transitions and relationships
  - Loss of family, friends, pets
  - School graduation
  - Work setting changes
  - Physical relocation
  - Response to hospitalization or medical condition
  - COVID-19

Autoimmune disorders

Additional evidence required

- Hashimoto’s encephalopathy
- Pediatric autoimmune neuropsychiatric disorders associated with streptococcal infections (PANDAS)
- Central nervous system manifestation of celiac disease
- Central nervous system manifestation of systemic lupus erythematosus (SLE)
- Autoimmune encephalopathy
- Limbic encephalitis

Down Syndrome Regression Disorder (DSRD)

DSRD

- First described in 1946 by Rollin – “catatonic psychoses”
- Names
  - Down syndrome disintegrative disorder
  - Regression
  - Adult regression syndrome
  - Down syndrome regression disorder
- Continues to be studied and discussed

DSRD

- Published in July 2022
- 27 panelists who previously published on regression in DS or were involved in national or international working groups
- Reached consensus on name, diagnostic work up, and diagnostic criteria
- Link to abstract

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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 heterogenous 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

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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 Down syndrome
- **Exclusion of other causes**

Diagnostic criteria

- **Symptoms:**
  - Altered mental status or behavioral dysregulation
  - Cognitive decline
  - Developmental regression with or 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

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

How is DSRD different than other forms of decline in skills?

- A sub-category?
- DSRD tends to be more severe and more pervasive.
- The cause can be the same in some instances.

Alzheimer’s Disease

What is Alzheimer's disease (AD)?

- Progressive neurological condition
- Affects the brain
- Is a type of dementia
- Plaques and tangles = the microscopic change of the brain consistent with AD
  - Also referred to as neuropathologic changes

Association between DS and AD

- Nearly all people with DS have plaques and tangles by age 40.
- But NOT everyone gets symptoms of Alzheimer’s disease.

Incidence of clinical AD

- AD thought to be uncommon before age 40.
- Incidence estimated to be 55% in those between 50-59.
- Incidence estimated to be greater than 75% in those 60 years of age and older.

How is Alzheimer's disease similar to DSRD?

- Both involve decline in skills
- Both are (probably) neurological conditions that often have psychological symptoms
- Both are challenging for the individual and families
- Both need more research, including ways to support the individual and family

How is Alzheimer's disease different than DSRD?

- Alzheimer's disease
  - Age of onset = > 40
  - Not reversible
- DSRD
  - Age of onset = teens, early 20s
  - Sometimes reversible
- Not all decline in skills in those age ranges is either Alzheimer's disease or DSRD.

How do we determine the cause of decline?

Determining the cause

- Evaluate for contributing causes
- Ongoing evaluation for (additional) contributing causes

Diagnostic work up

- History and physical
- Neuroimaging
- Blood work
- Lumbar puncture
- EEG
- Urine studies
- Other

How is a decline in skills treated?

Treatment

- Treat diagnosable conditions
  - Some specific treatments (e.g., hypothyroidism, catatonia, autoimmune encephalopathy, sleep apnea)
- Treat related signs and symptoms
- Use therapies to help improve function
- Start with “safe” activities

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.

Medications for AD

- Cholinesterase inhibitors (e.g., donepezil / Aricept)
- NMDA receptor antagonist (memantine / Namenda)
- Aduhelm (aducanumab)
  - LuMind IDSC: website
  - National Task Group on Intellectual Disabilities and Dementia Practices (NTG): website and statement

Treatments for DSRD

- Medications
- ECT
- Therapy (physical, occupational)
- Counseling
- Immunotherapies

How can families support an individual with decline in skills?

Diagnosis and treatment

- Be observant
- Share your observations
  - Mood, behavior, function, motor activities, sleep, appetite, any other symptoms you notice
  - What is still “missing” from the person?
    - Continue to share observations on an ongoing basis

Diagnosis and treatment

- Treat condition and function
  - “Safe” activities
  - Re-teach
- Trust yourself

Non-medicinal strategies

- Sensory strategies
- Create schedules and routines
  - Sleep, healthy eating, physical activity
- Encourage safe social interactions

Case examples

Case 1

Case 1

- 22-year-old woman with Down syndrome
- Presented with:
  - Personality change
  - Difficulty performing at her job
  - Inability to care for herself or learn new skills
  - Agitated behavior initially, followed by decreased movement, eating, and interest in activities
- Progressive over 6 months

Case 1

- Diagnosis
  - Down syndrome regression disorder
- Treatment
  - Medication
  - ECT
  - Measured reintroduction into activities
    - Visuals – daily schedule

Case 2

Case 2

- 27-year-old man with Down syndrome
- Presented with:
  - Reduced interest in activities
  - Apathy
  - Altered sleep
  - Generally disagreeable (a change)

Case 2

- Diagnosis
  - Medication side effect – Dicyclomine (Bentyl)
- Treatment
  - Stopped medication
  - Return to “safe” activities, visuals

Case 3

Case 3

- 32-year-old man with Down syndrome
- Presented with:
  - Depressed mood
  - Loss of skills
  - Hallucinatory behavior
- Psychoses? Alzheimer’s disease? DSRD?
- Completed a sleep study

Case 3

- Diagnosis
  - Sleep apnea
- Treatment
  - CPAP

Case 4

Case 4

- 22-year-old woman with Down syndrome
- Presented with:
  - Depressed mood
  - Catatonia

Case 4

- Diagnosis
  - DSRD
    - Depression and catatonia
- Treatment
  - Treated with anti-depressant but no improvement. Stopped medication.
  - Started Lorazepam
  - Catatonia started to improve with Lorazepam

Case 4

- Treatment
  - Depression persisted
  - Started anti-depressant again after catatonia fully resolved
  - Depression significantly improved; catatonia continued to improve

Finding support

- Regression in Down Syndrome Facebook Support Group
- Down Syndrome and Alzheimer's Disease Facebook Support Group
- Information on finding a Down syndrome clinic or health care providers
- Down Syndrome Medical Interest Group (DSMIG-USA)

More information

- Resources on Decline in Skills/Regression
- Resources on Alzheimer's Disease & Dementia
- Resources on Mental Health

Resources:
adscresources.advocatehealth.com

Facebook:
facebook.com/adultdownsyndromecenter

Email Newsletter:
eepurl.com/c7uV1v

AdvocateAuroraHealth

Questions?

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