

Decline in Skills:

Determining the Cause and Finding the Solutions

National Down Syndrome Congress Webinar

May 14, 2019 | Brian Chicoine, MD

 Advocate Health Care

We are  AdvocateAuroraHealth

Adult Down Syndrome Center



Leo Foley

1907-1947



What is “Decline in Skills”?

- Cognition
- Behavior/Psychological changes
- Ability to perform activities of daily living



Our Brains respond to Many Conditions

- Metabolic
- Infectious
- Trauma
- Direct and Indirect



What about People with Down syndrome?

- What is the role of genetics?
- What about family genetics vs the extra chromosome?



What are the Causes for Decline in Skills?

- Reversible vs Irreversible
- Direct vs Indirect



Three Cause of Decline Across the Lifespan

- Early childhood: Autism
- Adolescence and young adult: Regression
- Older adult: Alzheimer's disease

Association between DS and AD

- Nearly all people with Down syndrome have plaques and tangles by age 40.
- All people with DS over age 60

Mann, D.M.A. (1997). Neuropathological changes of Alzheimer's disease in persons with Down's syndrome. In Esiri, M.M. & Morris, J.H. (Eds.), *The Neuropathology of dementia*. Cambridge: Cambridge University Press.

Does everyone with DS get Alzheimer's disease?

- Everyone with DS gets the neuropathologic changes eventually
- BUT, everyone does NOT get symptoms of Alzheimer's disease
- Why?



What is the incidence of clinical Alzheimer's disease?

- 9-23% between ages 35 and 49
- Some studies found it is rare before 40
- 55% in those between 50-59
- Estimates of greater than 75% in those 60 years of age and greater

Mann et al. 1984, McCarron et al. 2014, Coppus et al. 2006, Strydom et al. 2018

Why is AD more common in DS?

- Due to an extra chromosome, there are genes that are “triplicated.”
- Amyloid precursor protein (APP) gene is on chromosome 21.
- There is compelling evidence that increased gene dose for APP (and its products) is related to AD in DS (Prasher et al. 1998, Doran et al. 2017, Korbel et al. 2009).
- Other chromosome 21 genes may impact AD in DS as may genes on other chromosomes, including variants that also increase the probability of AD, but a uniquely important role is played by increased dose for APP (Wiseman et al. 2015, Ballard et al. 2016).

Symptoms

- Memory deterioration
- Loss of previously mastered skills
- Incontinence
- Gait apraxia
- Dysphagia (swallowing)
- Seizures
- Weight loss
- Psychological changes



Diagnosis

- Look for the pattern of decline
- Rule out other causes of dementia (including reversible dementia)
- Examples:
 - Vitamin B 12 deficiency
 - Depression
 - Sleep apnea
 - Hypothyroidism
 - Cataracts

What about my 19 y/o?

- Memory impairment
- Loss of skills
- Behavioral changes
- Diagnosed with Alzheimer's disease

Regression: Case Presentation

- Lucy is a 30 -year-old woman with Down syndrome
- Presented for her first annual evaluation at our Adult Down Syndrome Center
- Lucy did not speak during the assessment, was reported to have no verbal skills.
- Repetitive motions through much of the assessment, and had little or no eye contact or interaction with the clinician.
- At first glance, she appeared to have autism, but the history was not classic for that diagnosis.
- Lucy had been a typically developing girl with Down syndrome with mild cognitive impairment, good verbal skills, and good self-care skills who attended school and a variety of activities prior to age 17.

Case Presentation continued

- According to her mother, Lucy developed an upper respiratory infection at age 17, was treated with an antibiotic, and suddenly regressed.
- Over the following years, she had an extensive work-up including blood tests, imaging, and sleep study.
- She had been given a variety of diagnoses including depression, anxiety, late-onset autism, early Alzheimer's disease, and psychoses.
- She had been treated with several psychotropic medications and supplements with little or no improvement.
- However, she also did not display further cognitive decline.
- She had neither improved nor deteriorated over the 13-year period.

Working Definition

What it isn't

Alzheimer's disease

Autism

Working Definition

What it is

Core features	Variable features
Decline in cognitive (executive) function	Maladaptive behavior
Social Withdrawal	Psychiatric symptoms
Loss of acquired skills	Failure to acquire new skills
Loss of functional use of language	Inattention – disorganization
Duration > 3 months	Motor slowing
	Vegetative symptoms
Demographics	-appetite/weight loss, incontinence
Typically btwn 15-30 years, M=F	-sleep pattern disturbance

What Else Could it Be?

- The differential diagnosis
- Psychological
- Social
- Physical

Direct cause

- Lisa, 27 years old
- Reduced interest in activities
- Apathy
- Altered sleep
- Generally disagreeable
- Irritable bowel syndrome
- Dicyclomine (Bentyl)



Indirect Cause

- Sandy, age 26
- Depressed mood
- Congenital Shoulder injury
- Fall
- Anti-depressant
- Physical therapy



What Else Causes Decline in Skills?

- Hypothyroidism and hyperthyroidism
- Hearing impairment
- Vision problems including cataracts and keratoconus
- Celiac Disease
- Sleep apnea
- Auto-immune conditions
- Depression



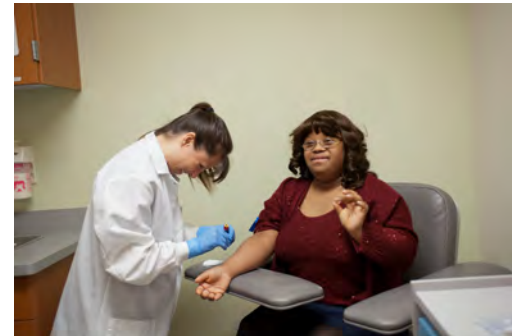
What Else Causes Decline in Skills?

- Cervical subluxation
- Vitamin B12 deficiency
- Gastroesophageal reflux disease
- Constipation
- Skin infections
- Osteoarthritis
- Obesity




The Interaction Between Physical and Mental Health

- George, age 32
- Depressed mood, loss of skills, and hallucinatory behavior
- Psychoses
- Alzheimer's disease?
- Regression?
- Sleep study
- Sleep apnea



Adult Down Syndrome Center

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Resources

We offer a variety of resources for people with Down syndrome, their families and caregivers and the professionals who care for and work with them. Choose a section for a list of helpful websites, articles, educational materials and more. Useful videos about a variety of health topics can be found in our [video gallery](#).



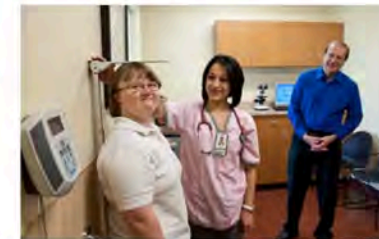
People with Down Syndrome

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