

Regression in Down Syndrome

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Affiliates

The Global Down Syndrome Foundation is part of a network of affiliate organizations that work closely together on a daily basis to deliver on our mission, vision, values, and goals:



Global & Affiliates

❖ **Global:** was established as a 501(c)3 in 2009 and is “Dedicated to significantly improving the lives of people with Down syndrome through Research, Medical Care, Education, and Advocacy”

❖ Affiliates are:

- Established with a lead gift from Anna & John J. Sie Foundation
- Must work closely together to benefit people with Down syndrome
- Must be self-sustaining financially



Objectives

- ❖ Define regression
- ❖ Discuss what we know and don't know
- ❖ Next steps



Historical Perspective

- ❖ There was a time when this decline was considered to be early onset Alzheimer's.
- ❖ This rare phenomenon was first described by Prasher in a publication in 2004.
 - He defined it in the context of adolescents and young adults
 - He gave it the name Young Adult Disintegrative Disorder



What is Regression?

“...Involves the relatively rapid, atypical loss of previously acquired skills in cognition, socialization, activities of daily living, and an increase in maladaptive behaviors. These declines occur after a trajectory of development that is described as consistent with Down syndrome.”

Devenny and Matthews, 2011



Regression



What we know...

❖ Symptom presentation

Bathing

Dressing

Feeding and Appetite

Toileting

Extrapyramidal

Gait

Communication

Self-talk

Echopraxia/Echolalia

Eye Contact

Thought Process, Associations, and Content

Attention Span/Concentration

Impulsivity

Memory and Fund of Knowledge

Facial grimacing

Stereotypy

Initiation-Motivation

Affect

Mood

Sleep

Social Engagement



What we know...

- ❖ It is rare but increasingly being identified
- ❖ Age of onset is typically in adolescence
- ❖ It is not autism
- ❖ Onset is rapid
- ❖ It is treatable
- ❖ Prognosis is variable



What we don't know...

❖ What causes it

- Immune issues
- Hormones
- Life events
- Anesthesia
- Life events of changes
- Trauma
- Medical factors

❖ Variations in definition

- Developmental regression
- Catatonia



What we don't know...

❖ Most effective treatment interventions

- IVIG
- Psychiatric medications
- ECT
- Transcranial magnetic stimulation
- Implementation of structure and routine
- Increased activity



Next Steps...

We are in the process of creating a collaboration between the Sie Center for Down Syndrome and the Linda Crnic Institute to evaluate regression and improve clinical care. Information will be gathered from cells to clinical presentation and include:

- Medical workups
- Diagnostic tools to evaluate pre-regression, treatment, and post-regression
- Biobank data to assess potential cellular risk factors differentiating those with and without regression

