

Down Syndrome Regression Disorder

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Down syndrome regression disorder (DSRD), also referred to as regression, is a rare but serious disorder that occurs in some adolescents and young adults with Down syndrome. Regression is used to describe the loss of skills an individual has previously learned. These skills can be daily living, language, movement, or social skills. The loss is often sudden and occurs over a period of weeks to months. Since DSRD may be caused by a variety of factors, the first priority is to determine the most likely cause. Individuals who are younger than 10 years or older than 30 years are very rarely diagnosed with DSRD. If these individuals are experiencing regression-like symptoms, a close look at all other factors must be taken before giving the diagnosis of DSRD.

In 2022, a group of medical experts with experience treating regression in patients with Down syndrome gathered to create the first criteria for diagnosing DSRD. These criteria provide guidance to physicians and families who do not have access to specialists in DSRD. There are eight clusters of symptoms linked to regression, and some are broken down into specific examples. Individuals who are experiencing four or more of the eight symptom clusters are considered “possible” cases of DSRD, meaning further investigation is needed. Individuals who have seven or eight of the eight symptom clusters are considered “probable” cases, meaning they are likely to receive a diagnosis of DSRD if no other cause can be found.

Testing

If you or your loved one are experiencing symptoms in the checklist (see page 3), it is recommended that you make an appointment with your primary care doctor or neurologist as soon as possible. The diagnosis of DSRD is mostly based on symptoms, but your doctor may order some of the following medical tests as part of the evaluation:

- Imaging of the brain with an MRI
- Blood work
- Urine tests (if necessary)
- Lumbar puncture (also known as a spinal tap)
- Electroencephalogram (EEG)
- Genetic or metabolic testing (if necessary)
- Polysomnogram (sleep study)

For more detailed testing recommendations, you and your doctor can read the Assessment and Diagnosis of Regression in Down Syndrome (Santoro et al., Front Neurol, 2022).

<https://www.frontiersin.org/articles/10.3389/fneur.2022.940175/full>

Treatment

There is currently no one-size-fits-all treatment for regression, and every patient requires a unique and personalized treatment plan. However, once you and your doctor have determined the most likely cause of the regression, the best treatment options can be discussed.

- It is recommended that you seek consultation with experts who work in a multi-disciplinary team and include psychiatrists, neurologists, and/or a provider who is familiar with treating individuals with Down syndrome and regression.
- If the regression is found to be due to a medical issue such as a psychiatric diagnosis, obstructive sleep apnea, inflammation or infection in the brain, vitamin deficiency, or toxin exposure, treating the issue may reduce the symptoms of regression.
- The most commonly used treatments in persons with DSRD include benzodiazepines, antidepressants, antipsychotics, immunotherapy, and electroconvulsive therapy. Although many of these treatments are used in other conditions, in DSRD they are often used for symptom control.

It is important to remember every patient is likely to have a different treatment plan and what works for one patient may not always be the best intervention for you or your loved one.

References:

Santoro JD, Patel L, Kammeyer R, Filipink RA, Gombolay GY, Cardinale KM, Real de Asua D, Zaman S, Santoro SL, Marzouk SM, Khoshnood M, Vogel BN, Tanna R, Pagarkar D, Dhanani S, Ortega MdC, Partridge R, Stanley MA, Sanders JS, Christy A, Sannar EM, Brown R, McCormick AA, Van Mater H, Franklin C, Worley G, Quinn EA, Capone GT, Chicoine B, Skotko BG and Rafii MS (2022) Assessment and Diagnosis of Down Syndrome Regression Disorder: International Expert Consensus. *Front. Neurol.* 13:940175. doi: 10.3389/fneur.2022.940175

Additional Resources:

Regression resources page from the Advocate Medical Group at the Adult Down Syndrome Center

<https://adscresources.advocatehealth.com/search/families-and-caregivers/?category=Decline%20in%20Skills%20and%20Regression>

Down syndrome 'regression' webpage with list of resources from the National Task Group on Intellectual Disabilities and Dementia Practices <https://www.the-ntg.org/down-syndrome-regression>

Regression in persons with Down Syndrome: Current consensus document from the Down Syndrome Medical Interest Group USA <https://www.dsmig->

[usa.org/resources/Documents/21DSMIG_Regression%20in%20Persons%20with%20Down%20Syndrome_0928.pdf](https://www.dsmig-usa.org/resources/Documents/21DSMIG_Regression%20in%20Persons%20with%20Down%20Syndrome_0928.pdf)

Children's Hospital Los Angeles blog post on Down syndrome regression disorder

<https://www.chla.org/blog/patient-stories/family-finds-hope-down-syndrome-regression-disorder>

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Down Syndrome Regression Disorder Symptoms Checklist

Date: _____

Symptoms occurring more frequently within the last three months (check all that apply):

1. Behavioral Changes

Date symptom(s) began: _____

- Eating much more or less than usual
- Confusion or disorientation
- Laughing or crying at inappropriate times
- Frequent changes in mood or rapid fluctuations between happiness, sadness, or anger

2. Changes in Thinking and Processing of Information

Date symptom(s) began: _____

- Decreased visible emotions and empathy
- Lack of motivation or lack of engagement
- Difficulty starting or finishing tasks
- Worsening memory

3. Loss of Functional and Social Skills

Date symptoms(s) began: _____

- Loss/worsening of previously learned skills (self-feeding, toileting, dressing, etc.)
- Decreased social interaction with friends, family, classmates, or coworkers
- Decreased eye contact
- Repetitive hand or body movements with no clear purpose

4. New seizures or neurological deficits (weakness, slurring of speech, etc.) determined by a physician

Date symptom(s) began: _____

5. Difficulty sleeping or sleeping at irregular times

Date symptom(s) began: _____

6. Language Difficulties

Date symptom(s) began: _____

- Difficulty producing speech or trouble reading and understanding speech
- No longer using speech or speaking only in a whisper

7. Irregular Movements

Date symptom(s) began: _____

- Lack of movement sometimes with stiff and rigid muscles
- Moving very slowly or using an unusual walk or run gait pattern

8. Mental Health Symptoms

Date symptom(s) began: _____

- New or worsened anxiety
- Delusions (untrue beliefs) or hallucinations (seeing things that are not there)
- Derealization (feeling detached from surroundings) or depersonalization (feeling of observing oneself from outside of the body)
- Obsessive compulsive tendencies like lining up items, only talking about specific topics of interest, and difficulty tolerating changes in routine
- Aggression or agitation toward others