

How do you diagnose adrenal insufficiency?

The adrenal gland is part of the endocrine system. It produces a series of hormones. Adrenal insufficiency occurs when there is inadequate production of adrenal hormones. Findings on the history and physical exam of adrenal insufficiency include decreased appetite, nausea, vomiting, abdominal pain, increased pigmentation of the skin, decreased blood pressure with change in position (such as from lying to sitting or standing or from sitting to standing), salt craving, weakness, fatigue, lethargy, decreased menses (periods), muscle and/or joint pain, fever, confusion, and psychiatric symptoms. In the most severe state, coma or shock can occur and, if undiagnosed in the severe state, adrenal insufficiency can be fatal. Common blood test abnormalities may include an elevated potassium level, a low sodium level, low blood sugar, and/or anemia.

If adrenal insufficiency is suspected based on the above, the usual next step is to draw a serum cortisol level (one of the adrenal hormones). This is usually drawn early in the morning. The cortisol level fluctuates throughout the day and the morning is the most recognized standard. It can also be drawn in the late afternoon. If the cortisol level is abnormal, additional hormonal tests include tests that measure the cortisol level after another hormone is given to stimulate the adrenal gland.

Diagnosis of metabolic dysfunction?? (not diabetes) other things like adrenal insufficiency, Lyme toxic overload??

For people who have a decline in skills including regression, a variety of tests may be considered. These may include lab evaluation for:

- Metabolic conditions
 - Kidney function
 - Liver function
- Endocrine disorders
 - Adrenal insufficiency
 - Thyroid dysfunction
- Infections
 - Lyme's disease
 - Meningitis/encephalitis
 - Syphilis
 - Sepsis
 - Urinary tract infection
 - pneumonia

This is not an exhaustive list. Other conditions may be considered. A very extensive list is included in [this article](#) by Jacobs et al. It is usually not indicated to test for all the conditions

noted in this article, but a selective assessment is made based on the history, physical, other labs, and the response to treatment.

Any suggestions on how to treat Neuro-inflammation?

Inflammation research??

What inflammation markers?

What research is being done to focus on neuro inflammation prevention?

Treating neuro-inflammation is an area that needs more study. Steroids and a variety of immunotherapy agents (medications that affect the immune system function) have been evaluated in a few small studies and are being used to treat regression in some situations. Further information is available in [this article](#) by Cardinale et al. There are limited data on the effectiveness in regression syndrome or Down syndrome disintegrative disorder but hopefully more information will become available. Review of the clinicaltrials.gov website (a site that lists all clinical trials in the United States) for studies under “Down syndrome” that may be related to inflammation include:

- a study on vitamin E in aging and Down syndrome
- a study looking at use of a medication that reduces interferon function (that can cause inflammation) and its effect on skin conditions.

I could not find any other studies registered that are looking at medications or supplements to address neuro-inflammation in people with Down syndrome.

When testing for inflammation, both blood and cerebral spinal fluid (CSF, collected from a spinal tap/lumbar puncture) may be evaluated. Tests may include:

Blood

- sedimentation rate (ESR)
- C-reactive protein (CRP)
- Autoimmune evaluation panel
- Anti-nuclear antibody (ANA)
- Thyroid antibodies
- Other antibodies

CSF

- Cell count, glucose, protein
- Oligoclonal bands
- Autoimmune Evaluation Panel

In DSDD is the regression always consistently progressive or can it kind of ebb and flow as they decline?

The decline associated with DSDD (or regression syndrome) can have several different patterns.

- With treatment, many will improve, some to the same functional level from before the onset of symptoms.
- Some will have fluctuations.

- Some will have a plateau. With treatment, they no longer decline but they do not regain their previous level of function. Even without treatment, some will plateau and not continue to decline (but not improve).
- We are not aware of anyone in our practice that had a spontaneous resolution without treatment (but we cannot say it is not possible)

My son was diagnosed at your clinic several years ago with PTSD. He is now showing new symptoms since the pandemic. Is regression syndrome more common in those with earlier diagnosis of psychological issues?

The answer to your question is unknown. However, here are some thoughts.

For many psychological conditions, having one episode puts one at greater risk for additional episodes. For example, with each additional episode of depression, additional episodes are more likely.

Whether having a previous diagnosis or episode of a psychological condition puts one at greater risk for regression syndrome is not known. We need to learn more to clearly understand. Nevertheless, theoretically there may be some logic to someone who has a previous psychological diagnosis being more at risk for regression because some individuals with regression syndrome have co-occurring mental health problems and since they do, increased recurrence might be expected.

However, it is not clear whether the mental illness caused the regression, or the regression caused the mental illness. Is regression a psychological condition, a neurological condition, an immune disorder, something else, or a combination? More study is needed.

Is it possible that late term effects of chemotherapy treatment years ago or experiencing massive bilateral pulmonary embolism may have contributed in some ways to one's regression later? My son's regression, showing no definite markers for disease and having come on extremely gradually, is given no particular recommendation for treatment beyond lorazepam and other medications that did not help him.

Trauma (of a variety of forms) and stressful situations are thought to contribute to the onset of regression in some individuals. An indirect effect of the medical illness, the potential side effects of medications, the psychological trauma of the illness, the treatment, and/or hospitalization can all have an impact.

While gradual onset is not the more typical progression, it can occur.

We have seen some individuals who did not respond to anti-depressants initially (even though they had depression as part of their regression symptoms). Later when the catatonia symptoms improved with lorazepam, re-introduction of an anti-depressant (in some cases one of the ones they did not respond to before) resulted in not only improvement in the depression symptoms but marked overall improvement in all their symptoms.

Are these regression cases treatable? What do you do to medically help these individuals?

Regression is treatable. However, while some individuals completely recover, some will only partially recover, some will plateau, and some show little if any recovery. Assessing for and treating underlying or co-occurring conditions is very important as well as symptom management. There may be conditions that contribute that aren't the main cause that also need treatment to optimize the success of treatment. For example, someone with regression with catatonia and depression who also has previously unrecognized and untreated celiac disease will likely need treatment for all 3 conditions- catatonia, depression, and celiac disease.

In addition, physical therapy, occupational therapy, and speech therapy may be needed to help the individual regain skills. Addressing sensory issues as well as providing counseling in one or more of a variety of possibilities may also be part of the treatment.

Re-introducing the individual to activities, school, work, family gatherings, etc. in a gradual, careful manner can also be a key to improvement. Pushing the person back into activities too much, too soon can be frightening and even result in loss of skills that have been regained or even loss of more skills. "Safe re-introduction" is key but exactly what that means for each individual varies greatly. Careful observation as activities are re-introduced is usually the best way to assess and then adjust activities as warranted by the person's response.

Will giving ivermectin to someone with downs on a daily basis help them

I am not aware of any study assessing ivermectin use in people with Down syndrome for COVID or any other indication.

The data for the use of ivermectin for COVID-19 in people without Down syndrome is based on small studies that were typically not double-blinded (the gold standard for clinical trials). In a double-blind study, neither the person receiving the treatment, nor the researcher know what is being given (the drug or the placebo) until the study ends. That study design eliminates a lot of potential bias. In addition, some of the studies measured factors, such as blood tests, have not been studied to determine that the change in the test is actually significant as far as preventing or treating COVID-19. Therefore, the CDC has concluded as of August 2021 that ivermectin is not recommended for treatment of COVID-19 at this time.

As far as other uses, it is approved for treatment for a variety of parasitic infections but not for other uses in people with or without Down syndrome.

Can you say more about how ECT is potentially a useful treatment for regression? I find that families hear ECT and are fearful of this approach. Thank you!

Electroconvulsive therapy (ECT) is an effective treatment for some individuals with catatonia, and/or severe depression as well as some other mental health conditions. More information can be found on the American Psychiatric Association [website](#). In our experience and in the experience of some of the doctors from the Down Syndrome Medical Interest Group- USA, treatment with ECT is more likely to be beneficial if it is done earlier in the course of the regression and catatonia. However, at our Center, we have patients who had catatonia and regression symptoms for several years before undergoing ECT who still responded well to ECT.

ECT can be frightening to some. Hollywood has done a good job of helping us remember disturbing, outdated ECT procedures, such as is seen in the movie *One Flew over the Cuckoo's Nest*. ECT does cause the person to have a seizure and that can be frightening, too. However, today it is typically done with the person under anesthesia, so the person is not aware of the procedure, does not remember it, and is safe during the procedure. In addition, some other brain stimulation procedures are available and brief descriptions are provided on the American Psychiatric Association [website](#).

My son's doctor put him on clonazepam instead of lorazepam for catatonia saying it was a similar drug. Have you seen improvement using clonazepam or should we ask for lorazepam instead?

It is a similar medication, however, the case series that have been published describing treatment of catatonia in several individuals with Down syndrome used lorazepam. Therefore, we have used lorazepam instead of any similar medications such as clonazepam for our patients with Down syndrome and catatonia.

I have a son who is 31 he is nonverbal. He has regressed from a 2-step skill now to a 1 step skill and sometimes doesn't do well with that. He's easy going. I really can't tell any difference but that. I had him tested by a neurological doctor. He decided he has the onset of dementia. He has put him on Memantine. I'm not happy with the medication. What are your thoughts?

Please say more about working with the health care provider on not jumping to the automatic conclusion that any loss in an adult with DS must have Alzheimer's disease. Practical Suggestions?

Alzheimer's dementia has been found to be uncommon in people with Down syndrome before the age of 40. It is not impossible for a person with Down syndrome to develop symptomatic Alzheimer's dementia at age 31 years, but it is unlikely. For any individual with Down syndrome who has a decline in skills, a thorough evaluation for causes is recommended. Recommendation #3 of the GLOBAL Medical Care Guidelines for Adults with Down Syndrome published in 2020 states: "Caution is needed when diagnosing age-related, Alzheimer's-type dementia in adults with Down syndrome younger than age 40 due to its low prevalence before this age." Further details can be found in the guidelines document at [this link](#).

Available data have not demonstrated benefit from treating people with Down syndrome with Alzheimer's disease with memantine. Interestingly, memantine can be used to treat catatonia, sometimes as an additional treatment to lorazepam.

Recommendation #4 in the GLOBAL Guidelines states:

“Medical professionals should assess adults with Down syndrome and interview their primary caregivers about changes from baseline function annually beginning at age 40. Decline in the following six domains as per the National Task Group – Early Detection Screen for Dementia (NTG-EDSD), should be used to identify early-stage age-related Alzheimer's-type dementia and/or a potentially reversible medical condition:

- Cognition, memory, and executive function
- Behavior and personality
- Communication
- Adaptive functioning
- Ambulation and motor skills
- General decline in established skills”

The guideline points out that the evaluation should focus on both the pattern of decline being consistent with that of Alzheimer's disease as well as ruling out other potentially reversible medical conditions. While Alzheimer's disease is more common in people with Down syndrome, there are many reasons people with Down syndrome can have a decline in skills and many of them are reversible. Even if the person with Down syndrome has Alzheimer's disease, treatment for other causes, if found, has the potential to temporarily improve the individual's level of function.

With these unique symptom manifestations with people with DS, would it be beneficial for my son (almost 12 years old) to be established at a DS clinic?

Should any of these symptoms start, what doctors should we go to? Our regular gp isn't qualified

The familiarity of providing health care for people with Down syndrome can improve the care of those individuals. However, research indicates only about 5% of people with Down syndrome in the United States have access to a clinic for people with Down syndrome. You can find information on the location of clinics in the United States for people with Down syndrome on [this site](#).

Unfortunately, with limited clinics available, it may still be challenging find a clinic in your area or that is available to you. Several organizations are working to provide information for your knowledge but also to share with your provider. We recommend:

- [LuMind IDSC myDSC](#)
- [Adult Down Syndrome Center Online Resource Library](#)

For providers, we encourage membership in the [Down Syndrome Medical Interest Group-USA](#). It is a professional organization that shares wonderful resources as well as opportunities to

interact with other health care providers. We encourage people with Down syndrome and their families to tell their providers about DSMIG-USA.

Do you recommend a baseline MRI or anything else for a 39-year-old with no issues other than thyroid and celiac?

I have a 56yr old male with Down syndrome; is there anything we can do regarding Alzheimer's

Currently there is no recommendation for screening MRIs for people with Down syndrome. The GLOBAL Medical Care Guidelines for Adults with Down Syndrome published in 2020 recommends:

“Medical professionals should assess adults with Down syndrome and interview their primary caregivers about changes from baseline function annually beginning at age 40. Decline in the following six domains as per the National Task Group – Early Detection Screen for Dementia (NTG-EDSD), should be used to identify early-stage age-related Alzheimer’s-type dementia and/or a potentially reversible medical condition:

- Cognition, memory, and executive function
- Behavior and personality
- Communication
- Adaptive functioning
- Ambulation and motor skills
- General decline in established skills”

Further information on the guidelines can be found at [this link](#).

Screening for Alzheimer’s disease with the evaluation noted above is recommended. If someone gets Alzheimer’s disease, there are no cures at this time but there are many things that can be done for the individual including:

- Assessing for and treating co-occurring conditions (such as depression, hypothyroidism, etc.)
- Treating symptoms
- Working with the individual in a variety of ways to improve or maintain their level of function as long as possible and to reduce psychological and behavioral symptoms.

More information on Alzheimer’s disease and dementia in people with Down syndrome can be found at [this link](#).

What vitamins need to be taken on a daily basis that will help to not get regression

This is a question for which there is not a definite answer. Unfortunately, there are not data that inform us as to how supplements may or may not prevent or reduce the chance of regression. We recommend a healthy diet that includes a variety of foods. Maintaining a healthy weight and regular exercise promote health in general. We know that poor health can trigger regression in

some individuals so, although research has not been done to prove these prevent regression, improving overall health is likely to be of benefit:

- [Regular and healthy sleep](#)
- [Healthy diet](#)
- [Regular exercise](#)

So we need more research, then right??

Yes, more research for people with DS is needed to improve our understanding of decline in skills in general as well as Regression (Down Syndrome Disintegrative Disorder) and Alzheimer's disease. The participation of people with Down syndrome and their families is a key to this process.