

Ataxia with Vitamin B12 Deficiency

Vitamin B12 deficiency happens when someone does not consume enough vitamin B12 in their diet, or their body isn't absorbing it properly. Vitamin B12 deficiency can cause physical, neurological, and psychological symptoms. Some people with vitamin B12 deficiency develop ataxia. Vitamin B12 deficiency is a treatable condition.

It is estimated that between 2-3% of adults in the United States have a vitamin B12 deficiency. Some people can be at a higher risk of developing a vitamin B12 deficiency. Risk factors include being over 75 years old, following a vegan or vegetarian diet, taking certain medications such as metformin, having digestive system disorders, having Sjögren's syndrome, or consuming excessive amounts of alcohol.

Most causes of Ataxia with Vitamin B12 Deficiency are acquired. This means that the ataxia symptoms are the result of an injury or illness. However, rare forms of Ataxia with Vitamin B12 Deficiency are caused by mutations in the MTFHR gene or TCN2 gene.

Symptoms

Ataxia with Vitamin B12 Deficiency causes poor balance and coordination. In fact, the word Ataxia means incoordination. There can also be problems coordinating muscles that control speech and swallowing. Other common symptoms include numbness or tingling in the hands or feet, vision problems, fatigue, pale or jaundiced skin, memory problems, and mood changes. These symptoms usually develop slowly over time.

If left untreated, Vitamin B12 Deficiency can cause long-term complications. This can include neuropathy, paralysis, incontinence, erectile dysfunction, paranoia, delusions, or memory loss.

Prognosis

Ataxia with Vitamin B12 Deficiency is treatable with vitamin B12 supplements. Options for vitamin B12 treatment include oral medications, injections, or a nasal spray. Depending on the severity of symptoms, a doctor may prescribe cyanocobalamin or hydroxocobalamin, which are manufactured forms of vitamin B12. Patients may also be recommended to eat more foods that are rich in B12, such as meat, fish, eggs, or dairy products. Vitamin B12 is only naturally found in animal products. However, vegan and vegetarian foods that are supplemented with vitamin B12 can be an alternative.

The underlying cause of the Vitamin B12 Deficiency determines the length of treatment. Some people may only need treatment until their vitamin B12 levels are back to normal. Others may need vitamin B12 therapy for the rest of their life.

The underlying cause of Vitamin B12 Deficiency and the amount of time the patient was untreated can impact the effectiveness of treatment. Most people with Ataxia with Vitamin B12 Deficiency see improvement in symptoms. However, if significant damage occurred to the body before treatment, people with Ataxia with Vitamin B12 Deficiency may experience some long-term symptoms.

People with Ataxia with Vitamin B12 Deficiency may benefit from exercise, physical therapy, and occupational therapy. Consulting a Dietitian can be helpful for dietary recommendations.

Diagnosis

A neurologic examination can determine whether a person has symptoms typical of Ataxia with Vitamin B12 Deficiency. A neurologist is often the most helpful specialist in recognizing symptoms and diagnosing the disease that causes Ataxia. Doctors will order blood tests to determine whether someone has a vitamin B12 deficiency. These tests may include a vitamin B12 level test, complete blood count (CBC), methylmalonic acid (MMA) test, or Homocysteine test. Another potential follow-up test is an MRI of the brain or spine.



Prognosis

Ataxia caused by chronic alcohol use typically requires the long-term daily consumption of significant quantities of alcohol. However, the exact amount may vary person to person based on underlying factors. This can include nutrition (especially Thiamine/Vitamin B1), other substance use (tobacco and cannabis), biological sex, genetic factors, and overall health.

There is limited demographic information available on people who have Ataxia caused by chronic alcohol use. These disorders tend to affect more men than women.

Alcohol cessation, meaning stopping or significantly reducing alcohol consumption, is recommended to help manage ataxia symptoms. Suddenly stopping the consumption of alcohol after chronic use can cause severe, life-threatening withdrawal symptoms. Medical support is recommended during the alcohol withdrawal process. Supportive treatments including Thiamine supplementation, physical therapy, and social support can also significantly improve the lives of people with Ataxia caused by chronic alcohol use.

Wernicke encephalopathy can be life-threatening. Emergency treatment with Thiamine is critical to prevent long-term brain damage.

Some recovery from Ataxia caused by chronic alcohol use is possible. This depends on the amount of damage to the cerebellum that has occurred. There is a better prognosis if someone is diagnosed and begins treatment early. It is more challenging to recover after significant damage has occurred.

Diagnosis

Early diagnosis is critical for A-T symptom management. Depending on when or in what order symptoms begin to show, different clinical specialists will be involved in the diagnosis process. Clinical examinations from a doctor can help assess balance, coordination, vision, and speech impairments. MRI scans are often used to confirm cerebellar atrophy or degeneration. Blood tests can be used to detect elevated levels of molecules that suggest someone has A-T. However, a specific diagnosis of A-T can only be made by a genetic test. Genetic testing can identify the specific mutations present in the ATM gene.