LHON & Vitamin B12 Deficiency

Vitamin B12[1] is an essential vitamin found in animal products such as meat, fish, and dairy. It can also be made in a lab and is often taken with other B vitamins. Vitamin B12 is required for the function and development of many parts of the body, including the brain,[2] nerves, and blood cells.[3]

It is recommended that individuals with an LHON mutation, with and without vision loss, be aware of the increased risk for vitamin B12 deficiency and the potential benefit of screening.

Some research indicates that some individuals with an LHON mutation may be more likely to have low B12 than the general population, so it may be helpful to share the information in this handout with one's physician.

What is vitamin B12 deficiency?

Large quantities of vitamin B12 are stored in the liver, so a deficiency can take months or years to develop. Risk factors for vitamin B12 deficiency may be linked to reduced dietary intake or impaired absorption. Reduced dietary intake may be linked to a vegan diet or malnutrition. Impaired absorption may be linked to excessive alcohol intake, reduced stomach acid secretion (e.g., antacid use), gastric surgery, pernicious anemia, and gastrointestinal disorders such as inflammatory bowel disease or Celiac disease.

What are the symptoms?

Symptoms of vitamin B12 deficiency include anemia and other blood count abnormalities, neuropathy, and cognitive impairment; however, people may be asymptomatic early in vitamin B12 deficiency.

How is B12 evaluated?

Vitamin B12 levels can be evaluated with a simple blood test. If levels are in the low-to-normal range, a doctor may also check homocysteine or methylmalonic acid levels to confirm the deficiency.

How is low B12 treated?

Vitamin B12 deficiency can be treated with oral tablets or intramuscular injections (shots). Even in cases of impaired absorption, oral tablets can be effective. It's important to follow up with the doctor after treatment to ensure a normal level has been achieved and is maintained.

How can I adjust my risk of having a vitamin B12 deficiency?

Some risk factors that can lead to vitamin B12 deficiency cannot be changed, such as a disease. Some lifestyle factors can be modified to help prevent or treat vitamin B12 deficiency, such as increasing the intake of animal products in the diet (or taking appropriate supplements if vegetarian or vegan) or reducing alcohol consumption.

What research has been done in the LHON population?

A large study of vitamin B12 deficiency in 244 people carrying an LHON mutation (with and without vision loss) was published in 2022.[4] Key findings include:

- The prevalence of vitamin B12 deficiency among those under age 65 was about 20%, compared to only 6% in the general population in the same age range.
- Among those in the study with vitamin B12 deficiency, a higher-than-normal percentage also had neuropathy.
- The study showed a strong correlation between high alcohol consumption and vitamin B12 deficiency.

The study authors recommend that individuals with an LHON mutation, with and without vision loss, be aware of the increased risk for vitamin B12 deficiency and the potential benefit of screening.

Relevant Research

These articles may be of interest to you and your health care providers:

1 Vitamin B12. The Nutrition Source, Harvard T.H. Chan School of Public Health. Accessed October 17, 2022. https://www.hsph.harvard.edu/nutritionsource/vitamin-b12

2 Hoffman M. Picture of the Brain, Human Anatomy. WebMD. Medically reviewed June 23, 2021. Accessed October 17, 2022. https://www.webmd.com/brain/picture-of-the-brain

3 Hoffman M. Picture of Blood, Human Anatomy. WebMD. Medically reviewed June 23, 2021. Accessed October 17, 2022. https://www.webmd.com/heart/anatomy-picture-of-blood

4 Zibold J, von Livonius B, Kolarova H, Rudolph G, Priglinger CS, Klopstock T, Catarino CB. Vitamin B12 in Leber hereditary optic neuropathy mutation carriers: a prospective cohort study. Orphanet Journal of Rare Diseases. 2022; 17:310. doi:10.1186/s13023-022-02453-z

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