Published on *My46* (https://www.my46.org) 01/23/2022

My46 Trait Profile

Biotinidase deficiency

Biotinidase deficiency is genetic condition characterized by the body's inability to reuse and recycle a vitamin called biotin. It is caused by mutations in the BTD gene that make an enzyme called biotinidase.

Characteristics of Biotinidase deficiency

Biotinidase deficiency is a condition characterized by a lack of the biotinidase enzyme. This enzyme is responsible for cutting biotin out of food. Biotin is an important vitamin that helps the body function (grow and develop) properly. Most individuals usually get biotin from the foods they eat because the biotinidase enzyme cuts it out of the food into a form that can be used and recycled in the body. There are two types of biotinidase deficiency: profound and partial.

Individuals with profound biotinidase deficiency who are not treated can have a range of signs and symptoms. These can include seizures, weak muscle tone (hypotonia), breathing problems, and delayed development. If left untreated, lifelong complications such as hearing loss, vision loss, problems with movement and balance (ataxia), skin rashes, and hair loss can result. However, individuals who are diagnosed and treated shortly after birth with biotin supplements avoid most if not all complications with lifelong treatment.

Individuals with partial biotinidase deficiency have a milder form of the condition. This is because they still have some residual (remaining) biotinidase enzyme in their bodies. Symptoms typically only appear during times of illness, infection, or other metabolic stress. These symptoms may include hypotonia, skin rashes, and hair loss. Many individuals with partial biotinidase deficiency may take biotin supplements in infancy and childhood. However, there is not a clear consensus as to whether lifelong treatment is beneficial or necessary for these individuals.

Diagnosis/Testing

Individuals with biotinidase deficiency have changes or mutations in a gene called BTD. This gene makes the biotinidase enzyme. This enzyme is responsible for cutting biotin from food that is eaten. Mutations in the BTD gene can cause partial deficiency (enzyme works a little) or profound deficiency (enzyme does not work at all).

Many babies with biotinidase deficiency are diagnosed early in life through newborn screening (NBS). NBS tests a spot of blood from the baby's heel, and looks to see if the biotinidase enzyme is working properly. NBS test results are confirmed with additional blood tests, and possibly genetic testing of the BTD gene.

Management/Surveillance

Management of biotinidase deficiency is very simple. It is recommended that these individuals be treated with a special biotin supplement called "unbound" biotin (typically 5-10 mg/day) to meet the body's needs. Usually with the treatment of unbound biotin supplementation, all clinical symptoms can be prevented and individuals should have normal growth and development.

Mode of inheritance

Biotinidase deficiency is inherited in an autosomal recessive pattern. This means that an individual has to inherit two BTD mutations (i.e., one from each parent) to be affected with biotinidase deficiency. If both parents are carriers of a BTD mutation, they have a 1 in 4 (25%) chance with each pregnancy of having a child with biotinidase deficiency. Babies born in the United States are screened for biotinidase deficiency by newborn screening.

Risk to family members

Parents of a child with biotinidase deficiency are carriers of biotinidase deficiency. If a sibling of a child with biotinidase deficiency is unaffected, he/she has a 2 in 3 (66%) chance of being a carrier of biotinidase deficiency.

Special considerations

None

Resources

Genetics Home Reference: Biotinidase deficiency
http://ghr.nlm.nih.gov/condition/biotinidase-deficiency
Biotinidase deficiency Family Support Group
http://biotinidasedeficiency.20m.com

References

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