BTD Gene

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biotinidase

genes

1. Normal Function

The *BTD* gene provides instructions for making an enzyme called biotinidase. This enzyme recycles biotin, a B vitamin found in foods such as liver, egg yolks, and milk. Biotinidase removes biotin that is bound to proteins in food, leaving the vitamin in its free (unbound) state. The body needs free biotin to activate enzymes called biotin-dependent carboxylases. These carboxylases are involved in many critical cellular functions, including the breakdown of proteins, fats, and carbohydrates.

In addition to processing biotin obtained from the diet, biotinidase recycles biotin within the body. As biotin-dependent carboxylases are broken down, they release a molecule called biocytin. Biocytin is a complex made of up biotin and a protein building block (amino acid) called lysine. Biotinidase splits this complex, making free biotin available for reuse by other carboxylase enzymes.

Researchers suspect that biotinidase may have several additional functions. This enzyme may transport free biotin through the bloodstream. It might also have the ability to attach biotin to certain proteins through a process called biotinylation. Within the nucleus, biotinylation of DNA-associated proteins called histones may help determine whether certain genes are turned on or off. It is unclear, however, whether biotinidase plays a role in regulating gene activity.

2. Health Conditions Related to Genetic Changes

2.1. Biotinidase Deficiency

More than 150 mutations in the *BTD* gene have been identified in people with biotinidase deficiency. This disorder, if untreated, can affect many parts of the body and cause delayed development. Most of the mutations that cause biotinidase deficiency change single amino acids in the biotinidase enzyme. These changes occur in critical regions of the enzyme and reduce or eliminate the enzyme's activity.

Most *BTD* gene mutations cause profound biotinidase deficiency. This severe form of the disorder results when the activity of biotinidase is reduced to less than 10 percent of normal. Other mutations cause a milder form of the condition called partial biotinidase deficiency. These mutations reduce biotinidase activity to between 10 percent and 30 percent of normal. Without enough of this enzyme, biotin cannot be recycled. The resulting shortage of free biotin impairs the activity of biotin-dependent carboxylases, leading to a buildup of potentially toxic compounds in the body. If the condition is not treated promptly, this buildup damages various cells and tissues, causing the signs and symptoms associated with biotinidase deficiency.

2.2. Leigh Syndrome

Leigh syndrome

3. Other Names for This Gene

- Biocytin Hydrolase

References

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