MOCS1 Gene

Subjects: Genetics & Heredity
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molybdenum cofactor synthesis 1

genes

1. Introduction

The *MOCS1* gene provides instructions for making two different proteins, MOCS1A and MOCS1B. Both are involved in the formation (biosynthesis) of a molecule called molybdenum cofactor. Specifically, MOCS1A and MOCS1B perform the first of a series of reactions that produce the cofactor, although the function of MOCS1B in this process is not understood. Molybdenum cofactor, which contains the element molybdenum, is essential to the function of several enzymes called sulfite oxidase, aldehyde oxidase, xanthine dehydrogenase, and mitochondrial amidoxime reducing component (mARC). These enzymes help break down (metabolize) different substances in the body, some of which are toxic if not metabolized.

2. Health Conditions Related to Genetic Changes

2.1. Molybdenum cofactor deficiency

MOCS1 gene mutations cause a disorder called molybdenum cofactor deficiency. This disorder is characterized by seizures that begin early in life and brain dysfunction that worsens over time (encephalopathy); the condition is usually fatal by early childhood. At least 32 mutations in the *MOCS1* gene have been found to cause a form of the disorder designated type A or complementation group A. This is the most common form of the condition, accounting for approximately two-thirds of cases.

The *MOCS1* gene mutations involved in molybdenum cofactor deficiency likely eliminate the function of MOCS1A, MOCS1B, or both, although in rare cases that are less severe, some protein function may remain. Without the activity of one or both of these proteins, molybdenum cofactor biosynthesis is impaired. Loss of the cofactor impedes the function of the metabolic enzymes that rely on it.

The resulting loss of enzyme activity leads to buildup of certain chemicals, including sulfite, S-sulfocysteine, xanthine, and hypoxanthine, and low levels of another chemical called uric acid. (Testing for these chemicals can help in the diagnosis of this condition.) Sulfite, which is normally broken down by sulfite oxidase, is toxic, especially

to the brain. Researchers suggest that damage caused by the abnormally high levels of sulfite (and possibly other chemicals) leads to encephalopathy, seizures, and the other features of molybdenum cofactor deficiency.

3. Other Names for This Gene

- cell migration-inducing gene 11 protein
- MIG11
- migration-inducing gene 11 protein
- MOCOD
- MOCODA
- MOCS1A enzyme
- molybdenum cofactor biosynthesis protein 1
- molybdenum cofactor biosynthesis protein A
- molybdenum cofactor synthesis-step 1 protein A-B

References

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