MOCS2 gene

molybdenum cofactor synthesis 2

Normal Function

The *MOCS2* gene provides instructions for making two different proteins, MOCS2A and MOCS2B, which combine to form an enzyme called molybdopterin synthase. Molybdopterin synthase performs the second of a series of reactions in the formation (biosynthesis) of a molecule called molybdenum cofactor. 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.

Health Conditions Related to Genetic Changes

Molybdenum cofactor deficiency

MOCS2 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 a dozen mutations in the *MOCS2* gene have been found to cause a form of the disorder designated type B or complementation group B.

The *MOCS2* gene mutations involved in molybdenum cofactor deficiency likely eliminate the function of MOCS2A, MOCS2B, or both, although in rare cases that are less severe, some protein function may remain. Without either piece of molybdopterin synthase, 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.

Other Names for This Gene

- MCBPE
- MOCO1
- MOCODB
- molybdenum cofactor biosynthesis protein E
- molybdopterin synthase catalytic subunit large subunit MOCS2B
- molybdopterin synthase small and large subunit
- molybdopterin synthase sulfur carrier subunit
- molybdopterin synthase sulfur carrier subunit small subunit MOCS2A
- MPTS

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of MOCS2 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=4338[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28MOCS2%5BTIAB%5D%29+OR+%28MOCO1%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- MOLYBDENUM COFACTOR SYNTHESIS GENE 2; MOCS2 (https://omim.org/entry/603708)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/4338)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=MOCS2[gene])

References

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- Reiss J, Johnson JL. Mutations in the molybdenum cofactor biosynthetic genesMOCS1, MOCS2, and GEPH. Hum Mutat. 2003 Jun;21(6):569-76. doi:10.1002/humu.10223. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/12754701)

Genomic Location

The *MOCS2* gene is found on chromosome 5 (https://medlineplus.gov/genetics/chromosome/5/).

Last updated March 1, 2014