MOCOS gene

molybdenum cofactor sulfurase

Normal Function

The *MOCOS* gene provides instructions for making an enzyme called molybdenum cofactor sulfurase. This enzyme is necessary for the function of two other enzymes, xanthine dehydrogenase and aldehyde oxidase. Xanthine dehydrogenase is involved in the normal breakdown of purines, which are building blocks of DNA and its chemical cousin, RNA. Specifically, it carries out the final two steps in the process: the conversion of a molecule called hypoxanthine to another molecule called xanthine, and the conversion of xanthine to uric acid, a waste product that is normally excreted in urine and feces. Less is known about the function of aldehyde oxidase, although it appears to play a role in the breakdown (metabolism) of many different compounds.

Molybdenum cofactor sulfurase carries out a chemical reaction that adds sulfur to a molecule called the molybdenum cofactor. This molecule is required for xanthine dehydrogenase and aldehyde oxidase to be turned on (activated) and carry out their functions.

Health Conditions Related to Genetic Changes

Hereditary xanthinuria

At least four mutations in the *MOCOS* gene have been found to cause hereditary xanthinuria type II, a condition that most often affects the kidneys. Most of these mutations change a single protein building block (amino acid) in molybdenum cofactor sulfurase. The effects of these mutations are not fully understood, but they likely alter the shape and function of the enzyme. If molybdenum cofactor sulfurase is unable to add sulfur to the molybdenum cofactor, xanthine dehydrogenase and aldehyde oxidase are not activated. The loss of aldehyde oxidase activity does not appear to cause any signs or symptoms. However, the loss of xanthine dehydrogenase activity prevents the conversion of xanthine to uric acid, leading to an accumulation of xanthine in the kidneys and other tissues. The excess xanthine can form tiny crystals that accumulate in the kidneys, occasionally leading to the formation of stones that can impair kidney function and ultimately cause kidney failure. Less commonly, xanthine crystals build up in the muscles, causing pain and cramping. In some people with hereditary xanthinuria type II, the condition does not cause any health problems.

Other Names for This Gene

- FLJ20733
- HMCS
- MCS
- MOS

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of MOCOS (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=55034[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28%28MOCOS%5BTIAB%5D%29+OR+%28molybdenum+cofactor+sulfurase%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D)

Catalog of Genes and Diseases from OMIM

- MOLYBDENUM COFACTOR SULFURASE; MOCOS (https://omim.org/entry/613274)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/55034)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=MOCOS[gene])

References

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terminal domain of the Human Molybdenum Cofactor Sulfurase (HMCS)associated with type II classical xanthinuria. Mol Genet Metab. 2007May;91(1):23-9. doi: 10.1016/j.ymgme.2007.02.005. Epub 2007 Mar 23. Citation on PubMed (https://pubmed.ncbi.nlm.nih.gov/17368066)

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Genomic Location

The *MOCOS* gene is found on chromosome 18 (https://medlineplus.gov/genetics/chromosome/18/).

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