**XDH gene**

xanthine dehydrogenase

**Normal Function**

The *XDH* gene provides instructions for making an enzyme called xanthine dehydrogenase. This enzyme 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.

Xanthine dehydrogenase has been studied extensively because it can be involved in the production of molecules called superoxide radicals. Specifically, xanthine dehydrogenase is sometimes converted to another form called xanthine oxidase, which produces superoxide radicals. These molecules are byproducts of normal cell processes, and they must be broken down regularly to avoid damaging cells. Superoxide radicals are thought to play a role in many diseases, including heart disease and high blood pressure (hypertension).

Researchers suspect that xanthine dehydrogenase plays a role in milk production (lactation) in women. However, the enzyme's role in lactation is unclear.

**Health Conditions Related to Genetic Changes**

**Hereditary xanthinuria**

At least 12 mutations in the *XDH* gene have been found to cause hereditary xanthinuria type I, a condition that most often affects the kidneys. These mutations reduce or eliminate the activity of xanthine dehydrogenase. As a result, the enzyme is not available to carry out the last two steps of purine breakdown. Because xanthine is not converted to uric acid, affected individuals have high levels of xanthine and very low levels of uric acid in their blood and urine. The excess xanthine can accumulate in the kidneys and other tissues. In the kidneys, xanthine forms tiny crystals that occasionally build up to create kidney stones. These stones 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, the condition does not cause any health problems.
Chromosomal Location

Cytogenetic Location: 2p23.1, which is the short (p) arm of chromosome 2 at position 23.1

Molecular Location: base pairs 31,334,320 to 31,414,777 on chromosome 2 (Homo sapiens Updated Annotation Release 109.20200228, GRCh38.p13) (NCBI)

Other Names for This Gene

• xanthine dehydrogenase/oxidase
• xanthine oxidoreductase
• XO
• XOR

Additional Information & Resources

Educational Resources

• Basic Neurochemistry (sixth edition, 1999): Purine Release and Metabolism
  https://www.ncbi.nlm.nih.gov/books/NBK28118/

Scientific Articles on PubMed

• PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28xanthine+oxidase%5BTI%5D%29+OR+%28xanthine+dehydrogenase%5BTI%5D%29%29+OR+%28xanthine+oxidoreductase%5BTI%5D%29+AND+english%5Blanguage%5D+AND+human%5Bspecies%5D+AND+last+720+days+AND+last+720+days%22+AND+last+720+days%22+AND+last+720+days%22+AND+last+720+days%22

Catalog of Genes and Diseases from OMIM

• XANTHINE DEHYDROGENASE
  http://omim.org/entry/607633
Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_XDH.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=XDH%5Bgene%5D
- HGNC Gene Symbol Report
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:7498
- NCBI Gene
- UniProt
  https://www.uniprot.org/uniprot/P47989

Sources for This Summary

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/9153281
  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC508078/

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/8224915

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/23203137
  Free article on PubMed Central: https://www.ncbi.nlm.nih.gov/pmc/articles/PMC3509653/

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- OMIM: XANTHINE DEHYDROGENASE
  http://omim.org/entry/607633

  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/8661045

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