SUOX gene

sulfite oxidase

Normal Function

The *SUOX* gene provides instructions for making an enzyme called sulfite oxidase, which helps break down protein building blocks (amino acids) that contain sulfur when they are no longer needed. Specifically, sulfite oxidase is involved in the final step of this process, in which sulfur-containing molecules called sulfites are converted to other molecules called sulfates by adding an oxygen atom (a process called oxidation).

Health Conditions Related to Genetic Changes

Isolated sulfite oxidase deficiency

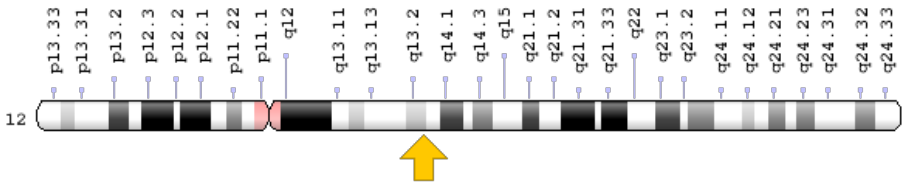
At least 27 *SUOX* gene mutations have been identified in people with isolated sulfite oxidase deficiency (ISOD), a severe disorder that causes brain damage and is generally fatal in the first months or years of life. The *SUOX* gene mutations that cause ISOD impair the function of sulfite oxidase, preventing complete breakdown of sulfur-containing amino acids. As a result, sulfites and other compounds left over from the partial breakdown process abnormally accumulate in the body. Researchers suggest that the nervous system is especially sensitive to this abnormal accumulation, and excessive levels of sulfite compounds that are toxic to the brain are thought to result in the brain damage that occurs in ISOD.

Polycystic ovary syndrome

Chromosomal Location

Cytogenetic Location: 12q13.2, which is the long (q) arm of chromosome 12 at position 13.2

Molecular Location: base pairs 55,992,547 to 56,005,525 on chromosome 12 (Homo sapiens Updated Annotation Release 109.20200522, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- sulfite oxidase, mitochondrial

Additional Information & Resources

Clinical Information from GeneReviews

- Isolated Sulfite Oxidase Deficiency
  https://www.ncbi.nlm.nih.gov/books/NBK453433

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28SUOX%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+2160+days%22%5Bdp%5D

Catalog of Genes and Diseases from OMIM

- SULFITE OXIDASE
  http://omim.org/entry/606887

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_SUOX.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=SUOX%5Bgene%5D

- HGNC Gene Symbol Report
  https://www.genenames.org/data/gene-symbol-report/#!/hgnc_id/HGNC:11460
- Monarch Initiative
  https://monarchinitiative.org/gene/NCBIGene:6821
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/6821
- UniProt
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