



SOD1 gene

superoxide dismutase 1

Normal Function

The *SOD1* gene provides instructions for making an enzyme called superoxide dismutase, which is abundant in cells throughout the body. This enzyme attaches (binds) to molecules of copper and zinc to break down toxic, charged oxygen molecules called superoxide radicals. The molecules are byproducts of normal cell processes, and they must be broken down regularly to avoid damaging cells.

Health Conditions Related to Genetic Changes

Amyotrophic lateral sclerosis

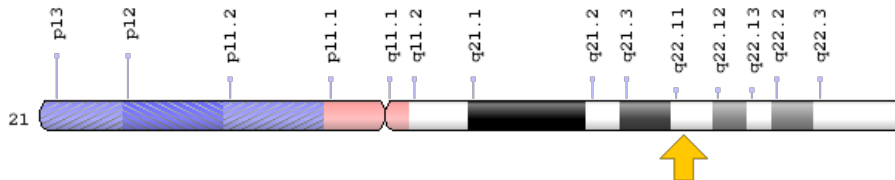
At least 200 mutations in the *SOD1* gene have been found to cause amyotrophic lateral sclerosis (ALS), a condition characterized by progressive muscle weakness, a loss of muscle mass, and an inability to control movement. Most of these mutations change one of the protein building blocks (amino acids) in the superoxide dismutase enzyme. About half of all Americans with ALS caused by *SOD1* gene mutations have a particular mutation that replaces the amino acid alanine with the amino acid valine at position 5 in the enzyme, written as Ala5Val or A5V. (Because of variations in the ways amino acids are counted in proteins, this mutation is sometimes called Ala4Val or A4V.) ALS caused by the A5V mutation is generally associated with a shorter life expectancy compared with ALS caused by other genetic mutations.

ALS is caused by the death of nerve cells that control muscle movement (motor neurons). It is unclear why these cells are particularly sensitive to *SOD1* gene mutations. Researchers have suggested several ways in which the altered enzyme may cause the death of motor neurons. These possibilities include an increase in harmful superoxide radicals, increased production of other types of toxic radicals, increased cell death, or accumulation of clumps (aggregates) of misfolded superoxide dismutase that may be toxic to cells.

Chromosomal Location

Cytogenetic Location: 21q22.11, which is the long (q) arm of chromosome 21 at position 22.11

Molecular Location: base pairs 31,659,622 to 31,668,931 on chromosome 21 (Homo sapiens Updated Annotation Release 109.20190607, GRCh38.p13) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- ALS1
- Cu/Zn superoxide dismutase
- indophenoloxidase A
- IPOA
- SODC_HUMAN
- superoxide dismutase 1, soluble
- superoxide dismutase-1, soluble
- superoxide dismutase 1, soluble (amyotrophic lateral sclerosis 1 (adult))
- superoxide dismutase, cystolic

Additional Information & Resources

Educational Resources

- Biochemistry (fifth edition, 2002): Toxic Derivatives of Molecular Oxygen Such as Superoxide Radical are Scavenged by Protective Enzymes
<https://www.ncbi.nlm.nih.gov/books/NBK22505/#A2522>
- Washington University, St. Louis Neuromuscular Disease Center
<https://neuromuscular.wustl.edu/synmot.html#so>

Clinical Information from GeneReviews

- Amyotrophic Lateral Sclerosis Overview
<https://www.ncbi.nlm.nih.gov/books/NBK1450>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28SOD1%5BTI%5D%29+OR+%28superoxide+dismutase+1%5BTI%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5BIa%5D+AND+human%5Bmh%5D+AND+%22last+720+days%22%5Bdp%5D>

Catalog of Genes and Diseases from OMIM

- SUPEROXIDE DISMUTASE 1
<http://omim.org/entry/147450>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_SOD1.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=SOD1%5Bgene%5D>
- HGNC Gene Symbol Report
https://www.genenames.org/data/gene-symbol-report#!/hgnc_id/HGNC:11179
- Monarch Initiative
<https://monarchinitiative.org/gene/NCBIGene:6647>
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/6647>
- UniProt
<https://www.uniprot.org/uniprot/P00441>

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