

## **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.

### **Other Names for This Gene**

- ALS1
- Cu/Zn superoxide dismutase
- indophenoloxidase A

- IPOA
- SODC\_HUMAN
- superoxide dismutase 1, soluble
- superoxide dismutase 1, soluble (amyotrophic lateral sclerosis 1 (adult))
- superoxide dismutase, cytosolic
- superoxide dismutase-1, soluble

## Additional Information & Resources

### Tests Listed in the Genetic Testing Registry

- Tests of SOD1 ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=6647\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=6647[geneid]))

### Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?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%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+720+days%22%5Bdp%5D%29%29%29>)

### Catalog of Genes and Diseases from OMIM

- SUPEROXIDE DISMUTASE 1; SOD1 (<https://omim.org/entry/147450>)

### Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/6647>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=SOD1\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=SOD1[gene]))

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## Genomic Location

The *SOD1* gene is found on chromosome 21 (<https://medlineplus.gov/genetics/chromosome/21/>).

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