

kgXref.geneSymbol

knownGene.name

## Human Gene **SOD1** (**uc002ypa.2**) Description and Page Index

pulled from RefSeq page based on kgXref.refseq

**Description:** superoxide dismutase 1, soluble

kgXref.description

**RefSeq Summary (NM\_000454):** The protein encoded by this gene binds copper and zinc ions and is one of two isozymes responsible for free superoxide radicals in the body. The encoded isozyme is a soluble cytoplasmic protein, acting as a homodimer that normally-occurring but harmful superoxide radicals to molecular oxygen and hydrogen peroxide. The other isozyme is a mitochondrial protein. Mutations in this gene have been implicated as causes of familial amyotrophic lateral sclerosis. Rare transcript variants have been reported for this gene. [provided by RefSeq].

**Strand:** + **Genomic Size:** 9307 **Exon Count:** 5 **Coding Exon Count:** 5

<b>Page Index</b>	Sequence and Links	UniProtKB Comments	CTD	Microarray	RNA Structure
Protein Structure	Other Species	GO Annotations	mRNA Descriptions	Pathways	Other Names
Model Information	Methods				

### - Sequence and Links to Tools and Databases

Genomic Sequence (chr21:33,031,935-33,041,241)		mRNA (may differ from genome)		Protein (154 aa)	
Gene Sorter	Genome Browser	Protein FASTA	VisiGene	Table Schema	CGAP
Ensembl	Entrez Gene	ExonPrimer	GeneCards	Gepis Tissue	HGNC
HPRD	Human Cortex Gene Expression	Jackson Lab	OMIM	PubMed	Stanford SOURCE
Treefam	UniProtKB	User annotations			

### - Comments and Description Text from UniProtKB

**ID:** [SODC\\_HUMAN](#)

**DESCRIPTION:** RecName: Full=Superoxide dismutase [Cu-Zn]; EC=1.15.1.1;

**FUNCTION:** Destroys radicals which are normally produced within the cells and which are toxic to biological systems.

**CATALYTIC ACTIVITY:** 2 superoxide + 2 H(+) = O(2) + H(2)O(2).

**COFACTOR:** Binds 1 copper ion per subunit.

**COFACTOR:** Binds 1 zinc ion per subunit.

**SUBUNIT:** Homodimer. The pathogenic variants ALS1 Arg-38, Arg-47, Arg-86 and Ala-94 interact with RNF19A, whereas wild-type protein does not.

**INTERACTION:** P26339:Chga (xeno); NbExp=2; IntAct=EBI-990792, EBI-990900; P16014:Chgb (xeno); NbExp=2; IntAct=EBI-990792, EBI-990820; Q62313:Tgoln1 (xeno); NbExp=1; IntAct=EBI-990792, EBI-991369;

**SUBCELLULAR LOCATION:** Cytoplasm.

**PTM:** Unlike wild-type protein, the pathogenic variants ALS1 Arg-38, Arg-47, Arg-86 and Ala-94 are polyubiquitinated by RNF19A; which leads to their proteasomal degradation.

**DISEASE:** Defects in SOD1 are the cause of amyotrophic lateral sclerosis type 1 (ALS1) [[MIM:105400](#)]. ALS1 is a familial form of amyotrophic lateral sclerosis, a neurodegenerative disorder affecting upper and lower motor neurons and resulting in fatal paralysis. Sensory abnormalities are absent. Death usually occurs within 2 to 5 years. The etiology of amyotrophic lateral sclerosis is likely to be multifactorial, involving both genetic and environmental factors. The disease is inherited in 5-10% of cases leading to familial forms.

**MISCELLANEOUS:** The protein (both wild-type and ALS1 variants) has a tendency to form fibrillar aggregates in the absence of the intramolecular disulfide bond or of bound zinc ions. These aggregates may have cytotoxic effects. Zinc binding promotes dimerization and stabilizes the native form.

**SIMILARITY:** Belongs to the Cu-Zn superoxide dismutase family.

**WEB RESOURCE:** Name=Alsod; Note=ALS genetic mutations db; URL="http://alsod.iop.kcl.ac.uk/Als/";

**WEB RESOURCE:** Name=GeneReviews; URL="http://www.genetests.org/query?gene=SOD1";

**WEB RESOURCE:** Name=NIEHS-SNPs; URL="http://egp.gs.washington.edu/data/sod1/";

**WEB RESOURCE:** Name=Wikipedia; Note=Superoxide dismutase entry; URL="http://en.wikipedia.org/wiki/Superoxide\_dismutase";

## - Comparative Toxicogenomics Database (CTD)

The following chemicals interact with this gene

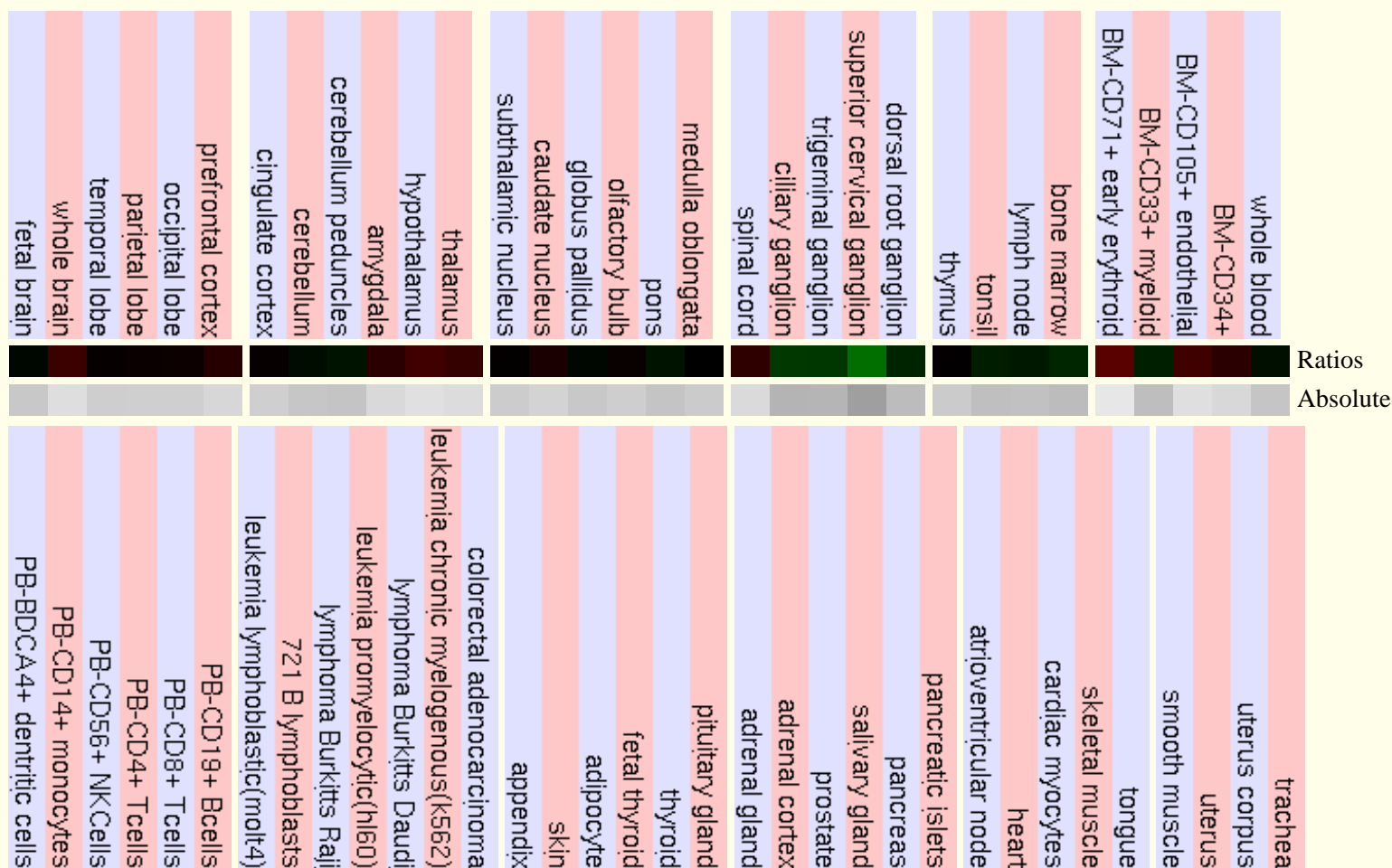
- [D010269](#) Paraquat
- [D003300](#) Copper
- [C467567](#) lenalidomide
- [D019256](#) Cadmium Chloride
- [D013792](#) Thalidomide
- [D015032](#) Zinc
- [D003314](#) Corn Oil
- [D011078](#) Polychlorinated Biphenyls
- [C017160](#) cypermethrin
- [C017947](#) sodium arsenite

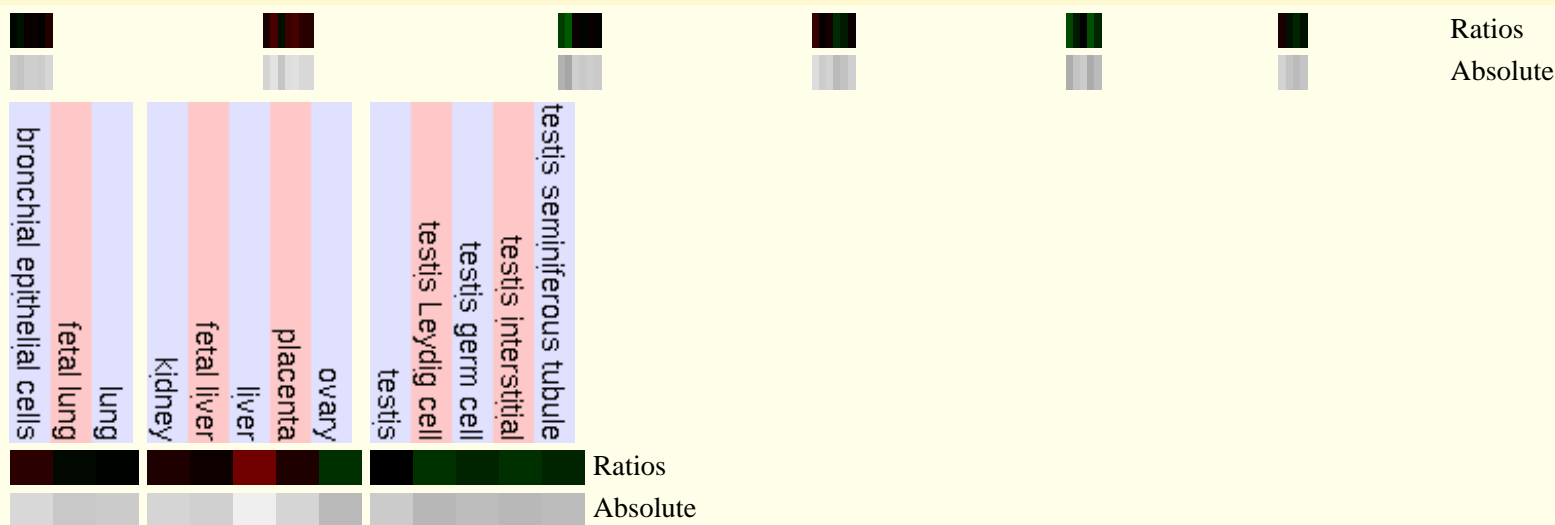
more ... [click here to view the complete list](#)

## - Microarray Expression Data

Expression ratio colors:

### GNF Expression Atlas 2 Data from U133A and GNF1H Chips





## mRNA Secondary Structure of 3' and 5' UTRs

Region	Fold Energy	Bases	Energy/Base	Display As		
5' UTR	-58.92	148	-0.398	Picture	PostScript	Text
3' UTR	-68.14	350	-0.195	Picture	PostScript	Text

The RNAfold program from the [Vienna RNA Package](#) is used to perform the secondary structure predictions and folding calculations. The estimated folding energy is in kcal/mol. The more negative the energy, the more secondary structure the RNA is likely to have.

## Protein Domain and Structure Information

**InterPro Domains:** [Graphical view of domain structure](#)

[IPR018152](#) - SOD\_Cu/Zn\_BS

[IPR001424](#) - SOD\_Cu\_Zn

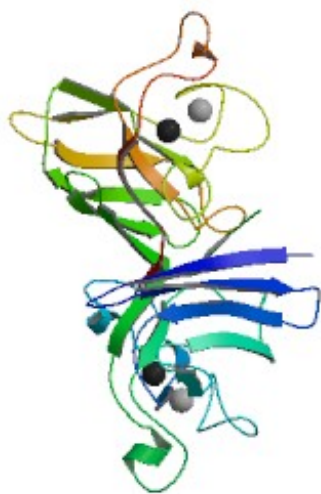
**Pfam Domains:**

[PF00080](#) - Copper/zinc superoxide dismutase (SODC)

**SCOP Domains:**

[49329](#) - Cu,Zn superoxide dismutase-like

**Protein Data Bank (PDB) 3-D Structure**



[1AZV](#) - X-ray [Chimera](#)



[1BA9](#) - NMR [Chimera](#)



[1DSW](#) - NMR [Chimera](#)

*To conserve bandwidth, only the images from the first 3 structures are shown.*

[1FUN](#) - X-ray [Chimera](#)

[1HL4](#) - X-ray [Chimera](#)

[1HL5](#) - X-ray [Chimera](#)

[1KMG](#) - NMR [Chimera](#)

[1L3N](#) - NMR [Chimera](#)

[1MFM](#) - X-ray [Chimera](#)

[1N18](#) - X-ray [Chimera](#)

[1N19](#) - X-ray [Chimera](#)

[1OEZ](#) - X-ray [Chimera](#)

[1OZT](#) - X-ray [Chimera](#)

[1OZU](#) - X-ray [Chimera](#)

[1P1V](#) - X-ray [Chimera](#)

[1PTZ](#) - X-ray [Chimera](#)

[1PU0](#) - X-ray [Chimera](#)

[1RK7](#) - NMR [Chimera](#)

[1SOS](#) - X-ray [Chimera](#)

[1SPD](#) - X-ray [Chimera](#)

[1UXL](#) - X-ray [Chimera](#)

[1UXM](#) - X-ray [Chimera](#)

[2AF2](#) - NMR [Chimera](#)

[2C9S](#) - X-ray [Chimera](#)

[2C9U](#) - X-ray [Chimera](#)

[2C9V](#) - X-ray [Chimera](#)

[2GBT](#) - X-ray [Chimera](#)

[2GBU](#) - X-ray [Chimera](#)

[2GBV](#) - X-ray [Chimera](#)

[2NNX](#) - X-ray [Chimera](#)

[2R27](#) - X-ray [Chimera](#)

[2V0A](#) - X-ray [Chimera](#)

[2VR6](#) - X-ray [Chimera](#)

[2VR7](#) - X-ray [Chimera](#)

[2VR8](#) - X-ray [Chimera](#)

[2ZKW](#) - X-ray [Chimera](#)

[2ZKX](#) - X-ray [Chimera](#)

[2ZKY](#) - X-ray [Chimera](#)

[3CQP](#) - X-ray [Chimera](#)

[3CQQ](#) - X-ray [Chimera](#)

[3ECU](#) - X-ray [Chimera](#)

[3ECV](#) - X-ray [Chimera](#)

[3ECW](#) - X-ray [Chimera](#)

[3GQF](#) - X-ray [Chimera](#)

[3HFF](#) - X-ray [Chimera](#)

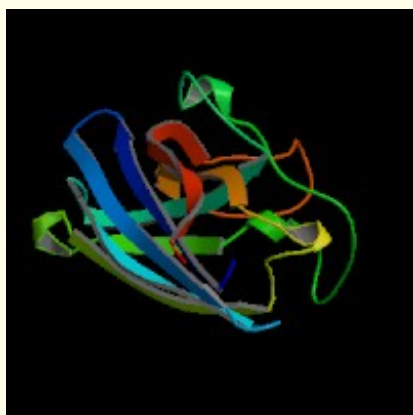
[4SOD](#) - Model [Chimera](#)

[Chimera help](#)

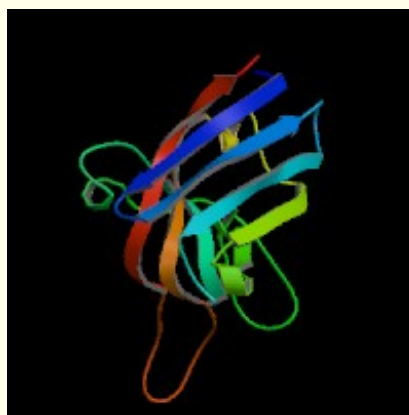
### ModBase Predicted Comparative 3D Structure on [P00441](#)



Front



Top



Side

*The pictures above may be empty if there is no ModBase structure for the protein. The ModBase structure frequently covers just a fragment of the protein. You may be asked to log onto ModBase the first time you click on the pictures. It is simplest after logging in to just click on the picture again to get to the specific info on that model.*

## - Orthologous Genes in Other Species

Orthologies between human, mouse, and rat are computed by taking the best BLASTP hit, and filtering out non-syntenic hits. For more distant species reciprocal-best BLASTP hits are used. Note that the absence of an ortholog in the table below may reflect incomplete annotations in the other species rather than a true absence of the orthologous gene.

Mouse	Rat	Zebrafish	D. melanogaster	C. elegans	S. cerevisiae
<a href="#">Genome Browser</a>	<a href="#">Genome Browser</a>	<a href="#">Genome Browser</a>	<a href="#">Genome Browser</a>	<a href="#">Genome Browser</a>	<a href="#">Genome Browser</a>
<a href="#">Gene Details</a>	<a href="#">Gene Details</a>		<a href="#">Gene Details</a>	<a href="#">Gene Details</a>	<a href="#">Gene Details</a>
<a href="#">Gene Sorter</a>	<a href="#">Gene Sorter</a>		<a href="#">Gene Sorter</a>	<a href="#">Gene Sorter</a>	<a href="#">Gene Sorter</a>
<a href="#">Jackson Lab</a>	<a href="#">RGD</a>	<a href="#">Ensembl</a>	<a href="#">FlyBase</a>	<a href="#">WormBase</a>	<a href="#">SGD</a>
<a href="#">Protein Sequence</a>	<a href="#">Protein Sequence</a>	<a href="#">Protein Sequence</a>	<a href="#">Protein Sequence</a>	<a href="#">Protein Sequence</a>	<a href="#">Protein Sequence</a>
<a href="#">Alignment</a>	<a href="#">Alignment</a>	<a href="#">Alignment</a>	<a href="#">Alignment</a>	<a href="#">Alignment</a>	<a href="#">Alignment</a>

## - Gene Ontology (GO) Annotations with Structured Vocabulary

### Molecular Function:

[GO:0004784](#) superoxide dismutase activity  
[GO:0004785](#) copper, zinc superoxide dismutase activity  
[GO:0005507](#) copper ion binding  
[GO:0005515](#) protein binding  
[GO:0008270](#) zinc ion binding  
[GO:0016209](#) antioxidant activity  
[GO:0016491](#) oxidoreductase activity  
[GO:0030346](#) protein phosphatase 2B binding  
[GO:0042803](#) protein homodimerization activity  
[GO:0046872](#) metal ion binding  
[GO:0051087](#) chaperone binding

### Biological Process:

[GO:0000187](#) activation of MAPK activity  
[GO:0000303](#) response to superoxide  
[GO:0001541](#) ovarian follicle development  
[GO:0001819](#) positive regulation of cytokine production  
[GO:0001890](#) placenta development  
[GO:0001895](#) retinal homeostasis  
[GO:0002262](#) myeloid cell homeostasis  
[GO:0006302](#) double-strand break repair  
[GO:0006309](#) DNA fragmentation during apoptosis  
[GO:0006749](#) glutathione metabolic process  
[GO:0006801](#) superoxide metabolic process  
[GO:0006879](#) cellular iron ion homeostasis  
[GO:0006979](#) response to oxidative stress  
[GO:0007283](#) spermatogenesis  
[GO:0007566](#) embryo implantation  
[GO:0007569](#) cell aging  
[GO:0007605](#) sensory perception of sound  
[GO:0007626](#) locomotory behavior  
[GO:0008217](#) regulation of blood pressure

[GO:0009408](#) response to heat  
[GO:0010033](#) response to organic substance  
[GO:0019226](#) transmission of nerve impulse  
[GO:0019430](#) removal of superoxide radicals  
[GO:0032287](#) myelin maintenance in the peripheral nervous system  
[GO:0033081](#) regulation of T cell differentiation in the thymus  
[GO:0040014](#) regulation of multicellular organism growth  
[GO:0042493](#) response to drug  
[GO:0042542](#) response to hydrogen peroxide  
[GO:0043065](#) positive regulation of apoptosis  
[GO:0043085](#) positive regulation of catalytic activity  
[GO:0043524](#) negative regulation of neuron apoptosis  
[GO:0045471](#) response to ethanol  
[GO:0045541](#) negative regulation of cholesterol biosynthetic process  
[GO:0045859](#) regulation of protein kinase activity  
[GO:0046620](#) regulation of organ growth  
[GO:0046716](#) muscle maintenance  
[GO:0048538](#) thymus development  
[GO:0048678](#) response to axon injury  
[GO:0050665](#) hydrogen peroxide biosynthetic process  
[GO:0051881](#) regulation of mitochondrial membrane potential  
[GO:0060047](#) heart contraction  
[GO:0060052](#) neurofilament cytoskeleton organization and biogenesis  
[GO:0060087](#) relaxation of vascular smooth muscle  
[GO:0060088](#) auditory receptor cell stereocilium organization and biogenesis

**Cellular Component:**

[GO:0005886](#) plasma membrane  
[GO:0005615](#) extracellular space  
[GO:0005634](#) nucleus  
[GO:0005737](#) cytoplasm  
[GO:0005739](#) mitochondrion  
[GO:0005759](#) mitochondrial matrix  
[GO:0005777](#) peroxisome  
[GO:0005829](#) cytosol  
[GO:0031012](#) extracellular matrix  
[GO:0031410](#) cytoplasmic vesicle  
[GO:0032839](#) dendrite cytoplasm  
[GO:0043025](#) cell soma  
[GO:0043234](#) protein complex



## **- Descriptions from all associated GenBank mRNAs**

- [CR621637](#) - full-length cDNA clone CS0DC029YC06 of Neuroblastoma Cot 25-normalized of Homo sapiens (human).
- [CR613850](#) - full-length cDNA clone CS0DI013YA08 of Placenta Cot 25-normalized of Homo sapiens (human).
- [AK312116](#) - Homo sapiens cDNA, FLJ92398, Homo sapiens superoxide dismutase 1, soluble (amyotrophic lateral sclerosis 1 (adult)) (SOD1), mRNA.
- [BC001034](#) - Homo sapiens superoxide dismutase 1, soluble, mRNA (cDNA clone MGC:2325 IMAGE:3140145), complete cds.
- [X02317](#) - Human mRNA for Cu/Zn superoxide dismutase (SOD).
- [CR624343](#) - full-length cDNA clone CS0DC018YI22 of Neuroblastoma Cot 25-normalized of Homo sapiens (human).
- [EF151142](#) - Homo sapiens superoxide dismutase 1 (SOD1) mRNA, complete cds.
- [CR594020](#) - full-length cDNA clone CS0DI054YA23 of Placenta Cot 25-normalized of Homo sapiens (human).
- [CR600386](#) - full-length cDNA clone CS0DK008YL11 of HeLa cells Cot 25-normalized of Homo sapiens (human).
- [EF143990](#) - Homo sapiens superoxide dismutase 1 (SOD1) mRNA, partial cds.
- [CR450355](#) - Homo sapiens full open reading frame cDNA clone RZPDo834A053D for gene SOD1, superoxide dismutase 1, soluble (amyotrophic lateral sclerosis 1 (adult)); complete cds; without stopcodon.
- [AY450286](#) - Homo sapiens superoxide dismutase (SOD) mRNA, complete cds.
- [CR541742](#) - Homo sapiens full open reading frame cDNA clone RZPDo834E0529D for gene SOD1, superoxide dismutase 1, soluble (amyotrophic lateral sclerosis 1 (adult)); complete cds, incl. stopcodon.
- [BT006676](#) - Homo sapiens superoxide dismutase 1, soluble (amyotrophic lateral sclerosis 1 (adult)) mRNA, complete cds.
- [AY049787](#) - Homo sapiens soluble superoxide dismutase 1 (SOD1) gene, complete cds.
- [AB464254](#) - Synthetic construct DNA, clone: pF1KB8213, Homo sapiens SOD1 gene for superoxide dismutase 1, soluble, without stop codon, in Flexi system.
- [CU674512](#) - Synthetic construct Homo sapiens gateway clone IMAGE:100017938 5' read SOD1 mRNA.

## **- Biochemical and Signaling Pathways**

### **KEGG - Kyoto Encyclopedia of Genes and Genomes**

[hsa05020](#) - Prion diseases

### **BioCyc Knowledge Library**

[DETOX1-PWY](#) - removal of superoxide radicals

### **BioCarta from NCI Cancer Genome Anatomy Project**

[h\\_flumazenilPathway](#) - Cardiac Protection Against ROS

[h\\_freePathway](#) - Free Radical Induced Apoptosis

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

**Alternate Gene Symbols:** A6NHJ0, NM\_000454, NP\_000445, P00441, Q16669, Q16711, Q16838, Q16839, Q16840, Q6NR85, SODC\_HUMAN, uc002ypa.1

**UCSC ID:** uc002ypa.2

**RefSeq Accession:** [NM\\_000454](#)

**Protein:** [P00441](#) (aka SODC\_HUMAN)

**CCDS:** [CCDS33536.1](#)

## - Gene Model Information

<b>category:</b>	coding	<b>nonsense-mediated-decay:</b>	no	<b>RNA accession:</b>	NM_000454.4
<b>exon count:</b>	5	<b>CDS single in 3' UTR:</b>	no	<b>RNA size:</b>	981
<b>ORF size:</b>	465	<b>CDS single in intron:</b>	no	<b>Alignment % ID:</b>	100.00
<b>txCdsPredict score:</b>	1105.00	<b>frame shift in genome:</b>	no	<b>% Coverage:</b>	98.17
<b>has start codon:</b>	yes	<b>stop codon in genome:</b>	no	<b># of Alignments:</b>	1
<b>has end codon:</b>	yes	<b>retained intron:</b>	no	<b># AT/AC introns</b>	0
<b>selenocysteine:</b>	no	<b>end bleed into intron:</b>	0	<b># strange splices:</b>	0

Click [here](#) for a detailed description of the fields of the table above.

## - Methods, Credits, and Use Restrictions

Click [here](#) for details on how this gene model was made and data restrictions if any.