kgXref.geneSymbol knownGene.

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

Description: superoxide dismutase 1, soluble (kgXref.description)

pulled from RefSeq page based on kgXref.refseq

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 to rapidly-occuring but harmful superoxide radicals to molecular oxygen and hydrogen peroxide. The other is a mitochondrial protein. Mutations in this gene have been implicated as causes of familial amyotrophic lateral sclerosis. Multiple pseudogenes variants have been reported for this gene. [provided by RefSeq].

kgXref.refseq

knownGene.strand

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

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

links.ra files

Sequence and Links to Tools and Databases

knownToEnsembl knownToLocusLink knownGeneMrna knownToVisiGene

Genomic Sequence (chr21:33,031,935-33,041,24 (multi_way)			mRNA (may differ from genome)		Protein (154 aa)
Gene Sorter	Genome Browser	Protein FASTA	VisiGene	Table Schema	CGAP (cgapAlias & kgXref)
Ensembl	Entrez Gene (allenBrainGene)	ExonPrimer	GeneCards	Gepis Tissue	HGNC
HPRD	Human Cortex Gene Expression	Jackson Lab	OMIM	PubMed	Stanford SOURCE
Treefam	UniProtKB (kgXref.spID)	User annotations			

knownToHprd knownToTreefam knownGene.name kgXref.geneSymbol

Comments and Description Text from UniProtKB

Displaying name is from kgXref.spDisplayID. Link uses kgXref.spID

From sp###### database, e.g. hg19 uses sp090821

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 pathogenics 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 pathogenics 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

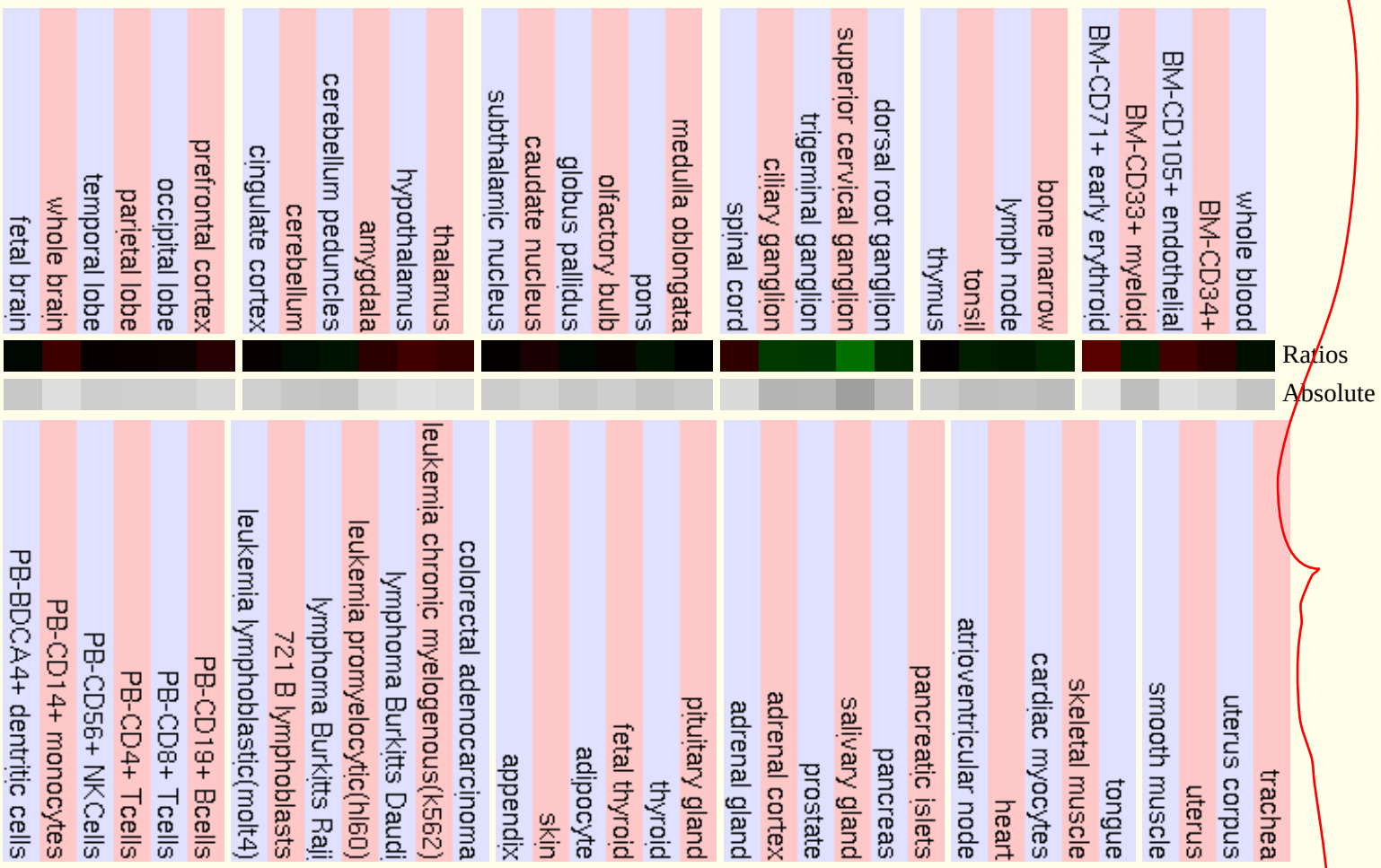
more ... click here to view the complete list

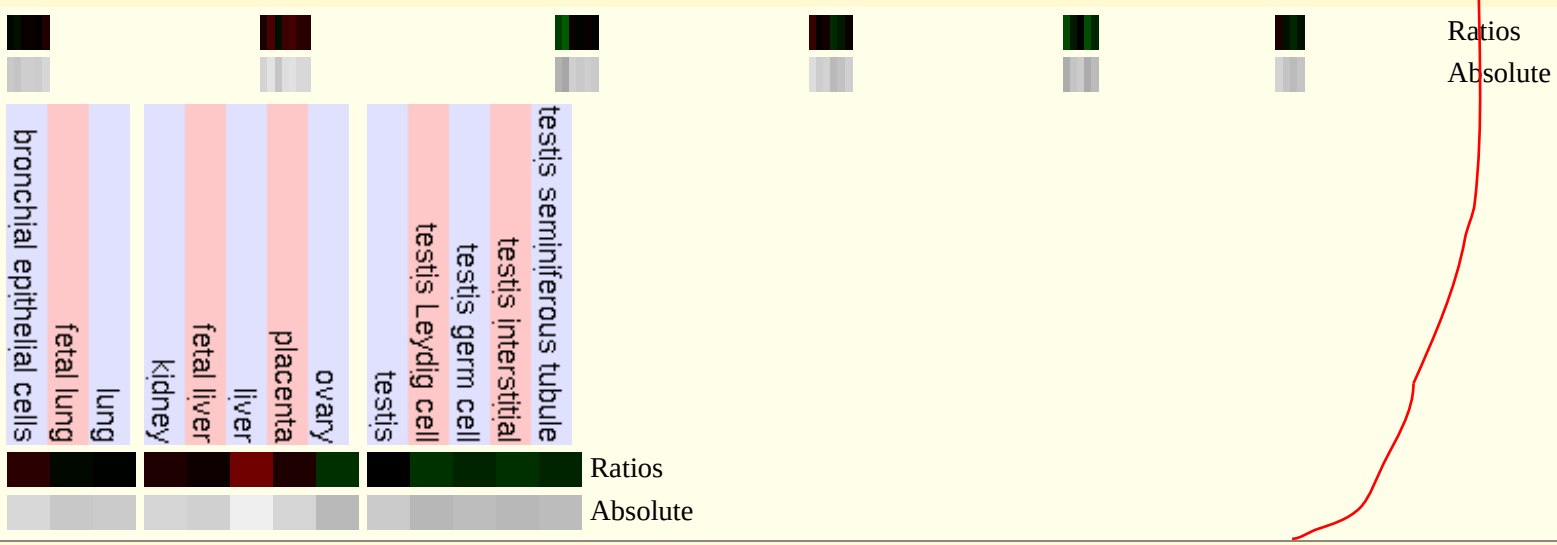
Microarray Expression Data

Expression ratio colors: red high/green low Submit

knownToGnfAtlas2

GNF Expression Atlas 2 Data from U133A and GNF1H Chips





mRNA Secondary Structure of 3' and 5' UTRs

calculated

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

foldUtr5

foldUtr4

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

baed on sp###### database
e.g. hg19 is sp090821

Pfam Domains:

knownToPfam

PF00080 - Copper/zinc superoxide dismutase (SODC)

pfamDesc.

SCOP Domains:

scopDesc.acc

thru ucscScop

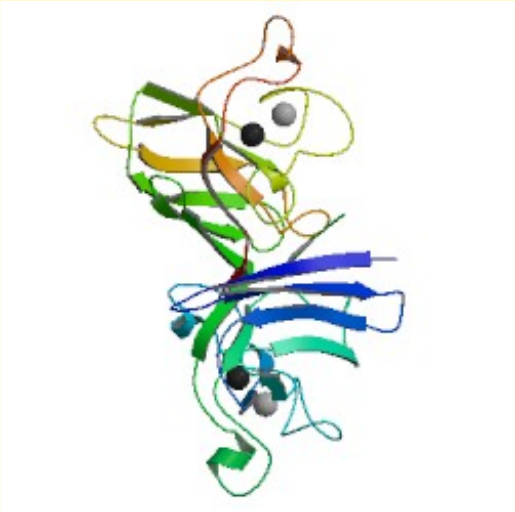
49329 - Cu,Zn superoxide dismutase-like

scopDesc.description

also dependent on kgProtMap2

Protein Data Bank (PDB) 3-D Structure

based on sp###### database; e.g. hg19 is sp090821



1AZV - X-ray Chimera | 1BA9 - NMR Chimera | 1DSW - NMR Chimera

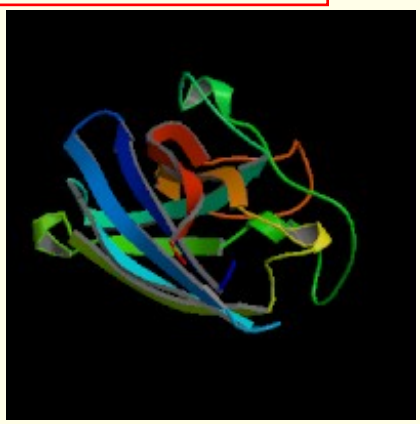
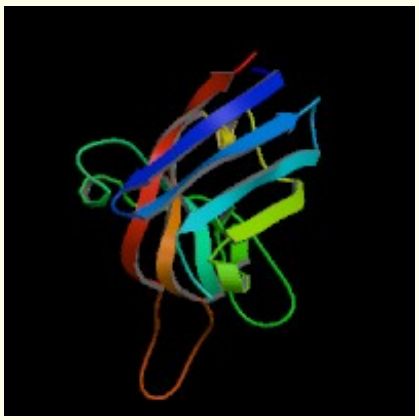
To conserve bandwidth, only the images from the first 3 structures are shown.

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

based on sp####### database, e.g. hg19 is sp090821

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
Genome Browser	Genome Browser	Genome Browser	Genome Browser	Genome Browser	Genome Browser
Gene Details	Gene Details		Gene Details	Gene Details	Gene Details
Gene Sorter	Gene Sorter		Gene Sorter	Gene Sorter	Gene Sorter
Jackson Lab	RGD	Ensembl	FlyBase	WormBase	SGD
Protein Sequence	Protein Sequence	Protein Sequence	Protein Sequence	Protein Sequence	Protein Sequence
Alignment	Alignment	Alignment	Alignment	Alignment	Alignment

otherOrgs.ra file

links.ra files

mmBlastTab rnBlastTab drBlastTab dmBlastTab ceBlastTab scBlastTab

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

keggPathway

KEGG - Kyoto Encyclopedia of Genes and Genomes

hsa05020 - Prion diseases — keggMapDesc

BioCyc Knowledge Library

DETOX1-PWY - removal of superoxide radicals — bioCycMapDesc

bioCycPathway

BioCarta from NCI Cancer Genome Anatomy Project

h_flumazenilPathway - Cardiac Protection Against ROS — cpgBiocDesc

h_freePathway - Free Radical Induced Apoptosis

kgXref.geneSymbol -> cgpAlias -> cpgBiocPathway

Other Names for This Gene

kgAlias (1 record/alias)

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 — kgXref.refseq

Protein: **P00441** (aka SODC_HUMAN) — kgXref.spID (aka kgXref.spDisplayID)

CCDS: CCDS33536.1 — ccdskgMap.ccdsID

If no RefSeq, then the you will see:
Representative RNA: _____
which is controlled by kgXref.mRNA

Gene Model Information

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

Click here for a detailed description of the fields of the table above.

kgTxInfo

Methods, Credits, and Use Restrictions

Click here for details on how this gene model was made and data restrictions if any.