

## GEPHE SUMMARY

<p>squalene synthase (<a href="https://www.gephebase.org/search-criteria?/and+Gene">https://www.gephebase.org/search-criteria?/and+Gene</a> Gephebase=<sup>^</sup>squalene synthase<sup>^</sup>#gephebase-summary-title)</p> <p>Published</p>	<p>Gephebase Gene</p> <p>Entry Status</p>	<p>GP00001947</p> <p>Courtier</p>	<p>GepheID</p> <p>Main curator</p>
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## PHENOTYPIC CHANGE

<p>Physiology (<a href="https://www.gephebase.org/search-criteria?/and+Trait">https://www.gephebase.org/search-criteria?/and+Trait</a> Category=<sup>^</sup>Physiology<sup>^</sup>#gephebase-summary-title)</p> <p>Cholesterol metabolism (cholesterol biosynthesis) (<a href="https://www.gephebase.org/search-criteria?/and+Trait">https://www.gephebase.org/search-criteria?/and+Trait</a> =<sup>^</sup>Cholesterol metabolism (cholesterol biosynthesis)<sup>^</sup>#gephebase-summary-title)</p> <p>able to synthesise cholesterol de novo</p> <p>unable to synthesise cholesterol de novo</p> <p>Taxon A</p> <p>Intergeneric or Higher (<a href="https://www.gephebase.org/search-criteria?/and+Taxonomic">https://www.gephebase.org/search-criteria?/and+Taxonomic</a> Status=<sup>^</sup>Intergeneric or Higher<sup>^</sup>#gephebase-summary-title)</p>	<p>Trait Category</p> <p>Trait</p> <p>Trait State in Taxon A</p> <p>Trait State in Taxon B</p> <p>Ancestral State</p> <p>Taxonomic Status</p>	<p>Taxon A</p> <p>Latin Name</p> <p>Homo sapiens (<a href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms</a> =<sup>^</sup>Homo sapiens<sup>^</sup>#gephebase-summary-title)</p> <p>Common Name</p> <p>human</p> <p>Synonyms</p> <p>human; man; Homo sapiens Linnaeus, 1758; Home sapiens; Homo sapiens; Homo sapeins; Homo sapien; Homo sapiens; Homo sapience; Homo sapiense; Homo sapients; Homo sapines; Homo spaiens; Homo spiens; Humo sapiens</p> <p>Rank</p> <p>species</p> <p>Lineage</p> <p>cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria; Euarchontoglires; Primates; Haplorrhini; Simiiformes; Catarrhini; Hominoidea; Hominidae; Homininae; Homo</p> <p>Parent</p> <p>Homo () - (Rank: genus) (<a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9605">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9605</a>)</p> <p>9606 (<a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9606">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9606</a>)</p> <p>No is Taxon A an Intraspecies?</p>	<p>Taxon B</p> <p>Latin Name</p> <p>Caenorhabditis elegans (<a href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms</a> =<sup>^</sup>Caenorhabditis elegans<sup>^</sup>#gephebase-summary-title)</p> <p>Common Name</p> <p>-</p> <p>Synonyms</p> <p>roundworm; Rhabditis elegans; Caenorhabditis elegans (Maupas, 1900); Rhabditis elegans Maupas, 1900</p> <p>Rank</p> <p>species</p> <p>Lineage</p> <p>cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Protostomia; Ecdysozoa; Nematoda; Chromadorea; Rhabditida; Rhabditina; Rhabditomorpha; Rhabditoidea; Rhabditidae; Peloderinae; Caenorhabditis</p> <p>Parent</p> <p>Caenorhabditis () - (Rank: genus) (<a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=6237">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=6237</a>)</p> <p>6239 (<a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=6239">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=6239</a>)</p> <p>No is Taxon B an Intraspecies?</p>
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## GENOTYPIC CHANGE

<p>FDFT1</p> <p>SS; SQS; DGPT; ERG9; SQSD</p> <p>9606.ENSPO0000480828 (<a href="http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSPO0000480828">http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSPO0000480828</a>)</p> <p>Belongs to the phytoene/squalene synthase family.</p> <p>GO:0046872 : metal ion binding (<a href="https://www.ebi.ac.uk/QuickGO/term/GO:0046872">https://www.ebi.ac.uk/QuickGO/term/GO:0046872</a>) GO:0004310 : farnesyl-diphosphate farnesyltransferase activity (<a href="https://www.ebi.ac.uk/QuickGO/term/GO:0004310">https://www.ebi.ac.uk/QuickGO/term/GO:0004310</a>)</p>	<p>Generic Gene Name</p> <p>Synonyms</p> <p>String</p> <p>Sequence Similarities</p> <p>GO - Molecular Function</p>	<p>UniProtKB Homo sapiens</p> <p>P37268 (<a href="http://www.uniprot.org/uniprot/P37268">http://www.uniprot.org/uniprot/P37268</a>)</p> <p>GenebankID or UniProtKB</p> <p>()</p>
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GO:0051996 : squalene synthase activity  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0051996>)

GO - Biological Process

GO:0006695 : cholesterol biosynthetic process  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0006695>)

GO:0006696 : ergosterol biosynthetic process  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0006696>)

GO:0019216 : regulation of lipid metabolic process  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0019216>)

GO:0006694 : steroid biosynthetic process  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0006694>)

GO:0045338 : farnesyl diphosphate metabolic process  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0045338>)

GO:0045540 : regulation of cholesterol biosynthetic process  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0045540>)

GO - Cellular Component

GO:0016021 : integral component of membrane  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0016021>)

GO:0005783 : endoplasmic reticulum  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0005783>)

GO:0005789 : endoplasmic reticulum membrane  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0005789>)

Presumptive Null

Yes ([https://www.gephebase.org/search-criteria?/and+Presumptive Null=~Yes^#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Presumptive+Null=~Yes^#gephebase-summary-title))

Molecular Type

Gene Loss ([https://www.gephebase.org/search-criteria?/and+Molecular Type=~Gene Loss^#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Molecular+Type=~Gene+Loss^#gephebase-summary-title))

Aberration Type

Deletion ([https://www.gephebase.org/search-criteria?/and+Aberration Type=~Deletion^#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Aberration+Type=~Deletion^#gephebase-summary-title))

Deletion Size

unknown

Molecular Details of the Mutation

gene absent in the genome

Experimental Evidence

Candidate Gene ([https://www.gephebase.org/search-criteria?/and+Experimental Evidence=~Candidate Gene^#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Experimental+Evidence=~Candidate+Gene^#gephebase-summary-title))

Main Reference

Why do worms need cholesterol?. (2003) (<https://pubmed.ncbi.nlm.nih.gov/12894170>)

Authors

Kurzchalia TV; Ward S

Abstract

Cholesterol is a structural component of animal membranes that influences fluidity, permeability and formation of lipid microdomains. It is also a precursor to signalling molecules, including mammalian steroid hormones and insect ecdysones. The nematode *Caenorhabditis elegans* requires too little cholesterol for it to have a major role in membrane structure. Instead, its most probable signalling functions are to control molting and induce a specialized non-feeding larval stage, although no cholesterol-derived signalling molecule has yet been identified for these or any other functions.

Additional References

Preservation of genes involved in sterol metabolism in cholesterol auxotrophs: facts and hypotheses. (2008) (<https://pubmed.ncbi.nlm.nih.gov/18682733>)

RELATED GEPHE

Related Genes

3 (lanosterol c14 demethylase, lanosterol synthase, sterol C5 desaturase) ([https://www.gephebase.org/search-criteria?/or+Taxon ID=~9606^/and+Trait=Cholesterol metabolism/or+Taxon ID=~6239^/and+Trait=Cholesterol metabolism/and+groupHaplotypes=true#gephebase-summary-title](https://www.gephebase.org/search-criteria?/or+Taxon+ID=~9606^/and+Trait=Cholesterol+metabolism/or+Taxon+ID=~6239^/and+Trait=Cholesterol+metabolism/and+groupHaplotypes=true#gephebase-summary-title))

Related Haplotypes

1 ([https://www.gephebase.org/search-criteria?/or+Gene Gephebase=~squalene synthase^/and+Taxon ID=~9606^/or+Gene Gephebase=~squalene synthase^/and+Taxon ID=~6239^#gephebase-summary-title](https://www.gephebase.org/search-criteria?/or+Gene+Gephebase=~squalene+synthase^/and+Taxon+ID=~9606^/or+Gene+Gephebase=~squalene+synthase^/and+Taxon+ID=~6239^#gephebase-summary-title))

EXTERNAL LINKS

COMMENTS

