

GEPHE SUMMARY

<p>squalene synthase (<a +squalene+synthase+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=">https://www.gephebase.org/search-criteria?/and+Gene+Gephebase="+squalene+synthase+"#gephebase-summary-title)</p> <p>Published</p>	<p>Gephebase Gene</p> <p>Entry Status</p>	<p>GP00001948</p> <p>Courtier</p>	<p>GepheID</p> <p>Main curator</p>
---	---	-----------------------------------	------------------------------------

PHENOTYPIC CHANGE

<p>Physiology (<a +physiology+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Trait+Category=">https://www.gephebase.org/search-criteria?/and+Trait+Category="+Physiology+"#gephebase-summary-title)</p> <p>Cholesterol metabolism (cholesterol biosynthesis) (<a +cholesterol+metabolism+(cholesterol+biosynthesis)+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Trait=">https://www.gephebase.org/search-criteria?/and+Trait="+Cholesterol+metabolism+(cholesterol+biosynthesis)+"#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 +intergeneric+or+higher+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=">https://www.gephebase.org/search-criteria?/and+Taxonomic+Status="+Intergeneric+or+Higher+"#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 +homo+sapiens+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms="+Homo+sapiens+"#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) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9605)</p> <p>NCBI Taxonomy ID</p> <p>9606 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9606)</p> <p>is Taxon A an Intraspecies?</p> <p>No</p>	<p>Taxon B</p> <p>Latin Name</p> <p>Drosophila melanogaster (<a +drosophila+melanogaster+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms="+Drosophila+melanogaster+"#gephebase-summary-title)</p> <p>Common Name</p> <p>fruit fly</p> <p>Synonyms</p> <p>Sophophora melanogaster; fruit fly; Drosophila melanogaster Meigen, 1830; Sophophora melanogaster (Meigen, 1830); Drosophila melangaster</p> <p>Rank</p> <p>species</p> <p>Lineage</p> <p>cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Protostomia; Ecdysozoa; Panarthropoda; Arthropoda; Mandibulata; Pancrustacea; Hexapoda; Insecta; Dicondylia; Pterygota; Neoptera; Holometabola; Diptera; Brachycera; Muscomorpha; Eremoneura; Cyclorrhapha; Schizophora; Acalypterae; Ephydroidea; Drosophilidae; Drosophilinae; Drosophilini; Drosophila; Sophophora; melanogaster group; melanogaster subgroup</p> <p>Parent</p> <p>melanogaster subgroup () - (Rank: species subgroup) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=32351)</p> <p>NCBI Taxonomy ID</p> <p>7227 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=7227)</p> <p>is Taxon B an Intraspecies?</p> <p>No</p>
---	---	--	---

GENOTYPIC CHANGE

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

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=~7227^/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=~7227^/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=~7227^#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=~7227^#gephebase-summary-title))

EXTERNAL LINKS

COMMENTS

