GEPHE SUMMARY

Gephebase Gene
squalene synthase (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^squalene synthase^#gephebase-summary-title)

GepheID
GP00001947

Entry Status
Published

Main curator
Courtier

PHENOTYPIC CHANGE

Trait Category
Physiology (https://www.gephebase.org/search-criteria?/and+Trait Category=^Physiology^#gephebase-summary-title)

Trait
Cholesterol metabolism (cholesterol biosynthesis) (https://www.gephebase.org/search-criteria?/and+Trait=^Cholesterol metabolism (cholesterol biosynthesis)^#gephebase-summary-title)

Trait State in Taxon A
able to synthesise cholesterol de novo

Trait State in Taxon B
unable to synthesise cholesterol de novo

Ancestral State
Taxon A

Taxonomic Status
Intergeneric or Higher (https://www.gephebase.org/search-criteria?/and+Taxonomic Status=^Intergeneric or Higher^#gephebase-summary-title)

Taxon A

Latin Name
Homo sapiens (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Homo sapiens^#gephebase-summary-title)

Common Name
human

Synonyms
human; man; Homo sapiens Linnaeus, 1758; Home sapiens; Homo sampiens; Homo sapeins; Homo sapian; Homo sapians; Homo sapien; Homo sapience; Homo sapiense; Homo sapients; Homo sapines; Homo spaiens; Homo spiens; Humo sapiens

Rank
species

Lineage
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

Parent
Homo () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9605)

NCBI Taxonomy ID
9606 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9606)

is Taxon A an Infraspecies?
No

Taxon B

Latin Name
Caenorhabditis elegans (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Caenorhabditis elegans^#gephebase-summary-title)

Common Name
-

Synonyms
roundworm; Rhabditis elegans; Caenorhabditis elegans (Maupas, 1900); Rhabditis elegans Maupas, 1900

Rank
species

Lineage
cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Protostomia; Ecdysozoa; Nematoda; Chromadorea; Rhabditida; Rhabditina; Rhabditomorpha; Rhabditoidea; Rhabditidae; Peloderinae; Caenorhabditis

Parent
Caenorhabditis () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 6237)

NCBI Taxonomy ID
6239 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 6239)

is Taxon B an Infraspecies?
No

GENOTYPIC CHANGE

Generic Gene Name
FDFT1

Synonyms
SS; SQS; DGPT; ERG9; SQSD

String
9606.ENSP00000480828 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9606.ENSP00000480828)

Sequence Similarities
Belongs to the phytoene/squalene synthase family.

GO - Molecular Function
GO:0046872 : metal ion binding (https://www.ebi.ac.uk/QuickGO/term/GO:0046872)
GO:0004310 : farnesyl-diphosphate farnesyltransferase activity (https://www.ebi.ac.uk/QuickGO/term/GO:0004310)

UniProtKB Homo sapiens
P37268 (http://www.uniprot.org/uniprot/P37268)

GenebankID or UniProtKB
()

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)

Molecular Type

Gene Loss (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)

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)

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)

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)

EXTERNAL LINKS

COMMENTS