

GEPHE SUMMARY

lanosterol c14 demethylase (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^lanosterol c14 demethylase^#gephebase-summary-title)	Gephebase Gene	GP00001952	GepheID
Published	Entry Status	Courtier	Main curator

PHENOTYPIC CHANGE

Physiology (https://www.gephebase.org/search-criteria?/and+Trait Category=^Physiology^#gephebase-summary-title)	Trait Category
Cholesterol metabolism (cholesterol biosynthesis) (https://www.gephebase.org/search-criteria?/and+Trait=^Cholesterol+metabolism+(cholesterol+biosynthesis)^#gephebase-summary-title)	Trait
able to synthesise cholesterol de novo	Trait State in Taxon A
unable to synthesise cholesterol de novo	Trait State in Taxon B
Taxon A	Ancestral State
Intergeneric or Higher (https://www.gephebase.org/search-criteria?/and+Taxonomic Status=^Intergeneric or Higher^#gephebase-summary-title)	Taxonomic Status

Taxon A	Latin Name	Taxon B	Latin Name
Homo sapiens (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Homo+sapiens^#gephebase-summary-title)	Common Name	Drosophila melanogaster (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Drosophila+melanogaster^#gephebase-summary-title)	Common Name
human	Synonyms	fruit fly	Synonyms
human; man; Homo sapiens Linnaeus, 1758; Homo sapiens; Homo sapiens		Sophophora melanogaster; fruit fly; Drosophila melanogaster Meigen, 1830; Sophophora melanogaster (Meigen, 1830); Drosophila melangaster	
species	Rank	species	Rank
cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria; Euchortoglires; Primates; Haplorrhini; Simiiformes; Catarrhini; Hominoidea; Hominidae; Homininae; Homo	Lineage	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; Acalyptratae; Ephdroidea; Drosophilidae; Drosophilinae; Drosophilini; Drosophila; Sophophora; melanogaster group; melanogaster subgroup	Lineage
Homo () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9605)	Parent	melanogaster subgroup () - (Rank: species subgroup) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 32351)	Parent
9606 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9606)	NCBI Taxonomy ID	7227 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 7227)	NCBI Taxonomy ID
No	is Taxon A an Infraspecies?	No	is Taxon B an Infraspecies?

GENOTYPIC CHANGE

CYP51A1	Generic Gene Name	UniProtKB Homo sapiens
CYP51; LDM; CP51; CYPL1; P450L1; P450-14DM	Synonyms	GenebankID or UniProtKB
9606.ENSP00000003100 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9606.ENSP00000003100)	String	0
Belongs to the cytochrome P450 family.	Sequence Similarities	
GO:0020037 : heme binding (https://www.ebi.ac.uk/QuickGO/term/GO:0020037)	GO - Molecular Function	
GO:0005506 : iron ion binding (https://www.ebi.ac.uk/QuickGO/term/GO:0005506)		
GO:0008398 : sterol 14-demethylase activity		

GO:0055114 : oxidation-reduction process
(<https://www.ebi.ac.uk/QuickGO/term/GO:0055114>)GO:0006695 : cholesterol biosynthetic process
(<https://www.ebi.ac.uk/QuickGO/term/GO:0006695>)GO:0016126 : sterol biosynthetic process
(<https://www.ebi.ac.uk/QuickGO/term/GO:0016126>)GO:0016125 : sterol metabolic process
(<https://www.ebi.ac.uk/QuickGO/term/GO:0016125>)GO:0006694 : steroid biosynthetic process
(<https://www.ebi.ac.uk/QuickGO/term/GO:0006694>)GO:0042177 : negative regulation of protein catabolic process
(<https://www.ebi.ac.uk/QuickGO/term/GO:0042177>)GO:0045540 : regulation of cholesterol biosynthetic process
(<https://www.ebi.ac.uk/QuickGO/term/GO:0045540>)GO:0033488 : cholesterol biosynthetic process via 24,25-dihydrolanosterol
(<https://www.ebi.ac.uk/QuickGO/term/GO:0033488>)GO:1900222 : negative regulation of amyloid-beta clearance
(<https://www.ebi.ac.uk/QuickGO/term/GO:1900222>)GO:0050709 : negative regulation of protein secretion
(<https://www.ebi.ac.uk/QuickGO/term/GO:0050709>)GO:0016021 : integral component of membrane
(<https://www.ebi.ac.uk/QuickGO/term/GO:0016021>)GO:0005886 : plasma membrane (<https://www.ebi.ac.uk/QuickGO/term/GO:0005886>)GO:0016020 : membrane (<https://www.ebi.ac.uk/QuickGO/term/GO:0016020>)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 ([#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Presumptive Null=^Yes))

Molecular Type

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

Aberration Type

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

Deletion Size

unknown

Molecular Details of the Mutation

gene absent in the genome

Experimental Evidence

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

Main Reference

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

Authors

Vinci G; Xia X; Veitia RA

Abstract

It is known that primary sequences of enzymes involved in sterol biosynthesis are well conserved in organisms that produce sterols de novo. However, we provide evidence for a preservation of the corresponding genes in two animals unable to synthesize cholesterol (auxotrophs): *Drosophila melanogaster* and *Caenorhabditis elegans*.

We have been able to detect bona fide orthologs of several ERG genes in both organisms using a series of complementary approaches. We have detected strong sequence divergence between the orthologs of the nematode and of the fruitfly; they are also very divergent with respect to the orthologs in organisms able to synthesize sterols de novo (prototrophs). Interestingly, the orthologs in both the nematode and the fruitfly are still under selective pressure. It is possible that these genes, which are not involved in cholesterol synthesis anymore, have been recruited to perform different new functions. We propose a more parsimonious way to explain their accelerated evolution and subsequent stabilization. The products of ERG genes in prototrophs might be involved in several biological roles, in addition to sterol synthesis. In the case of the nematode and the fruitfly, the relevant genes would have lost their ancestral function in cholesterogenesis but would have retained the other function(s), which keep them under pressure.

By exploiting microarray data we have noticed a strong expressional correlation between the orthologs of ERG24 and ERG25 in *D. melanogaster* and genes encoding factors involved in intracellular protein trafficking and folding and with Start1 involved in ecdysteroid synthesis. These potential functional connections are worth being explored not only in *Drosophila*, but also in *Caenorhabditis* as well as in sterol prototrophs.

Additional References

RELATED GEPHE

Related Genes

3 (lanosterol synthase, squalene synthase, sterol C5 desaturase) ([#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))

Related Haplotypes

1 ([#gephebase-summary-title](https://www.gephebase.org/search-criteria?/or+Gene Gephebase=^lanosterol c14 demethylase^/and+Taxon ID=^9606^/or+Gene Gephebase=^lanosterol c14 demethylase^/and+Taxon ID=^7227^))

EXTERNAL LINKS

COMMENTS

@ParrallelEvolution in nematods