

GEPHE SUMMARY

	Gephebase Gene		GepheID
lanosterol c14 demethylase (https://www.gephebase.org/search-criteria?/and+Gene Gephebase= [^] lanosterol c14 demethylase [^] #gephebase-summary-title)		GP00001952	Main curator
Published	Entry Status	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)			
able to synthesise cholesterol de novo	Trait State in Taxon A		
unable to synthesise cholesterol de novo	Trait State in Taxon B		
	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		Taxon B	
	Latin Name		Latin Name
Homo sapiens (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms= [^] Homo sapiens [^] #gephebase-summary-title)		Drosophila melanogaster (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms= [^] Drosophila melanogaster [^] #gephebase-summary-title)	
	Common Name		Common Name
human		fruit fly	
	Synonyms		Synonyms
human; man; Homo sapiens Linnaeus, 1758; Home sapiens; Homo sapiens; Homo sapeins; Homo sapien; Homo sapians; Homo sapien; Homo sapience; Homo sapiense; Homo sapients; Homo sapines; Homo spaiens; Homo spiens; Humo sapiens		Sophophora melanogaster; fruit fly; Drosophila melanogaster Meigen, 1830; Sophophora melanogaster (Meigen, 1830); Drosophila melangaster	
	Rank		Rank
species		species	
	Lineage		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		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; Ephydroidea; Drosophilidae; Drosophilinae; Drosophilini; Drosophila; Sophophora; melanogaster group; melanogaster subgroup	
	Parent		Parent
Homo () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9605)		melanogaster subgroup () - (Rank: species subgroup) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=32351)	
	NCBI Taxonomy ID		NCBI Taxonomy ID
9606 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9606)		7227 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=7227)	
	is Taxon A an Intraspecies?		is Taxon B an Intraspecies?
No		No	

GENOTYPIC CHANGE

	Generic Gene Name		UniProtKB Homo sapiens
CYP51A1		Q16850 (http://www.uniprot.org/uniprot/Q16850)	GenebankID or UniProtKB
	Synonyms		
CYP51; LDM; CP51; CYPL1; P450L1; P450-14DM		()	
	String		
9606.ENSP00000003100 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSP00000003100)			
	Sequence Similarities		
Belongs to the cytochrome P450 family.			
	GO - Molecular Function		
GO:0020037 : heme binding (https://www.ebi.ac.uk/QuickGO/term/GO:0020037)			
GO:0005506 : iron ion binding (https://www.ebi.ac.uk/QuickGO/term/GO:0005506)			
GO:0008398 : sterol 14-demethylase activity			

(<https://www.ebi.ac.uk/QuickGO/term/GO:0008398>)

GO - Biological Process

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 - Cellular Component

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 ([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

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

Authors

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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 cholesterologenesis 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) ([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=^lanosterol c14 demethylase^/and+Taxon ID=^9606^/or+Gene Gephebase=^lanosterol c14 demethylase^/and+Taxon ID=^7227^#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^#gephebase-summary-title))

EXTERNAL LINKS

COMMENTS

@ParrallelEvolution in nematods