

GEPHE SUMMARY

Gulo (https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=Gulo^#gephebase-summary-title)	Gephebase Gene	GP00000423	GepheID
Published	Entry Status	Martin	Main curator

PHENOTYPIC CHANGE

Physiology (https://www.gephebase.org/search-criteria?/and+Trait+Category=Physiology^#gephebase-summary-title)	Trait Category		
Vitamin-C synthesis (loss) (https://www.gephebase.org/search-criteria?/and+Trait=Vitamin-C+synthesis+(loss)^#gephebase-summary-title)	Trait		
Other mammals	Trait State in Taxon A		
Pteropus vampyrus	Trait State in Taxon B		
Data not curated	Ancestral State		
Intergenic or Higher (https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=Intergenic+or+Higher^#gephebase-summary-title)	Taxonomic Status		
	Taxon A	Taxon B	
Mammalia (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=Mammalia^#gephebase-summary-title)	Latin Name	Pteropus vampyrus (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=Pteropus+vampyrus^#gephebase-summary-title)	Latin Name
mammals	Common Name	large flying fox	Common Name
mammals	Synonyms	large flying fox	Synonyms
class	Rank	species	Rank
cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota	Lineage	cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria; Laurasiatheria; Chiroptera; Megachiroptera; Pteropodidae; Pteropodinae; Pteropus	Lineage
Amniota (amniotes) - (Rank: no rank) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=32524)	Parent	Pteropus () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9401)	Parent
40674 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=40674)	NCBI Taxonomy ID	132908 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=132908)	NCBI Taxonomy ID
No	is Taxon A an Intraspecies?	No	is Taxon B an Intraspecies?

GENOTYPIC CHANGE

Gulo	Generic Gene Name	P58710 (http://www.uniprot.org/uniprot/P58710)	UniProtKB Mus musculus
GLO; LGO; sfx; unh; unhip; AU018375; BC028822; 5730581M22	Synonyms	()	GenebankID or UniProtKB
10090.ENSMUSP00000060912 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=10090.ENSMUSP00000060912)	String		
Belongs to the oxygen-dependent FAD-linked oxidoreductase family.	Sequence Similarities		
GO:0071949 : FAD binding (https://www.ebi.ac.uk/QuickGO/term/GO:0071949)	GO - Molecular Function		
GO:0050660 : flavin adenine dinucleotide binding (https://www.ebi.ac.uk/QuickGO/term/GO:0050660)			
GO:0003885 : D-arabinono-1,4-lactone oxidase activity (https://www.ebi.ac.uk/QuickGO/term/GO:0003885)			
GO:0050105 : L-gulonolactone oxidase activity (https://www.ebi.ac.uk/QuickGO/term/GO:0050105)			

GO - Biological Process

GO:0019853 : L-ascorbic acid biosynthetic process
(<https://www.ebi.ac.uk/QuickGO/term/GO:0019853>)

GO - Cellular Component

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

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

GO:0031090 : organelle membrane (<https://www.ebi.ac.uk/QuickGO/term/GO:0031090>)

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

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

Aberration Type

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

Molecular Details of the Mutation

pseudogenization

Experimental Evidence

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

Main Reference

A "forward genomics" approach links genotype to phenotype using independent phenotypic losses among related species. (2012) (<https://pubmed.ncbi.nlm.nih.gov/23022484>)

Authors

Hiller M; Schaar BT; Indjeian VB; Kingsley DM; Hagey LR; Bejerano G

Abstract

Genotype-phenotype mapping is hampered by countless genomic changes between species. We introduce a computational "forward genomics" strategy that-given only an independently lost phenotype and whole genomes-matches genomic and phenotypic loss patterns to associate specific genomic regions with this phenotype. We conducted genome-wide screens for two metabolic phenotypes. First, our approach correctly matches the inactivated Gulo gene exactly with the species that lost the ability to synthesize vitamin C. Second, we attribute naturally low biliary phospholipid levels in guinea pigs and horses to the inactivated phospholipid transporter Abcb4. Human ABCB4 mutations also result in low phospholipid levels but lead to a severe liver disease, suggesting compensatory mechanisms in guinea pig and horse. Our simulation studies, counts of independent changes in existing phenotype surveys, and the forthcoming availability of many new genomes all suggest that forward genomics can be applied to many phenotypes, including those relevant for human evolution and disease.

Copyright © 2012 The Authors. Published by Elsevier Inc. All rights reserved.

Additional References

RELATED GEPHE

Related Genes

No matches found.

Related Haplotypes

3 ([https://www.gephebase.org/search-criteria?/or+Gene Gephebase=~Gulo^/and+Taxon ID=~40674^/or+Gene Gephebase=~Gulo^/and+Taxon ID=~132908^#gephebase-summary-title](https://www.gephebase.org/search-criteria?/or+Gene+Gephebase=~Gulo^/and+Taxon+ID=~40674^/or+Gene+Gephebase=~Gulo^/and+Taxon+ID=~132908^#gephebase-summary-title))

EXTERNAL LINKS

COMMENTS

Consecutive invalidating mutations