

GEPHE SUMMARY

	Gephebase Gene	GephelD
Gulo (https://www.gephebase.org/search-criteria/?and+Gene Gephebase="Gulo">#gephebase-summary-title)	GP00000420	
	Entry Status	Main curator
Published	Martin	

PHENOTYPIC CHANGE

	Trait Category		
Physiology (https://www.gephebase.org/search-criteria/?and+Trait Category="Physiology">#gephebase-summary-title)	Trait		
Vitamin-C synthesis (loss) (https://www.gephebase.org/search-criteria/?and+Trait=^Vitamin-C+synthesis+(loss)^#gephebase-summary-title)	Trait State in Taxon A		
Other mammals	Trait State in Taxon B		
Cavia porcellus	Ancestral State		
Data not curated	Taxonomic Status		
Intergeneric or Higher (https://www.gephebase.org/search-criteria/?and+Taxonomic Status="Intergeneric or Higher">#gephebase-summary-title)			
Taxon A		Taxon B	
Mammalia	Latin Name	Cavia porcellus	Latin Name
(#gephebase-summary-title">https://www.gephebase.org/search-criteria/?and+Taxon+and+Synonyms=^Mammalia">#gephebase-summary-title)		(#gephebase-summary-title">https://www.gephebase.org/search-criteria/?and+Taxon+and+Synonyms=^Cavia+porcellus">#gephebase-summary-title)	
mammals	Common Name	domestic guinea pig	Common Name
mammals	Synonyms	Cavia aperea porcellus; Cavia cobaya; domestic guinea pig; guinea pig; Cavia cobaya	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; Euarchontoglires; Glires; Rodentia; Hystricomorpha; Caviidae; Cavia	Lineage
Amniota (amniotes) - (Rank: no rank)	Parent	Cavia (guinea pigs) - (Rank: genus)	Parent
(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=32524)		(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=10140)	
40674	NCBI Taxonomy ID	10141	NCBI Taxonomy ID
(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=40674)	is Taxon A an Infraspecies?	(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=10141)	is Taxon B an Infraspecies?
No		No	

GENOTYPIC CHANGE

	Generic Gene Name		
Gulo	P58710 (http://www.uniprot.org/uniprot/P58710)		UniProtKB Mus musculus
GLO; LGO; sfx; unh; unhip; AJ018375; BC028822; 5730581M22	Synonyms	0	GenebankID or UniProtKB
10090.ENSMUSP00000060912	String		
(http://string-db.org/newstring_cgi/show_network_section.pl?identifier=10090.ENSMUSP00000060912)	Sequence Similarities		
Belongs to the oxygen-dependent FAD-linked oxidoreductase family.			
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=%27Yes%27#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Presumptive%20Null=%27Yes%27#gephebase-summary-title))

Molecular Type

Coding ([https://www.gephebase.org/search-criteria?/and+Molecular Type=%27Coding%27#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Molecular%20Type=%27Coding%27#gephebase-summary-title))

Aberration Type

Complex Change ([https://www.gephebase.org/search-criteria?/and+Aberration Type=%27Complex Change%27#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Aberration%20Type=%27Complex%20Change%27#gephebase-summary-title))

Molecular Details of the Mutation

pseudogenization involving invalidating mutations at most exons

Experimental Evidence

Association Mapping ([https://www.gephebase.org/search-criteria?/and+Experimental Evidence=%27Association Mapping%27#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Experimental%20Evidence=%27Association%20Mapping%27#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 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 \(\[https://www.gephebase.org/search-criteria?/or+Gene Gephebase=%27Gulo%27/and+Taxon ID=%2740674%27/or+Gene Gephebase=%27Gulo%27/and+Taxon ID=%2710141%27#gephebase-summary-title\]\(https://www.gephebase.org/search-criteria?/or+Gene%20Gephebase=%27Gulo%27/and+Taxon%20ID=%2740674%27/or+Gene%20Gephebase=%27Gulo%27/and+Taxon%20ID=%2710141%27#gephebase-summary-title\)\)](https://www.gephebase.org/search-criteria?/or+Gene%20Gephebase=%27Gulo%27/and+Taxon%20ID=%2740674%27/or+Gene%20Gephebase=%27Gulo%27/and+Taxon%20ID=%2710141%27#gephebase-summary-title)

EXTERNAL LINKS

COMMENTS

Consecutive invalidating mutations