

GEPHE SUMMARY

Gephebase Gene
Gulo

Entry Status
Published

GepheID
GP00000420

Main curator
Martin

PHENOTYPIC CHANGE

Trait Category
Physiology

Trait
Vitamin-C synthesis (loss)

Trait State in Taxon A
Other mammals

Trait State in Taxon B
Cavia porcellus

Ancestral State
Data not curated

Taxonomic Status
Intergeneric or Higher

	Taxon A	Taxon B
Latin Name	<i>Mammalia</i>	<i>Cavia porcellus</i>
Common Name	mammals	domestic guinea pig
Synonyms	mammals	Cavia aperea porcellus; Cavia cobaya; domestic guinea pig; guinea pig; Cavia cobyia
Rank	class	species
Lineage	cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota	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
Parent	Amniota (amniotes) - (Rank: no rank)	Cavia (guinea pigs) - (Rank: genus)
NCBI Taxonomy ID	40674	10141
is Taxon A an Intraspecies?	No	No

GENOTYPIC CHANGE

Generic Gene Name
Gulo

Synonyms
GLO; LGO; sfx; unh; unhip; AU018375; BC028822; 5730581M22

String
10090.ENSMUSP00000060912

Sequence Similarities
Belongs to the oxygen-dependent FAD-linked oxidoreductase family.

GO - Molecular Function
GO:0071949 : FAD binding
GO:0050660 : flavin adenine dinucleotide binding
GO:0003885 : D-arabinono-1,4-lactone oxidase activity
GO:0050105 : L-gulonolactone oxidase activity

GO - Biological Process
GO:0019853 : L-ascorbic acid biosynthetic process

GO - Cellular Component
GO:0016021 : integral component of membrane
GO:0005789 : endoplasmic reticulum membrane
GO:0031090 : organelle membrane

UniProtKB Mus musculus
P58710

GenebankID or UniProtKB

Presumptive Null

Yes

Molecular Type

Coding

Aberration Type

Complex Change

Molecular Details of the Mutation

pseudogenization involving invalidating mutations at most exons

Experimental Evidence

Association Mapping

Main Reference

A "forward genomics" approach links genotype to phenotype using independent phenotypic losses among related species. (2012)

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.

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[Additional References](#)

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Related Genes

No matches found.

Related Haplotypes

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COMMENTS

Consecutive invalidating mutations