GEPHE SUMMARY

Gephebase Gene

Gulo (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^Gulo^#gephebase-summary-title)

GepheID

GP00000423

Entry Status

Published

Main curator

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

Pteropus vampyrus

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

Latin Name

Mammalia (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Mammalia^#gephebase-summary-title)

Common Name

mammals

Synonyms

mammals

Rank

class

Lineage

cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota

Parent

Amniota (amniotes) - (Rank: no rank) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 32524)

NCBI Taxonomy ID

40674 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 40674)

is Taxon A an Infraspecies?

No

Taxon B

Latin Name

Pteropus vampyrus (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Pteropus vampyrus^#gephebase-summary-title)

Common Name

large flying fox

Synonyms

large flying fox

Rank

species

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

Parent

Pteropus () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9401)

NCBI Taxonomy ID

132908 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 132908)

is Taxon B an Infraspecies?

No

GENOTYPIC CHANGE

Generic Gene Name

Gulo

Synonyms

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

String

10090.ENSMUSP00000060912 (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 - Molecular Function

GO:0071949 : FAD binding (https://www.ebi.ac.uk/QuickGO/term/GO:0071949)
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)

UniProtKB Mus musculus

P58710 (http://www.uniprot.org/uniprot/P58710)

GenebankID or UniProtKB

()

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)

Molecular Type

Coding (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)

Molecular Details of the Mutation

pseudogenization

Experimental Evidence

Association Mapping (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 toÂ 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

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)

EXTERNAL LINKS

COMMENTS

Consecutive invalidating mutations