

GEPHE SUMMARY

Abcb4 (https://www.gephebase.org/search-criteria?/and+GeneGephebase=~Abcb4~#gephebase-summary-title)	Gephebase Gene	GP00000008	GepheID
	Entry Status	Martin	Main curator
	Published		

PHENOTYPIC CHANGE

Physiology (https://www.gephebase.org/search-criteria?/and+TraitCategory=~Physiology~#gephebase-summary-title)		Trait Category	
Biliary Phospholipid Level (low) (https://www.gephebase.org/search-criteria?/and+Trait=~Biliary Phospholipid Level (low)~#gephebase-summary-title)		Trait	
Other mammals		Trait State in Taxon A	
Cavia porcellus		Trait State in Taxon B	
Data not curated		Ancestral State	
Intergeneric or Higher (https://www.gephebase.org/search-criteria?/and+TaxonomicStatus=~Intergeneric 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)		Cavia porcellus (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=~Cavia porcellus~#gephebase-summary-title)	
mammals		domestic guinea pig	
mammals		Cavia aperea porcellus; Cavia cobaya; domestic guinea pig; guinea pig; Cavia cobya	
class		species	
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	
Amniota (amniotes) - (Rank: no rank) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 32524)		Cavia (guinea pigs) - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 10140)	
40674 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 40674)		10141 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 10141)	
No		No	

GENOTYPIC CHANGE

Abcb4	Generic Gene Name	UniProtKB Mus musculus
	Synonyms	P21440 (http://www.uniprot.org/uniprot/P21440)
	String	XP_014594494 (https://www.ncbi.nlm.nih.gov/nuccore/XP_014594494)
10090.ENSMUSP00000003717 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=10090.ENSMUSP00000003717)		
Belongs to the ABC transporter superfamily. ABCB family. Multidrug resistance exporter (TC 3.A.1.201) subfamily.		
GO - Molecular Function		
GO:0005524 : ATP binding (https://www.ebi.ac.uk/QuickGO/term/GO:0005524)		
GO:0042626 : ATPase activity, coupled to transmembrane movement of substances (https://www.ebi.ac.uk/QuickGO/term/GO:0042626)		
GO:0008525 : phosphatidylcholine transporter activity (https://www.ebi.ac.uk/QuickGO/term/GO:0008525)		
GO:0090554 : phosphatidylcholine-translocating ATPase activity		

(<https://www.ebi.ac.uk/QuickGO/term/GO:0090554>)

GO - Biological Process

GO:0032782 : bile acid secretion (<https://www.ebi.ac.uk/QuickGO/term/GO:0032782>)
GO:1903413 : cellular response to bile acid
(<https://www.ebi.ac.uk/QuickGO/term/GO:1903413>)
GO:0055088 : lipid homeostasis (<https://www.ebi.ac.uk/QuickGO/term/GO:0055088>)
GO:0045332 : phospholipid translocation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0045332>)
GO:0032376 : positive regulation of cholesterol transport
(<https://www.ebi.ac.uk/QuickGO/term/GO:0032376>)
GO:0061092 : positive regulation of phospholipid translocation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0061092>)
GO:2001140 : positive regulation of phospholipid transport
(<https://www.ebi.ac.uk/QuickGO/term/GO:2001140>)
GO:1901557 : response to fenofibrate (<https://www.ebi.ac.uk/QuickGO/term/GO:1901557>)

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:0015629 : actin cytoskeleton (<https://www.ebi.ac.uk/QuickGO/term/GO:0015629>)
GO:0016324 : apical plasma membrane
(<https://www.ebi.ac.uk/QuickGO/term/GO:0016324>)
GO:0030136 : clathrin-coated vesicle (<https://www.ebi.ac.uk/QuickGO/term/GO:0030136>)
GO:0005737 : cytoplasm (<https://www.ebi.ac.uk/QuickGO/term/GO:0005737>)
GO:0005829 : cytosol (<https://www.ebi.ac.uk/QuickGO/term/GO:0005829>)
GO:0005925 : focal adhesion (<https://www.ebi.ac.uk/QuickGO/term/GO:0005925>)
GO:0000139 : Golgi membrane (<https://www.ebi.ac.uk/QuickGO/term/GO:0000139>)
GO:0046581 : intercellular canalculus
(<https://www.ebi.ac.uk/QuickGO/term/GO:0046581>)
GO:0016020 : membrane (<https://www.ebi.ac.uk/QuickGO/term/GO:0016020>)
GO:0045121 : membrane raft (<https://www.ebi.ac.uk/QuickGO/term/GO:0045121>)
GO:0005654 : nucleoplasm (<https://www.ebi.ac.uk/QuickGO/term/GO:0005654>)

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

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

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Additional References

RELATED GEPHE

Related Genes

No matches found.

Related Haplotypes

1 ([https://www.gephebase.org/search-criteria?/or+Gene Gephebase=~Abcb4^/and+Taxon ID=~40674^/or+Gene Gephebase=~Abcb4^/and+Taxon ID=~10141^#gephebase-summary-title](https://www.gephebase.org/search-criteria?/or+Gene+Gephebase=~Abcb4^/and+Taxon+ID=~40674^/or+Gene+Gephebase=~Abcb4^/and+Taxon+ID=~10141^#gephebase-summary-title))

EXTERNAL LINKS

COMMENTS

