

GEPHE SUMMARY

	Gephebase Gene	GepheID
hemoglobin; HBA-T1 and T2 paralogues (<a +hemoglobin;+hba-t1+and+t2+paralogues`#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=">https://www.gephebase.org/search-criteria?/and+Gene+Gephebase="+hemoglobin;+HBA-T1+and+T2+paralogues`#gephebase-summary-title)	GP00000456	
	Martin	Main curator
	Entry Status	
Published		

PHENOTYPIC CHANGE

	Trait Category		
Physiology (<a +physiology`#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Trait+Category=">https://www.gephebase.org/search-criteria?/and+Trait+Category="+Physiology`#gephebase-summary-title)		Trait	
Hypoxia response (<a +hypoxia+response`#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Trait=">https://www.gephebase.org/search-criteria?/and+Trait="+Hypoxia+response`#gephebase-summary-title)			
	Trait State in Taxon A		
Peromyscus maniculatus -low elevation		Trait State in Taxon B	
Peromyscus maniculatus - high elevation			
	Ancestral State		
Taxon A		Taxonomic Status	
Intraspecific (<a +intraspecific`#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=">https://www.gephebase.org/search-criteria?/and+Taxonomic+Status="+Intraspecific`#gephebase-summary-title)			
	Taxon A		Taxon B
	Latin Name		Latin Name
Peromyscus maniculatus (<a +peromyscus+maniculatus`#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms="+Peromyscus+maniculatus`#gephebase-summary-title)		Peromyscus maniculatus (<a +peromyscus+maniculatus`#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms="+Peromyscus+maniculatus`#gephebase-summary-title)	
	Common Name		Common Name
North American deer mouse		North American deer mouse	
	Synonyms		Synonyms
North American deer mouse; Peromyscus maniculatus (Wagner, 1845); MSB:Mamm:74965; MSB:collector:Mamm:74965; Peromyscus maniculatis		North American deer mouse; Peromyscus maniculatus (Wagner, 1845); MSB:Mamm:74965; MSB:collector:Mamm:74965; Peromyscus maniculatis	
	Rank		Rank
species		species	
	Lineage		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; Myomorpha; Muroidea; Cricetidae; Neotominae; Peromyscus		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; Myomorpha; Muroidea; Cricetidae; Neotominae; Peromyscus	
	Parent		Parent
Peromyscus () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=10040)		Peromyscus () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=10040)	
	NCBI Taxonomy ID		NCBI Taxonomy ID
10042 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=10042)		10042 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=10042)	
	is Taxon A an Intraspecies?		is Taxon B an Intraspecies?
No		No	

GENOTYPIC CHANGE

	Generic Gene Name	UniProtKB Homo sapiens
HBA1		P69905 (http://www.uniprot.org/uniprot/P69905)
	Synonyms	GenebankID or UniProtKB
HBH; ECYT7; HBA-T3; METHBA		ABN71228 (https://www.ncbi.nlm.nih.gov/nucleotide/ABN71228)
	String	
9606.ENSPP00000322421 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSPP00000322421)		
	Sequence Similarities	
Belongs to the globin family.		
	GO - Molecular Function	
GO:0020037 : heme binding (https://www.ebi.ac.uk/QuickGO/term/GO:0020037)		
GO:0005506 : iron ion binding (https://www.ebi.ac.uk/QuickGO/term/GO:0005506)		
GO:0005344 : oxygen carrier activity (https://www.ebi.ac.uk/QuickGO/term/GO:0005344)		
GO:0043177 : organic acid binding (https://www.ebi.ac.uk/QuickGO/term/GO:0043177)		

GO:0019825 : oxygen binding (<https://www.ebi.ac.uk/QuickGO/term/GO:0019825>)
GO - Biological Process

GO:0006898 : receptor-mediated endocytosis
(<https://www.ebi.ac.uk/QuickGO/term/GO:0006898>)
GO:0042542 : response to hydrogen peroxide
(<https://www.ebi.ac.uk/QuickGO/term/GO:0042542>)
GO:0015701 : bicarbonate transport (<https://www.ebi.ac.uk/QuickGO/term/GO:0015701>)
GO:0098869 : cellular oxidant detoxification
(<https://www.ebi.ac.uk/QuickGO/term/GO:0098869>)
GO:0042744 : hydrogen peroxide catabolic process
(<https://www.ebi.ac.uk/QuickGO/term/GO:0042744>)
GO:0015671 : oxygen transport (<https://www.ebi.ac.uk/QuickGO/term/GO:0015671>)
GO:0010942 : positive regulation of cell death
(<https://www.ebi.ac.uk/QuickGO/term/GO:0010942>)
GO:0051291 : protein heterooligomerization
(<https://www.ebi.ac.uk/QuickGO/term/GO:0051291>)

GO - Cellular Component

GO:0005829 : cytosol (<https://www.ebi.ac.uk/QuickGO/term/GO:0005829>)
GO:0016020 : membrane (<https://www.ebi.ac.uk/QuickGO/term/GO:0016020>)
GO:0070062 : extracellular exosome (<https://www.ebi.ac.uk/QuickGO/term/GO:0070062>)
GO:0005576 : extracellular region (<https://www.ebi.ac.uk/QuickGO/term/GO:0005576>)
GO:0005615 : extracellular space (<https://www.ebi.ac.uk/QuickGO/term/GO:0005615>)
GO:0072562 : blood microparticle (<https://www.ebi.ac.uk/QuickGO/term/GO:0072562>)
GO:0071682 : endocytic vesicle lumen
(<https://www.ebi.ac.uk/QuickGO/term/GO:0071682>)
GO:0022627 : cytosolic small ribosomal subunit
(<https://www.ebi.ac.uk/QuickGO/term/GO:0022627>)
GO:0031838 : haptoglobin-hemoglobin complex
(<https://www.ebi.ac.uk/QuickGO/term/GO:0031838>)
GO:0005833 : hemoglobin complex (<https://www.ebi.ac.uk/QuickGO/term/GO:0005833>)

Mutation #1

No ([https://www.gephebase.org/search-criteria?/and+Presumptive Null=`No`#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Presumptive+Null=))

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

SNP ([https://www.gephebase.org/search-criteria?/and+Aberration Type=`SNP`#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Aberration+Type=))

Nonsynonymous

His50Pro; Gly57Ala; Ala60Gly; Asp64Gly; Gly71Ser

Candidate Gene ([https://www.gephebase.org/search-criteria?/and+Experimental Evidence=`Candidate Gene`#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Experimental+Evidence=))

Taxon A

Taxon B

Position

Codon

-

-

-

Amino-acid

His

Pro

50

The molecular basis of high-altitude adaptation in deer mice. (2007) (<https://pubmed.ncbi.nlm.nih.gov/17397259>)

Storz JF; Sabatino SJ; Hoffmann FG; Gering EJ; Moriyama H; Ferrand N; Monteiro B; Nachman MW

Elucidating genetic mechanisms of adaptation is a goal of central importance in evolutionary biology, yet few empirical studies have succeeded in documenting causal links between molecular variation and organismal fitness in natural populations. Here we report a population genetic analysis of a two-locus alpha-globin polymorphism that underlies physiological adaptation to high-altitude hypoxia in natural populations of deer mice, *Peromyscus maniculatus*. This system provides a rare opportunity to examine the molecular underpinnings of fitness-related variation in protein function that can be related to a well-defined selection pressure. We surveyed DNA sequence variation in the duplicated alpha-globin genes of *P. maniculatus* from high- and low-altitude localities (i) to identify the specific mutations that may be responsible for the divergent fine-tuning of hemoglobin function and (ii) to test whether the genes exhibit the expected signature of diversifying selection between populations that inhabit different elevational zones. Results demonstrate that functionally distinct protein alleles are maintained as a long-term balanced polymorphism and that adaptive modifications of hemoglobin function are produced by the independent or joint effects of five amino acid mutations that modulate oxygen-binding affinity.

Effects of spatially varying selection on nucleotide diversity and linkage disequilibrium: insights from deer mouse globin genes. (2008) (<https://pubmed.ncbi.nlm.nih.gov/18716337>)
Epistasis among adaptive mutations in deer mouse hemoglobin. (2013) (<https://pubmed.ncbi.nlm.nih.gov/23766324>)

Mutation #2

No ([https://www.gephebase.org/search-criteria?/and+Presumptive Null=`No`#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Presumptive+Null=))

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

SNP ([https://www.gephebase.org/search-criteria?/and+Aberration Type=`SNP`#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Aberration+Type=))

Nonsynonymous

SNP Coding Change

His50Pro; Gly57Ala; Ala60Gly; Asp64Gly; Gly71Ser

Molecular Details of the Mutation

Experimental Evidence

Candidate Gene (<https://www.gephebase.org/search-criteria?/and+Experimental Evidence=~Candidate Gene~#gephebase-summary-title>)

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	Gly	Ala	57

Main Reference

The molecular basis of high-altitude adaptation in deer mice. (2007) (<https://pubmed.ncbi.nlm.nih.gov/17397259>)

Authors

Storz JF; Sabatino SJ; Hoffmann FG; Gering EJ; Moriyama H; Ferrand N; Monteiro B; Nachman MW

Abstract

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Additional References

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Mutation #3

Presumptive Null

No (<https://www.gephebase.org/search-criteria?/and+Presumptive Null=~No~#gephebase-summary-title>)

Molecular Type

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

Aberration Type

SNP (<https://www.gephebase.org/search-criteria?/and+Aberration Type=~SNP~#gephebase-summary-title>)

SNP Coding Change

Nonsynonymous

Molecular Details of the Mutation

His50Pro; Gly57Ala; Ala60Gly; Asp64Gly; Gly71Ser

Experimental Evidence

Candidate Gene (<https://www.gephebase.org/search-criteria?/and+Experimental Evidence=~Candidate Gene~#gephebase-summary-title>)

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	Ala	Gly	60

Main Reference

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Mutation #4

Presumptive Null

No (<https://www.gephebase.org/search-criteria?/and+Presumptive Null=~No~#gephebase-summary-title>)

Molecular Type

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

Aberration Type

SNP (<https://www.gephebase.org/search-criteria?/and+Aberration+Type=~SNP~#gephebase-summary-title>)

SNP Coding Change

Nonsynonymous

Molecular Details of the Mutation

His50Pro; Gly57Ala; Ala60Gly; Asp64Gly; Gly71Ser

Experimental Evidence

Candidate Gene (<https://www.gephebase.org/search-criteria?/and+Experimental+Evidence=~Candidate+Gene~#gephebase-summary-title>)

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	Asp	Gly	64

Main Reference

The molecular basis of high-altitude adaptation in deer mice. (2007) (<https://pubmed.ncbi.nlm.nih.gov/17397259>)

Authors

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Mutation #5

Presumptive Null

No (<https://www.gephebase.org/search-criteria?/and+Presumptive+Null=~No~#gephebase-summary-title>)

Molecular Type

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

Aberration Type

SNP (<https://www.gephebase.org/search-criteria?/and+Aberration+Type=~SNP~#gephebase-summary-title>)

SNP Coding Change

Nonsynonymous

Molecular Details of the Mutation

His50Pro; Gly57Ala; Ala60Gly; Asp64Gly; Gly71Ser

Experimental Evidence

Candidate Gene (<https://www.gephebase.org/search-criteria?/and+Experimental+Evidence=~Candidate+Gene~#gephebase-summary-title>)

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	Gly	Ser	71

Main Reference

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Authors

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Epistasis among adaptive mutations in deer mouse hemoglobin. (2013) (<https://pubmed.ncbi.nlm.nih.gov/23766324>)

RELATED GEPHE

2 (EPAS1, hemoglobin; HBB-T1 and T2 paralogues) (<https://www.gephebase.org/search-criteria?/or+Taxon ID=^10042^/and+Trait=Hypoxia response/and+groupHaplotypes=true#gephebase-summary-title>)

No matches found.

Related Genes

Related Haplotypes

EXTERNAL LINKS

COMMENTS

@SeveralMutationsWithEffect