GEPHE SUMMARY

Gephebase Gene
hemoglobin; HBA-T1 and T2 paralogues (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^hemoglobin; HBA-T1 and T2 paralogues^#gephebase-summary-title)

GepheID
GP00000456

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
Hypoxia response (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 (https://www.gephebase.org/search-criteria?/and+Taxonomic Status=^Intraspecific^#gephebase-summary-title)

Taxon A

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

Common Name
North American deer mouse

Synonyms
North American deer mouse; Peromyscus maniculatus (Wagner, 1845); MSB Mamm 74965; MSB:collector:Mamm:74965; Peromyscus maniculatis

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; Euarchontoglires; Glires; Rodentia; Myomorpha; Muroidea; Cricetidae; Neotominae; Peromyscus

Parent
Peromyscus () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 10040)

NCBI Taxonomy ID
10042 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 10042)

is Taxon A an Infraspecies?
No

Taxon B

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

Common Name
North American deer mouse

Synonyms
North American deer mouse; Peromyscus maniculatus (Wagner, 1845); MSB Mamm 74965; MSB:collector:Mamm:74965; Peromyscus maniculatis

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; Euarchontoglires; Glires; Rodentia; Myomorpha; Muroidea; Cricetidae; Neotominae; Peromyscus

Parent
Peromyscus () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 10040)

NCBI Taxonomy ID
10042 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 10042)

is Taxon B an Infraspecies?
No

GENOTYPIC CHANGE

Generic Gene Name
HBA1

Synonyms
HBH; ECYT7; HBA-T3; METHBA

String
9606.ENSP00000322421 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9606.ENSP00000322421)

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)

UniProtKB Homo sapiens
P69905 (http://www.uniprot.org/uniprot/P69905)

GenebankID or UniProtKB
ABN71228 (https://www.ncbi.nlm.nih.gov/nuccore/ABN71228)

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

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	His	Pro	50

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

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.

Additional References

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

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	Ala	57

Main Reference

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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

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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

Related Genes

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)

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS

@SeveralMutationsWithEffect