

GEPHE SUMMARY

Gephebase Gene
hemoglobin; HBB/HBD fusion gene

Entry Status
Published

GepheID
GP00000477

Main curator
Martin

PHENOTYPIC CHANGE

Trait Category
Physiology

Trait
Temperature tolerance (cold)

Trait State in Taxon A
Elephantids

Trait State in Taxon B
Mammuthus primigenius

Ancestral State
Data not curated

Taxonomic Status
Intergeneric or Higher

Taxon A

Latin Name
Elephantidae

Common Name
elephants

Synonyms
elephants

Rank
family

Lineage
cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Afrotheria; Proboscidea

Parent
Proboscidea (elephants) - (Rank: order)

NCBI Taxonomy ID
9780

is Taxon A an Intraspecies?
No

Taxon B

Latin Name
Mammuthus primigenius

Common Name
woolly mammoth

Synonyms
woolly mammoth; mammoth

Rank
species

Lineage
cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Afrotheria; Proboscidea; Elephantidae; Mammuthus

Parent
Mammuthus () - (Rank: genus)

NCBI Taxonomy ID
37349

is Taxon B an Intraspecies?
No

GENOTYPIC CHANGE

Generic Gene Name
HBB

Synonyms
ECYT6; CD113t-C; beta-globin

String
9606.ENSP00000333994

Sequence Similarities
Belongs to the globin family.

GO - Molecular Function
GO:0046872 : metal ion binding
GO:0020037 : heme binding
GO:0005344 : oxygen carrier activity
GO:0043177 : organic acid binding
GO:0019825 : oxygen binding
GO:0031721 : hemoglobin alpha binding
GO:0030492 : hemoglobin binding

GO - Biological Process
GO:0006898 : receptor-mediated endocytosis
GO:0007596 : blood coagulation

UniProtKB Homo sapiens
P68871

GenebankID or UniProtKB

GO:0008217 : regulation of blood pressure
 GO:0042542 : response to hydrogen peroxide
 GO:0043312 : neutrophil degranulation
 GO:0015701 : bicarbonate transport
 GO:0098869 : cellular oxidant detoxification
 GO:0042744 : hydrogen peroxide catabolic process
 GO:0015671 : oxygen transport
 GO:0010942 : positive regulation of cell death
 GO:0051291 : protein heterooligomerization
 GO:0030185 : nitric oxide transport
 GO:0070527 : platelet aggregation
 GO:0045429 : positive regulation of nitric oxide biosynthetic process
 GO:0050880 : regulation of blood vessel size
 GO:0070293 : renal absorption

GO - Cellular Component

GO:0005829 : cytosol
 GO:0070062 : extracellular exosome
 GO:0005576 : extracellular region
 GO:0005615 : extracellular space
 GO:0072562 : blood microparticle
 GO:0071682 : endocytic vesicle lumen
 GO:0031838 : haptoglobin-hemoglobin complex
 GO:0005833 : hemoglobin complex
 GO:1904813 : ficolin-1-rich granule lumen
 GO:1904724 : tertiary granule lumen

Mutation #1

Presumptive Null

No

Molecular Type

Coding

Aberration Type

SNP

SNP Coding Change

Nonsynonymous

Molecular Details of the Mutation

Thr12Ala

Experimental Evidence

Candidate Gene

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	Thr	Ala	12

Main Reference

Substitutions in woolly mammoth hemoglobin confer biochemical properties adaptive for cold tolerance. (2010)

Authors

Campbell KL; Roberts JE; Watson LN; Stetefeld J; Sloan AM; Signore AV; Howatt JW; Tame JR; Rohland N; Shen TJ; Austin JJ; Hofreiter M; Ho C; Weber RE; Cooper A

Abstract

We have genetically retrieved, resurrected and performed detailed structure-function analyses on authentic woolly mammoth hemoglobin to reveal for the first time both the evolutionary origins and the structural underpinnings of a key adaptive physiochemical trait in an extinct species. Hemoglobin binds and carries O₂; however, its ability to offload O₂ to respiring cells is hampered at low temperatures, as heme deoxygenation is inherently endothermic (that is, hemoglobin-O₂ affinity increases as temperature decreases). We identify amino acid substitutions with large phenotypic effect on the chimeric beta/delta-globin subunit of mammoth hemoglobin that provide a unique solution to this problem and thereby minimize energetically costly heat loss. This biochemical specialization may have been involved in the exploitation of high-latitude environments by this African-derived elephantid lineage during the Pleistocene period. This powerful new approach to directly analyze the genetic and structural basis of physiological adaptations in an extinct species adds an important new dimension to the study of natural selection.

Additional References

Mutation #2

Presumptive Null

No

Molecular Type

Coding

Aberration Type

SNP

SNP Coding Change

Nonsynonymous

Molecular Details of the Mutation

Ala86Ser

Experimental Evidence

Candidate Gene

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	Ala	Ser	86

Main Reference

Substitutions in woolly mammoth hemoglobin confer biochemical properties adaptive for cold tolerance. (2010)

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Additional References

Mutation #3

Presumptive Null

No

Molecular Type

Coding

Aberration Type

SNP

SNP Coding Change

Nonsynonymous

Molecular Details of the Mutation

Glu101Gln

Experimental Evidence

Candidate Gene

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	Glu	Gln	101

Main Reference

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Additional References

RELATED GEPHE

Related Genes

No matches found.

Related Haplotypes

No matches found.

COMMENTS

Needs curation @SeveralMutationsWithEffect

