GEPHE SUMMARY

Gephebase Gene

hemoglobin; HBB/HBD fusion gene (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^hemoglobin; HBB/HBD fusion gene^#gephebase-summary-title)

Entry Status

Published

GepheID

GP00000477

Main curator

Martin

PHENOTYPIC CHANGE

Trait Category

Physiology (https://www.gephebase.org/search-criteria?/and+Trait Category=^Physiology^#gephebase-summary-title)

Trait

Temperature tolerance (cold) (https://www.gephebase.org/search-criteria?/and+Trait=^Temperature tolerance (cold)^#gephebase-summary-title)

Trait State in Taxon A

Elephantids

Trait State in Taxon B

Mammuthus primigenius

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

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

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) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9779)

NCBI Taxonomy ID

9780 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9780)

is Taxon A an Infraspecies?

No

Taxon B

Latin Name

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

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) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 37348)

NCBI Taxonomy ID

37349 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 37349)

is Taxon B an Infraspecies?

No

GENOTYPIC CHANGE

Generic Gene Name

HBB

Synonyms

ECYT6; CD113t-C; beta-globin

String

9606.ENSP00000333994 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9606.ENSP00000333994)

Sequence Similarities

Belongs to the globin family.

GO - Molecular Function

GO:0046872 : metal ion binding (https://www.ebi.ac.uk/QuickGO/term/GO:0046872)
GO:0020037 : heme binding (https://www.ebi.ac.uk/QuickGO/term/GO:0020037)
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:0031721 : hemoglobin alpha binding

UniProtKB Homo sapiens

P68871 (http://www.uniprot.org/uniprot/P68871)

GenebankID or UniProtKB

()

(https://www.ebi.ac.uk/QuickGO/term/GO:0031721)
GO:0030492 : hemoglobin binding (https://www.ebi.ac.uk/QuickGO/term/GO:0030492)

GO - Biological Process

GO:0006898 : receptor-mediated endocytosis
(https://www.ebi.ac.uk/QuickGO/term/GO:0006898)
GO:0007596 : blood coagulation (https://www.ebi.ac.uk/QuickGO/term/GO:0007596)
GO:0008217 : regulation of blood pressure
(https://www.ebi.ac.uk/QuickGO/term/GO:0008217)
GO:0042542 : response to hydrogen peroxide
(https://www.ebi.ac.uk/QuickGO/term/GO:0042542)
GO:0043312 : neutrophil degranulation
(https://www.ebi.ac.uk/QuickGO/term/GO:0043312)
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:0030185 : nitric oxide transport (https://www.ebi.ac.uk/QuickGO/term/GO:0030185)
GO:0070527 : platelet aggregation (https://www.ebi.ac.uk/QuickGO/term/GO:0070527)
GO:0045429 : positive regulation of nitric oxide biosynthetic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0045429)
GO:0050880 : regulation of blood vessel size
(https://www.ebi.ac.uk/QuickGO/term/GO:0050880)
GO:0070293 : renal absorption (https://www.ebi.ac.uk/QuickGO/term/GO:0070293)

GO - Cellular Component

GO:0005829 : cytosol (https://www.ebi.ac.uk/QuickGO/term/GO:0005829)
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: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)
GO:1904813 : ficolin-1-rich granule lumen
(https://www.ebi.ac.uk/QuickGO/term/GO:1904813)
GO:1904724 : tertiary granule lumen (https://www.ebi.ac.uk/QuickGO/term/GO:1904724)

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

Thr12Ala

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	Thr	Ala	12

Main Reference

Substitutions in woolly mammoth hemoglobin confer biochemical properties adaptive for cold tolerance. (2010) (https://pubmed.ncbi.nlm.nih.gov/20436470)

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 O(2); however, its ability to offload O(2) to respiring cells is hampered at low temperatures, as heme deoxygenation is inherently endothermic (that is, hemoglobin-O(2) 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 (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

Ala86Ser

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	Ser	86

Main Reference

Substitutions in woolly mammoth hemoglobin confer biochemical properties adaptive for cold tolerance. (2010) (https://pubmed.ncbi.nlm.nih.gov/20436470)

Authors

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Additional References

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

Glu101Gln

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	Glu	Gln	101

Main Reference

Substitutions in woolly mammoth hemoglobin confer biochemical properties adaptive for cold tolerance. (2010) (https://pubmed.ncbi.nlm.nih.gov/20436470)

Authors

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Additional References

RELATED GEPHE

Related Genes

No matches found.

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS

Needs curation @SeveralMutationsWithEffect