

GEPHE SUMMARY

hemoglobin; HBB/HBD fusion gene (https://www.gephebase.org/search-criteria?/and+Gene=Gephebase=^hemoglobin; HBB/HBD fusion gene^#gephebase-summary-title)	Gephebase Gene GP00000477	GepheID Main curator
Published	Entry Status Martin	

PHENOTYPIC CHANGE

Physiology (https://www.gephebase.org/search-criteria?/and+Trait=Category=^Physiology^#gephebase-summary-title)	Trait Category		
Temperature tolerance (cold) (https://www.gephebase.org/search-criteria?/and+Trait=^Temperature tolerance (cold)^#gephebase-summary-title)	Trait		
Elephantids	Trait State in Taxon A		
Mammuthus primigenius	Trait State in Taxon B		
Data not curated	Ancestral State		
Intergenic or Higher (https://www.gephebase.org/search-criteria?/and+Taxonomic=^Intergenic or Higher^#gephebase-summary-title)	Taxonomic Status		
	Taxon A	Taxon B	
Elephantidae (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Elephantidae^#gephebase-summary-title)	Latin Name	Mammuthus primigenius (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Mammuthus primigenius^#gephebase-summary-title)	Latin Name
elephants	Common Name	woolly mammoth	Common Name
elephants	Synonyms	woolly mammoth; mammoth	Synonyms
family	Rank	species	Rank
cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Afrotheria; Proboscidea	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	Lineage
Proboscidea (elephants) - (Rank: order) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9779)	Parent	Mammuthus () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=37348)	Parent
9780 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9780)	NCBI Taxonomy ID	37349 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=37349)	NCBI Taxonomy ID
	is Taxon A an Intraspecies?		is Taxon B an Intraspecies?
No		No	

GENOTYPIC CHANGE

HBB	Generic Gene Name	P68871 (http://www.uniprot.org/uniprot/P68871)	UniProtKB Homo sapiens
ECYT6; CD113t-C; beta-globin	Synonyms	()	GenebankID or UniProtKB
9606.ENSPP00000333994 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSPP00000333994)	String		
Belongs to the globin family.	Sequence Similarities		
GO:0046872 : metal ion binding (https://www.ebi.ac.uk/QuickGO/term/GO:0046872)	GO - Molecular Function		
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			

(<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

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

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

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

Nonsynonymous

Thr12Ala

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

Presumptive Null

Molecular Type

Aberration Type

SNP Coding Change

Molecular Details of the Mutation

Experimental Evidence

	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 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

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

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=~Coding~#gephebase-summary-title))

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=~SNP~#gephebase-summary-title))

Aberration Type

Nonsynonymous

SNP Coding Change

Ala86Ser

Molecular Details of the Mutation

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=~Candidate+Gene~#gephebase-summary-title))

Experimental Evidence

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

Main Reference

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Authors

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Additional References

Mutation #3

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

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=~Coding~#gephebase-summary-title))

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=~SNP~#gephebase-summary-title))

Aberration Type

Nonsynonymous

SNP Coding Change

Glu101Gln

Molecular Details of the Mutation

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=~Candidate+Gene~#gephebase-summary-title))

Experimental Evidence

	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

No matches found.

No matches found.

Related Genes

Related Haplotypes

EXTERNAL LINKS

COMMENTS

Needs curation @SeveralMutationsWithEffect