GEPHE SUMMARY

Gephebase Gene
hemoglobin; HBA and HBB

Entry Status
Published

GepheID
GP00000455

Main curator
Martin

PHENOTYPIC CHANGE

Trait Category
Physiology

Trait
Hypoxia response

Trait State in Taxon A
Homo sapiens

Trait State in Taxon B
Crocodyilus niloticus

Ancestral State
Data not curated

Taxonomic Status
Intergeneric or Higher

Taxon A

Latin Name
Homo sapiens

Common Name
human

Synonyms
human; man; Homo sapiens Linnaeus, 1758; Home sapiens; Homo sampiens; Homo sapeins; Homo sapian; Homo sapians; Homo sapien; Homo sapience; Homo sapiense; Homo sapients; Homo sapines; Homo spaiens; Homo spiens; Humo sapiens

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; Primates; Haplorrhini; Simiiformes; Catarrhini; Hominoidea; Hominidae; Homininae; Homo

Parent
Homo () - (Rank: genus)

NCBI Taxonomy ID
9606

is Taxon A an Infraspecies?
No

Taxon B

Latin Name
Crocodylus niloticus

Common Name
Nile crocodile

Synonyms
Nile crocodile; African crocodile; Crocodylus niloticus Laurenti, 1768

Rank
species

Lineage
cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Sauropsida; Sauria; Archelosauria; Archosauria; Crocodylia; Longirostres; Crocodylidae; Crocodylus

Parent
Crocodylus () - (Rank: genus)

NCBI Taxonomy ID
8501

is Taxon B an Infraspecies?
No

GENOTYPIC CHANGE

Generic Gene Name
HBA1

Synonyms
HBH; ECYT7; HBA-T3; METHBA

String
9606.ENSP00000322421

Sequence Similarities
Belongs to the globin family.

GO - Molecular Function
GO:0020037 : heme binding
GO:0005506 : iron ion binding
GO:0005344 : oxygen carrier activity
GO:0043177 : organic acid binding
GO:0019825 : oxygen binding

GO - Biological Process
GO:0006898 : receptor-mediated endocytosis

UniProtKB Homo sapiens
P69905

GenebankID or UniProtKB

GO:0042542 : response to hydrogen peroxide
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 - Cellular Component
GO:0005829 : cytosol
GO:0016020 : membrane
GO:0070062 : extracellular exosome
GO:0005576 : extracellular region
GO:0005615 : extracellular space
GO:0072562 : blood microparticle
GO:0071682 : endocytic vesicle lumen
GO:0022627 : cytosolic small ribosomal subunit
GO:0031838 : haptoglobin-hemoglobin complex
GO:0005833 : hemoglobin complex

Presumptive Null
No

Molecular Type
Coding

Aberration Type
SNP

SNP Coding Change
Nonsynonymous

Molecular Details of the Mutation
No more than 12 amino acid substitutions required for providing crocodile-like properties in engineered human Hb - effect of single amino acid changes not tested

Experimental Evidence
Candidate Gene

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	-	-	-

Main Reference
Transplanting a unique allosteric effect from crocodile into human haemoglobin. (1995)

Authors
Komiyama NH; Miyazaki G; Tame J; Nagai K

Abstract
Crocodiles are able to remain under water for more than one hour without surfacing to breathe and often kill their prey by drowning it. How do crocodiles stay under water for a long time? When they hold their breath, bicarbonate ions, the final product of respiration, accumulate and drastically reduce the oxygen affinity of haemoglobin, releasing a large fraction of haemoglobin-bound oxygen into the tissues. We have now located the bicarbonate-ion-binding site at the alpha 1 beta 2-subunit interface by making various human-crocodile chimaeric haemoglobins. Furthermore, we have been able to transplant the bicarbonate effect into human haemoglobin by replacing only a few residues, even though the amino-acid sequence identity between crocodile (Crocodylus niloticus) and human haemoglobins is only 68% for the alpha- and 51% for the beta-subunit. These results indicate that an entirely new function which enables species to adapt to a new environment could evolve in a protein by a relatively small number of amino-acid substitutions in key positions.

Additional References

RELATED GEPHE

Related Genes
4 (BHLHE41, EGLN1, EPAS1, PPAR-alpha)
Related Haplotypes
No matches found.

COMMENTS