GEPHE SUMMARY

Gephebase Gene

EPAS1 (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^EPAS1^#gephebase-summary-title)

Entry Status

Published

GepheID

GP00001357

Main curator

Prigent

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

Panthera spp + Neofilis clouded leopard

Trait State in Taxon B

snow leopard

Ancestral State

Taxon A

Taxonomic Status

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

Taxon A

Latin Name

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

Common Name

-

Synonyms

Leo

Rank

genus

Lineage

cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria; Laurasiatheria; Carnivora; Feliformia; Felidae; Pantherinae

Parent

Pantherinae () - (Rank: subfamily) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 338153)

NCBI Taxonomy ID

9688 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9688)

is Taxon A an Infraspecies?

No

Taxon B

Latin Name

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

Common Name

snow leopard

Synonyms

Felis uncia; Uncia uncia; snow leopard; Unica unica

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; Laurasiatheria; Carnivora; Feliformia; Felidae; Pantherinae; Panthera

Parent

Panthera () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9688)

NCBI Taxonomy ID

29064 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 29064)

is Taxon B an Infraspecies?

No

GENOTYPIC CHANGE

Generic Gene Name

EPAS1

Synonyms

HLF; MOP2; ECYT4; HIF2A; PASD2; bHLHe73; BHLHE73

String

9606.ENSP00000263734 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier= 9606.ENSP00000263734)

Sequence Similarities

-

GO - Molecular Function

GO:0046982 : protein heterodimerization activity (https://www.ebi.ac.uk/QuickGO/term/GO:0046982)
GO:0043565 : sequence-specific DNA binding (https://www.ebi.ac.uk/QuickGO/term/GO:0043565)
GO:0008134 : transcription factor binding (https://www.ebi.ac.uk/QuickGO/term/GO:0008134)
GO:0003677 : DNA binding (https://www.ebi.ac.uk/QuickGO/term/GO:0003677)

UniProtKB Homo sapiens

Q99814 (http://www.uniprot.org/uniprot/Q99814)

GenebankID or UniProtKB

()

GO:0000981 : DNA-binding transcription factor activity, RNA polymerase II-specific (https://www.ebi.ac.uk/QuickGO/term/GO:0000981)
GO:0001077 : proximal promoter DNA-binding transcription activator activity, RNA polymerase II-specific (https://www.ebi.ac.uk/QuickGO/term/GO:0001077)
GO:0035035 : histone acetyltransferase binding (https://www.ebi.ac.uk/QuickGO/term/GO:0035035)

GO - Biological Process

GO:0007165 : signal transduction (https://www.ebi.ac.uk/QuickGO/term/GO:0007165)
GO:0045944 : positive regulation of transcription by RNA polymerase II (https://www.ebi.ac.uk/QuickGO/term/GO:0045944)
GO:0006357 : regulation of transcription by RNA polymerase II (https://www.ebi.ac.uk/QuickGO/term/GO:0006357)
GO:0043687 : post-translational protein modification (https://www.ebi.ac.uk/QuickGO/term/GO:0043687)
GO:0030324 : lung development (https://www.ebi.ac.uk/QuickGO/term/GO:0030324)
GO:0071456 : cellular response to hypoxia (https://www.ebi.ac.uk/QuickGO/term/GO:0071456)
GO:0061418 : regulation of transcription from RNA polymerase II promoter in response to hypoxia (https://www.ebi.ac.uk/QuickGO/term/GO:0061418)
GO:0001666 : response to hypoxia (https://www.ebi.ac.uk/QuickGO/term/GO:0001666)
GO:0001525 : angiogenesis (https://www.ebi.ac.uk/QuickGO/term/GO:0001525)
GO:0001974 : blood vessel remodeling (https://www.ebi.ac.uk/QuickGO/term/GO:0001974)
GO:0048469 : cell maturation (https://www.ebi.ac.uk/QuickGO/term/GO:0048469)
GO:0001892 : embryonic placenta development (https://www.ebi.ac.uk/QuickGO/term/GO:0001892)
GO:0030218 : erythrocyte differentiation (https://www.ebi.ac.uk/QuickGO/term/GO:0030218)
GO:0055072 : iron ion homeostasis (https://www.ebi.ac.uk/QuickGO/term/GO:0055072)
GO:0007005 : mitochondrion organization (https://www.ebi.ac.uk/QuickGO/term/GO:0007005)
GO:0048625 : myoblast fate commitment (https://www.ebi.ac.uk/QuickGO/term/GO:0048625)
GO:0042415 : norepinephrine metabolic process (https://www.ebi.ac.uk/QuickGO/term/GO:0042415)
GO:0120162 : positive regulation of cold-induced thermogenesis (https://www.ebi.ac.uk/QuickGO/term/GO:0120162)
GO:0002027 : regulation of heart rate (https://www.ebi.ac.uk/QuickGO/term/GO:0002027)
GO:0043619 : regulation of transcription from RNA polymerase II promoter in response to oxidative stress (https://www.ebi.ac.uk/QuickGO/term/GO:0043619)
GO:0043129 : surfactant homeostasis (https://www.ebi.ac.uk/QuickGO/term/GO:0043129)
GO:0006366 : transcription by RNA polymerase II (https://www.ebi.ac.uk/QuickGO/term/GO:0006366)
GO:0007601 : visual perception (https://www.ebi.ac.uk/QuickGO/term/GO:0007601)

GO - Cellular Component

GO:0005829 : cytosol (https://www.ebi.ac.uk/QuickGO/term/GO:0005829)
GO:0005654 : nucleoplasm (https://www.ebi.ac.uk/QuickGO/term/GO:0005654)
GO:0005634 : nucleus (https://www.ebi.ac.uk/QuickGO/term/GO:0005634)
GO:0005667 : transcription factor complex (https://www.ebi.ac.uk/QuickGO/term/GO:0005667)
GO:0016607 : nuclear speck (https://www.ebi.ac.uk/QuickGO/term/GO:0016607)

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

two species-specific amino acid changes: Val663Ile and Cys794Arg - whether both or only one is affecting the phenotype is unknown

Experimental Evidence

Association Mapping (https://www.gephebase.org/search-criteria?/and+Experimental Evidence=^Association Mapping^#gephebase-summary-title)

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

Main Reference

The tiger genome and comparative analysis with lion and snow leopard genomes. (2013) (https://pubmed.ncbi.nlm.nih.gov/24045858)

Authors

Cho YS; Hu L; Hou H; Lee H; Xu J; Kwon S; Oh S; Kim HM; Jho S; Kim S; Shin YA; Kim BC; Kim H; Kim CU; Luo SJ; Johnson WE; Koepfli KP; Schmidt-KÃ¼ntzel A; Turner JA; Marker L; Harper C; Miller SM; Jacobs W; Bertola LD; Kim TH; Lee S; Zhou Q; Jung HJ; Xu X; Gadhvi P; Xu P; Xiong Y; Luo Y; Pan S; Gou C; Chu X; Zhang J; Liu S; He J; Chen Y; Yang L; Yang Y; He J; Liu S; Wang J; Kim CH; Kwak H; Kim JS; Hwang S; Ko J; Kim CB; Kim S; Bayarlkhagva D; Paek WK; Kim SJ; O'Brien SJ; Wang J; Bhak J

Abstract

Tigers and their close relatives (Panthera) are some of the world's most endangered species. Here we report the de novo assembly of an Amur tiger whole-genome sequence as well as the genomic sequences of a white Bengal tiger, African lion, white African lion and snow leopard. Through comparative genetic analyses of these genomes, we find genetic signatures that may reflect molecular adaptations consistent with the big cats' hypercarnivorous diet and muscle strength. We report a snow leopard-specific genetic determinant in EGLN1 (Met39>Lys39), which is likely to be associated with adaptation to high altitude. We also detect a TYR260G>A mutation likely responsible for the white lion coat colour. Tiger and cat genomes show similar repeat composition and an appreciably conserved synteny. Genomic data from the five big cats provide an invaluable resource for resolving easily identifiable phenotypes evident in very close, but distinct, species.

Additional References

RELATED GEPHE

Related Genes

1 (EGLN1) (https://www.gephebase.org/search-criteria?/or+Taxon ID=^9688^/and+Trait=Hypoxia response/or+Taxon ID=^29064^/and+Trait=Hypoxia response/and+groupHaplotypes=true#gephebase-summary-title)

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS

putative candidate