

GEPHE SUMMARY

EPAS1 (https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=^EPAS1^#gephebase-summary-title)	Gephebase Gene	GP00001357	GepheID
Published	Entry Status	Prigent	Main curator

PHENOTYPIC CHANGE

Physiology (https://www.gephebase.org/search-criteria?/and+Trait+Category=^Physiology^#gephebase-summary-title)	Trait Category		
Hypoxia response (https://www.gephebase.org/search-criteria?/and+Trait=^Hypoxia+response^#gephebase-summary-title)	Trait		
Panthera spp + Neofelis clouded leopard	Trait State in Taxon A		
snow leopard	Trait State in Taxon B		
Taxon A	Ancestral State		
Interspecific (https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=^Interspecific^#gephebase-summary-title)	Taxonomic Status		
	Taxon A	Taxon B	
Panthera (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Panthera^#gephebase-summary-title)	Latin Name	Panthera uncia (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Panthera+uncia^#gephebase-summary-title)	Latin Name
-	Common Name	snow leopard	Common Name
Leo	Synonyms	Felis uncia; Uncia uncia; snow leopard; Unica unica	Synonyms
genus	Rank	species	Rank
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	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	Lineage
Pantherinae () - (Rank: subfamily) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=338153)	Parent	Panthera () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9688)	Parent
9688 (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)	NCBI Taxonomy ID
No	is Taxon A an Intraspecies?	No	is Taxon B an Intraspecies?

GENOTYPIC CHANGE

EPAS1	Generic Gene Name	Q99814 (http://www.uniprot.org/uniprot/Q99814)	UniProtKB Homo sapiens
HLF; MOP2; ECYT4; HIF2A; PASD2; bHLHe73; BHLHE73	Synonyms	()	GenebankID or UniProtKB
9606.ENSPP00000263734 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9606.ENSPP00000263734)	String		
-	Sequence Similarities		
GO:0046982 : protein heterodimerization activity (https://www.ebi.ac.uk/QuickGO/term/GO:0046982)	GO - Molecular Function		
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)			

GO:000981 : DNA-binding transcription factor activity, RNA polymerase II-specific
 (https://www.ebi.ac.uk/QuickGO/term/GO:000981)
 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-KÄ¼ntzel 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

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

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 Genes

No matches found.

Related Haplotypes

EXTERNAL LINKS

COMMENTS

putative candidate