

## GEPHE SUMMARY

EGLN1 ( <a href="https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=EGLN1#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=EGLN1#gephebase-summary-title</a> )	Gephebase Gene	GP00001358	GepheID
Published	Entry Status	Prigent	Main curator

## PHENOTYPIC CHANGE

Physiology ( <a href="https://www.gephebase.org/search-criteria?/and+Trait+Category=Physiology#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Trait+Category=Physiology#gephebase-summary-title</a> )	Trait Category		
Hypoxia response ( <a href="https://www.gephebase.org/search-criteria?/and+Trait=Hypoxia+response#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Trait=Hypoxia+response#gephebase-summary-title</a> )	Trait		
Panthera spp + Neofelis clouded leopard	Trait State in Taxon A		
snow leopard	Trait State in Taxon B		
Taxon A	Ancestral State		
Interspecific ( <a href="https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=Interspecific#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=Interspecific#gephebase-summary-title</a> )	Taxonomic Status		
	Taxon A		Taxon B
Panthera	Latin Name	Panthera uncia	Latin Name
( <a href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=Panthera#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=Panthera#gephebase-summary-title</a> )	Common Name	( <a href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=Panthera+uncia#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=Panthera+uncia#gephebase-summary-title</a> )	Common Name
-	Synonyms	snow leopard	Synonyms
Leo	Rank	Felis uncia; Uncia uncia; snow leopard; Unica unica	Rank
genus	Lineage	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	Parent	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
Pantherinae () - (Rank: subfamily)	NCBI Taxonomy ID	Panthera () - (Rank: genus)	NCBI Taxonomy ID
( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=338153">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=338153</a> )	9688	( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9688">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9688</a> )	29064
( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9688">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9688</a> )	is Taxon A an Intraspecies?	( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=29064">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=29064</a> )	is Taxon B an Intraspecies?
No		No	

## GENOTYPIC CHANGE

EGLN1	Generic Gene Name	Q9GZT9 ( <a href="http://www.uniprot.org/uniprot/Q9GZT9">http://www.uniprot.org/uniprot/Q9GZT9</a> )	UniProtKB Homo sapiens
HPH2; PHD2; SM20; ECYT3; HALAH; HPH-2; HIFPH2; ZMYND6; C1orf12; HIF-PH2; PNAS-118; PNAS-137	Synonyms	()	GenebankID or UniProtKB
9606.ENSPP00000355601	String		
( <a href="http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSPP00000355601">http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSPP00000355601</a> )	Sequence Similarities		
-	GO - Molecular Function		
GO:0016706 : oxidoreductase activity, acting on paired donors, with incorporation or reduction of molecular oxygen, 2-oxoglutarate as one donor, and incorporation of one atom each of oxygen into both donors ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0016706">https://www.ebi.ac.uk/QuickGO/term/GO:0016706</a> )			
GO:0019899 : enzyme binding ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0019899">https://www.ebi.ac.uk/QuickGO/term/GO:0019899</a> )			
GO:0008198 : ferrous iron binding ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0008198">https://www.ebi.ac.uk/QuickGO/term/GO:0008198</a> )			
GO:0031418 : L-ascorbic acid binding ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0031418">https://www.ebi.ac.uk/QuickGO/term/GO:0031418</a> )			

GO:0031545 : peptidyl-proline 4-dioxygenase activity  
 (https://www.ebi.ac.uk/QuickGO/term/GO:0031545)  
 GO:0031543 : peptidyl-proline dioxygenase activity  
 (https://www.ebi.ac.uk/QuickGO/term/GO:0031543)

GO - Biological Process

GO:0045944 : positive regulation of transcription by RNA polymerase II  
 (https://www.ebi.ac.uk/QuickGO/term/GO:0045944)  
 GO:1901214 : regulation of neuron death  
 (https://www.ebi.ac.uk/QuickGO/term/GO:1901214)  
 GO:0006879 : cellular iron ion homeostasis  
 (https://www.ebi.ac.uk/QuickGO/term/GO:0006879)  
 GO:0055008 : cardiac muscle tissue morphogenesis  
 (https://www.ebi.ac.uk/QuickGO/term/GO:0055008)  
 GO:0060347 : heart trabecula formation  
 (https://www.ebi.ac.uk/QuickGO/term/GO:0060347)  
 GO:0060711 : labyrinthine layer development  
 (https://www.ebi.ac.uk/QuickGO/term/GO:0060711)  
 GO:0051344 : negative regulation of cyclic-nucleotide phosphodiesterase activity  
 (https://www.ebi.ac.uk/QuickGO/term/GO:0051344)  
 GO:0043433 : negative regulation of DNA-binding transcription factor activity  
 (https://www.ebi.ac.uk/QuickGO/term/GO:0043433)  
 GO:0032364 : oxygen homeostasis (https://www.ebi.ac.uk/QuickGO/term/GO:0032364)  
 GO:0018401 : peptidyl-proline hydroxylation to 4-hydroxy-L-proline  
 (https://www.ebi.ac.uk/QuickGO/term/GO:0018401)  
 GO:0045765 : regulation of angiogenesis  
 (https://www.ebi.ac.uk/QuickGO/term/GO:0045765)  
 GO:0099175 : regulation of postsynapse organization  
 (https://www.ebi.ac.uk/QuickGO/term/GO:0099175)  
 GO:0099576 : regulation of protein catabolic process at postsynapse, modulating synaptic transmission (https://www.ebi.ac.uk/QuickGO/term/GO:0099576)  
 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:0071731 : response to nitric oxide (https://www.ebi.ac.uk/QuickGO/term/GO:0071731)  
 GO:0060412 : ventricular septum morphogenesis  
 (https://www.ebi.ac.uk/QuickGO/term/GO:0060412)

GO - Cellular Component

GO:0005737 : cytoplasm (https://www.ebi.ac.uk/QuickGO/term/GO:0005737)  
 GO:0005829 : cytosol (https://www.ebi.ac.uk/QuickGO/term/GO:0005829)  
 GO:0005634 : nucleus (https://www.ebi.ac.uk/QuickGO/term/GO:0005634)  
 GO:0098978 : glutamatergic synapse  
 (https://www.ebi.ac.uk/QuickGO/term/GO:0098978)  
 GO:0014069 : postsynaptic density (https://www.ebi.ac.uk/QuickGO/term/GO:0014069)

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

p.Met39Lys

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

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

1 (EPAS1) (<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>)

No matches found.

Related Genes

Related Haplotypes

## EXTERNAL LINKS

## COMMENTS

putative candidate