GEPHE SUMMARY

Gephebase Gene
EGLN1 (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^EGLN1^#gephebase-summary-title)

Entry Status
Published

GepheID
GP00001358

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
EGLN1

Synonyms
HPH2; PHD2; SM20; ECYT3; HALAH; HPH-2; HIFPH2; ZMYND6; C1orf12; HIF-PH2; PNAS-118; PNAS-137

String
9606.ENSP00000355601
(http://string-db.org/newstring_cgi/show_network_section.pl?identifier= 9606.ENSP00000355601)

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 (https://www.ebi.ac.uk/QuickGO/term/GO:0016706)
GO:0019899 : enzyme binding (https://www.ebi.ac.uk/QuickGO/term/GO:0019899)
GO:0008198 : ferrous iron binding (https://www.ebi.ac.uk/QuickGO/term/GO:0008198)
GO:0031418 : L-ascorbic acid binding (https://www.ebi.ac.uk/QuickGO/term/GO:0031418)

UniProtKB Homo sapiens
Q9GZT9 (http://www.uniprot.org/uniprot/Q9GZT9)

GenebankID or UniProtKB
()

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-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 (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)

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS

putative candidate