GEPHE SUMMARY

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

GepheID
GP00001759

Entry Status
Published

Main curator
Courtier

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
-

Trait State in Taxon B
naked mole rat - resistance to hypoxia

Ancestral State
Taxon A

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

Taxon A

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

Common Name
rodent

Synonyms
rodent

Rank
order

Lineage
cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria; Euarchontoglires; Glires

Parent
Glires (Rodents and rabbits) - (Rank: no rank) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 314147)

NCBI Taxonomy ID
9989 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9989)

is Taxon A an Infraspecies?
No

Taxon B

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

Common Name
naked mole-rat

Synonyms
naked mole-rat; naked mole rat

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; Glires; Rodentia; Hystricomorpha; Bathyergidae; Heterocephalus

Parent
Heterocephalus () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 10180)

NCBI Taxonomy ID
10181 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 10181)

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

unique amino-acid changes in different positions of EGLN1 (Pro15, Arg17 and Arg36)

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

Genome sequencing reveals insights into physiology and longevity of the naked mole rat. (2011) (https://pubmed.ncbi.nlm.nih.gov/21993625)

Authors

Kim EB; Fang X; Fushan AA; Huang Z; Lobanov AV; Han L; Marino SM; Sun X; Turanov AA; Yang P; Yim SH; Zhao X; Kasaikina MV; Stoletzki N; Peng C; Polak P; Xiong Z; Kiezun A; Zhu Y; Chen Y; Kryukov GV; Zhang Q; Peshkin L; Yang L; Bronson RT; Buffenstein R; Wang B; Han C; Li Q; Chen L; Zhao W; Sunyaev SR; Park TJ; Zhang G; Wang J; Gladyshev VN

Abstract

The naked mole rat (Heterocephalus glaber) is a strictly subterranean, extraordinarily long-lived eusocial mammal. Although it is the size of a mouse, its maximum lifespan exceeds 30 years, making this animal the longest-living rodent. Naked mole rats show negligible senescence, no age-related increase in mortality, and high fecundity until death. In addition to delayed ageing, they are resistant to both spontaneous cancer and experimentally induced tumorigenesis. Naked mole rats pose a challenge to the theories that link ageing, cancer and redox homeostasis. Although characterized by significant oxidative stress, the naked mole rat proteome does not show age-related susceptibility to oxidative damage or increased ubiquitination. Naked mole rats naturally reside in large colonies with a single breeding female, the 'queen', who suppresses the sexual maturity of her subordinates. They also live in full darkness, at low oxygen and high carbon dioxide concentrations, and are unable to sustain thermogenesis nor feel certain types of pain. Here we report the sequencing and analysis of the naked mole rat genome, which reveals unique genome features and molecular adaptations consistent with cancer resistance, poikilothermy, hairlessness and insensitivity to low oxygen, and altered visual function, circadian rythms and taste sensing. This information provides insights into the naked mole rat's exceptional longevity and ability to live in hostile conditions, in the dark and at low oxygen. The extreme traits of the naked mole rat, together with the reported genome and transcriptome information, offer opportunities for understanding ageing and advancing other areas of biological and biomedical

research.

Additional References

RELATED GEPHE

Related Genes

3 (EPAS1, hemoglobin; HBA-T1 and T2 paralogues, hemoglobin; HBB-T1 and T2 paralogues) (https://www.gephebase.org/search-criteria?/or+Taxon ID=^9989^/and+Trait=Hypoxia response/or+Taxon ID=^10181^/and+Trait=Hypoxia response/and+groupHaplotypes=true#gephebase-summary-title)

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS