

GEPHE SUMMARY

Gephebase Gene		GepheID
Myogenic factor 5 (Myf5) (https://www.gephebase.org/search-criteria?and+Gene=Gephebase+Myogenic+factor+5+(Myf5)+#gephebase-summary-title)	GP00001717	Main curator
Published	Entry Status	Courtier

PHENOTYPIC CHANGE

Trait Category		Trait	
Morphology (https://www.gephebase.org/search-criteria?/and+Trait+Category=^Morphology^#gephebase-summary-title)		Trait	
Rib morphology (skeleton) (https://www.gephebase.org/search-criteria?/and+Trait=^Rib morphology (skeleton)^#gephebase-summary-title)		Trait State in Taxon A	
no ribs in the lumbar region		Trait State in Taxon B	
ribs in the lumbar region		Ancestral State	
Taxon A		Taxonomic Status	
Intergeneric or Higher (https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=^Intergeneric or Higher^#gephebase-summary-title)		Taxon A	
Eutheria (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Eutheria^#gephebase-summary-title)		Taxon B #1	
placentals		Trichechus manatus (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Trichechus manatus^#gephebase-summary-title)	
Placentalia; placentals; eutherian mammals; placental mammals		West Indian manatee	
no rank		West Indian manatee; Trichechus manatus Linnaeus, 1758	
cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria		species	
Theria () - (Rank: no rank) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 32525)		cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Afrotheria; Sirenia; Trichechidae; Trichechus	
9347 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9347)		Trichechus () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9776)	
No		9778 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9778)	
		No	

Taxon B #2	
Procavia capensis (<a +procavia+capensis`#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms="+Procavia+capensis`#gephebase-summary-title)	Latin Name
Cape rock hyrax	Common Name
Cape rock hyrax; Cape hyrax; large-toothed rock hyrax; rock dassie; rock hyrax	Synonyms
species	Rank
cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Afrotheria; Hyracoidea; Procaviidae; Procavia	Lineage
Procavia () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9812)	Parent
9813 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9813)	NCBI Taxonomy ID

No

is Taxon B an Intraspecies?

Taxon B #3

Latin Name

Loxodonta africana
([https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms="+Loxodonta africana^#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=))

Common Name

African savanna elephant

Synonyms

Loxodonta africana africana; African savanna elephant; African bush elephant; African savannah elephant

Rank

species

Lineage

cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Afrotheria; Proboscidea; Elephantidae; Loxodonta

Parent

Loxodonta () - (Rank: genus)
(<https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9784>)

NCBI Taxonomy ID

9785
(<https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9785>)

is Taxon B an Intraspecies?

No

GENOTYPIC CHANGE

Myf5

Myf-5; bHLHc2; B130010.J22Rik

10090.ENSMUSP00000000445
(http://string-db.org/newstring.cgi/show_network_section.pl?identifier=10090.ENSMUSP00000000445)

-

Generic Gene Name

Synonyms

String

Sequence Similarities

GO - Molecular Function

GO - Biological Process

UniProtKB Mus musculus

GenebankID or UniProtKB

P24699 (<http://www.uniprot.org/uniprot/P24699>)

JX154077 (<https://www.ncbi.nlm.nih.gov/nucore/JX154077>)

GO:0046983 : protein dimerization activity
(<https://www.ebi.ac.uk/QuickGO/term/GO:0046983>)
GO:0001228 : DNA-binding transcription activator activity, RNA polymerase II-specific
(<https://www.ebi.ac.uk/QuickGO/term/GO:0001228>)
GO:0043565 : sequence-specific DNA binding
(<https://www.ebi.ac.uk/QuickGO/term/GO:0043565>)
GO:0000981 : DNA-binding transcription factor activity, RNA polymerase II-specific
(<https://www.ebi.ac.uk/QuickGO/term/GO:0000981>)
GO:0000978 : RNA polymerase II proximal promoter sequence-specific DNA binding
(<https://www.ebi.ac.uk/QuickGO/term/GO:0000978>)

GO:0045944 : positive regulation of transcription by RNA polymerase II
(<https://www.ebi.ac.uk/QuickGO/term/GO:0045944>)
GO:0001502 : cartilage condensation (<https://www.ebi.ac.uk/QuickGO/term/GO:0001502>)
GO:0030198 : extracellular matrix organization
(<https://www.ebi.ac.uk/QuickGO/term/GO:0030198>)
GO:0048704 : embryonic skeletal system morphogenesis
(<https://www.ebi.ac.uk/QuickGO/term/GO:0048704>)
GO:0001756 : somitogenesis (<https://www.ebi.ac.uk/QuickGO/term/GO:0001756>)
GO:0001503 : ossification (<https://www.ebi.ac.uk/QuickGO/term/GO:0001503>)
GO:1901741 : positive regulation of myoblast fusion
(<https://www.ebi.ac.uk/QuickGO/term/GO:1901741>)
GO:0035914 : skeletal muscle cell differentiation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0035914>)
GO:0043010 : camera-type eye development
(<https://www.ebi.ac.uk/QuickGO/term/GO:0043010>)
GO:0007519 : skeletal muscle tissue development
(<https://www.ebi.ac.uk/QuickGO/term/GO:0007519>)
GO:0007517 : muscle organ development
(<https://www.ebi.ac.uk/QuickGO/term/GO:0007517>)
GO:0060415 : muscle tissue morphogenesis

(<https://www.ebi.ac.uk/QuickGO/term/GO:0060415>)
GO:0048644 : muscle organ morphogenesis
(<https://www.ebi.ac.uk/QuickGO/term/GO:0048644>)
GO:0045663 : positive regulation of myoblast differentiation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0045663>)
GO:0048743 : positive regulation of skeletal muscle fiber development
(<https://www.ebi.ac.uk/QuickGO/term/GO:0048743>)
GO:0001952 : regulation of cell-matrix adhesion
(<https://www.ebi.ac.uk/QuickGO/term/GO:0001952>)

GO - Cellular Component

GO:0005654 : nucleoplasm (<https://www.ebi.ac.uk/QuickGO/term/GO:0005654>)
GO:0005634 : nucleus (<https://www.ebi.ac.uk/QuickGO/term/GO:0005634>)

Presumptive Null

No (<https://www.gephebase.org/search-criteria?/and+Presumptive Null=~No~#gephebase-summary-title>)

Molecular Type

Cis-regulatory (<https://www.gephebase.org/search-criteria?/and+Molecular Type=~Cis-regulatory~#gephebase-summary-title>)

Aberration Type

SNP (<https://www.gephebase.org/search-criteria?/and+Aberration Type=~SNP~#gephebase-summary-title>)

Molecular Details of the Mutation

one nucleotide change at a Hox binding site (CTAATTG to CCAATTG) - mouse transgenics and protein-DNA binding assay

Experimental Evidence

Candidate Gene (<https://www.gephebase.org/search-criteria?/and+Experimental Evidence=~Candidate Gene~#gephebase-summary-title>)

Main Reference

Role of a polymorphism in a Hox/Pax-responsive enhancer in the evolution of the vertebrate spine. (2013) (<https://pubmed.ncbi.nlm.nih.gov/23674686>)

Authors

Guerreiro I; Nunes A; Woltering JM; Casaca A; NÃ³voa A; Vinagre T; Hunter ME; Duboule D; Mallo M

Abstract

Patterning of the vertebrate skeleton requires the coordinated activity of Hox genes. In particular, Hox10 proteins are essential to set the transition from thoracic to lumbar vertebrae because of their rib-repressing activity. In snakes, however, the thoracic region extends well into Hox10-expressing areas of the embryo, suggesting that these proteins are unable to block rib formation. Here, we show that this is not a result of the loss of rib-repressing properties by the snake proteins, but rather to a single base pair change in a Hox/Paired box (Pax)-responsive enhancer, which prevents the binding of Hox proteins. This polymorphism is also found in Paenungulata, such as elephants and manatees, which have extended rib cages. In vivo, this modified enhancer failed to respond to Hox10 activity, supporting its role in the extension of rib cages. In contrast, the enhancer could still interact with Hoxb6 and Pax3 to promote rib formation. These results suggest that a polymorphism in the Hox/Pax-responsive enhancer may have played a role in the evolution of the vertebrate spine by differently modulating its response to rib-suppressing and rib-promoting Hox proteins.

Additional References

RELATED GEPHE

Related Genes

No matches found.

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS

@Parallelism - same nucleotide change occurred in snakes