## GEPHE SUMMARY

Gephebase Gene
Myogenic factor 5 (Myf5) (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^Myogenic factor 5 (Myf5)^#gephebase-summary-title)

GepheID
GP00001716

Entry Status
Published

Main curator
Courtier

## PHENOTYPIC CHANGE

Trait Category
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

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

Common Name
squamates

Synonyms
squamates

Rank
order

Lineage
cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Sauropsida; Sauria; Lepidosauria

Parent
Lepidosauria (lepidosaurs) - (Rank: superorder) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 8504 )

NCBI Taxonomy ID
8509 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 8509 )

is Taxon A an Infraspecies?
No

### Taxon B #1

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

Common Name
-

Synonyms
Elaphe guttatus guttatus

Rank
subspecies

Lineage
cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Sauropsida; Sauria; Lepidosauria; Squamata; Bifurcata; Unidentata; Episquamata; Toxicofera; Serpentes; Colubroidea; Colubridae; Colubrinae; Pantherophis; Pantherophis guttatus

Parent
Pantherophis guttatus () - (Rank: species) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 94885 )

NCBI Taxonomy ID
412765 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 412765 )

is Taxon B an Infraspecies?
No

### Taxon B #2

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

Common Name
reticulated python

Synonyms
Boa reticulata; Broghammerus reticulatus; Python reticulatus; reticulated python; reticulate python; Python reticulatus (Schneider, 1801); ZFMK 32378; ZFMK:32378

Rank
species

Lineage
cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Sauropsida; Sauria; Lepidosauria; Squamata; Bifurcata; Unidentata; Episquamata; Toxicofera; Serpentes; Henophidia; Pythonidae; Malayopython

Parent
Malayopython () - (Rank: genus)

(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 1496304 )

NCBI Taxonomy ID

1496311
(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 1496311 )

is Taxon B an Infraspecies?

No

Taxon B #3

Latin Name

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

Common Name

boa

Synonyms

Constrictor constrictor; boa; Boa constrictor Linnaeus, 1758

Rank

species

Lineage

cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Sauropsida; Sauria; Lepidosauria; Squamata; Bifurcata; Unidentata; Episquamata; Toxicofera; Serpentes; Henophidia; Boidae; Boinae; Boa

Parent

Boa () - (Rank: genus)
(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 8573 )

NCBI Taxonomy ID

8574
(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 8574 )

is Taxon B an Infraspecies?

No

## GENOTYPIC CHANGE

Generic Gene Name

Myf5

Synonyms

Myf-5; bHLHc2; B130010J22Rik

String

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

Sequence Similarities

-

GO - Molecular Function

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 - Biological Process

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

UniProtKB Mus musculus

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

GenebankID or UniProtKB

JX154077 (https://www.ncbi.nlm.nih.gov/nuccore/JX154077)

(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 tenrecs; elephants; hyrax and manatees