GEPHE SUMMARY

Gephebase Gene
GDF6 (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^GDF6^#gephebase-summary-title)

GepheID
GP00000389

Entry Status
Published

Main curator
Martin

PHENOTYPIC CHANGE

Trait Category
Morphology (https://www.gephebase.org/search-criteria?/and+Trait Category=^Morphology^#gephebase-summary-title)

Trait
Limb morphology (hindlimb;skeleton) (https://www.gephebase.org/search-criteria?/and+Trait=^Limb morphology (hindlimb;skeleton)^#gephebase-summary-title)

Trait State in Taxon A
Homo sapiens

Trait State in Taxon B
Pan troglodytes; other primates

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

Common Name
human

Synonyms
human; man; Homo sapiens Linnaeus, 1758; Home sapiens; Homo sampiens; Homo sapeins; Homo sapian; Homo sapians; Homo sapien; Homo sapience; Homo sapiense; Homo sapients; Homo sapines; Homo spaiens; Homo spiens; Humo sapiens

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; Primates; Haplorrhini; Simiiformes; Catarrhini; Hominoidea; Hominidae; Homininae; Homo

Parent
Homo () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9605)

NCBI Taxonomy ID
9606 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9606)

is Taxon A an Infraspecies?
No

Taxon B

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

Common Name
-

Synonyms
Homo/Pan/Gorilla group

Rank
subfamily

Lineage
cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria; Euarchontoglires; Primates; Haplorrhini; Simiiformes; Catarrhini; Hominoidea; Hominidae

Parent
Hominidae (great apes) - (Rank: family) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9604)

NCBI Taxonomy ID
207598 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 207598)

is Taxon B an Infraspecies?
No

GENOTYPIC CHANGE

Generic Gene Name
GDF6

Synonyms
KFM; KFS; KFS1; KFSL; SGM1; BMP13; CDMP2; SYNS4; BMP-13; GDF16

String
9606.ENSP00000287020 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9606.ENSP00000287020)

Sequence Similarities
Belongs to the TGF-beta family.

GO - Molecular Function
GO:0042803 : protein homodimerization activity (https://www.ebi.ac.uk/QuickGO/term/GO:0042803)
GO:0005125 : cytokine activity (https://www.ebi.ac.uk/QuickGO/term/GO:0005125)
GO:0008083 : growth factor activity (https://www.ebi.ac.uk/QuickGO/term/GO:0008083)

UniProtKB Homo sapiens
Q6KF10 (http://www.uniprot.org/uniprot/Q6KF10)

GenebankID or UniProtKB
()

GO:0005160 : transforming growth factor beta receptor binding (https://www.ebi.ac.uk/QuickGO/term/GO:0005160)

GO - Biological Process

GO:0045893 : positive regulation of transcription, DNA-templated (https://www.ebi.ac.uk/QuickGO/term/GO:0045893)
GO:0030509 : BMP signaling pathway (https://www.ebi.ac.uk/QuickGO/term/GO:0030509)
GO:0048468 : cell development (https://www.ebi.ac.uk/QuickGO/term/GO:0048468)
GO:0010862 : positive regulation of pathway-restricted SMAD protein phosphorylation (https://www.ebi.ac.uk/QuickGO/term/GO:0010862)
GO:0042981 : regulation of apoptotic process (https://www.ebi.ac.uk/QuickGO/term/GO:0042981)
GO:0043408 : regulation of MAPK cascade (https://www.ebi.ac.uk/QuickGO/term/GO:0043408)
GO:0060395 : SMAD protein signal transduction (https://www.ebi.ac.uk/QuickGO/term/GO:0060395)
GO:0032332 : positive regulation of chondrocyte differentiation (https://www.ebi.ac.uk/QuickGO/term/GO:0032332)
GO:0045666 : positive regulation of neuron differentiation (https://www.ebi.ac.uk/QuickGO/term/GO:0045666)
GO:0006915 : apoptotic process (https://www.ebi.ac.uk/QuickGO/term/GO:0006915)
GO:1900745 : positive regulation of p38MAPK cascade (https://www.ebi.ac.uk/QuickGO/term/GO:1900745)
GO:0032924 : activin receptor signaling pathway (https://www.ebi.ac.uk/QuickGO/term/GO:0032924)
GO:0045444 : fat cell differentiation (https://www.ebi.ac.uk/QuickGO/term/GO:0045444)
GO:0060389 : pathway-restricted SMAD protein phosphorylation (https://www.ebi.ac.uk/QuickGO/term/GO:0060389)
GO:1990009 : retinal cell apoptotic process (https://www.ebi.ac.uk/QuickGO/term/GO:1990009)

GO - Cellular Component

GO:0005615 : extracellular space (https://www.ebi.ac.uk/QuickGO/term/GO:0005615)

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

Deletion (https://www.gephebase.org/search-criteria?/and+Aberration Type=^Deletion^#gephebase-summary-title)

Deletion Size

1-10 kb

Molecular Details of the Mutation

Loss of a limb specific enhancer, the deletion is 5.6 kb

Experimental Evidence

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

Main Reference

Evolving New Skeletal Traits by cis-Regulatory Changes in Bone Morphogenetic Proteins. (2016) (https://pubmed.ncbi.nlm.nih.gov/26774823)

Authors

Indjeian VB; Kingman GA; Jones FC; Guenther CA; Grimwood J; Schmutz J; Myers RM; Kingsley DM

Abstract

Changes in bone size and shape are defining features of many vertebrates. Here we use genetic crosses and comparative genomics to identify specific regulatory DNA alterations controlling skeletal evolution. Armor bone-size differences in sticklebacks map to a major effect locus overlapping BMP family member GDF6. Freshwater fish express more GDF6 due in part to a transposon insertion, and transgenic overexpression of GDF6 phenocopies evolutionary changes in armor-plate size. The human GDF6 locus also has undergone distinctive regulatory evolution, including complete loss of an enhancer that is otherwise highly conserved between chimps and other mammals. Functional tests show that the ancestral enhancer drives expression in hindlimbs but not forelimbs, in locations that have been specifically modified during the human transition to bipedalism. Both gain and loss of regulatory elements can localize BMP changes to specific anatomical locations, providing a flexible regulatory basis for evolving species-specific changes in skeletal form.

Copyright Â© 2016 Elsevier Inc. All rights reserved.

Additional References

RELATED GEPHE

Related Genes

No matches found.

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS

Proposed role in bipedalism