GEPHE SUMMARY

Gephebase Gene

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

GepheID

GP00000388

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

Dermal bone size (armor plates) (https://www.gephebase.org/search-criteria?/and+Trait=^Dermal bone size (armor plates)^#gephebase-summary-title)

Trait State in Taxon A

Gasterosteus aculeatus (marine)

Trait State in Taxon B

Gasterosteus aculeatus (freshwater)

Ancestral State

Taxon A

Taxonomic Status

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

Taxon A

Latin Name

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

Common Name

three-spined stickleback

Synonyms

three-spined stickleback; three spined stickleback; Gasterosteus aculeatus Linnaeus, 1758

Rank

species

Lineage

cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Actinopterygii; Actinopteri; Neopterygii; Teleostei; Osteoglossocephalai; Clupeocephala; Euteleosteomorpha; Neoteleostei; Eurypterygia; Ctenosquamata; Acanthomorphata; Euacanthomorphacea; Percomorphaceae; Eupercaria; Perciformes; Cottioidei; Gasterosteales; Gasterosteidae; Gasterosteus

Parent

Gasterosteus () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 69292)

NCBI Taxonomy ID

69293 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 69293)

is Taxon A an Infraspecies?

Yes

Taxon A Description

Gasterosteus aculeatus (marine)

Taxon B

Latin Name

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

Common Name

three-spined stickleback

Synonyms

three-spined stickleback; three spined stickleback; Gasterosteus aculeatus Linnaeus, 1758

Rank

species

Lineage

cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Actinopterygii; Actinopteri; Neopterygii; Teleostei; Osteoglossocephalai; Clupeocephala; Euteleosteomorpha; Neoteleostei; Eurypterygia; Ctenosquamata; Acanthomorphata; Euacanthomorphacea; Percomorphaceae; Eupercaria; Perciformes; Cottioidei; Gasterosteales; Gasterosteidae; Gasterosteus

Parent

Gasterosteus () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 69292)

NCBI Taxonomy ID

69293 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 69293)

is Taxon B an Infraspecies?

Yes

Taxon B Description

Gasterosteus aculeatus (freshwater)

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)

UniProtKB Homo sapiens

Q6KF10 (http://www.uniprot.org/uniprot/Q6KF10)

GenebankID or UniProtKB

()

GO:0008083 : growth factor activity (https://www.ebi.ac.uk/QuickGO/term/GO:0008083)
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

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

Molecular Details of the Mutation

gain of a flank enhancer activity involving a complex repeat region; 1.2kb L2 LINE insertion; 18 small indels and 89 SNPs - the LINE insertion is required but not sufficient, to produce increased flank expression

Experimental Evidence

Linkage Mapping (https://www.gephebase.org/search-criteria?/and+Experimental Evidence=^Linkage Mapping^#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.

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Additional References

RELATED GEPHE

Related Genes

No matches found.

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS