

GEPHE SUMMARY

GDF6 (https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=GDF6^#gephebase-summary-title)	Gephebase Gene	GP00000388	GepheID
Published	Entry Status	Courtier	Main curator

PHENOTYPIC CHANGE

Morphology (https://www.gephebase.org/search-criteria?/and+Trait+Category=Morphology^#gephebase-summary-title)	Trait Category		
Dermal bone size (armor plates) (https://www.gephebase.org/search-criteria?/and+Trait=Dermal+bone+size+(armor+plates)^#gephebase-summary-title)	Trait		
Gasterosteus aculeatus (marine)	Trait State in Taxon A		
Gasterosteus aculeatus (freshwater)	Trait State in Taxon B		
Taxon A	Ancestral State		
Intraspecific (https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=Intraspecific^#gephebase-summary-title)	Taxonomic Status		
	Taxon A		Taxon B
Gasterosteus aculeatus (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=Gasterosteus+aculeatus^#gephebase-summary-title)	Latin Name	Gasterosteus aculeatus (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=Gasterosteus+aculeatus^#gephebase-summary-title)	Latin Name
three-spined stickleback	Common Name	three-spined stickleback	Common Name
three-spined stickleback; three spined stickleback; Gasterosteus aculeatus Linnaeus, 1758	Synonyms	three-spined stickleback; three spined stickleback; Gasterosteus aculeatus Linnaeus, 1758	Synonyms
species	Rank	species	Rank
cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Actinopterygii; Actinopteri; Neopterygii; Teleostei; Osteoglossocephalai; Clupeocephala; Euteleostei; Neoteleostei; Eurypterygia; Ctenosquamata; Acanthomorpha; Euacanthomorpha; Perciformes; Cottoidei; Gasterosteales; Gasterosteidae; Gasterosteus	Lineage	cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Actinopterygii; Actinopteri; Neopterygii; Teleostei; Osteoglossocephalai; Clupeocephala; Euteleostei; Neoteleostei; Eurypterygia; Ctenosquamata; Acanthomorpha; Euacanthomorpha; Perciformes; Cottoidei; Gasterosteales; Gasterosteidae; Gasterosteus	Lineage
Gasterosteus () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=69292)	Parent	Gasterosteus () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=69292)	Parent
69293 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=69293)	NCBI Taxonomy ID	69293 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=69293)	NCBI Taxonomy ID
Yes	is Taxon A an Intraspecies?	Yes	is Taxon B an Intraspecies?
Gasterosteus aculeatus (marine)	Taxon A Description	Gasterosteus aculeatus (freshwater)	Taxon B Description

GENOTYPIC CHANGE

GDF6	Generic Gene Name	Q6KF10 (http://www.uniprot.org/uniprot/Q6KF10)	UniProtKB Homo sapiens
KFM; KFS; KFS1; KFSL; SGM1; BMP13; CDMP2; SYNS4; BMP-13; GDF16	Synonyms	()	GenebankID or UniProtKB
9606.ENSPO0000287020 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSPO0000287020)	String		
Belongs to the TGF-beta family.	Sequence Similarities		
GO:0042803 : protein homodimerization activity (https://www.ebi.ac.uk/QuickGO/term/GO:0042803)	GO - Molecular Function		
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>)

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](https://www.gephebase.org/search-criteria?/and+Presumptive+Null=))

Molecular Type

Cis-regulatory ([https://www.gephebase.org/search-criteria?/and+Molecular Type="+Cis-regulatory+"#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Molecular+Type=))

Aberration Type

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

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](https://www.gephebase.org/search-criteria?/and+Experimental+Evidence=))

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

