

GEPHE SUMMARY

pitx1 (https://www.gephebase.org/search-criteria?/and+GeneGephebase=^pitx1^#gephebase-summary-title)	Gephebase Gene	GP00000887	GepheID
	Entry Status	Courtier	Main curator
	Published		

PHENOTYPIC CHANGE

Morphology (https://www.gephebase.org/search-criteria?/and+TraitCategory=~Morphology^#gephebase-summary-title)		Trait Category	
Pelvis morphology (loss of ventral spines) (https://www.gephebase.org/search-criteria?/and+Trait=~Pelvis morphology (loss of ventral spines)^#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+TaxonomicStatus=~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)		Gasterosteus aculeatus (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=~Gasterosteus aculeatus^#gephebase-summary-title)	
three-spined stickleback		three-spined stickleback	
three-spined stickleback; three spined stickleback; Gasterosteus aculeatus Linnaeus, 1758		three-spined stickleback; three spined stickleback; Gasterosteus aculeatus Linnaeus, 1758	
species		species	
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; Percomorphaceae; Eupercaria; Perciformes; Cottioidei; Gasterosteales; Gasterosteidae; Gasterosteus		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; Percomorphaceae; Eupercaria; Perciformes; Cottioidei; Gasterosteales; Gasterosteidae; Gasterosteus	
Gasterosteus () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 69292)		Gasterosteus () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 69292)	
69293 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 69293)		69293 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 69293)	
Yes		Yes	
Gasterosteus aculeatus - marine		Gasterosteus aculeatus - freshwater	

GENOTYPIC CHANGE

Pitx1	Generic Gene Name	P70314 (http://www.uniprot.org/uniprot/P70314)	UniProtKB Mus musculus
Bft; Potx; Ptx1; P-OTX	Synonyms	ABH01138 (https://www.ncbi.nlm.nih.gov/nucore/ABH01138)	GenebankID or UniProtKB
10090.ENSMUSP00000021968 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=10090.ENSMUSP00000021968)	String		
Belongs to the paired homeobox family. Bicoid subfamily.	Sequence Similarities		
GO:0003700 : DNA-binding transcription factor activity (https://www.ebi.ac.uk/QuickGO/term/GO:0003700)	GO - Molecular Function		
GO:0043565 : sequence-specific DNA binding			

(<https://www.ebi.ac.uk/QuickGO/term/GO:0043565>)
GO:0001077 : proximal promoter DNA-binding transcription activator activity, RNA polymerase II-specific (<https://www.ebi.ac.uk/QuickGO/term/GO:0001077>)
GO:0000978 : RNA polymerase II proximal promoter sequence-specific DNA binding (<https://www.ebi.ac.uk/QuickGO/term/GO:0000978>)
GO:0001085 : RNA polymerase II transcription factor binding (<https://www.ebi.ac.uk/QuickGO/term/GO:0001085>)

GO - Biological Process

GO:0009653 : anatomical structure morphogenesis (<https://www.ebi.ac.uk/QuickGO/term/GO:0009653>)
GO:0045944 : positive regulation of transcription by RNA polymerase II (<https://www.ebi.ac.uk/QuickGO/term/GO:0045944>)
GO:0006355 : regulation of transcription, DNA-templated (<https://www.ebi.ac.uk/QuickGO/term/GO:0006355>)
GO:0001501 : skeletal system development (<https://www.ebi.ac.uk/QuickGO/term/GO:0001501>)
GO:0045892 : negative regulation of transcription, DNA-templated (<https://www.ebi.ac.uk/QuickGO/term/GO:0045892>)
GO:0051216 : cartilage development (<https://www.ebi.ac.uk/QuickGO/term/GO:0051216>)
GO:0048625 : myoblast fate commitment (<https://www.ebi.ac.uk/QuickGO/term/GO:0048625>)
GO:0035116 : embryonic hindlimb morphogenesis (<https://www.ebi.ac.uk/QuickGO/term/GO:0035116>)
GO:0035137 : hindlimb morphogenesis (<https://www.ebi.ac.uk/QuickGO/term/GO:0035137>)
GO:0014707 : branchiomic skeletal muscle development (<https://www.ebi.ac.uk/QuickGO/term/GO:0014707>)
GO:0021983 : pituitary gland development (<https://www.ebi.ac.uk/QuickGO/term/GO:0021983>)

GO - Cellular Component

GO:0005737 : cytoplasm (<https://www.ebi.ac.uk/QuickGO/term/GO:0005737>)
GO:0005634 : nucleus (<https://www.ebi.ac.uk/QuickGO/term/GO:0005634>)
GO:0005667 : transcription factor complex (<https://www.ebi.ac.uk/QuickGO/term/GO:0005667>)
GO:0005730 : nucleolus (<https://www.ebi.ac.uk/QuickGO/term/GO:0005730>)

No (https://www.gephebase.org/search-criteria?/and+Presumptive Null=^No^#gephebase-summary-title)	Presumptive Null
Cis-regulatory (https://www.gephebase.org/search-criteria?/and+Molecular Type=^Cis-regulatory^#gephebase-summary-title)	Molecular Type
Deletion (https://www.gephebase.org/search-criteria?/and+Aberration Type=^Deletion^#gephebase-summary-title)	Aberration Type
100-999 bp	Deletion Size
488bp promoter region : deletion 2 ; 757bp deletion present in the pelvic-reduced BEPA population from Alaska	Molecular Details of the Mutation
Linkage Mapping (https://www.gephebase.org/search-criteria?/and+Experimental Evidence=^Linkage Mapping^#gephebase-summary-title)	Experimental Evidence
Genetic and developmental basis of evolutionary pelvic reduction in threespine sticklebacks. (2004) (https://pubmed.ncbi.nlm.nih.gov/15085123)	Main Reference
Shapiro MD; Marks ME; Peichel CL; Blackman BK; Nereng KS; JÄ³nsson B; Schluter D; Kingsley DM	Authors
Hindlimb loss has evolved repeatedly in many different animals by means of molecular mechanisms that are still unknown. To determine the number and type of genetic changes underlying pelvic reduction in natural populations, we carried out genetic crosses between threespine stickleback fish with complete or missing pelvic structures. Genome-wide linkage mapping shows that pelvic reduction is controlled by one major and four minor chromosome regions. Pitx1 maps to the major chromosome region controlling most of the variation in pelvic size. Pelvic-reduced fish show the same left-right asymmetry seen in Pitx1 knockout mice, but do not show changes in Pitx1 protein sequence. Instead, pelvic-reduced sticklebacks show site-specific regulatory changes in Pitx1 expression, with reduced or absent expression in pelvic and caudal fin precursors. Regulatory mutations in major developmental control genes may provide a mechanism for generating rapid skeletal changes in natural populations, while preserving the essential roles of these genes in other processes.	Abstract
Adaptive evolution of pelvic reduction in sticklebacks by recurrent deletion of a Pitx1 enhancer. (2010) (https://pubmed.ncbi.nlm.nih.gov/20007865)	Additional References

RELATED GEPHE

No matches found.	Related Genes
8 (https://www.gephebase.org/search-criteria?/or+Gene Gephebase=^pitx1^/and+Taxon ID=^69293^/or+Gene Gephebase=^pitx1^/and+Taxon ID=^69293^#gephebase-summary-title)	Related Haplotypes

EXTERNAL LINKS

COMMENTS

@Parallelism @Multi-allelic