GEPHE SUMMARY

Gephebase Gene

pitx1 (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^pitx1^#gephebase-summary-title)

GepheID

GP00000892

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

Pelvis morphology (loss of ventral spines) (https://www.gephebase.org/search-criteria?/and+Trait=^Pelvis morphology (loss of ventral spines)^#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

Pitx1

Synonyms

Bft; Potx; Ptx1; P-OTX

String

10090.ENSMUSP00000021968 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=10090.ENSMUSP00000021968)

Sequence Similarities

Belongs to the paired homeobox family. Bicoid subfamily.

GO - Molecular Function

GO:0003700 : DNA-binding transcription factor activity (https://www.ebi.ac.uk/QuickGO/term/GO:0003700)
GO:0043565 : sequence-specific DNA binding

UniProtKB Mus musculus

P70314 (http://www.uniprot.org/uniprot/P70314)

GenebankID or UniProtKB

ABH01138 (https://www.ncbi.nlm.nih.gov/nuccore/ABH01138)

(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 : branchiomeric 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)

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

488bp promoter region : deletion 7 ; 1868bp deletion

Experimental Evidence

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

Main Reference

Genetic and developmental basis of evolutionary pelvic reduction in threespine sticklebacks. (2004) (https://pubmed.ncbi.nlm.nih.gov/15085123)

Authors

Shapiro MD; Marks ME; Peichel CL; Blackman BK; Nereng KS; JÃ³nsson B; Schluter D; Kingsley DM

Abstract

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.

Additional References

Adaptive evolution of pelvic reduction in sticklebacks by recurrent deletion of a Pitx1 enhancer. (2010) (https://pubmed.ncbi.nlm.nih.gov/20007865)

RELATED GEPHE

Related Genes

No matches found.

Related Haplotypes

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)

EXTERNAL LINKS

COMMENTS

@Parallelism @Multi-allelic