

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

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

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](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

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