

GEPHE SUMMARY

	Gephebase Gene	GephelD
Ibh (limb bud and heart homolog) (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^Ibh (limb bud and heart homolog)^#gephebase-summary-title)	GP00001387	
	Entry Status	Main curator
Published	Prigent	

PHENOTYPIC CHANGE

	Trait Category	
Morphology (https://www.gephebase.org/search-criteria?/and+Trait Category=^Morphology^#gephebase-summary-title)	Trait	
Cranio-facial morphology (mandible) (https://www.gephebase.org/search-criteria?/and+Trait=^Cranio-facial morphology (mandible)^#gephebase-summary-title)	Trait State in Taxon A	
Maylandia zebra	Trait State in Taxon B	
Labeotropheus fuelleborni	Ancestral State	
Taxon A	Taxonomic Status	
Interspecific (https://www.gephebase.org/search-criteria?/and+Taxonomic Status=^Interspecific^#gephebase-summary-title)		
Taxon A	Latin Name	Taxon B
Maylandia zebra (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Maylandia zebra^#gephebase-summary-title)	Labeotropheus fuelleborni (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Labeotropheus fuelleborni^#gephebase-summary-title)	Latin Name
zebra mbuna	Common Name	Common Name
Metriaclima zebra; Pseudotropheus sp. 'Pseudotropheus zebra complex'; Pseudotropheus zebra; zebra mbuna; Maylandia zebra (Boulenger, 1899)	Synonyms	Synonyms
species	Rank	Rank
cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Actinopterygii; Actinopteri; Neopterygii; Teleostei; Osteoglossocephalai; Clupeocephala; Euteleosteomorpha; Neoteleoste; Eurypterygia; Ctenosquamata; Acanthomorpha; Euacanthomorphae; Percomorphaceae; Ovalentaria; Cichlomorphae; Cichliformes; Cichlidae; African cichlids; Pseudocrenilabrinae; Haplochromini; Maylandia; Maylandia zebra complex	Lineage	Lineage
Maylandia zebra complex () - (Rank: species group) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 57445)	Parent	Parent
106582 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 106582)	NCBI Taxonomy ID	NCBI Taxonomy ID
No	is Taxon A an Infraspecies?	is Taxon B an Infraspecies?

GENOTYPIC CHANGE

	Generic Gene Name	UniProtKB Homo sapiens
LBH	Q53QV2 (http://www.uniprot.org/uniprot/Q53QV2)	
	Synonyms	GenebankID or UniProtKB
-	0	
	String	
9606.ENSP00000378733 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9606.ENSP00000378733)		
	Sequence Similarities	
Belongs to the LBH family.		
	GO - Molecular Function	
-		
	GO - Biological Process	
GO:0045893 : positive regulation of transcription, DNA-templated		

(<https://www.ebi.ac.uk/QuickGO/term/GO:0045893>)
 GO:0060644 : mammary gland epithelial cell differentiation
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0060644>)
 GO:0033147 : negative regulation of intracellular estrogen receptor signaling pathway
 (<https://www.ebi.ac.uk/QuickGO/term/GO:0033147>)
 GO:2000737 : negative regulation of stem cell differentiation
 (<https://www.ebi.ac.uk/QuickGO/term/GO:2000737>)
 GO:2000103 : positive regulation of mammary stem cell proliferation
 (<https://www.ebi.ac.uk/QuickGO/term/GO:2000103>)
 GO:1904677 : positive regulation of somatic stem cell division
 (<https://www.ebi.ac.uk/QuickGO/term/GO:1904677>)
 GO:1904674 : positive regulation of somatic stem cell population maintenance
 (<https://www.ebi.ac.uk/QuickGO/term/GO:1904674>)

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>)

Presumptive Null

No ([#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Presumptive Null=^No))

Molecular Type

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

Aberration Type

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

SNP Coding Change

Nonsynonymous

Molecular Details of the Mutation

G>A p.R17Q

Experimental Evidence

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

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	-	-	-

Main Reference

A nonsynonymous mutation in the transcriptional regulator *Ibh* is associated with cichlid craniofacial adaptation and neural crest cell development. (2014)
 (<https://pubmed.ncbi.nlm.nih.gov/25234704>)

Authors

Powder KE; Cousin H; McLinden GP; Craig Albertson R

Abstract

Since the time of Darwin, biologists have sought to understand the origins and maintenance of life's diversity of form. However, the nature of the exact DNA mutations and molecular mechanisms that result in morphological differences between species remains unclear. Here, we characterize a nonsynonymous mutation in a transcriptional coactivator, limb bud and heart homolog (*Ibh*), which is associated with adaptive variation in the lower jaw of cichlid fishes. Using both zebrafish and *Xenopus*, we demonstrate that *Ibh* mediates migration of cranial neural crest cells, the cellular source of the craniofacial skeleton. A single amino acid change that is alternatively fixed in cichlids with differing facial morphologies results in discrete shifts in migration patterns of this multipotent cell type that are consistent with both embryological and adult craniofacial phenotypes. Among animals, this polymorphism in *Ibh* represents a rare example of a coding change that is associated with continuous morphological variation. This work offers novel insights into the development and evolution of the craniofacial skeleton, underscores the evolutionary potential of neural crest cells, and extends our understanding of the genetic nature of mutations that underlie divergence in complex phenotypes.

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 journals.permissions@oup.com.

Additional References

RELATED GEPHE

1 (BMP4 (uncertain)) ([#gephebase-summary-title](https://www.gephebase.org/search-criteria?/or+Taxon ID=^106582^/and+Trait=Cranio-facial morphology/or+Taxon ID=^57307^/and+Trait=Cranio-facial morphology/and+groupHaplotypes=true))

Related Genes

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS

