

GEPHE SUMMARY

BMP3 or PRKG2 (https://www.gephebase.org/search-criteria?/and+GeneGephebase=~BMP3 or PRKG2^#gephebase-summary-title)	Gephebase Gene	GP00000152	GepheID
	Entry Status	Martin	Main curator
	Published		

PHENOTYPIC CHANGE

Morphology (https://www.gephebase.org/search-criteria?/and+TraitCategory=~Morphology^#gephebase-summary-title)		Trait Category	
Cranio-facial morphology (https://www.gephebase.org/search-criteria?/and+Trait=~Cranio-facial morphology^#gephebase-summary-title)		Trait	
Canis familiaris - various breeds		Trait State in Taxon A	
Canis familiaris - various breeds		Trait State in Taxon B	
Taxon A		Ancestral State	
Domesticated (https://www.gephebase.org/search-criteria?/and+TaxonomicStatus=~Domesticated^#gephebase-summary-title)		Taxonomic Status	
Taxon A		Taxon B	
Canis lupus (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=~Canis lupus^#gephebase-summary-title)		Canis lupus familiaris (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=~Canis lupus familiaris^#gephebase-summary-title)	
gray wolf		dog	
gray wolf; grey wolf; Canis lupus Linnaeus, 1758		Canis canis; Canis domesticus; Canis familiaris; dog; dogs; Canis familiaris Linnaeus, 1758; Canis lupus familiaris Linnaeus, 1758	
species		subspecies	
cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria; Laurasiatheria; Carnivora; Caniformia; Canidae; Canis		cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria; Laurasiatheria; Carnivora; Caniformia; Canidae; Canis; Canis lupus	
Canis () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9611)		Canis lupus (gray wolf) - (Rank: species) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9612)	
9612 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9612)		9615 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9615)	
No is Taxon A an Intraspecies?		No is Taxon B an Intraspecies?	

GENOTYPIC CHANGE

Bmp3		Q8BHE5 (http://www.uniprot.org/uniprot/Q8BHE5)	
9530029 04Rik		0	
10090.ENSMUSP00000031278 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=10090.ENSMUSP00000031278)			
Belongs to the TGF-beta family.			
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:0070700 : BMP receptor binding (https://www.ebi.ac.uk/QuickGO/term/GO:0070700)			

GO - Biological Process

GO:0045944 : positive regulation of transcription by RNA polymerase II
(<https://www.ebi.ac.uk/QuickGO/term/GO:0045944>)
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:0051216 : cartilage development (<https://www.ebi.ac.uk/QuickGO/term/GO:0051216>)
GO:0001649 : osteoblast differentiation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0001649>)

GO - Cellular Component

GO:0031982 : vesicle (<https://www.ebi.ac.uk/QuickGO/term/GO:0031982>)
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>)

Molecular Type

Coding (<https://www.gephebase.org/search-criteria?/and+Molecular Type=~Coding~#gephebase-summary-title>)

Aberration Type

SNP (<https://www.gephebase.org/search-criteria?/and+Aberration Type=~SNP~#gephebase-summary-title>)

SNP Coding Change

Nonsynonymous

Molecular Details of the Mutation

F452L in BMP3 ; this missense mutation is in a conserved position and correlates well with small breeds. However; the functional data (zebrafish BMP3 morpholino knockdown) proposed in this article is inconsistent with the mice BMP3 null-mutation

Experimental Evidence

Association Mapping (<https://www.gephebase.org/search-criteria?/and+Experimental Evidence=~Association Mapping~#gephebase-summary-title>)

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

Main Reference

Variation of BMP3 contributes to dog breed skull diversity. (2012) (<https://pubmed.ncbi.nlm.nih.gov/22876193>)

Authors

Schoenebeck JJ; Hutchinson SA; Byers A; Beale HC; Carrington B; Faden DL; Rimbault M; Decker B; Kidd JM; Sood R; Boyko AR; Fondon JW; Wayne RK; Bustamante CD; Ciruna B; Ostrander EA

Abstract

Since the beginnings of domestication, the craniofacial architecture of the domestic dog has morphed and radiated to human whims. By beginning to define the genetic underpinnings of breed skull shapes, we can elucidate mechanisms of morphological diversification while presenting a framework for understanding human cephalic disorders. Using intrabreed association mapping with museum specimen measurements, we show that skull shape is regulated by at least five quantitative trait loci (QTLs). Our detailed analysis using whole-genome sequencing uncovers a missense mutation in BMP3. Validation studies in zebrafish show that Bmp3 function in cranial development is ancient. Our study reveals the causal variant for a canine QTL contributing to a major morphologic trait.

Additional References

RELATED GEPHE

Related Genes

No matches found.

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS

