GEPHE SUMMARY

Gephebase Gene
BMP3 or PRKG2 (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^BMP3 or PRKG2^#gephebase-summary-title)

GepheID
GP00000152

Entry Status
Published

Main curator
Martin

PHENOTYPIC CHANGE

Trait Category
Morphology (https://www.gephebase.org/search-criteria?/and+Trait Category=^Morphology^#gephebase-summary-title)

Trait
Cranio-facial morphology (https://www.gephebase.org/search-criteria?/and+Trait=^Cranio-facial morphology^#gephebase-summary-title)

Trait State in Taxon A
Canis familiaris - various breeds

Trait State in Taxon B
Canis familiaris - various breeds

Ancestral State
Taxon A

Taxonomic Status
Domesticated (https://www.gephebase.org/search-criteria?/and+Taxonomic Status=^Domesticated^#gephebase-summary-title)

Taxon A

Latin Name
Canis lupus (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Canis lupus^#gephebase-summary-title)

Common Name
gray wolf

Synonyms
gray wolf; grey wolf; Canis lupus Linnaeus, 1758

Rank
species

Lineage
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

Parent
Canis () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9611)

NCBI Taxonomy ID
9612 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9612)

is Taxon A an Infraspecies?
No

Taxon B

Latin Name
Canis lupus familiaris (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Canis lupus familiaris^#gephebase-summary-title)

Common Name
dog

Synonyms
Canis canis; Canis domesticus; Canis familiaris; dog; dogs; Canis familiaris Linnaeus, 1758; Canis lupus familiaris Linnaeus, 1758

Rank
subspecies

Lineage
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

Parent
Canis lupus (gray wolf) - (Rank: species) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9612)

NCBI Taxonomy ID
9615 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9615)

is Taxon B an Infraspecies?
No

GENOTYPIC CHANGE

Generic Gene Name
Bmp3

Synonyms
9530029I04Rik

String
10090.ENSMUSP00000031278 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=10090.ENSMUSP00000031278)

Sequence Similarities
Belongs to the TGF-beta family.

GO - Molecular Function
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)

UniProtKB Mus musculus
Q8BHE5 (http://www.uniprot.org/uniprot/Q8BHE5)

GenebankID or UniProtKB
()

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