GEPHE SUMMARY

Gephebase Gene
FGF4 retrogene (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^FGF4 retrogene^#gephebase-summary-title)

GepheID
GP00000309

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
Limb size (https://www.gephebase.org/search-criteria?/and+Trait=^Limb size^#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?
Yes

Taxon B Description
Basset Hound ; Cairn Terrier ; Cardigan Welsh Corgi ; Dachshund; Dandie Dinmont Terrier ; Lancashire heeler; Norwich Terrier ; Pekingese ; Pembroke Welsh Corgi ; Petit Basset ; Griffon VendÃ©en ; Shih-Tzu ; Skye Terrier ; Swedish Vallhund ; Tibetan spaniel ; West Highland Terrier

GENOTYPIC CHANGE

Generic Gene Name
FGF4

Synonyms
-

String
9615.ENSCAFP00000040116 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9615.ENSCAFP00000040116)

Sequence Similarities
Belongs to the heparin-binding growth factors family.

GO - Molecular Function
GO:0008083 : growth factor activity (https://www.ebi.ac.uk/QuickGO/term/GO:0008083)

UniProtKB Canis lupus familiaris
J9P3I7 (http://www.uniprot.org/uniprot/J9P3I7)

GenebankID or UniProtKB
AAEX03011593 (https://www.ncbi.nlm.nih.gov/nuccore/AAEX03011593)

GO:0005104 : fibroblast growth factor receptor binding
(https://www.ebi.ac.uk/QuickGO/term/GO:0005104)

GO - Biological Process

GO:0043066 : negative regulation of apoptotic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0043066)
GO:0045944 : positive regulation of transcription by RNA polymerase II
(https://www.ebi.ac.uk/QuickGO/term/GO:0045944)
GO:0001502 : cartilage condensation
(https://www.ebi.ac.uk/QuickGO/term/GO:0001502)
GO:0070374 : positive regulation of ERK1 and ERK2 cascade
(https://www.ebi.ac.uk/QuickGO/term/GO:0070374)
GO:0008284 : positive regulation of cell proliferation
(https://www.ebi.ac.uk/QuickGO/term/GO:0008284)
GO:0042475 : odontogenesis of dentin-containing tooth
(https://www.ebi.ac.uk/QuickGO/term/GO:0042475)
GO:0019827 : stem cell population maintenance
(https://www.ebi.ac.uk/QuickGO/term/GO:0019827)
GO:0060561 : apoptotic process involved in morphogenesis
(https://www.ebi.ac.uk/QuickGO/term/GO:0060561)
GO:1990830 : cellular response to leukemia inhibitory factor
(https://www.ebi.ac.uk/QuickGO/term/GO:1990830)
GO:0060591 : chondroblast differentiation
(https://www.ebi.ac.uk/QuickGO/term/GO:0060591)
GO:0060363 : cranial suture morphogenesis
(https://www.ebi.ac.uk/QuickGO/term/GO:0060363)
GO:0035116 : embryonic hindlimb morphogenesis
(https://www.ebi.ac.uk/QuickGO/term/GO:0035116)
GO:0008543 : fibroblast growth factor receptor signaling pathway
(https://www.ebi.ac.uk/QuickGO/term/GO:0008543)
GO:0010463 : mesenchymal cell proliferation
(https://www.ebi.ac.uk/QuickGO/term/GO:0010463)
GO:2000544 : regulation of endothelial cell chemotaxis to fibroblast growth factor
(https://www.ebi.ac.uk/QuickGO/term/GO:2000544)

GO - Cellular Component

GO:0005576 : extracellular region (https://www.ebi.ac.uk/QuickGO/term/GO:0005576)

Presumptive Null

Unknown (https://www.gephebase.org/search-criteria?/and+Presumptive Null=^Unknown^#gephebase-summary-title)

Molecular Type

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

Aberration Type

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

Insertion Size

10-100 kb

Molecular Details of the Mutation

Gene retroposition : 5kb insertion containing a FGF4 retrogene; 25Mb away from the complete (original) FGF4 gene

Experimental Evidence

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

Main Reference

An expressed fgf4 retrogene is associated with breed-defining chondrodysplasia in domestic dogs. (2009) (https://pubmed.ncbi.nlm.nih.gov/19608863)

Authors

Parker HG; VonHoldt BM; Quignon P; Margulies EH; Shao S; Mosher DS; Spady TC; Elkahloun A; Cargill M; Jones PG; Maslen CL; Acland GM; Sutter NB; Kuroki K; Bustamante CD; Wayne RK; Ostrander EA

Abstract

Retrotransposition of processed mRNAs is a common source of novel sequence acquired during the evolution of genomes. Although the vast majority of retroposed gene copies, or retrogenes, rapidly accumulate debilitating mutations that disrupt the reading frame, a small percentage become new genes that encode functional proteins. By using a multibreed association analysis in the domestic dog, we demonstrate that expression of a recently acquired retrogene encoding fibroblast growth factor 4 (fgf4) is strongly associated with chondrodysplasia, a short-legged phenotype that defines at least 19 dog breeds including dachshund, corgi, and basset hound. These results illustrate the important role of a single evolutionary event in constraining and directing phenotypic diversity in the domestic dog.

Additional References

RELATED GEPHE

Related Genes

No matches found.

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS

https://omia.org/OMIA000187/9615/