GEPHE SUMMARY

Gephebase Gene

FOXI3 (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^FOXI3^#gephebase-summary-title)

GepheID

GP00000351

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

Hair (hypotrichosis) (https://www.gephebase.org/search-criteria?/and+Trait=^Hair (hypotrichosis)^#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

FOXI3

Synonyms

-

String

9615.ENSCAFP00000041322 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9615.ENSCAFP00000041322)

Sequence Similarities

-

GO - Molecular Function

GO:0043565 : sequence-specific DNA binding (https://www.ebi.ac.uk/QuickGO/term/GO:0043565)
GO:0000981 : DNA-binding transcription factor activity, RNA polymerase II-specific (https://www.ebi.ac.uk/QuickGO/term/GO:0000981)

GO - Biological Process

GO:0009653 : anatomical structure morphogenesis

UniProtKB Canis lupus familiaris

B5RHS5 (http://www.uniprot.org/uniprot/B5RHS5)

GenebankID or UniProtKB

AM998820 (https://www.ncbi.nlm.nih.gov/nuccore/AM998820)

(https://www.ebi.ac.uk/QuickGO/term/GO:0009653)
GO:0006357 : regulation of transcription by RNA polymerase II
(https://www.ebi.ac.uk/QuickGO/term/GO:0006357)
GO:0030154 : cell differentiation (https://www.ebi.ac.uk/QuickGO/term/GO:0030154)

GO - Cellular Component

GO:0005634 : nucleus (https://www.ebi.ac.uk/QuickGO/term/GO:0005634)

Presumptive Null

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

Molecular Type

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

Aberration Type

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

Insertion Size

1-9 bp

Molecular Details of the Mutation

Frameshift; 7-bp duplication within exon 1

Experimental Evidence

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

Main Reference

A mutation in hairless dogs implicates FOXI3 in ectodermal development. (2008) (https://pubmed.ncbi.nlm.nih.gov/18787161)

Authors

DrÃ¶gemÃ¼ller C; Karlsson EK; HytÃ¶nen MK; Perloski M; Dolf G; Sainio K; Lohi H; Lindblad-Toh K; Leeb T

Abstract

Mexican and Peruvian hairless dogs and Chinese crested dogs are characterized by missing hair and teeth, a phenotype termed canine ectodermal dysplasia (CED). CED is inherited as a monogenic autosomal semidominant trait. With genomewide association analysis we mapped the CED mutation to a 102-kilo-base pair interval on chromosome 17. The associated interval contains a previously uncharacterized member of the forkhead box transcription factor family (FOXI3), which is specifically expressed in developing hair and teeth. Mutation analysis revealed a frameshift mutation within the FOXI3 coding sequence in hairless dogs. Thus, we have identified FOXI3 as a regulator of ectodermal development.

Additional References

RELATED GEPHE

Related Genes

4 (FGF5, R-spondin-2 (RSPO2), SGK3, KRT71) (https://www.gephebase.org/search-criteria?/or+Taxon ID=^9612^/and+Trait=Hair/or+Taxon ID=^9615^/and+Trait=Hair/and+groupHaplotypes=true#gephebase-summary-title)

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS

@HeterozygoteAdvantage (homozygous embryonic lethal) https://omia.org/OMIA000323/9615/