GEPHE SUMMARY

Gephebase Gene

KRT71 (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^KRT71^#gephebase-summary-title)

Entry Status

Published

GepheID

GP00002231

Main curator

Martin

PHENOTYPIC CHANGE

Trait Category

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

Trait

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

Trait State in Taxon A

Canis familiaris - various breeds

Trait State in Taxon B

Various breeds with curly hair including Bichon Frise ; Chesapeake Bay Retriever ; Curly-coated retriever ; Irish Terrier ; Lagotto Romagnolo ; Spanish water dog

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

Krt71

Synonyms

Ca; Cu; Cal4; mK6irs; Krt2-6g; mK6irs1; AA589543; K6irs1; Kb34; Krt6g

String

10090.ENSMUSP00000023710 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=10090.ENSMUSP00000023710)

Sequence Similarities

Belongs to the intermediate filament family.

GO - Molecular Function

GO:0005198 : structural molecule activity (https://www.ebi.ac.uk/QuickGO/term/GO:0005198)

GO - Biological Process

GO:0031069 : hair follicle morphogenesis (https://www.ebi.ac.uk/QuickGO/term/GO:0031069)

UniProtKB Mus musculus

Q9R0H5 (http://www.uniprot.org/uniprot/Q9R0H5)

GenebankID or UniProtKB

ADA57168 (https://www.ncbi.nlm.nih.gov/nuccore/ADA57168)

GO:0045109 : intermediate filament organization (https://www.ebi.ac.uk/QuickGO/term/GO:0045109)

GO - Cellular Component

GO:0005737 : cytoplasm (https://www.ebi.ac.uk/QuickGO/term/GO:0005737)
GO:0045095 : keratin filament (https://www.ebi.ac.uk/QuickGO/term/GO:0045095)

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

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

Indel Size

1-9 bp

Molecular Details of the Mutation

c.1266_1273delinsACA p.Ser422ArgfsTer?

Experimental Evidence

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

Main Reference

A second KRT71 allele in curly coated dogs. (2019) (https://pubmed.ncbi.nlm.nih.gov/30444027)

Authors

Bauer A; Hadji Rasouliha S; Brunner MT; Jagannathan V; Bucher I; Bannoehr J; Varjonen K; Bond R; Bergvall K; Welle MM; Roosje P; Leeb T

Abstract

Major characteristics of coat variation in dogs can be explained by variants in only a few genes. Until now, only one missense variant in the KRT71 gene, p.Arg151Trp, has been reported to cause curly hair in dogs. However, this variant does not explain the curly coat in all breeds as the mutant Trp allele, for example, is absent in Curly Coated Retrievers. We sequenced the genome of a Curly Coated Retriever at 22Ã— coverage and searched for variants in the KRT71 gene. Only one protein-changing variant was present in a homozygous state in the Curly Coated Retriever and absent or present in a heterozygous state in 221 control dogs from different dog breeds. This variant, NM_001197029.1:c.1266_1273delinsACA, was an indel variant in exon 7 that caused a frameshift and an altered and probably extended C-terminus of the KRT71 protein NP_001183958.1:p.(Ser422ArgfsTer?). Using Sanger sequencing, we found that the variant was fixed in a cohort of 125 Curly Coated Retrievers and segregating in five of 14 additionally tested breeds with a curly or wavy coat. KRT71 variants cause curly hair in humans, mice, rats, cats and dogs. Specific KRT71 variants were further shown to cause alopecia. Based on this knowledge from other species and the predicted molecular consequence of the newly identified canine KRT71 variant, it is a compelling candidate causing a second curly hair allele in dogs. It might cause a slightly different coat phenotype than the previously published p.Arg151Trp variant and could potentially be associated with follicular dysplasia in dogs.

Additional References

A novel KRT71 variant in curly-coated dogs. (2019) (https://pubmed.ncbi.nlm.nih.gov/30456859)

RELATED GEPHE

Related Genes

No matches found.

Related Haplotypes

1 (https://www.gephebase.org/search-criteria?/or+Gene Gephebase=^KRT71^/and+Taxon ID=^9612^/or+Gene Gephebase=^KRT71^/and+Taxon ID=^9615^#gephebase-summary-title)

EXTERNAL LINKS

COMMENTS

@AllelicSeries https://omia.org/OMIA000245/9615/