

GEPHE SUMMARY

<p>KRT71 (<a +krt71+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=">https://www.gephebase.org/search-criteria?/and+Gene+Gephebase="+KRT71+"#gephebase-summary-title)</p> <p>Published</p>	<p>Gephebase Gene</p> <p>Entry Status</p>	<p>GP00002231</p> <p>Martin</p>	<p>GepheID</p> <p>Main curator</p>
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PHENOTYPIC CHANGE

<p>Morphology (<a +morphology+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Trait+Category=">https://www.gephebase.org/search-criteria?/and+Trait+Category="+Morphology+"#gephebase-summary-title)</p> <p>Hair type (curly) (<a +hair+type+(curly)+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Trait=">https://www.gephebase.org/search-criteria?/and+Trait="+Hair+type+(curly)+"#gephebase-summary-title)</p> <p>Canis familiaris - various breeds</p> <p>Various breeds with curly hair including Bichon Frise ; Chesapeake Bay Retriever ; Curly-coated retriever ; Irish Terrier ; Lagotto Romagnolo ; Spanish water dog</p> <p>Taxon A</p> <p>Domesticated (<a +domesticated+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=">https://www.gephebase.org/search-criteria?/and+Taxonomic+Status="+Domesticated+"#gephebase-summary-title)</p>	<p>Trait Category</p> <p>Trait</p> <p>Trait State in Taxon A</p> <p>Trait State in Taxon B</p> <p>Ancestral State</p> <p>Taxonomic Status</p>	<p>Canis lupus (<a +canis+lupus+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms="+Canis+lupus+"#gephebase-summary-title)</p> <p>gray wolf</p> <p>gray wolf; grey wolf; Canis lupus Linnaeus, 1758</p> <p>species</p> <p>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</p> <p>Canis () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9611)</p> <p>9612 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9612)</p> <p>No is Taxon A an Intraspecies?</p>	<p>Taxon A</p> <p>Latin Name</p> <p>Common Name</p> <p>Synonyms</p> <p>Rank</p> <p>Lineage</p> <p>Parent</p> <p>NCBI Taxonomy ID</p> <p>is Taxon A an Intraspecies?</p>	<p>Canis lupus familiaris (<a +canis+lupus+familiaris+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms="+Canis+lupus+familiaris+"#gephebase-summary-title)</p> <p>dog</p> <p>Canis canis; Canis domesticus; Canis familiaris; dog; dogs; Canis familiaris Linnaeus, 1758; Canis lupus familiaris Linnaeus, 1758</p> <p>subspecies</p> <p>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</p> <p>Canis lupus (gray wolf) - (Rank: species) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9612)</p> <p>9615 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9615)</p> <p>No is Taxon B an Intraspecies?</p>	<p>Taxon B</p> <p>Latin Name</p> <p>Common Name</p> <p>Synonyms</p> <p>Rank</p> <p>Lineage</p> <p>Parent</p> <p>NCBI Taxonomy ID</p> <p>is Taxon B an Intraspecies?</p>
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GENOTYPIC CHANGE

<p>Krt71</p> <p>Ca; Cu; Cal4; mK6irs; Krt2-6g; mK6irs1; AA589543; K6irs1; Kb34; Krt6g</p> <p>10090.ENSMUSP00000023710 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=10090.ENSMUSP00000023710)</p> <p>Belongs to the intermediate filament family.</p> <p>GO:0005198 : structural molecule activity (https://www.ebi.ac.uk/QuickGO/term/GO:0005198)</p> <p>GO:0031069 : hair follicle morphogenesis (https://www.ebi.ac.uk/QuickGO/term/GO:0031069)</p>	<p>Generic Gene Name</p> <p>Synonyms</p> <p>String</p> <p>Sequence Similarities</p> <p>GO - Molecular Function</p> <p>GO - Biological Process</p>	<p>Q9R0H5 (http://www.uniprot.org/uniprot/Q9R0H5)</p> <p>ADA57168 (https://www.ncbi.nlm.nih.gov/nuccore/ADA57168)</p>	<p>UniProtKB Mus musculus</p> <p>GenebankID or UniProtKB</p>
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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>)

No (<https://www.gephebase.org/search-criteria?/and+Presumptive Null=^No^#gephebase-summary-title>) Presumptive Null

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

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

1-9 bp Indel Size

c.1266_1273delinsACA p.Ser422ArgfsTer? Molecular Details of the Mutation

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

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

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 Authors

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 22Å— 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.

© 2018 Stichting International Foundation for Animal Genetics. Additional References

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

RELATED GEPHE

No matches found. Related Genes

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>) Related Haplotypes

EXTERNAL LINKS

COMMENTS

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