GEPHE SUMMARY

Gephebase Gene

RALY (hnRNP associated with lethal yellow) (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^RALY (hnRNP associated with lethal yellow)^#gephebase-summary-title)

Entry Status

Published

GepheID

GP00001366

Main curator

Prigent

PHENOTYPIC CHANGE

Trait Category

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

Trait

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

Trait State in Taxon A

Dog (Basset Hounds and Pembroke Welsh Corgis) and wolf and coyotte ; saddle tan

Trait State in Taxon B

Basset hounds and Pembroke Welsh Corgis ; Black and tan

Ancestral State

Taxon A

Taxonomic Status

Intraspecific (https://www.gephebase.org/search-criteria?/and+Taxonomic Status=^Intraspecific^#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?

Yes

Taxon A Description

Dog (Basset Hounds and Pembroke Welsh Corgis) and wolf and coyotte ; saddle tan

Taxon B

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 B an Infraspecies?

Yes

Taxon B Description

Basset hounds and Pembroke Welsh Corgis ; Black and tan

GENOTYPIC CHANGE

Generic Gene Name

RALY

Synonyms

P542; HNRPCL2

String

9606.ENSP00000246194 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9606.ENSP00000246194)

Sequence Similarities

Belongs to the RRM HNRPC family. RALY subfamily.

GO - Molecular Function

GO:0003723 : RNA binding (https://www.ebi.ac.uk/QuickGO/term/GO:0003723)
GO:0003712 : transcription coregulator activity (https://www.ebi.ac.uk/QuickGO/term/GO:0003712)

GO - Biological Process

UniProtKB Homo sapiens

Q9UKM9 (http://www.uniprot.org/uniprot/Q9UKM9)

GenebankID or UniProtKB

()

GO:0042632 : cholesterol homeostasis
(https://www.ebi.ac.uk/QuickGO/term/GO:0042632)
GO:0000398 : mRNA splicing, via spliceosome
(https://www.ebi.ac.uk/QuickGO/term/GO:0000398)
GO:1903506 : regulation of nucleic acid-templated transcription
(https://www.ebi.ac.uk/QuickGO/term/GO:1903506)

GO - Cellular Component

GO:0005634 : nucleus (https://www.ebi.ac.uk/QuickGO/term/GO:0005634)
GO:0071013 : catalytic step 2 spliceosome
(https://www.ebi.ac.uk/QuickGO/term/GO:0071013)

Presumptive Null

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

Molecular Type

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

Aberration Type

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

Insertion Size

10-99 bp

Molecular Details of the Mutation

16 bpduplication g.1875_1890dupCCCCAGGTCAGAGTTT in intron

Experimental Evidence

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

Main Reference

Identification of a mutation that is associated with the saddle tan and black-and-tan phenotypes in Basset Hounds and Pembroke Welsh Corgis. (2013 May-Jun)
(https://pubmed.ncbi.nlm.nih.gov/23519866)

Authors

Dreger DL; Parker HG; Ostrander EA; Schmutz SM

Abstract

The causative mutation for the black-and-tan (a (t)) phenotype in dogs was previously shown to be a SINE insertion in the 5′ region of Agouti Signaling Protein (ASIP). Dogs with the black-and-tan phenotype, as well as dogs with the saddle tan phenotype, genotype as a (t) /_ at this locus. We have identified a 16-bp duplication (g.1875_1890dupCCCCAGGTCAGAGTTT) in an intron of hnRNP associated with lethal yellow (RALY), which segregates with the black-and-tan phenotype in a group of 99 saddle tan and black-and-tan Basset Hounds and Pembroke Welsh Corgis. In these breeds, all dogs with the saddle tan phenotype had RALY genotypes of +/+ or +/dup, whereas dogs with the black-and-tan phenotype were homozygous for the duplication. The presence of an a (y) /_ fawn or e/e red genotype is epistatic to the +/_ saddle tan genotype. Genotypes from 10 wolves and 1 coyote indicated that the saddle tan (+) allele is the ancestral allele, suggesting that black-and-tan is a modification of saddle tan. An additional 95 dogs from breeds that never have the saddle tan phenotype have all three of the possible RALY genotypes. We suggest that a multi-gene interaction involving ASIP, RALY, MC1R, DEFB103, and a yet-unidentified modifier gene is required for expression of saddle tan.

Additional References

RELATED GEPHE

Related Genes

13 (Agouti (ASIP), GPR22, MFSD12, PMEL17, SLC45A2=MATP, FGF3; FGF4; FGF19; ORAOV1, Kit, MC1R, Melanophilin (MLPH), Microphtalmia-associated transcription factor, PSMB7, tyrosinase-related protein 1 (TYRP1), beta-defensin 103 (CBD103)) (https://www.gephebase.org/search-criteria?/or+Taxon ID=^9612^/and+Trait=Coloration/and+groupHaplotypes=true#gephebase-summary-title)

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS

@Epistasis - recessive ; https://omia.org/OMIA001806/9615/