

GEPHE SUMMARY

Gephebase Gene
RALY (hnRNP associated with lethal yellow)

Entry Status
Published

GepheID
GP00001366

Main curator
Prigent

PHENOTYPIC CHANGE

Trait Category
Morphology

Trait
Coloration (coat)

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

Taxon A

Latin Name
Canis lupus

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)

NCBI Taxonomy ID
9612

is Taxon A an Intraspecies?
Yes

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

Taxon B

Latin Name
Canis lupus

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)

NCBI Taxonomy ID
9612

is Taxon B an Intraspecies?
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

Sequence Similarities
Belongs to the RRM HNRPC family. RALY subfamily.

GO - Molecular Function
GO:0003723 : RNA binding
GO:0003712 : transcription coregulator activity

GO - Biological Process
GO:0042632 : cholesterol homeostasis
GO:0000398 : mRNA splicing, via spliceosome
GO:1903506 : regulation of nucleic acid-templated transcription

GO - Cellular Component

UniProtKB Homo sapiens
Q9UKM9

GenebankID or UniProtKB

GO:0005634 : nucleus
GO:0071013 : catalytic step 2 spliceosome

Presumptive Null

No

Molecular Type

Cis-regulatory

Aberration Type

Insertion

Insertion Size

10-99 bp

Molecular Details of the Mutation

16 bpduplication g.1875_1890dupCCCCAGGTCAGAGTTT in intron

Experimental Evidence

Association Mapping

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)

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

12 (Agouti (ASIP), MFSD12, PMEL17, SLC45A2-MATP, FGF3; FGF4; FGF19; ORAOV1, Kit, MC1R, Melanophilin (MLPH), Microphthalmia-associated transcription factor, PSMB7, tyrosinase-related protein 1 (TYRP1), beta-defensin 103 (CBD103))

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS

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