

GEPHE SUMMARY

<p>RALY (hnRNP associated with lethal yellow) (https://www.gephebase.org/search-criteria?/and+Gene=Gephebase+RALY+(hnRNP+associated+with+lethal+yellow)+#gephebase-summary-title)</p> <p>Published</p>	<p>Gephebase Gene</p> <p>GP00001366</p> <p>Prigent</p> <p>Entry Status</p>	<p>GepheID</p> <p>Main curator</p>
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PHENOTYPIC CHANGE

<p>Morphology (#gephebase-summary-title)</p> <p>Coloration (coat) (https://www.gephebase.org/search-criteria?/and+Trait+Coloration+(coat)+#gephebase-summary-title)</p> <p>Dog (Basset Hounds and Pembroke Welsh Corgis) and wolf and coyotte ; saddle tan</p> <p>Basset hounds and Pembroke Welsh Corgis ; Black and tan</p> <p>Taxon A</p> <p>Intraspecific (#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>Taxon B</p>
<p>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>is Taxon A an Intraspecies?</p> <p>Yes</p> <p>Dog (Basset Hounds and Pembroke Welsh Corgis) and wolf and coyotte ; saddle tan</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>Taxon A Description</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> <p>Taxon B Description</p>
		<p>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>is Taxon B an Intraspecies?</p> <p>Yes</p> <p>Basset hounds and Pembroke Welsh Corgis ; Black and tan</p>

GENOTYPIC CHANGE

<p>RALY</p> <p>P542; HNRPCL2</p> <p>9606.ENSP00000246194 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSP00000246194)</p> <p>Belongs to the RRM HNRPC family. RALY subfamily.</p> <p>GO:0003723 : RNA binding (https://www.ebi.ac.uk/QuickGO/term/GO:0003723)</p> <p>GO:0003712 : transcription coregulator activity (https://www.ebi.ac.uk/QuickGO/term/GO:0003712)</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>UniProtKB Homo sapiens</p> <p>GenebankID or UniProtKB</p>
		<p>Q9UKM9 (http://www.uniprot.org/uniprot/Q9UKM9)</p> <p>0</p>

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

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)) (<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/>