

## GEPHE SUMMARY

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

## PHENOTYPIC CHANGE

	Trait Category		
Morphology ( <a href="https://www.gephebase.org/search-criteria/?and+Trait Category=^Morphology">#gephebase-summary-title)</a>	Trait		
Coloration (coat) ( <a href="https://www.gephebase.org/search-criteria/?and+Trait=^Coloration (coat)">#gephebase-summary-title)</a>	Trait State in Taxon A		
Dog (Basset Hounds and Pembroke Welsh Corgis) and wolf and coyote ; saddle tan	Trait State in Taxon B		
Basset hounds and Pembroke Welsh Corgis ; Black and tan	Ancestral State		
Taxon A	Taxonomic Status		
Intraspecific ( <a href="https://www.gephebase.org/search-criteria/?and+Taxonomic Status=^Intraspecific">#gephebase-summary-title)</a>			
Taxon A	Latin Name	Taxon B	Latin Name
Canis lupus ( <a href="https://www.gephebase.org/search-criteria/?and+Taxon and Synonyms=^Canis lupus">#gephebase-summary-title)</a>	Canis lupus ( <a href="https://www.gephebase.org/search-criteria/?and+Taxon and Synonyms=^Canis lupus">#gephebase-summary-title)</a>	Canis lupus ( <a href="https://www.gephebase.org/search-criteria/?and+Taxon and Synonyms=^Canis lupus">#gephebase-summary-title)</a>	Canis lupus ( <a href="https://www.gephebase.org/search-criteria/?and+Taxon and Synonyms=^Canis lupus">#gephebase-summary-title)</a>
gray wolf	Common Name	gray wolf	Common Name
gray wolf; grey wolf; Canis lupus Linnaeus, 1758	Synonyms	gray wolf; grey wolf; Canis lupus Linnaeus, 1758	Synonyms
species	Rank	species	Rank
	Lineage		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		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 () - (Rank: genus) ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9611">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9611</a> )	Parent	Canis () - (Rank: genus) ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9611">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9611</a> )	Parent
9612 ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9612">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9612</a> )	NCBI Taxonomy ID	9612 ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9612">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9612</a> )	NCBI Taxonomy ID
Yes	is Taxon A an Infraspecies?	Yes	is Taxon B an Infraspecies?
	Taxon A Description		Taxon B Description
Dog (Basset Hounds and Pembroke Welsh Corgis) and wolf and coyote ; saddle tan		Basset hounds and Pembroke Welsh Corgis ; Black and tan	

## GENOTYPIC CHANGE

	Generic Gene Name	UniProtKB Homo sapiens
RALY		
P542; HNRPCL2	Synonyms	GenebankID or UniProtKB
9606.ENSP00000246194 ( <a href="http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9606.ENSP00000246194">http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9606.ENSP00000246194</a> )	String	0
	Sequence Similarities	
Belongs to the RRM HNRP family. RALY subfamily.		
	GO - Molecular Function	
GO:0003723 : RNA binding ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0003723">https://www.ebi.ac.uk/QuickGO/term/GO:0003723</a> )		
GO:0003712 : transcription coregulator activity ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0003712">https://www.ebi.ac.uk/QuickGO/term/GO:0003712</a> )		
	GO - Biological Process	

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\_1890dupCCCCAGGTCAAGAGTTT 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\_1890dupCCCCAGGTCAAGAGTTT) 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), 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/>