GEPHE SUMMARY

Gephebase Gene
SLC45A2=MATP

GephelD
GP00002297

Entry Status
Published

Main curator
Martin

PHENOTYPIC CHANGE

Trait Category
Morphology

Trait
Coloration (coat; albinism)

Trait State in Taxon A
WT melanin content

Trait State in Taxon B
Oculocutaneous albinism

Ancestral State
Taxon A

Taxonomic Status
Domesticated

Taxon A

Latin Name
Canis lupus familiaris

Common Name
dog

Synonyms
Canis canis; Canis domesticus; Canis familiaris; dog; dogs; Canis familiaris Linnaeus, 1758; Canis lupus familiaris Linnaeus, 1758

Rank
subspecies

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; Canis lupus

Parent
Canis lupus (gray wolf) - (Rank: species)

NCBI Taxonomy ID
9615

is Taxon A an Infraspecies?
No

Taxon B

Latin Name
Canis lupus familiaris

Common Name
dog

Synonyms
Canis canis; Canis domesticus; Canis familiaris; dog; dogs; Canis familiaris Linnaeus, 1758; Canis lupus familiaris Linnaeus, 1758

Rank
subspecies

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; Canis lupus

Parent
Canis lupus (gray wolf) - (Rank: species)

NCBI Taxonomy ID
9615

is Taxon B an Infraspecies?
Yes

Taxon B Description
Lhasa Apso ;Mixed breed ;Pekingese ;Pomeranian

GENOTYPIC CHANGE

Generic Gene Name
SLC45A2

Synonyms
1A1; AIM1; MATP; OCA4; SHEP5

String
9606.ENSP00000296589

Sequence Similarities
Belongs to the glycoside-pentoside-hexuronide (GPH) cation symporter transporter (TC 2.A.2) family.

GO - Molecular Function
GO:0008506 : sucrose:proton symporter activity

GO - Biological Process
GO:0042438 : melanin biosynthetic process
GO:0048066 : developmental pigmentation
GO:0007601 : visual perception
GO:0050896 : response to stimulus

UniProtKB Homo sapiens
Q9UMX9

GenebankID or UniProtKB

GO:0015770 : sucrose transport

GO - Cellular Component
GO:0016021 : integral component of membrane
GO:0033162 : melanosome membrane

Presumptive Null
Yes

Molecular Type
Coding

Aberration Type
SNP

SNP Coding Change
Nonsynonymous

Molecular Details of the Mutation
c.1478G>A p.G493D

Experimental Evidence
Candidate Gene

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	Gly	Asp	493

Main Reference
A Missense Mutation in SLC45A2 Is Associated with Albinism in Several Small Long Haired Dog Breeds. (2015 May-Jun)

Authors
Wijesena HR; Schmutz SM

Abstract
Homozygosity for a large deletion in the solute carrier family 45, member 2 (SLC45A2) gene causes oculocutaneous albinism (OCA) in the Doberman Pinscher breed. An albino Lhasa Apso did not have this g.27141_31223del (CanFam2) deletion in her SLC45A2 sequence. Therefore, SLC45A2 was investigated in this female Lhasa Apso to search for other possible variants that caused her albinism. The albino Lhasa Apso was homozygous for a nonsynonymous substitution in the seventh exon, a c.1478G>A base change that resulted in a glycine to aspartic acid substitution (p.G493D). This mutation was not found in a wolf, a coyote, or any of the 15 other Lhasa Apso dogs or 32 other dogs of breeds related to the Lhasa Apso. However, an albino Pekingese, 2 albino Pomeranians, and an albino mixed breed dog that was small and long haired were also homozygous for the 493D allele. The colored puppies of the albino Lhasa Apso and the colored dam of the 2 albino Pomeranians were heterozygous for this allele. However, an albino Pug was homozygous for the 493G allele and therefore although we suggest the 493D allele causes albinism when homozygous in several small, long haired dog breeds, it does not explain all albinism in dogs. A variant effect prediction for the albino Lhasa Apso confirms that p.G493D is a deleterious substitution, and a topology prediction for SLC45A2 suggests that the 11th transmembrane domain where the 493rd amino acid was located, has an altered structure.

Additional References

RELATED GEPHE

Related Genes
11 (Agouti (ASIP), MFSD12, PMEL17, FGF3; FGF4; FGF19; ORAOV1, Kit, MC1R, Melanophilin (MLPH), Microphtalmia-associated transcription factor, PSMB7, tyrosinase-related protein 1 (TYRP1), beta-defensin 103 (CBD103))
Related Haplotypes
2

EXTERNAL LINKS

COMMENTS

@AllelicSeries @Parallelism https://omia.org/OMIA001821/9615/