

## GEPHE SUMMARY

SLC45A2=MATP ( <a href="https://www.gephebase.org/search-criteria?/and+Gene">https://www.gephebase.org/search-criteria?/and+Gene</a> Gephebase= <sup>^</sup> SLC45A2=MATP <sup>^</sup> #gephebase-summary-title)	Gephebase Gene	GP00002297	GepheID
Published	Entry Status	Martin	Main curator

## PHENOTYPIC CHANGE

Morphology ( <a href="https://www.gephebase.org/search-criteria?/and+Trait">https://www.gephebase.org/search-criteria?/and+Trait</a> Category= <sup>^</sup> Morphology <sup>^</sup> #gephebase-summary-title)	Trait Category		
Coloration (coat; albinism) ( <a href="https://www.gephebase.org/search-criteria?/and+Trait=&lt;sup&gt;^&lt;/sup&gt;Coloration (coat; albinism)&lt;sup&gt;^&lt;/sup&gt;#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Trait=<sup>^</sup>Coloration (coat; albinism)<sup>^</sup>#gephebase-summary-title</a> )	Trait		
WT melanin content	Trait State in Taxon A		
Oculocutaneous albinism	Trait State in Taxon B		
Taxon A	Ancestral State		
Domesticated ( <a href="https://www.gephebase.org/search-criteria?/and+Taxonomic">https://www.gephebase.org/search-criteria?/and+Taxonomic</a> Status= <sup>^</sup> Domesticated <sup>^</sup> #gephebase-summary-title)	Taxonomic Status		
	Taxon A	Taxon B	
Canis lupus familiaris ( <a href="https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=&lt;sup&gt;^&lt;/sup&gt;Canis lupus familiaris&lt;sup&gt;^&lt;/sup&gt;#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=<sup>^</sup>Canis lupus familiaris<sup>^</sup>#gephebase-summary-title</a> )	Latin Name	Canis lupus familiaris ( <a href="https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=&lt;sup&gt;^&lt;/sup&gt;Canis lupus familiaris&lt;sup&gt;^&lt;/sup&gt;#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=<sup>^</sup>Canis lupus familiaris<sup>^</sup>#gephebase-summary-title</a> )	Latin Name
dog	Common Name	dog	Common Name
Canis canis; Canis domesticus; Canis familiaris; dog; dogs; Canis familiaris Linnaeus, 1758; Canis lupus familiaris Linnaeus, 1758	Synonyms	Canis canis; Canis domesticus; Canis familiaris; dog; dogs; Canis familiaris Linnaeus, 1758; Canis lupus familiaris Linnaeus, 1758	Synonyms
subspecies	Rank	subspecies	Rank
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	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	Lineage
Canis lupus (gray wolf) - (Rank: species) ( <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> )	Parent	Canis lupus (gray wolf) - (Rank: species) ( <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> )	Parent
9615 ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9615">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9615</a> )	NCBI Taxonomy ID	9615 ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9615">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9615</a> )	NCBI Taxonomy ID
No	is Taxon A an Intraspecies?	Yes	is Taxon B an Intraspecies?
		Lhasa Apso ;Mixed breed ;Pekingese ;Pomeranian	Taxon B Description

## GENOTYPIC CHANGE

SLC45A2	Generic Gene Name	Q9UMX9 ( <a href="http://www.uniprot.org/uniprot/Q9UMX9">http://www.uniprot.org/uniprot/Q9UMX9</a> )	UniProtKB Homo sapiens
1A1; AIM1; MATP; OCA4; SHEP5	Synonyms	0	GenebankID or UniProtKB
9606.ENSP00000296589 ( <a href="http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9606.ENSP00000296589">http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9606.ENSP00000296589</a> )	String		
Belongs to the glycoside-pentoside-hexuronide (GPH) cation symporter transporter (TC 2.A.2) family.	Sequence Similarities		
GO:0008506 : sucrose:proton symporter activity ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0008506">https://www.ebi.ac.uk/QuickGO/term/GO:0008506</a> )	GO - Molecular Function		
	GO - Biological Process		

GO:0042438 : melanin biosynthetic process  
 (https://www.ebi.ac.uk/QuickGO/term/GO:0042438)  
 GO:0048066 : developmental pigmentation  
 (https://www.ebi.ac.uk/QuickGO/term/GO:0048066)  
 GO:0007601 : visual perception (https://www.ebi.ac.uk/QuickGO/term/GO:0007601)  
 GO:0050896 : response to stimulus (https://www.ebi.ac.uk/QuickGO/term/GO:0050896)  
 GO:0015770 : sucrose transport (https://www.ebi.ac.uk/QuickGO/term/GO:0015770)  
 GO - Cellular Component

GO:0016021 : integral component of membrane  
 (https://www.ebi.ac.uk/QuickGO/term/GO:0016021)  
 GO:0033162 : melanosome membrane  
 (https://www.ebi.ac.uk/QuickGO/term/GO:0033162)

Yes (https://www.gephebase.org/search-criteria?/and+Presumptive Null=^Yes^#gephebase-summary-title)

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

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

Nonsynonymous

c.1478G>A p.G493D

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

Presumptive Null

Molecular Type

Aberration Type

SNP Coding Change

Molecular Details of the Mutation

Experimental Evidence

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

A Missense Mutation in SLC45A2 Is Associated with Albinism in Several Small Long Haired Dog Breeds. (2015 May-Jun) (https://pubmed.ncbi.nlm.nih.gov/25790827)

Wijesena HR; Schmutz SM

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.

© The American Genetic Association 2015. All rights reserved. For permissions, please e-mail: journals.permissions@oup.com.

Additional References

RELATED GEPHE

12 (Agouti (ASIP), GPR22, MFSD12, PMEL17, 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=^9615^/and+Trait=Coloration/and+groupHaplotypes=true#gephebase-summary-title)

2 (https://www.gephebase.org/search-criteria?/or+Gene Gephebase=^SLC45A2=MATP^/and+Taxon ID=^9615^/or+Gene Gephebase=^SLC45A2=MATP^/and+Taxon ID=^9615^#gephebase-summary-title)

EXTERNAL LINKS

COMMENTS

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

