

GEPHE SUMMARY

SLC45A2=MATP (https://www.gephebase.org/search-criteria?/and+Gene Gephebase= [^] SLC45A2=MATP [^] #gephebase-summary-title)	Gephebase Gene	GP00002298	GepheID
Published	Entry Status	Martin	Main curator

PHENOTYPIC CHANGE

Morphology (https://www.gephebase.org/search-criteria?/and+Trait Category= [^] Morphology [^] #gephebase-summary-title)	Trait Category		
Coloration (coat; albinism) (<a href="https://www.gephebase.org/search-criteria?/and+Trait=<sup>^</sup>Coloration (coat; albinism)<sup>^</sup>#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Trait=[^]Coloration (coat; albinism)[^]#gephebase-summary-title)	Trait		
WT melanin content	Trait State in Taxon A		
Oculocutaneous albinism	Trait State in Taxon B		
Taxon A	Ancestral State		
Domesticated (https://www.gephebase.org/search-criteria?/and+Taxonomic Status= [^] Domesticated [^] #gephebase-summary-title)	Taxonomic Status		
	Taxon A	Taxon B	
Canis lupus familiaris (<a href="https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=<sup>^</sup>Canis lupus familiaris<sup>^</sup>#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=[^]Canis lupus familiaris[^]#gephebase-summary-title)	Latin Name	Canis lupus familiaris (<a href="https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=<sup>^</sup>Canis lupus familiaris<sup>^</sup>#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=[^]Canis lupus familiaris[^]#gephebase-summary-title)	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) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9612)	Parent	Canis lupus (gray wolf) - (Rank: species) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9612)	Parent
9615 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9615)	NCBI Taxonomy ID	9615 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9615)	NCBI Taxonomy ID
No	is Taxon A an Intraspecies?	Yes	is Taxon B an Intraspecies?
		Bull Mastiff	Taxon B Description

GENOTYPIC CHANGE

SLC45A2	Generic Gene Name	Q9UMX9 (http://www.uniprot.org/uniprot/Q9UMX9)	UniProtKB Homo sapiens
1A1; AIM1; MATP; OCA4; SHEP5	Synonyms	0	GenebankID or UniProtKB
9606.ENSP00000296589 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9606.ENSP00000296589)	String		
Belongs to the glycoside-pentoside-hexuronide (GPH) cation symporter transporter (TC 2.A.2) family.	Sequence Similarities		
GO:0008506 : sucrose:proton symporter activity (https://www.ebi.ac.uk/QuickGO/term/GO:0008506)	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](https://www.gephebase.org/search-criteria?/and+Presumptive+Null=^Yes^#gephebase-summary-title))

Presumptive Null

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

Molecular Type

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

Aberration Type

1-9 bp

Deletion Size

c.1287delC p.Met430CysfsTer4

Molecular Details of the Mutation

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

Experimental Evidence

A single base deletion in the SLC45A2 gene in a Bullmastiff with oculocutaneous albinism. (2017) (<https://pubmed.ncbi.nlm.nih.gov/28737247>)

Main Reference

Caduff M; Bauer A; Jagannathan V; Leeb T

Authors

Oculocutaneous albinism type 4 (OCA4) in humans and similar phenotypes in many animal species are caused by variants in the SLC45A2 gene, encoding a putative sugar transporter. In dog, two independent SLC45A2 variants are known that cause oculocutaneous albinism in Doberman Pinschers and several small dog breeds respectively. For the present study, we investigated a Bullmastiff with oculocutaneous albinism. The affected dog was highly inbred and resulted from the mating of a sire to its own grandmother. We obtained whole genome sequence data from the affected dog and searched specifically for variants in candidate genes known to cause albinism. We detected a single base deletion in exon 6 of the SLC45A2 gene (NM_001037947.1:c.1287delC) that has not been reported thus far. This deletion is predicted to result in an early premature stop codon. It was confirmed by Sanger sequencing and perfectly co-segregated with the phenotype in the available family members. We genotyped 174 unrelated dogs from diverse breeds, all of which were homozygous wildtype. We therefore suggest that SLC45A2:c.1287delC causes the observed oculocutaneous albinism in the affected Bullmastiff.

Abstract

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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>)

Related Genes

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](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))

Related Haplotypes

EXTERNAL LINKS

COMMENTS

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