

GEPHE SUMMARY

SLC45A2=MATP (<a +slc45a2='MATP"#gephebase-summary-title"' href="https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=">https://www.gephebase.org/search-criteria?/and+Gene+Gephebase="+SLC45A2=MATP"#gephebase-summary-title)	Gephebase Gene	GP00002296	GepheID
Published	Entry Status	Martin	Main curator

PHENOTYPIC CHANGE

Morphology (<a +morphology"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Trait+Category=">https://www.gephebase.org/search-criteria?/and+Trait+Category="+Morphology"#gephebase-summary-title)	Trait Category		
Coloration (coat; albinism) (<a +coloration+(coat;+albinism)"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Trait=">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 (<a +domesticated"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=">https://www.gephebase.org/search-criteria?/and+Taxonomic+Status="+Domesticated"#gephebase-summary-title)	Taxonomic Status		
	Taxon A		Taxon B
Canis lupus familiaris (<a +canis+lupus+familiaris"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms="+Canis+lupus+familiaris"#gephebase-summary-title)	Latin Name	Canis lupus familiaris (<a +canis+lupus+familiaris"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=">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?
		Doberman Pinscher	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) Presumptive Null

Coding (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) Aberration Type

10-100 kb Deletion Size

4081 base pair deletion resulting in loss of the terminus of exon seven of SLC45A2 Molecular Details of the Mutation

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

A partial gene deletion of SLC45A2 causes oculocutaneous albinism in Doberman pinscher dogs. (2014) (https://pubmed.ncbi.nlm.nih.gov/24647637) Main Reference

Winkler PA; Gornik KR; Ramsey DT; Dubielzig RR; Venta PJ; Petersen-Jones SM; Bartoe JT Authors

The first white Doberman pinscher (WDP) dog was registered by the American Kennel Club in 1976. The novelty of the white coat color resulted in extensive line breeding of this dog and her offspring. The WDP phenotype closely resembles human oculocutaneous albinism (OCA) and clinicians noticed a seemingly high prevalence of pigmented masses on these dogs. This study had three specific aims: (1) produce a detailed description of the ocular phenotype of WDPs, (2) objectively determine if an increased prevalence of ocular and cutaneous melanocytic tumors was present in WDPs, and (3) determine if a genetic mutation in any of the genes known to cause human OCA is causal for the WDP phenotype. WDPs have a consistent ocular phenotype of photophobia, hypopigmented adnexal structures, blue irides with a tan periphery and hypopigmented retinal pigment epithelium and choroid. WDPs have a higher prevalence of cutaneous melanocytic neoplasms compared with control standard color Doberman pinschers (SDPs); cutaneous tumors were noted in 12/20 WDP (<5 years of age: 4/12; >5 years of age: 8/8) and 1/20 SDPs (p<0.00001). Using exclusion analysis, four OCA causative genes were investigated for their association with WDP phenotype: TYR, OCA2, TYRP1 and SLC45A2. SLC45A2 was found to be linked to the phenotype and gene sequencing revealed a 4,081 base pair deletion resulting in loss of the terminus of exon seven of SLC45A2 (chr4^177,062,968-77,067,051). This mutation is highly likely to be the cause of the WDP phenotype and is supported by a lack of detectable SLC45A2 transcript levels by reverse transcriptase PCR. The WDP provides a valuable model for studying OCA4 visual disturbances and melanocytic neoplasms in a large animal model. Abstract

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) Related Haplotypes

EXTERNAL LINKS

COMMENTS

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