

GEPHE SUMMARY

	Gephebase Gene	GephelD
SLC45A2=MATP (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^SLC45A2=MATP^#gephebase-summary-title)	GP00002296	
	Entry Status	Main curator
Published	Martin	

PHENOTYPIC CHANGE

	Trait Category	
Morphology (https://www.gephebase.org/search-criteria?/and+Trait Category=^Morphology^#gephebase-summary-title)	Trait	
Coloration (coat; albinism) (https://www.gephebase.org/search-criteria?/and+Trait=^Coloration+(coat;+albinism)^#gephebase-summary-title)	Trait State in Taxon A	
WT melanin content	Trait State in Taxon B	
Oculocutaneous albinism	Ancestral State	
Taxon A	Taxonomic Status	
Domesticated (https://www.gephebase.org/search-criteria?/and+Taxonomic Status=^Domesticated^#gephebase-summary-title)		
Taxon A	Latin Name	Latin Name
Canis lupus familiaris (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Canis+lupus+familiaris^#gephebase-summary-title)	Canis lupus familiaris (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Canis+lupus+familiaris^#gephebase-summary-title)	
dog	Common Name	Common Name
Canis canis; Canis domesticus; Canis familiaris; dog; dogs; Canis familiaris Linnaeus, 1758; Canis lupus familiaris Linnaeus, 1758	Synonyms	Synonyms
	Rank	Rank
subspecies	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; Canis lupus		
Canis lupus (gray wolf) - (Rank: species) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9612)	Parent	Parent
9615 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9615)	NCBI Taxonomy ID	NCBI Taxonomy ID
No	is Taxon A an Infraspecies?	is Taxon B an Infraspecies?
	Yes	Yes
	Doberman Pinscher	Taxon B Description

GENOTYPIC CHANGE

	Generic Gene Name	UniProtKB Homo sapiens
SLC45A2		
1A1; AIM1; MATP; OCA4; SHEP5	Synonyms	GenebankID or UniProtKB
9606.ENSP00000296589 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9606.ENSP00000296589)	String	
	Sequence Similarities	
Belongs to the glycoside-pentoside-hexuronide (GPH) cation symporter transporter (TC 2.A.2) family.		
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)	
	Presumptive Null
Yes (https://www.gephebase.org/search-criteria?/and+Presumptive Null=^Yes^#gephebase-summary-title)	
	Molecular Type
Coding (https://www.gephebase.org/search-criteria?/and+Molecular Type=^Coding^#gephebase-summary-title)	
	Aberration Type
Deletion (https://www.gephebase.org/search-criteria?/and+Aberration Type=^Deletion^#gephebase-summary-title)	
	Deletion Size
10-100 kb	
	Molecular Details of the Mutation
4081 base pair deletion resulting in loss of the terminus of exon seven of SLC45A2	
	Experimental Evidence
Candidate Gene (https://www.gephebase.org/search-criteria?/and+Experimental Evidence=^Candidate Gene^#gephebase-summary-title)	
	Main Reference
A partial gene deletion of SLC45A2 causes oculocutaneous albinism in Doberman pinscher dogs. (2014) (https://pubmed.ncbi.nlm.nih.gov/24647637)	
	Authors
Winkler PA; Gornik KR; Ramsey DT; Dubielzig RR; Venta PJ; Petersen-Jones SM; Bartoe JT	
	Abstract
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.	
	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+Gene Gephebase=^SLC45A2=MATP^/and+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/>