GEPHE SUMMARY

Gephebase Gene
SLC45A2=MATP

GephelD
GP00002296

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
Doberman Pinscher

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
Deletion

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

Main Reference
A partial gene deletion of SLC45A2 causes oculocutaneous albinism in Doberman pinscher dogs. (2014)

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 (chr4â^¶77,062,968-77,067,051). 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

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/