GEPHE SUMMARY

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

GepheID
GP00001060

Entry Status
Published

Main curator
Martin

PHENOTYPIC CHANGE

Trait Category
Morphology (https://www.gephebase.org/search-criteria?/and+Trait Category=^Morphology^#gephebase-summary-title)

Trait
Coloration (eyes; fur) (https://www.gephebase.org/search-criteria?/and+Trait=^Coloration (eyes; fur)^#gephebase-summary-title)

Trait State in Taxon A
Mus musculus

Trait State in Taxon B
Mus musculus

Ancestral State
Data not curated

Taxonomic Status
Domesticated (https://www.gephebase.org/search-criteria?/and+Taxonomic Status=^Domesticated^#gephebase-summary-title)

Taxon A

Latin Name
Mus musculus (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Mus musculus^#gephebase-summary-title)

Common Name
house mouse

Synonyms
house mouse; mouse; Mus musculus Linnaeus, 1758; mice C57BL/6xCBA/CaJ hybrid

Rank
species

Lineage
cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria; Euarchontoglires; Glires; Rodentia; Myomorpha; Muroidea; Muridae; Murinae; Mus; Mus

Parent
Mus () - (Rank: subgenus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 862507)

NCBI Taxonomy ID
10090 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 10090)

is Taxon A an Infraspecies?
No

Taxon B

Latin Name
Mus musculus (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Mus musculus^#gephebase-summary-title)

Common Name
house mouse

Synonyms
house mouse; mouse; Mus musculus Linnaeus, 1758; mice C57BL/6xCBA/CaJ hybrid

Rank
species

Lineage
cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria; Euarchontoglires; Glires; Rodentia; Myomorpha; Muroidea; Muridae; Murinae; Mus; Mus

Parent
Mus () - (Rank: subgenus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 862507)

NCBI Taxonomy ID
10090 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 10090)

is Taxon B an Infraspecies?
No

GENOTYPIC CHANGE

Generic Gene Name
SLC45A2

Synonyms
1A1; AIM1; MATP; OCA4; SHEP5

String
9606.ENSP00000296589 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=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 (https://www.ebi.ac.uk/QuickGO/term/GO:0008506)

GO - Biological Process
GO:0042438 : melanin biosynthetic process (https://www.ebi.ac.uk/QuickGO/term/GO:0042438)
GO:0048066 : developmental pigmentation

UniProtKB Homo sapiens
Q9UMX9 (http://www.uniprot.org/uniprot/Q9UMX9)

GenebankID or UniProtKB
()

(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

No (https://www.gephebase.org/search-criteria?/and+Presumptive Null=^No^#gephebase-summary-title)

Molecular Type

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

Aberration Type

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

SNP Coding Change

Nonsynonymous

Molecular Details of the Mutation

N153D

Experimental Evidence

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

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	-	-	-

Main Reference

Identification of Aim-1 as the underwhite mouse mutant and its transcriptional regulation by MITF. (2002) (https://pubmed.ncbi.nlm.nih.gov/11700328)

Authors

Du J; Fisher DE

Abstract

Animal pigmentation mutants have provided rich models for the identification of genes modulating pathways from melanocyte development to melanoma. One mouse model is the underwhite locus, alleles of which manifest altered pigmentation of both eye and fur, sometimes in an age-dependent fashion. Here we show that the mouse homolog of a recently identified gene whose mutation produces Japanese gold-colored fish, medaka b, maps to the mouse underwhite locus. We identify distinct mutations of this gene, known as Aim-1, in three underwhite mouse alleles and find that structure/function differences correlate with recessive versus dominant inheritance. The human ortholog of AIM-1 was originally identified as a melanocyte-restricted antigen that is recognized by autologous T cells from a patient with melanoma. We also provide evidence that AIM-1 is transcriptionally modulated by MITF, a melanocyte-specific transcription factor essential to pigmentation and a clinical diagnostic marker in human melanoma. Although AIM-1 appears to reside downstream of MITF, chromatin immunoprecipitations do not reveal binding of MITF to a 5′-flanking region containing histone 3 acetylation, indicating that MITF either acts indirectly on AIM-1 or it binds to a remote regulatory sequence. Nevertheless, MITF links AIM-1 expression and the underwhite phenotype to a transcriptional network central to pigmentation in mammals.

Additional References

RELATED GEPHE

Related Genes

4 (Agouti, Agouti (ASIP), MC1R, PMEL17) (https://www.gephebase.org/search-criteria?/or+Taxon ID=^10090^/and+Trait=Coloration/and+groupHaplotypes=true#gephebase-summary-title)

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS