

GEPHE SUMMARY

SLC45A2=MATP (https://www.gephebase.org/search-criteria?/and+Gene Gephebase= [^] SLC45A2=MATP [^] #gephebase-summary-title)	Gephebase Gene	GP00001060	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 (eyes; fur) (https://www.gephebase.org/search-criteria?/and+Trait = [^] Coloration (eyes; fur) [^] #gephebase-summary-title)	Trait
Mus musculus	Trait State in Taxon A
Mus musculus	Trait State in Taxon B
Data not curated	Ancestral State
Domesticated (https://www.gephebase.org/search-criteria?/and+Taxonomic Status= [^] Domesticated [^] #gephebase-summary-title)	Taxonomic Status

Taxon A	Latin Name	Taxon B	Latin Name
Mus musculus (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms= [^] Mus musculus [^] #gephebase-summary-title)		Mus musculus (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms= [^] Mus musculus [^] #gephebase-summary-title)	
house mouse	Common Name	house mouse	Common Name
house mouse; mouse; Mus musculus Linnaeus, 1758; mice C57BL/6xCBA/CaJ hybrid	Synonyms	house mouse; mouse; Mus musculus Linnaeus, 1758; mice C57BL/6xCBA/CaJ hybrid	Synonyms
species	Rank	species	Rank
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	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	Lineage
Mus () - (Rank: subgenus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=862507)	Parent	Mus () - (Rank: subgenus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=862507)	Parent
10090 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=10090)	NCBI Taxonomy ID	10090 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=10090)	NCBI Taxonomy ID
No	is Taxon A an Intraspecies?	No	is Taxon B an Intraspecies?

GENOTYPIC CHANGE

SLC45A2	Generic Gene Name	Q9UMX9 (http://www.uniprot.org/uniprot/Q9UMX9)	UniProtKB Homo sapiens
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		
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:0042438 : melanin biosynthetic process (https://www.ebi.ac.uk/QuickGO/term/GO:0042438)	GO - Biological Process		
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

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