

GEPHE SUMMARY

	Gephebase Gene	GephelD
SLC45A2=MATP (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^SLC45A2=MATP^#gephebase-summary-title)	GP00001060	
Published	Entry Status	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		Taxon B
Mus musculus	Latin Name	Latin Name
(https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Mus musculus^#gephebase-summary-title)		
house mouse	Common Name	Common Name
house mouse; mouse; Mus musculus Linnaeus, 1758; mice C57BL/6xCBA/CaJ hybrid	Synonyms	Synonyms
species	Rank	Rank
	Lineage	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		
Mus () - (Rank: subgenus)	Parent	Parent
(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 862507)		
10090	NCBI Taxonomy ID	NCBI Taxonomy ID
(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 10090)		
No	is Taxon A an Infraspecies?	is Taxon B an Infraspecies?
	No	

GENOTYPIC CHANGE

	Generic Gene Name		
SLC45A2			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	0	
	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: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>)

No (https://www.gephebase.org/search-criteria?/and+Presumptive Null=^No^#gephebase-summary-title)	Presumptive Null
Coding (https://www.gephebase.org/search-criteria?/and+Molecular Type=^Coding^#gephebase-summary-title)	Molecular Type
SNP (https://www.gephebase.org/search-criteria?/and+Aberration Type=^SNP^#gephebase-summary-title)	Aberration Type
Nonsynonymous	SNP Coding Change
N153D	Molecular Details of the Mutation
Linkage Mapping (https://www.gephebase.org/search-criteria?/and+Experimental Evidence=^Linkage Mapping^#gephebase-summary-title)	Experimental Evidence

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

Identification of Aim-1 as the underwhite mouse mutant and its transcriptional regulation by MITF. (2002) (https://pubmed.ncbi.nlm.nih.gov/11700328)	Main Reference
Du J; Fisher DE	Authors
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.	Abstract
Additional References	

RELATED GEPHE

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 Genes
No matches found.	Related Haplotypes

EXTERNAL LINKS

COMMENTS