

GEPHE SUMMARY

Gephebase Gene		GepheID
Agouti (ASIP) (<a +agouti+(asip)+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=">https://www.gephebase.org/search-criteria?/and+Gene+Gephebase="+Agouti+(ASIP)+"#gephebase-summary-title)	GP00000070	
Entry Status		Main curator
Published		

PHENOTYPIC CHANGE

Trait Category			
Morphology (<a +morphology+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Trait+Category=">https://www.gephebase.org/search-criteria?/and+Trait+Category="+Morphology+"#gephebase-summary-title)			
Trait			
Coloration (skin; hair) (<a +coloration+(skin;+hair)+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Trait=">https://www.gephebase.org/search-criteria?/and+Trait="+Coloration+(skin;+hair)+"#gephebase-summary-title)			
Trait State in Taxon A			
Homo sapiens			
Trait State in Taxon B			
Homo sapiens			
Ancestral State			
Data not curated			
Taxonomic Status			
Intraspecific (<a +intraspecific+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=">https://www.gephebase.org/search-criteria?/and+Taxonomic+Status="+Intraspecific+"#gephebase-summary-title)			
Taxon A		Taxon B	
Latin Name		Latin Name	
Homo sapiens (<a +homo+sapiens+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms="+Homo+sapiens+"#gephebase-summary-title)		Homo sapiens (<a +homo+sapiens+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms="+Homo+sapiens+"#gephebase-summary-title)	
Common Name		Common Name	
human		human	
Synonyms		Synonyms	
human; man; Homo sapiens Linnaeus, 1758; Home sapiens; Homo sampiens; Homo sapeins; Homo sapian; Homo sapians; Homo sapien; Homo sapience; Homo sapiense; Homo sapients; Homo sapines; Homo spaiens; Homo spiens; Humo sapiens		human; man; Homo sapiens Linnaeus, 1758; Home sapiens; Homo sampiens; Homo sapeins; Homo sapian; Homo sapians; Homo sapien; Homo sapience; Homo sapiense; Homo sapients; Homo sapines; Homo spaiens; Homo spiens; Humo sapiens	
Rank		Rank	
species		species	
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; Primates; Haplorrhini; Simiiformes; Catarrhini; Hominoidea; Hominidae; Homininae; Homo		cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria; Euarchontoglires; Primates; Haplorrhini; Simiiformes; Catarrhini; Hominoidea; Hominidae; Homininae; Homo	
Parent		Parent	
Homo () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9605)		Homo () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9605)	
NCBI Taxonomy ID		NCBI Taxonomy ID	
9606 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9606)		9606 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9606)	
is Taxon A an Intraspecies?		is Taxon B an Intraspecies?	
No		No	

GENOTYPIC CHANGE

Generic Gene Name		UniProtKB Mus musculus
Asip		Q03288 (http://www.uniprot.org/uniprot/Q03288)
Synonyms		GenebankID or UniProtKB
As; ASP; A<y>; ASIP; a		AAB61247 (https://www.ncbi.nlm.nih.gov/nuccore/AAB61247)
String		
10090.ENSMUSP00000029123 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=10090.ENSMUSP00000029123)		
Sequence Similarities		
-		
GO - Molecular Function		
GO:0031779 : melanocortin receptor binding (https://www.ebi.ac.uk/QuickGO/term/GO:0031779)		
GO:0031781 : type 3 melanocortin receptor binding (https://www.ebi.ac.uk/QuickGO/term/GO:0031781)		

GO:0031782 : type 4 melanocortin receptor binding
(<https://www.ebi.ac.uk/QuickGO/term/GO:0031782>)

GO - Biological Process

GO:0008343 : adult feeding behavior
(<https://www.ebi.ac.uk/QuickGO/term/GO:0008343>)
GO:0006091 : generation of precursor metabolites and energy
(<https://www.ebi.ac.uk/QuickGO/term/GO:0006091>)
GO:0071514 : genetic imprinting (<https://www.ebi.ac.uk/QuickGO/term/GO:0071514>)
GO:0009755 : hormone-mediated signaling pathway
(<https://www.ebi.ac.uk/QuickGO/term/GO:0009755>)
GO:0042438 : melanin biosynthetic process
(<https://www.ebi.ac.uk/QuickGO/term/GO:0042438>)
GO:0032438 : melanosome organization
(<https://www.ebi.ac.uk/QuickGO/term/GO:0032438>)
GO:0032402 : melanosome transport
(<https://www.ebi.ac.uk/QuickGO/term/GO:0032402>)
GO:0043473 : pigmentation (<https://www.ebi.ac.uk/QuickGO/term/GO:0043473>)
GO:0048023 : positive regulation of melanin biosynthetic process
(<https://www.ebi.ac.uk/QuickGO/term/GO:0048023>)
GO:0040030 : regulation of molecular function, epigenetic
(<https://www.ebi.ac.uk/QuickGO/term/GO:0040030>)

GO - Cellular Component

GO:0005576 : extracellular region (<https://www.ebi.ac.uk/QuickGO/term/GO:0005576>)
GO:0005623 : cell (<https://www.ebi.ac.uk/QuickGO/term/GO:0005623>)

No (https://www.gephebase.org/search-criteria?/and+Presumptive Null=^No^#gephebase-summary-title)	Presumptive Null
Cis-regulatory (https://www.gephebase.org/search-criteria?/and+Molecular Type=^Cis-regulatory^#gephebase-summary-title)	Molecular Type
Unknown (https://www.gephebase.org/search-criteria?/and+Aberration Type=^Unknown^#gephebase-summary-title)	Aberration Type
unknown	Molecular Details of the Mutation
Association Mapping (https://www.gephebase.org/search-criteria?/and+Experimental Evidence=^Association Mapping^#gephebase-summary-title)	Experimental Evidence
The 8818G allele of the agouti signaling protein (ASIP) gene is ancestral and is associated with darker skin color in African Americans. (2005) (https://pubmed.ncbi.nlm.nih.gov/15726415)	Main Reference
Bonilla C; Boxill LA; Donald SA; Williams T; Sylvester N; Parra EJ; Dios S; Norton HL; Shriver MD; Kittles RA	Authors
Skin color, a predictor of social interactions and risk factor for several types of cancer, is due to two contrasting forms of melanin, the darker eumelanin and lighter pheomelanin. The lighter pigment pheomelanin is the product of the antagonistic function of the agouti signaling protein (ASIP) on the alpha-melanocyte stimulating hormone receptor (MC1R). Studies have shown that a single-nucleotide polymorphism (SNP) in the 3'UTR of the ASIP gene is associated with dark hair and eyes; however, little is known about its role in inter-individual variation in skin color. Here we examine the relationship between the ASIP g.8818A>G SNP and skin color (M index) as assessed by reflectometry in 234 African Americans. Analyses of variance (ANOVA) were performed to evaluate the effects of ASIP genotypes, age, individual ancestry, and sex on skin color variation. Significant effects on M index variation were observed for ASIP genotypes (F(2,236)=4.37, P=0.01), ancestry (F(1,243)=37.2, P<0.001), and sex (F(1,244)=4.08, P=0.05). Subsequent analyses revealed a strong effect on M index from ASIP genotypes in African American females (P<0.001). Our study suggests that the ASIP G>A polymorphism exhibits a dominant effect leading to lighter skin color and that variation in the ASIP gene may have been one of several factors contributing to reductions in pigmentation in some populations. Further study is needed to reveal how interactions between ASIP and several other genes, such as MC1R and P, predict human pigmentation.	Abstract
Two newly identified genetic determinants of pigmentation in Europeans. (2008) (https://pubmed.ncbi.nlm.nih.gov/18488028) A polymorphism in the agouti signalling protein (ASIP) is associated with decreased levels of mRNA. (2006) (https://pubmed.ncbi.nlm.nih.gov/16704456)	Additional References

RELATED GEPHE

14 (EGFR, EIF2S2, GSS (glutathione synthetase), IRF4, Kit ligand, MC1R, MFSD12, Oca2, OPRM1, SLC24A5 (NCKX5), SLC45A2=MATP, TPCN2, tyrosinase (TYR), tyrosinase-related protein 1 (TYRP1)) (https://www.gephebase.org/search-criteria?/or+Taxon ID=^9606^/and+Trait=Coloration/and+groupHaplotypes=true#gephebase-summary-title)	Related Genes
No matches found.	Related Haplotypes

EXTERNAL LINKS

COMMENTS

Sulem et al. suggest confusing effect of rs6058017 (8818A>G) compared to an associated halpotype. Needs clarification

