GEPHE SUMMARY

Gephebase Gene
SLC24A5 (NCKX5) (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^SLC24A5 (NCKX5)^#gephebase-summary-title)

GepheID
GP00001056

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

Trait State in Taxon A
Homo sapiens

Trait State in Taxon B
Homo sapiens - European (see also Cape Verde 2013 study)

Ancestral State
Data not curated

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

Taxon A

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

Common Name
human

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

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; Primates; Haplorrhini; Simiiformes; Catarrhini; Hominoidea; Hominidae; Homininae; Homo

Parent
Homo () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9605)

NCBI Taxonomy ID
9606 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9606)

is Taxon A an Infraspecies?
No

Taxon B

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

Common Name
human

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

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; Primates; Haplorrhini; Simiiformes; Catarrhini; Hominoidea; Hominidae; Homininae; Homo

Parent
Homo () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9605)

NCBI Taxonomy ID
9606 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9606)

is Taxon B an Infraspecies?
No

GENOTYPIC CHANGE

Generic Gene Name
Slc24a5

Synonyms
JSX; NCX5; Oca6; NCKX5; F630045L20Rik; Nckx5

String
10090.ENSMUSP00000063887 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=10090.ENSMUSP00000063887)

Sequence Similarities
Belongs to the Ca(2+):cation antiporter (CaCA) (TC 2.A.19) family. SLC24A subfamily.

GO - Molecular Function
GO:0015293 : symporter activity (https://www.ebi.ac.uk/QuickGO/term/GO:0015293)
GO:0005262 : calcium channel activity (https://www.ebi.ac.uk/QuickGO/term/GO:0005262)
GO:0008273 : calcium, potassium:sodium antiporter activity

UniProtKB Mus musculus
Q8C261 (http://www.uniprot.org/uniprot/Q8C261)

GenebankID or UniProtKB
AAH73944 (https://www.ncbi.nlm.nih.gov/nuccore/AAH73944)

(https://www.ebi.ac.uk/QuickGO/term/GO:0008273)

GO - Biological Process

GO:0006874 : cellular calcium ion homeostasis
(https://www.ebi.ac.uk/QuickGO/term/GO:0006874)
GO:0070588 : calcium ion transmembrane transport
(https://www.ebi.ac.uk/QuickGO/term/GO:0070588)
GO:0034220 : ion transmembrane transport
(https://www.ebi.ac.uk/QuickGO/term/GO:0034220)
GO:0048022 : negative regulation of melanin biosynthetic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0048022)
GO:0050896 : response to stimulus (https://www.ebi.ac.uk/QuickGO/term/GO:0050896)

GO - Cellular Component

GO:0016021 : integral component of membrane
(https://www.ebi.ac.uk/QuickGO/term/GO:0016021)
GO:0042470 : melanosome (https://www.ebi.ac.uk/QuickGO/term/GO:0042470)
GO:0005802 : trans-Golgi network (https://www.ebi.ac.uk/QuickGO/term/GO:0005802)

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

Ala111Thr

Experimental Evidence

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

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

Main Reference

SLC24A5, a putative cation exchanger, affects pigmentation in zebrafish and humans. (2005) (https://pubmed.ncbi.nlm.nih.gov/16357253)

Authors

Lamason RL; Mohideen MA; Mest JR; Wong AC; Norton HL; Aros MC; Jurynec MJ; Mao X; Humphreville VR; Humbert JE; Sinha S; Moore JL; Jagadeeswaran P; Zhao W; Ning G; Makalowska I; McKeigue PM; O'donnell D; Kittles R; Parra EJ; Mangini NJ; Grunwald DJ; Shriver MD; Canfield VA; Cheng KC

Abstract

Lighter variations of pigmentation in humans are associated with diminished number, size, and density of melanosomes, the pigmented organelles of melanocytes. Here we show that zebrafish golden mutants share these melanosomal changes and that golden encodes a putative cation exchanger slc24a5 (nckx5) that localizes to an intracellular membrane, likely the melanosome or its precursor. The human ortholog is highly similar in sequence and functional in zebrafish. The evolutionarily conserved ancestral allele of a human coding polymorphism predominates in African and East Asian populations. In contrast, the variant allele is nearly fixed in European populations, is associated with a substantial reduction in regional heterozygosity, and correlates with lighter skin pigmentation in admixed populations, suggesting a key role for the SLC24A5 gene in human pigmentation.

Additional References

Genetic architecture of skin and eye color in an African-European admixed population. (2013) (https://pubmed.ncbi.nlm.nih.gov/23555287)
Diversity of pigmentation in cultured human melanocytes is due to differences in the type as well as quantity of melanin. (2006) (https://pubmed.ncbi.nlm.nih.gov/16524431)
SLC24A5 encodes a trans-Golgi network protein with potassium-dependent sodium-calcium exchange activity that regulates human epidermal melanogenesis. (2008) (https://pubmed.ncbi.nlm.nih.gov/18166528)
OPRM1 and EGFR contribute to skin pigmentation differences between Indigenous Americans and Europeans. (2012) (https://pubmed.ncbi.nlm.nih.gov/22198722)
The light skin allele of SLC24A5 in South Asians and Europeans shares identity by descent. (2013) (https://pubmed.ncbi.nlm.nih.gov/24244186)

RELATED GEPHE

Related Genes

14 (Agouti (ASIP), EGFR, EIF2S2, GSS (glutathione synthetase), IRF4, Kit ligand, MC1R, MFSD12, Oca2, OPRM1, 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 Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS