

GEPHE SUMMARY

Gephebase Gene		GepheID
Kit ligand (<a +kit+ligand+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=">https://www.gephebase.org/search-criteria?/and+Gene+Gephebase="+Kit+ligand+"#gephebase-summary-title)	GP00000522	
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 (coat) (<a +coloration+(coat)+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Trait=">https://www.gephebase.org/search-criteria?/and+Trait="+Coloration+(coat)+"#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 Homo sapiens
KITLG		P21583 (http://www.uniprot.org/uniprot/P21583)
Synonyms		GenebankID or UniProtKB
SF; MGF; SCF; SLF; DCUA; FPH2; FPHH; KL-1; Kitl; SHEP7; DFNA69		M59964 (https://www.ncbi.nlm.nih.gov/nuccore/M59964)
String		
9606.ENSPP00000228280 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSPP00000228280)		
Sequence Similarities		
Belongs to the SCF family.		
GO - Molecular Function		
GO:0005125 : cytokine activity (https://www.ebi.ac.uk/QuickGO/term/GO:0005125)		
GO:0008083 : growth factor activity (https://www.ebi.ac.uk/QuickGO/term/GO:0008083)		
GO:0046934 : phosphatidylinositol-4,5-bisphosphate 3-kinase activity (https://www.ebi.ac.uk/QuickGO/term/GO:0046934)		

GO:0005088 : Ras guanyl-nucleotide exchange factor activity
(<https://www.ebi.ac.uk/QuickGO/term/GO:0005088>)
GO:0005173 : stem cell factor receptor binding
(<https://www.ebi.ac.uk/QuickGO/term/GO:0005173>)

GO - Biological Process

GO:0007155 : cell adhesion (<https://www.ebi.ac.uk/QuickGO/term/GO:0007155>)
GO:0007165 : signal transduction (<https://www.ebi.ac.uk/QuickGO/term/GO:0007165>)
GO:0001755 : neural crest cell migration
(<https://www.ebi.ac.uk/QuickGO/term/GO:0001755>)
GO:0008283 : cell proliferation (<https://www.ebi.ac.uk/QuickGO/term/GO:0008283>)
GO:0035234 : ectopic germ cell programmed cell death
(<https://www.ebi.ac.uk/QuickGO/term/GO:0035234>)
GO:0035162 : embryonic hemopoiesis
(<https://www.ebi.ac.uk/QuickGO/term/GO:0035162>)
GO:0097192 : extrinsic apoptotic signaling pathway in absence of ligand
(<https://www.ebi.ac.uk/QuickGO/term/GO:0097192>)
GO:0008584 : male gonad development
(<https://www.ebi.ac.uk/QuickGO/term/GO:0008584>)
GO:0000165 : MAPK cascade (<https://www.ebi.ac.uk/QuickGO/term/GO:0000165>)
GO:0033026 : negative regulation of mast cell apoptotic process
(<https://www.ebi.ac.uk/QuickGO/term/GO:0033026>)
GO:0001541 : ovarian follicle development
(<https://www.ebi.ac.uk/QuickGO/term/GO:0001541>)
GO:0008284 : positive regulation of cell proliferation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0008284>)
GO:1902035 : positive regulation of hematopoietic stem cell proliferation
(<https://www.ebi.ac.uk/QuickGO/term/GO:1902035>)
GO:0002687 : positive regulation of leukocyte migration
(<https://www.ebi.ac.uk/QuickGO/term/GO:0002687>)
GO:0043406 : positive regulation of MAP kinase activity
(<https://www.ebi.ac.uk/QuickGO/term/GO:0043406>)
GO:0070668 : positive regulation of mast cell proliferation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0070668>)
GO:0045636 : positive regulation of melanocyte differentiation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0045636>)
GO:0002763 : positive regulation of myeloid leukocyte differentiation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0002763>)
GO:0050731 : positive regulation of peptidyl-tyrosine phosphorylation
(<https://www.ebi.ac.uk/QuickGO/term/GO:0050731>)
GO:0051897 : positive regulation of protein kinase B signaling
(<https://www.ebi.ac.uk/QuickGO/term/GO:0051897>)
GO:0046579 : positive regulation of Ras protein signal transduction
(<https://www.ebi.ac.uk/QuickGO/term/GO:0046579>)

GO - Cellular Component

GO:0016021 : integral component of membrane
(<https://www.ebi.ac.uk/QuickGO/term/GO:0016021>)
GO:0005886 : plasma membrane (<https://www.ebi.ac.uk/QuickGO/term/GO:0005886>)
GO:0005737 : cytoplasm (<https://www.ebi.ac.uk/QuickGO/term/GO:0005737>)
GO:0005576 : extracellular region (<https://www.ebi.ac.uk/QuickGO/term/GO:0005576>)
GO:0005615 : extracellular space (<https://www.ebi.ac.uk/QuickGO/term/GO:0005615>)
GO:0005856 : cytoskeleton (<https://www.ebi.ac.uk/QuickGO/term/GO:0005856>)
GO:0030175 : filopodium (<https://www.ebi.ac.uk/QuickGO/term/GO:0030175>)
GO:0030027 : lamellipodium (<https://www.ebi.ac.uk/QuickGO/term/GO:0030027>)

	Presumptive Null
Unknown (https://www.gephebase.org/search-criteria?/and+Presumptive Null=^Unknown^#gephebase-summary-title)	
	Molecular Type
Cis-regulatory (https://www.gephebase.org/search-criteria?/and+Molecular Type=^Cis-regulatory^#gephebase-summary-title)	
	Aberration Type
Unknown (https://www.gephebase.org/search-criteria?/and+Aberration Type=^Unknown^#gephebase-summary-title)	
	Molecular Details of the Mutation
unknown	
	Experimental Evidence
Association Mapping (https://www.gephebase.org/search-criteria?/and+Experimental Evidence=^Association Mapping^#gephebase-summary-title)	
	Main Reference
cis-Regulatory changes in Kit ligand expression and parallel evolution of pigmentation in sticklebacks and humans. (2007) (https://pubmed.ncbi.nlm.nih.gov/18083106)	
	Authors
Miller CT; Beleza S; Pollen AA; Schluter D; Kittles RA; Shriver MD; Kingsley DM	
	Abstract
Dramatic pigmentation changes have evolved within most vertebrate groups, including fish and humans. Here we use genetic crosses in sticklebacks to investigate the parallel origin of pigmentation changes in natural populations. High-resolution mapping and expression experiments show that light gills and light ventrums map to a divergent regulatory allele of the Kit ligand (Kitlg) gene. The divergent allele reduces expression in gill and skin tissue and is shared by multiple derived freshwater populations with reduced pigmentation. In humans, Europeans and East Asians also share derived alleles at the KITLG locus. Strong signatures of selection map to regulatory regions surrounding the gene, and admixture mapping shows that the KITLG genomic region has a significant effect on human skin color. These experiments suggest that regulatory changes in Kitlg contribute to natural variation in vertebrate pigmentation, and that similar genetic mechanisms may underlie rapid evolutionary change in fish and humans.	
	Additional References
Genetic determinants of hair, eye and skin pigmentation in Europeans. (2007) (https://pubmed.ncbi.nlm.nih.gov/17952075)	

RELATED GEPHE

Related Genes

14 (Agouti (ASIP), EGFR, EIF2S2, GSS (glutathione synthetase), IRF4, 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 Haplotypes

1 (<https://www.gephebase.org/search-criteria?/or+Gene Gephebase=~Kit ligand~/and+Taxon ID=~9606~/or+Gene Gephebase=~Kit ligand~/and+Taxon ID=~9606~#gephebase-summary-title>)

EXTERNAL LINKS

COMMENTS