

GEPHE SUMMARY

	Gephebase Gene		GepheID
Kit ligand ( <a href="https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=^Kit+ligand^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=^Kit+ligand^#gephebase-summary-title</a> )		GP00001349	
	Entry Status	Prigent	Main curator
Published			

PHENOTYPIC CHANGE

	Trait Category		
Morphology ( <a href="https://www.gephebase.org/search-criteria?/and+Trait+Category=^Morphology^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Trait+Category=^Morphology^#gephebase-summary-title</a> )			
	Trait		
Coloration (hair) ( <a href="https://www.gephebase.org/search-criteria?/and+Trait=^Coloration+(hair)^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Trait=^Coloration+(hair)^#gephebase-summary-title</a> )			
	Trait State in Taxon A		
Human-brown hair			
	Trait State in Taxon B		
Human northern Europeans ; blond hair			
	Ancestral State		
Taxon A			
	Taxonomic Status		
Intraspecific ( <a href="https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=^Intraspecific^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=^Intraspecific^#gephebase-summary-title</a> )			
Taxon A		Taxon B	
	Latin Name		Latin Name
Homo sapiens ( <a href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Homo+sapiens^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Homo+sapiens^#gephebase-summary-title</a> )		Homo sapiens ( <a href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Homo+sapiens^#gephebase-summary-title">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Homo+sapiens^#gephebase-summary-title</a> )	
	Common Name		Common Name
human		human	
	Synonyms		Synonyms
human; man; Homo sapiens Linnaeus, 1758; Home sapiens; Homo sampiens; Homo sapeins; Homo sapien; 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 sapien; 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) ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9605">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9605</a> )		Homo () - (Rank: genus) ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9605">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9605</a> )	
	NCBI Taxonomy ID		NCBI Taxonomy ID
9606 ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9606">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9606</a> )		9606 ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9606">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9606</a> )	
	is Taxon A an Intraspecies?		is Taxon B an Intraspecies?
No		No	

GENOTYPIC CHANGE

	Generic Gene Name		UniProtKB Homo sapiens
KITLG		P21583 ( <a href="http://www.uniprot.org/uniprot/P21583">http://www.uniprot.org/uniprot/P21583</a> )	
	Synonyms		GenebankID or UniProtKB
SF; MGF; SCF; SLF; DCUA; FPH2; FPHH; KL-1; Kitl; SHEP7; DFNA69		()	
	String		
9606.ENSPO0000228280 ( <a href="http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9606.ENSPO0000228280">http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9606.ENSPO0000228280</a> )			
	Sequence Similarities		
Belongs to the SCF family.			
	GO - Molecular Function		
GO:0005125 : cytokine activity ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0005125">https://www.ebi.ac.uk/QuickGO/term/GO:0005125</a> )			
GO:0008083 : growth factor activity ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0008083">https://www.ebi.ac.uk/QuickGO/term/GO:0008083</a> )			
GO:0046934 : phosphatidylinositol-4,5-bisphosphate 3-kinase activity ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0046934">https://www.ebi.ac.uk/QuickGO/term/GO:0046934</a> )			

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

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

Molecular Type

Cis-regulatory (<https://www.gephebase.org/search-criteria?/and+Molecular Type=^Cis-regulatory^#gephebase-summary-title>)

Aberration Type

SNP (<https://www.gephebase.org/search-criteria?/and+Aberration Type=^SNP^#gephebase-summary-title>)

Molecular Details of the Mutation

A>G SNP(rs12821256) 350kb upstream of transcription start

Experimental Evidence

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

Main Reference

A molecular basis for classic blond hair color in Europeans. (2014) (<https://pubmed.ncbi.nlm.nih.gov/24880339>)

Authors

Guenther CA; Tasic B; Luo L; Bedell MA; Kingsley DM

Abstract

Hair color differences are among the most obvious examples of phenotypic variation in humans. Although genome-wide association studies (GWAS) have implicated multiple loci in human pigment variation, the causative base-pair changes are still largely unknown. Here we dissect a regulatory region of the KITLG gene (encoding KIT ligand) that is significantly associated with common blond hair color in northern Europeans. Functional tests demonstrate that the region contains a regulatory enhancer that drives expression in developing hair follicles. This enhancer contains a common SNP (rs12821256) that alters a binding site for the lymphoid enhancer-binding factor 1 (LEF1) transcription factor, reducing LEF1 responsiveness and enhancer activity in cultured human keratinocytes. Mice carrying ancestral or derived variants of the human KITLG enhancer exhibit significant differences in hair pigmentation, confirming that altered regulation of an essential growth factor contributes to the classic blond hair phenotype found in northern Europeans.

Additional References

## 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

mutation alters a sequence that resembles a consensus LEF binding motif