GEPHE SUMMARY

Gephebase Gene
HERC2

GephelD
GP00001363

Entry Status
Published

Main curator
Prigent

PHENOTYPIC CHANGE

Trait Category
Morphology

Trait
Coloration (eye; skin; hair)

Trait State in Taxon A
Human-dark pigmentation

Trait State in Taxon B
human-light pigmentation

Ancestral State
Taxon A

Taxonomic Status
Intraspecific

Taxon A

Latin Name
Homo sapiens

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)

NCBI Taxonomy ID
9606

is Taxon A an Infraspecies?
No

Taxon B

Latin Name
Homo sapiens

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)

NCBI Taxonomy ID
9606

is Taxon B an Infraspecies?
No

GENOTYPIC CHANGE

Generic Gene Name
HERC2

Synonyms
jdf2; p528; MRT38; SHEP1; D15F37S1

String
9606.ENSP00000261609

Sequence Similarities
-

GO - Molecular Function
GO:0008270 : zinc ion binding
GO:0031625 : ubiquitin protein ligase binding
GO:0004842 : ubiquitin-protein transferase activity
GO:0005085 : guanyl-nucleotide exchange factor activity
GO:0032183 : SUMO binding

GO - Biological Process
GO:0016567 : protein ubiquitination

UniProtKB Homo sapiens
O95714

GenebankID or UniProtKB

GO:0007283 : spermatogenesis
GO:0006886 : intracellular protein transport
GO:0043161 : proteasome-mediated ubiquitin-dependent protein catabolic process
GO:0006974 : cellular response to DNA damage stimulus
GO:0006303 : double-strand break repair via nonhomologous end joining

GO - Cellular Component
GO:0005737 : cytoplasm
GO:0016020 : membrane
GO:0005654 : nucleoplasm
GO:0005634 : nucleus
GO:0005814 : centriole

Presumptive Null
No

Molecular Type
Cis-regulatory

Aberration Type
SNP

Molecular Details of the Mutation
c. A>G

Experimental Evidence
Association Mapping

Main Reference
Direct evidence for positive selection of skin, hair, and eye pigmentation in Europeans during the last 5,000 y. (2014)

Authors
Wilde S; Timpson A; Kirsanow K; Kaiser E; Kayser M; UnterlÃ¤nder M; Hollfelder N; Potekhina ID; Schier W; Thomas MG; Burger J

Abstract
Pigmentation is a polygenic trait encompassing some of the most visible phenotypic variation observed in humans. Here we present direct estimates of selection acting on functional alleles in three key genes known to be involved in human pigmentation pathways--HERC2, SLC45A2, and TYR--using allele frequency estimates from Eneolithic, Bronze Age, and modern Eastern European samples and forward simulations. Neutrality was overwhelmingly rejected for all alleles studied, with point estimates of selection ranging from around 2-10% per generation. Our results provide direct evidence that strong selection favoring lighter skin, hair, and eye pigmentation has been operating in European populations over the last 5,000 y.

Additional References

RELATED GEPHE

Related Genes
14 (Agouti (ASIP), EGFR, EIF2S2, GSS (glutathione synthetase), IRF4, Kit ligand, MC1R, Oca2, OPRM1, SLC24A5 (NCKX5), SLC45A2=MATP, TPCN2, tyrosinase (TYR), tyrosinase-related protein 1 (TYRP1))
Related Haplotypes
No matches found.

COMMENTS

derived mutation is recessive- under positive selection in European populations