



GO:0005507 : copper ion binding (<https://www.ebi.ac.uk/QuickGO/term/GO:0005507>)

GO:0004503 : monophenol monooxygenase activity  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0004503>)

GO - Biological Process

GO:0042438 : melanin biosynthetic process

(<https://www.ebi.ac.uk/QuickGO/term/GO:0042438>)

GO:0043473 : pigmentation (<https://www.ebi.ac.uk/QuickGO/term/GO:0043473>)

GO:0008283 : cell proliferation (<https://www.ebi.ac.uk/QuickGO/term/GO:0008283>)

GO:0033280 : response to vitamin D (<https://www.ebi.ac.uk/QuickGO/term/GO:0033280>)

GO:0051591 : response to cAMP (<https://www.ebi.ac.uk/QuickGO/term/GO:0051591>)

GO:0009411 : response to UV (<https://www.ebi.ac.uk/QuickGO/term/GO:0009411>)

GO:0048538 : thymus development (<https://www.ebi.ac.uk/QuickGO/term/GO:0048538>)

GO - Cellular Component

GO:0016021 : integral component of membrane

(<https://www.ebi.ac.uk/QuickGO/term/GO:0016021>)

GO:0005737 : cytoplasm (<https://www.ebi.ac.uk/QuickGO/term/GO:0005737>)

GO:0005829 : cytosol (<https://www.ebi.ac.uk/QuickGO/term/GO:0005829>)

GO:0005634 : nucleus (<https://www.ebi.ac.uk/QuickGO/term/GO:0005634>)

GO:0043231 : intracellular membrane-bounded organelle

(<https://www.ebi.ac.uk/QuickGO/term/GO:0043231>)

GO:0048471 : perinuclear region of cytoplasm

(<https://www.ebi.ac.uk/QuickGO/term/GO:0048471>)

GO:0042470 : melanosome (<https://www.ebi.ac.uk/QuickGO/term/GO:0042470>)

GO:0033162 : melanosome membrane

(<https://www.ebi.ac.uk/QuickGO/term/GO:0033162>)

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

c. C>A p.Ser192Tyr

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

Direct evidence for positive selection of skin, hair, and eye pigmentation in Europeans during the last 5,000 y. (2014) (<https://pubmed.ncbi.nlm.nih.gov/24616518>)

Authors

Wilde S; Timpson A; Kirsanow K; Kaiser E; Kayser M; Unterl  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, MFSD12, Oca2, OPRM1, SLC24A5 (NCKX5), SLC45A2=MATP, TPCN2, 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=~tyrosinase+\(TYR\)~/and+Taxon+ID=~9606~/or+Gene+Gephebase=~tyrosinase+\(TYR\)~/and+Taxon+ID=~9606~/#gephebase-summary-title](https://www.gephebase.org/search-criteria?/or+Gene+Gephebase=~tyrosinase+(TYR)~/and+Taxon+ID=~9606~/or+Gene+Gephebase=~tyrosinase+(TYR)~/and+Taxon+ID=~9606~/#gephebase-summary-title))

EXTERNAL LINKS

COMMENTS

codominance is assumed- under positive selection in European populations