## GEPHE SUMMARY

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

GepheID
GP00001348

Entry Status
Published

Main curator
Prigent

## PHENOTYPIC CHANGE

Trait Category
Morphology (https://www.gephebase.org/search-criteria?/and+Trait Category=^Morphology^#gephebase-summary-title)

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

Trait State in Taxon A
Human-dark pigmentation

Trait State in Taxon B
human-light pigmentation

Ancestral State
Taxon A

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
Tyr

Synonyms
c; Oca1; skc35; albino

String
10090.ENSMUSP00000004770 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=10090.ENSMUSP00000004770 )

Sequence Similarities
Belongs to the tyrosinase family.

GO - Molecular Function
GO:0042803 : protein homodimerization activity (https://www.ebi.ac.uk/QuickGO/term/GO:0042803)
GO:0046982 : protein heterodimerization activity (https://www.ebi.ac.uk/QuickGO/term/GO:0046982)

UniProtKB Mus musculus
P11344 (http://www.uniprot.org/uniprot/P11344)

GenebankID or UniProtKB
()

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; 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, 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)

## EXTERNAL LINKS

## COMMENTS

codominance is assumed- under positive selection in European populations