GEPHE SUMMARY

Gephebase Gene

Glucose-6-phosphate dehydrogenase (G6PD) (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^Glucose-6-phosphate dehydrogenase (G6PD)^#gephebase-summary-title)

Entry Status

Published

GepheID

GP00000402

Main curator

Martin

PHENOTYPIC CHANGE

Trait Category

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

Trait

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

Trait State in Taxon A

Homo sapiens-SouthEast Asia

Trait State in Taxon B

Homo sapiens-SouthEast Asia

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

G6PD

Synonyms

G6PD1

String

9606.ENSP00000377192 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9606.ENSP00000377192)

Sequence Similarities

Belongs to the glucose-6-phosphate dehydrogenase family.

GO - Molecular Function

GO:0042802 : identical protein binding (https://www.ebi.ac.uk/QuickGO/term/GO:0042802)
GO:0042803 : protein homodimerization activity

UniProtKB Homo sapiens

P11413 (http://www.uniprot.org/uniprot/P11413)

GenebankID or UniProtKB

AY158142 (https://www.ncbi.nlm.nih.gov/nuccore/AY158142)

(https://www.ebi.ac.uk/QuickGO/term/GO:0042803)
GO:0050661 : NADP binding (https://www.ebi.ac.uk/QuickGO/term/GO:0050661)
GO:0005536 : glucose binding (https://www.ebi.ac.uk/QuickGO/term/GO:0005536)
GO:0004345 : glucose-6-phosphate dehydrogenase activity
(https://www.ebi.ac.uk/QuickGO/term/GO:0004345)

GO - Biological Process

GO:0055114 : oxidation-reduction process
(https://www.ebi.ac.uk/QuickGO/term/GO:0055114)
GO:0006695 : cholesterol biosynthetic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0006695)
GO:0014070 : response to organic cyclic compound
(https://www.ebi.ac.uk/QuickGO/term/GO:0014070)
GO:0006629 : lipid metabolic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0006629)
GO:0045471 : response to ethanol (https://www.ebi.ac.uk/QuickGO/term/GO:0045471)
GO:0006749 : glutathione metabolic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0006749)
GO:0032094 : response to food (https://www.ebi.ac.uk/QuickGO/term/GO:0032094)
GO:0034599 : cellular response to oxidative stress
(https://www.ebi.ac.uk/QuickGO/term/GO:0034599)
GO:0043249 : erythrocyte maturation
(https://www.ebi.ac.uk/QuickGO/term/GO:0043249)
GO:0051156 : glucose 6-phosphate metabolic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0051156)
GO:0006006 : glucose metabolic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0006006)
GO:0006739 : NADP metabolic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0006739)
GO:0006740 : NADPH regeneration
(https://www.ebi.ac.uk/QuickGO/term/GO:0006740)
GO:0061052 : negative regulation of cell growth involved in cardiac muscle cell
development (https://www.ebi.ac.uk/QuickGO/term/GO:0061052)
GO:0010734 : negative regulation of protein glutathionylation
(https://www.ebi.ac.uk/QuickGO/term/GO:0010734)
GO:2000378 : negative regulation of reactive oxygen species metabolic process
(https://www.ebi.ac.uk/QuickGO/term/GO:2000378)
GO:0019322 : pentose biosynthetic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0019322)
GO:0006098 : pentose-phosphate shunt
(https://www.ebi.ac.uk/QuickGO/term/GO:0006098)
GO:0009051 : pentose-phosphate shunt, oxidative branch
(https://www.ebi.ac.uk/QuickGO/term/GO:0009051)
GO:1904879 : positive regulation of calcium ion transmembrane transport via high voltage-
gated calcium channel (https://www.ebi.ac.uk/QuickGO/term/GO:1904879)
GO:0043523 : regulation of neuron apoptotic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0043523)
GO:0010041 : response to iron(III) ion
(https://www.ebi.ac.uk/QuickGO/term/GO:0010041)
GO:0046390 : ribose phosphate biosynthetic process
(https://www.ebi.ac.uk/QuickGO/term/GO:0046390)
GO:0021762 : substantia nigra development
(https://www.ebi.ac.uk/QuickGO/term/GO:0021762)

GO - Cellular Component

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:0016020 : membrane (https://www.ebi.ac.uk/QuickGO/term/GO:0016020)
GO:0070062 : extracellular exosome (https://www.ebi.ac.uk/QuickGO/term/GO:0070062)
GO:0005634 : nucleus (https://www.ebi.ac.uk/QuickGO/term/GO:0005634)
GO:0009898 : cytoplasmic side of plasma membrane
(https://www.ebi.ac.uk/QuickGO/term/GO:0009898)

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

Ser163Gly

Experimental Evidence

Candidate Gene (https://www.gephebase.org/search-criteria?/and+Experimental Evidence=^Candidate Gene^#gephebase-summary-title)

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	-	-	-

Main Reference

Positively selected G6PD-Mahidol mutation reduces Plasmodium vivax density in Southeast Asians. (2009) (https://pubmed.ncbi.nlm.nih.gov/20007901)

Authors

Louicharoen C; Patin E; Paul R; Nuchprayoon I; Witoonpanich B; Peerapittayamongkol C; Casademont I; Sura T; Laird NM; Singhasivanon P; Quintana-Murci L; Sakuntabhai A

Abstract

Glucose-6-phosphate dehydrogenase (G6PD) deficiency--the most common known enzymopathy--is associated with neonatal jaundice and hemolytic anemia usually after exposure to certain infections, foods, or medications. Although G6PD-deficient alleles appear to confer a protective effect against malaria, the link with clinical protection from Plasmodium infection remains unclear. We investigated the effect of a common G6PD deficiency variant in Southeast Asia--the G6PD-Mahidol(487A) variant--on human survival related to vivax and falciparum malaria. Our results show that strong and recent positive selection has targeted the Mahidol variant over the past 1500 years. We found that the G6PD-Mahidol(487A) variant reduces vivax, but not falciparum, parasite density in humans, which indicates that Plasmodium vivax has been a driving force behind the strong selective advantage conferred by this mutation.

Additional References

RELATED GEPHE

Related Genes

10 (ATP2B4, CCL3L1, Duffy, Glycophorin GYPA-GYPB-GYPE cluster, hemoglobin; HBB, HLA-DRB1, Human Leukocyte Antigen-B (HLA-B), MARVELD3, SIGLEC13, SIGLEC17P (pseudogene)) (https://www.gephebase.org/search-criteria?/or+Taxon ID=^9606^/and+Trait=Pathogen resistance/and+groupHaplotypes=true#gephebase-summary-title)

Related Haplotypes

2 (https://www.gephebase.org/search-criteria?/or+Gene Gephebase=^Glucose-6-phosphate dehydrogenase (G6PD)^/and+Taxon ID=^9606^/or+Gene Gephebase=^Glucose-6-phosphate dehydrogenase (G6PD)^/and+Taxon ID=^9606^#gephebase-summary-title)

EXTERNAL LINKS

COMMENTS