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

GP00000401

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

AY158110 (https://www.ncbi.nlm.nih.gov/nuccore/AY158110)

(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

Ser188Phe C>T

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

Haplotype diversity and linkage disequilibrium at human G6PD: recent origin of alleles that confer malarial resistance. (2001) (https://pubmed.ncbi.nlm.nih.gov/11423617)

Authors

Tishkoff SA; Varkonyi R; Cahinhinan N; Abbes S; Argyropoulos G; Destro-Bisol G; Drousiotou A; Dangerfield B; Lefranc G; Loiselet J; Piro A; Stoneking M; Tagarelli A; Tagarelli G; Touma EH; Williams SM; Clark AG

Abstract

The frequencies of low-activity alleles of glucose-6-phosphate dehydrogenase in humans are highly correlated with the prevalence of malaria. These "deficiency" alleles are thought to provide reduced risk from infection by the Plasmodium parasite and are maintained at high frequency despite the hemopathologies that they cause. Haplotype analysis of "A-" and "Med" mutations at this locus indicates that they have evolved independently and have increased in frequency at a rate that is too rapid to be explained by random genetic drift. Statistical modeling indicates that the A- allele arose within the past 3840 to 11,760 years and the Med allele arose within the past 1600 to 6640 years. These results support the hypothesis that malaria has had a major impact on humans only since the introduction of agriculture within the past 10,000 years and provide a striking example of the signature of selection on the human genome.

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