

GEPHE SUMMARY

Gephebase Gene

Glucose-6-phosphate dehydrogenase (G6PD)

Entry Status

Published

GepheID

GP00000401

Main curator

Martin

PHENOTYPIC CHANGE

Trait Category

Physiology

Trait

Pathogen resistance (malaria)

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

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 Intraspecies?

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 Intraspecies?

No

GENOTYPIC CHANGE

Generic Gene Name

G6PD

Synonyms

G6PD1

String

9606.ENSF00000377192

Sequence Similarities

Belongs to the glucose-6-phosphate dehydrogenase family.

GO - Molecular Function

GO:0042802 : identical protein binding

GO:0042803 : protein homodimerization activity

GO:0050661 : NADP binding

GO:0005536 : glucose binding

GO:0004345 : glucose-6-phosphate dehydrogenase activity

GO - Biological Process

GO:0055114 : oxidation-reduction process

UniProtKB Homo sapiens

P11413

GenebankID or UniProtKB

AY158110

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

GO - Cellular Component

GO:0005737 : cytoplasm
 GO:0005829 : cytosol
 GO:0016020 : membrane
 GO:0070062 : extracellular exosome
 GO:0005634 : nucleus
 GO:0009898 : cytoplasmic side of plasma membrane

Presumptive Null

No

Molecular Type

Coding

Aberration Type

SNP

SNP Coding Change

Nonsynonymous

Molecular Details of the Mutation

Ser188Phe C>T

Experimental Evidence

Candidate Gene

	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)

Authors

Tishkoff SA; Varkonyi R; Cahinhan N; Abbas S; Argyropoulos G; Destro-Bisol G; Drouiotou 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))

Related Haplotypes

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COMMENTS