

GEPHE SUMMARY

	Gephebase Gene		GepheID
Glucose-6-phosphate dehydrogenase (G6PD) (<a +glucose-6-phosphate+dehydrogenase+(g6pd)+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=">https://www.gephebase.org/search-criteria?/and+Gene+Gephebase="+Glucose-6-phosphate+dehydrogenase+(G6PD)+"#gephebase-summary-title)	GP00000403		
	Martin		Main curator
Published	Entry Status		

PHENOTYPIC CHANGE

	Trait Category		
Physiology (<a +physiology+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Trait+Category=">https://www.gephebase.org/search-criteria?/and+Trait+Category="+Physiology+"#gephebase-summary-title)	Trait		
Pathogen resistance (malaria) (<a +pathogen+resistance+(malaria)+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Trait=">https://www.gephebase.org/search-criteria?/and+Trait="+Pathogen+resistance+(malaria)+"#gephebase-summary-title)	Trait State in Taxon A		
Homo sapiens-SubSaharan Africa	Trait State in Taxon B		
Homo sapiens-SubSaharan Africa	Ancestral State		
Taxon A	Taxonomic Status		
Intraspecific (<a +intraspecific+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=">https://www.gephebase.org/search-criteria?/and+Taxonomic+Status="+Intraspecific+"#gephebase-summary-title)			
	Taxon A		Taxon B
	Latin Name		Latin Name
Homo sapiens (<a +homo+sapiens+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=">https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms="+Homo+sapiens+"#gephebase-summary-title)	Common Name		Common Name
human	Synonyms		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		Rank
species	Lineage		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		Parent
Homo () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9605)	NCBI Taxonomy ID		NCBI Taxonomy ID
9606 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9606)	is Taxon A an Intraspecies?		is Taxon B an Intraspecies?
No			No

GENOTYPIC CHANGE

	Generic Gene Name		UniProtKB Homo sapiens
G6PD	Synonyms		GenebankID or UniProtKB
G6PD1	String		AY158110 (https://www.ncbi.nlm.nih.gov/nuccore/AY158110)
9606.ENSPO00000377192 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSPO00000377192)	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			

(<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

Val68Met G>A at nucleotide position 376 in exon 5 - OMIM code +305900.0002

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	Val	Met	68

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; 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)) (<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](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