

GEPHE SUMMARY

Gephebase Gene
hemoglobin; HBB

Entry Status
Published

GepheID
GP00000466

Main curator
Martin

PHENOTYPIC CHANGE

Trait Category
Physiology

Trait
Pathogen resistance (Plasmodium; malaria parasite)

Trait State in Taxon A
Homo sapiens - malarial sensitive

Trait State in Taxon B
Homo sapiens - malarial resistant

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
HBB

Synonyms
ECYT6; CD113t-C; beta-globin

String
9606.ENSP00000333994

Sequence Similarities
Belongs to the globin family.

GO - Molecular Function
GO:0046872 : metal ion binding
GO:0020037 : heme binding
GO:0005344 : oxygen carrier activity
GO:0043177 : organic acid binding
GO:0019825 : oxygen binding
GO:0031721 : hemoglobin alpha binding
GO:0030492 : hemoglobin binding

UniProtKB Homo sapiens
P68871

GenebankID or UniProtKB
BC039916

GO - Biological Process

GO:0006898 : receptor-mediated endocytosis
GO:0007596 : blood coagulation
GO:0008217 : regulation of blood pressure
GO:0042542 : response to hydrogen peroxide
GO:0043312 : neutrophil degranulation
GO:0015701 : bicarbonate transport
GO:0098869 : cellular oxidant detoxification
GO:0042744 : hydrogen peroxide catabolic process
GO:0015671 : oxygen transport
GO:0010942 : positive regulation of cell death
GO:0051291 : protein heterooligomerization
GO:0030185 : nitric oxide transport
GO:0070527 : platelet aggregation
GO:0045429 : positive regulation of nitric oxide biosynthetic process
GO:0050880 : regulation of blood vessel size
GO:0070293 : renal absorption

GO - Cellular Component

GO:0005829 : cytosol
GO:0070062 : extracellular exosome
GO:0005576 : extracellular region
GO:0005615 : extracellular space
GO:0072562 : blood microparticle
GO:0071682 : endocytic vesicle lumen
GO:0031838 : haptoglobin-hemoglobin complex
GO:0005833 : hemoglobin complex
GO:1904813 : ficolin-1-rich granule lumen
GO:1904724 : tertiary granule lumen

Presumptive Null

No

Molecular Type

Coding

Aberration Type

SNP

SNP Coding Change

Nonsynonymous

Molecular Details of the Mutation

Glu6Val

Experimental Evidence

Association Mapping

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

Main Reference

Genome-wide and fine-resolution association analysis of malaria in West Africa. (2009)

Authors

Jallow M; Teo YY; Small KS; Rockett KA; Deloukas P; Clark TG; Kivinen K; Bojang KA; Conway DJ; Pinder M; Sirugo G; Sisay-Joof F; Usen S; Auburn S; Bumpstead SJ; Campino S; Coffey A; Dunham A; Fry AE; Green A; Gwilliam R; Hunt SE; Inouye M; Jeffreys AE; Mendy A; Palotie A; Potter S; Ragoussis J; Rogers J; Rowlands K; Somaskantharajah E; Whittaker P; Widdens C; Donnelly P; Howie B; Marchini J; Morris A; SanJoaquin M; Achidi EA; Agbenyega T; Allen A; Amodu O; Corran P; Djimde A; Dolo A; Doumbo OK; Drakeley C; Dunstan S; Evans J; Farrar J; Fernando D; Hien TT; Horstmann RD; Ibrahim M; Karunaweera N; Kokwaro G; Koram KA; Lemnge M; Makani J; Marsh K; Michon P; Modiano D; Molyneux ME; Mueller I; Parker M; Peshu N; Plowe CV; Pujalon O; Reeder J; Reyburn H; Riley EM; Sakuntabhai A; Singhasivanon P; Sirima S; Tall A; Taylor TE; Thera M; Troye-Blomberg M; Williams TN; Wilson M; Kwiatkowski DP; ;

Abstract

We report a genome-wide association (GWA) study of severe malaria in The Gambia. The initial GWA scan included 2,500 children genotyped on the Affymetrix 500K GeneChip, and a replication study included 3,400 children. We used this to examine the performance of GWA methods in Africa. We found considerable population stratification, and also that signals of association at known malaria resistance loci were greatly attenuated owing to weak linkage disequilibrium (LD). To investigate possible solutions to the problem of low LD, we focused on the HbS locus, sequencing this region of the genome in 62 Gambian individuals and then using these data to conduct multipoint imputation in the GWA samples. This increased the signal of association, from $P = 4 \times 10^{-7}$ to $P = 4 \times 10^{-14}$, with the peak of the signal located precisely at the HbS causal variant. Our findings provide proof of principle that fine-resolution multipoint imputation, based on population-specific sequencing data, can substantially boost authentic GWA signals and enable fine mapping of causal variants in African populations.

Additional References

RELATED GEPHE

Related Genes

10 (ATP2B4, CCL3L1, Duffy, Glucose-6-phosphate dehydrogenase (G6PD), Glycophorin GYPA-GYPB-GYPE cluster, HLA-DRB1, Human Leukocyte Antigen-B (HLA-B), MARVELD3, SIGLEC13, SIGLEC17P (pseudogene))

Related Haplotypes

No matches found.

COMMENTS

Needs curation