GEPHE SUMMARY

Gephebase Gene

Glycophorin GYPA-GYPB-GYPE cluster (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^Glycophorin GYPA-GYPB-GYPE cluster^#gephebase-summary-title)

GepheID

GP00000406

Entry Status

Published

Main curator

Martin

PHENOTYPIC CHANGE

Trait Category

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

Trait

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

Trait State in Taxon A

Homo sapiens

Trait State in Taxon B

Homo sapiens

Ancestral State

Data not curated

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

GYPA

Synonyms

MN; GPA; MNS; GPSAT; PAS-2; CD235a; GPErik; HGpMiV; HGpMiXI; HGpSta(C)

String

9606.ENSP00000354003 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9606.ENSP00000354003)

Sequence Similarities

Belongs to the glycophorin A family.

GO - Molecular Function

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

UniProtKB Homo sapiens

P02724 (http://www.uniprot.org/uniprot/P02724)

GenebankID or UniProtKB

()

GO:0001618 : virus receptor activity (https://www.ebi.ac.uk/QuickGO/term/GO:0001618)

GO - Biological Process

GO:0050900 : leukocyte migration (https://www.ebi.ac.uk/QuickGO/term/GO:0050900)

GO - Cellular Component

GO:0016021 : integral component of membrane
(https://www.ebi.ac.uk/QuickGO/term/GO:0016021)
GO:0005886 : plasma membrane (https://www.ebi.ac.uk/QuickGO/term/GO:0005886)
GO:0016020 : membrane (https://www.ebi.ac.uk/QuickGO/term/GO:0016020)

Presumptive Null

Unknown (https://www.gephebase.org/search-criteria?/and+Presumptive Null=^Unknown^#gephebase-summary-title)

Molecular Type

Unknown (https://www.gephebase.org/search-criteria?/and+Molecular Type=^Unknown^#gephebase-summary-title)

Aberration Type

Unknown (https://www.gephebase.org/search-criteria?/and+Aberration Type=^Unknown^#gephebase-summary-title)

Molecular Details of the Mutation

unknown

Experimental Evidence

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

Main Reference

A novel locus of resistance to severe malaria in a region of ancient balancing selection. (2015) (https://pubmed.ncbi.nlm.nih.gov/26416757)

Authors

; Band G; Rockett KA; Spencer CC; Kwiatkowski DP

Abstract

The high prevalence of sickle haemoglobin in Africa shows that malaria has been a major force for human evolutionary selection, but surprisingly few other polymorphisms have been proven to confer resistance to malaria in large epidemiological studies. To address this problem, we conducted a multi-centre genome-wide association study (GWAS) of life-threatening Plasmodium falciparum infection (severe malaria) in over 11,000 African children, with replication data in a further 14,000 individuals. Here we report a novel malaria resistance locus close to a cluster of genes encoding glycophorins that are receptors for erythrocyte invasion by P. falciparum. We identify a haplotype at this locus that provides 33% protection against severe malaria (odds ratioâ€‰=â€‰0.67, 95% confidence intervalâ€‰=â€‰0.60-0.76, P valueâ€‰=â€‰9.5â€‰Ã—â€‰10(-11)) and is linked to polymorphisms that have previously been shown to have features of ancient balancing selection, on the basis of haplotype sharing between humans and chimpanzees. Taken together with previous observations on the malaria-protective role of blood group O, these data reveal that two of the strongest GWAS signals for severe malaria lie in or close to genes encoding the glycosylated surface coat of the erythrocyte cell membrane, both within regions of the genome where it appears that evolution has maintained diversity for millions of years. These findings provide new insights into the host-parasite interactions that are critical in determining the outcome of malaria infection.

Additional References

RELATED GEPHE

Related Genes

10 (ATP2B4, CCL3L1, Duffy, Glucose-6-phosphate dehydrogenase (G6PD), 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

No matches found.

EXTERNAL LINKS

COMMENTS

Ancient @BalancingSelection