GEPHE SUMMARY

Gephebase Gene

MARVELD3 (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^MARVELD3^#gephebase-summary-title)

Entry Status

Published

GepheID

GP00000564

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 -Ghana and Gambia

Trait State in Taxon B

Homo sapiens -Ghana and Gambia

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

MARVELD3

Synonyms

MARVD3; MRVLDC3

String

-

Sequence Similarities

-

GO - Molecular Function

GO:0031435 : mitogen-activated protein kinase kinase kinase binding (https://www.ebi.ac.uk/QuickGO/term/GO:0031435)

GO - Biological Process

GO:0050680 : negative regulation of epithelial cell proliferation (https://www.ebi.ac.uk/QuickGO/term/GO:0050680)

UniProtKB Homo sapiens

Q96A59 (http://www.uniprot.org/uniprot/Q96A59)

GenebankID or UniProtKB

AK292185 (https://www.ncbi.nlm.nih.gov/nuccore/AK292185)

GO:0006970 : response to osmotic stress
(https://www.ebi.ac.uk/QuickGO/term/GO:0006970)
GO:0010633 : negative regulation of epithelial cell migration
(https://www.ebi.ac.uk/QuickGO/term/GO:0010633)
GO:0070830 : bicellular tight junction assembly
(https://www.ebi.ac.uk/QuickGO/term/GO:0070830)
GO:0045216 : cell-cell junction organization
(https://www.ebi.ac.uk/QuickGO/term/GO:0045216)
GO:0046329 : negative regulation of JNK cascade
(https://www.ebi.ac.uk/QuickGO/term/GO:0046329)
GO:1902414 : protein localization to cell junction
(https://www.ebi.ac.uk/QuickGO/term/GO:1902414)

GO - Cellular Component

GO:0016021 : integral component of membrane
(https://www.ebi.ac.uk/QuickGO/term/GO:0016021)
GO:0031410 : cytoplasmic vesicle (https://www.ebi.ac.uk/QuickGO/term/GO:0031410)
GO:0005923 : bicellular tight junction
(https://www.ebi.ac.uk/QuickGO/term/GO:0005923)

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

Genome-wide association study indicates two novel resistance loci for severe malaria. (2012) (https://pubmed.ncbi.nlm.nih.gov/22895189)

Authors

Timmann C; Thye T; Vens M; Evans J; May J; Ehmen C; Sievertsen J; Muntau B; Ruge G; Loag W; Ansong D; Antwi S; Asafo-Adjei E; Nguah SB; Kwakye KO; Akoto AO; Sylverken J; Brendel M; Schuldt K; Loley C; Franke A; Meyer CG; Agbenyega T; Ziegler A; Horstmann RD

Abstract

Malaria causes approximately one million fatalities per year, mostly among African children. Although highlighted by the strong protective effect of the sickle-cell trait, the full impact of human genetics on resistance to the disease remains largely unexplored. Genome-wide association (GWA) studies are designed to unravel relevant genetic variants comprehensively; however, in malaria, as in other infectious diseases, these studies have been only partly successful. Here we identify two previously unknown loci associated with severe falciparum malaria in patients and controls from Ghana, West Africa. We applied the GWA approach to the diverse clinical syndromes of severe falciparum malaria, thereby targeting human genetic variants influencing any step in the complex pathogenesis of the disease. One of the loci was identified on chromosome 1q32 within the ATP2B4 gene, which encodes the main calcium pump of erythrocytes, the host cells of the pathogenic stage of malaria parasites. The second was indicated by an intergenic single nucleotide polymorphism on chromosome 16q22.2, possibly linked to a neighbouring gene encoding the tight-junction protein MARVELD3. The protein is expressed on endothelial cells and might therefore have a role in microvascular damage caused by endothelial adherence of parasitized erythrocytes. We also confirmed previous reports on protective effects of the sickle-cell trait and blood group O. Our findings underline the potential of the GWA approach to provide candidates for the development of control measures against infectious diseases in humans.

Additional References

RELATED GEPHE

Related Genes

10 (ATP2B4, CCL3L1, Duffy, Glucose-6-phosphate dehydrogenase (G6PD), Glycophorin GYPA-GYPB-GYPE cluster, hemoglobin; HBB, HLA-DRB1, Human Leukocyte Antigen-B (HLA-B), 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