

GEPHE SUMMARY

	Gephebase Gene		GepheID
MARVELD3 (https://www.gephebase.org/search-criteria?/and+GeneGephebase=~MARVELD3^#gephebase-summary-title)		GP00000564	
	Entry Status	Martin	Main curator
Published			

PHENOTYPIC CHANGE

	Trait Category		
Physiology (https://www.gephebase.org/search-criteria?/and+TraitCategory=~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+TaxonomicStatus=~Intraspecific^#gephebase-summary-title)			
	Taxon A		Taxon B
	Latin Name		Latin Name
Homo sapiens (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=~Homo sapiens^#gephebase-summary-title)		Homo sapiens (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=~Homo sapiens^#gephebase-summary-title)	
	Common Name		Common Name
human		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		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		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		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)		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)		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
MARVELD3		Q96A59 (http://www.uniprot.org/uniprot/Q96A59)	
	Synonyms		GenebankID or UniProtKB
MARVD3; MRVLDC3		AK292185 (https://www.ncbi.nlm.nih.gov/nuccore/AK292185)	
-	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)			

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>)

Unknown (https://www.gephebase.org/search-criteria?/and+Presumptive Null=^Unknown^#gephebase-summary-title)	Presumptive Null
Unknown (https://www.gephebase.org/search-criteria?/and+Molecular Type=^Unknown^#gephebase-summary-title)	Molecular Type
Unknown (https://www.gephebase.org/search-criteria?/and+Aberration Type=^Unknown^#gephebase-summary-title)	Aberration Type
unknown	Molecular Details of the Mutation
Association Mapping (https://www.gephebase.org/search-criteria?/and+Experimental Evidence=^Association Mapping^#gephebase-summary-title)	Experimental Evidence
Genome-wide association study indicates two novel resistance loci for severe malaria. (2012) (https://pubmed.ncbi.nlm.nih.gov/22895189)	Main Reference

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

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 Genes
No matches found.	Related Haplotypes

EXTERNAL LINKS

COMMENTS