

## GEPHE SUMMARY

	Gephebase Gene		GepheID
HLA-DRB1 ( <a +hla-drb1+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=">https://www.gephebase.org/search-criteria?/and+Gene+Gephebase="+HLA-DRB1+"#gephebase-summary-title</a> )		GP00000479	
Published	Entry Status	Martin	Main curator

## 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</a> )			
	Trait		
Pathogen resistance (typhoid fever) ( <a +pathogen+resistance+(typhoid+fever)+"#gephebase-summary-title"="" href="https://www.gephebase.org/search-criteria?/and+Trait=">https://www.gephebase.org/search-criteria?/and+Trait="+Pathogen+resistance+(typhoid+fever)+"#gephebase-summary-title</a> )			
	Trait State in Taxon A		
Homo sapiens			
	Trait State in Taxon B		
Homo sapiens			
	Ancestral State		
Data not curated			
	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</a> )			
	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</a> )		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</a> )	
	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) ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9605">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9605</a> )		Homo () - (Rank: genus) ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9605">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9605</a> )	
	NCBI Taxonomy ID		NCBI Taxonomy ID
9606 ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9606">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9606</a> )		9606 ( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9606">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9606</a> )	
	is Taxon A an Intraspecies?		is Taxon B an Intraspecies?
No		No	

## GENOTYPIC CHANGE

	Generic Gene Name		UniProtKB Homo sapiens
HLA-DRB1		Q30167 ( <a href="http://www.uniprot.org/uniprot/Q30167">http://www.uniprot.org/uniprot/Q30167</a> )	
	Synonyms		GenebankID or UniProtKB
-		0	
	String		
-			
	Sequence Similarities		
Belongs to the MHC class II family.			
	GO - Molecular Function		
-			
	GO - Biological Process		
GO:0019886 : antigen processing and presentation of exogenous peptide antigen via MHC class II ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0019886">https://www.ebi.ac.uk/QuickGO/term/GO:0019886</a> )			
GO:0060333 : interferon-gamma-mediated signaling pathway ( <a href="https://www.ebi.ac.uk/QuickGO/term/GO:0060333">https://www.ebi.ac.uk/QuickGO/term/GO:0060333</a> )			

GO:0050852 : T cell receptor signaling pathway  
(<https://www.ebi.ac.uk/QuickGO/term/GO:0050852>)

GO - Cellular Component

GO:0005886 : plasma membrane (<https://www.ebi.ac.uk/QuickGO/term/GO:0005886>)

GO:0000139 : Golgi membrane (<https://www.ebi.ac.uk/QuickGO/term/GO:0000139>)

GO:0030669 : clathrin-coated endocytic vesicle membrane

(<https://www.ebi.ac.uk/QuickGO/term/GO:0030669>)

GO:0030666 : endocytic vesicle membrane

(<https://www.ebi.ac.uk/QuickGO/term/GO:0030666>)

GO:0012507 : ER to Golgi transport vesicle membrane

(<https://www.ebi.ac.uk/QuickGO/term/GO:0012507>)

GO:0071556 : integral component of lumenal side of endoplasmic reticulum membrane

(<https://www.ebi.ac.uk/QuickGO/term/GO:0071556>)

GO:0005765 : lysosomal membrane (<https://www.ebi.ac.uk/QuickGO/term/GO:0005765>)

GO:0042613 : MHC class II protein complex

(<https://www.ebi.ac.uk/QuickGO/term/GO:0042613>)

GO:0032588 : trans-Golgi network membrane

(<https://www.ebi.ac.uk/QuickGO/term/GO:0032588>)

GO:0030658 : transport vesicle membrane

(<https://www.ebi.ac.uk/QuickGO/term/GO:0030658>)

GO:0031902 : late endosome membrane

(<https://www.ebi.ac.uk/QuickGO/term/GO:0031902>)

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

uncharacterized

Experimental Evidence

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

Main Reference

Variation at HLA-DRB1 is associated with resistance to enteric fever. (2014) (<https://pubmed.ncbi.nlm.nih.gov/25383971>)

Authors

Dunstan SJ; Hue NT; Han B; Li Z; Tram TT; Sim KS; Parry CM; Chinh NT; Vinh H; Lan NP; Thieu NT; Vinh PV; Koirala S; Dongol S; Arjyal A; Karkey A; Shilpakar O; Dolecek C; Foo JN; Phuong le T; Lanh MN; Do T; Aung T; Hon DN; Teo YY; Hibberd ML; Anders KL; Okada Y; Raychaudhuri S; Simmons CP; Baker S; de Bakker PI; Basnyat B; Hien TT; Farrar JJ; Khor CC

Abstract

Enteric fever affects more than 25 million people annually and results from systemic infection with *Salmonella enterica* serovar Typhi or Paratyphi pathovars A, B or C(1). We conducted a genome-wide association study of 432 individuals with blood culture-confirmed enteric fever and 2,011 controls from Vietnam. We observed strong association at rs7765379 (odds ratio (OR) for the minor allele = 0.18,  $P = 4.5 \times 10^{-10}$ ), a marker mapping to the HLA class II region, in proximity to HLA-DQB1 and HLA-DRB1. We replicated this association in 595 enteric fever cases and 386 controls from Nepal and also in a second independent collection of 151 cases and 668 controls from Vietnam. Imputation-based fine-mapping across the extended MHC region showed that the classical HLA-DRB1\*04:05 allele (OR = 0.14,  $P = 2.60 \times 10^{-11}$ ) could entirely explain the association at rs7765379, thus implicating HLA-DRB1 as a major contributor to resistance against enteric fever, presumably through antigen presentation.

Additional References

## RELATED GEPHE

Related Genes

10 (ATP2B4, CCL3L1, Duffy, Glucose-6-phosphate dehydrogenase (G6PD), Glycophorin GYPA-GYPB-GYPE cluster, hemoglobin; HBB, 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

