GEPHE SUMMARY

Gephebase Gene
HLA-DRB1 (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^HLA-DRB1^#gephebase-summary-title)

GepheID
GP00000479

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 (typhoid fever) (https://www.gephebase.org/search-criteria?/and+Trait=^Pathogen resistance (typhoid fever)^#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
HLA-DRB1

Synonyms
-

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 (https://www.ebi.ac.uk/QuickGO/term/GO:0019886)
GO:0060333 : interferon-gamma-mediated signaling pathway (https://www.ebi.ac.uk/QuickGO/term/GO:0060333)

UniProtKB Homo sapiens
Q30167 (http://www.uniprot.org/uniprot/Q30167)

GenebankID or UniProtKB
()

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 Ã— 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 Ã— 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