GEPHE SUMMARY

Gephebase Gene

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

GepheID

GP00000240

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

Trait State in Taxon A

Homo sapiens - malarial sensitive

Trait State in Taxon B

Homo sapiens - malarial resistant

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

ACKR1

Synonyms

FY; Dfy; GPD; DARC; GpFy; CCBP1; CD234; WBCQ1; DARC/ACKR1

String

9606.ENSP00000357103 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9606.ENSP00000357103)

Sequence Similarities

Belongs to the G-protein coupled receptor 1 family. Atypical chemokine receptor subfamily.

GO - Molecular Function

GO:0004888 : transmembrane signaling receptor activity (https://www.ebi.ac.uk/QuickGO/term/GO:0004888)
GO:0019957 : C-C chemokine binding

UniProtKB Homo sapiens

Q16570 (http://www.uniprot.org/uniprot/Q16570)

GenebankID or UniProtKB

AL035403 (https://www.ncbi.nlm.nih.gov/nuccore/AL035403)

(https://www.ebi.ac.uk/QuickGO/term/GO:0019957)
GO:0004930 : G protein-coupled receptor activity
(https://www.ebi.ac.uk/QuickGO/term/GO:0004930)
GO:0038023 : signaling receptor activity
(https://www.ebi.ac.uk/QuickGO/term/GO:0038023)

GO - Biological Process

GO:0006952 : defense response (https://www.ebi.ac.uk/QuickGO/term/GO:0006952)
GO:0006954 : inflammatory response
(https://www.ebi.ac.uk/QuickGO/term/GO:0006954)
GO:0070098 : chemokine-mediated signaling pathway
(https://www.ebi.ac.uk/QuickGO/term/GO:0070098)
GO:0032642 : regulation of chemokine production
(https://www.ebi.ac.uk/QuickGO/term/GO:0032642)

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:0005769 : early endosome (https://www.ebi.ac.uk/QuickGO/term/GO:0005769)
GO:0055037 : recycling endosome (https://www.ebi.ac.uk/QuickGO/term/GO:0055037)

Presumptive Null

No (https://www.gephebase.org/search-criteria?/and+Presumptive Null=^No^#gephebase-summary-title)

Molecular Type

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

Aberration Type

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

Molecular Details of the Mutation

T to C substitution in 5′ region at pos -46

Experimental Evidence

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

Main Reference

Disruption of a GATA motif in the Duffy gene promoter abolishes erythroid gene expression in Duffy-negative individuals. (1995) (https://pubmed.ncbi.nlm.nih.gov/7663520)

Authors

Tournamille C; Colin Y; Cartron JP; Le Van Kim C

Abstract

The mRNA for the Duffy blood group antigen, the erythrocyte receptor for the Plasmodium vivax malaria parasite, has recently been cloned and shown to encode a widely expressed chemokine receptor. Here, we show that the Duffy antigen/chemokine receptor gene (DARC) is composed of a single exon and that most Duffy-negative blacks carry a silent FY*B allele with a single T to C substitution at nucleotide -46. This mutation impairs the promoter activity in erythroid cells by disrupting a binding site for the GATA1 erythroid transcription factor. With the recent characterization of the FY*A and FY*B alleles, these findings provide the molecular basis of the Duffy blood group system and an explanation for the erythroid-specific repression of the DARC gene in Duffy-negative individuals.

Additional References

RELATED GEPHE

Related Genes

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