

GEPHE SUMMARY

Gephebase Gene
Duffy

Entry Status
Published

GepheID
GP00000240

Main curator
Martin

PHENOTYPIC CHANGE

Trait Category
Physiology

Trait
Pathogen resistance (Plasmodium; malaria parasite) (malaria)

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

Taxon A

Latin Name
Homo sapiens

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)

NCBI Taxonomy ID
9606

is Taxon A an Intraspecies?
No

Taxon B

Latin Name
Homo sapiens

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)

NCBI Taxonomy ID
9606

is Taxon B an Intraspecies?
No

GENOTYPIC CHANGE

Generic Gene Name
ACKR1

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

String
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
GO:0019957 : C-C chemokine binding
GO:0004930 : G protein-coupled receptor activity
GO:0038023 : signaling receptor activity

GO - Biological Process
GO:0006952 : defense response
GO:0006954 : inflammatory response

UniProtKB Homo sapiens
Q16570

GenebankID or UniProtKB
AL035403

GO:0070098 : chemokine-mediated signaling pathway
GO:0032642 : regulation of chemokine production

GO - Cellular Component

GO:0016021 : integral component of membrane
GO:0005886 : plasma membrane
GO:0005769 : early endosome
GO:0055037 : recycling endosome

Presumptive Null

No

Molecular Type

Cis-regulatory

Aberration Type

SNP

Molecular Details of the Mutation

T to C substitution in 5' region at pos -46

Experimental Evidence

Candidate Gene

Main Reference

Disruption of a GATA motif in the Duffy gene promoter abolishes erythroid gene expression in Duffy-negative individuals. (1995)

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))

Related Haplotypes

No matches found.

COMMENTS