

GEPHE SUMMARY

Duffy (https://www.gephebase.org/search-criteria?/and+Gene+Gephebase~Duffy^#gephebase-summary-title)	Gephebase Gene	GP00000240	GepheID
Published	Entry Status	Martin	Main curator

PHENOTYPIC CHANGE

Physiology (https://www.gephebase.org/search-criteria?/and+Trait+Category~Physiology^#gephebase-summary-title)	Trait Category		
Pathogen resistance (Plasmodium; malaria parasite) (malaria) (https://www.gephebase.org/search-criteria?/and+Trait~Pathogen+resistance+(Plasmodium;malaria+parasite)+(malaria)^#gephebase-summary-title)	Trait		
Homo sapiens - malarial sensitive	Trait State in Taxon A		
Homo sapiens - malarial resistant	Trait State in Taxon B		
Data not curated	Ancestral State		
Intraspecific (https://www.gephebase.org/search-criteria?/and+Taxonomic+Status~Intraspecific^#gephebase-summary-title)	Taxonomic Status		
	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 sapien; Homo sapiens; 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 sapien; Homo sapiens; 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

ACKR1	Generic Gene Name	UniProtKB Homo sapiens
	Synonyms	Q16570 (http://www.uniprot.org/uniprot/Q16570)
FY; Dfy; GPD; DARC; GpFy; CCBP1; CD234; WBCQ1; DARC/ACKR1		GenebankID or UniProtKB
	String	AL035403 (https://www.ncbi.nlm.nih.gov/nucore/AL035403)
9606.ENSPO0000357103 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSPO0000357103)	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		

(<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>)

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

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

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

T to C substitution in 5' region at pos -46 Molecular Details of the Mutation

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

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>) Main Reference

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

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. Abstract

Additional References

RELATED GEPHE

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 Genes

No matches found. Related Haplotypes

EXTERNAL LINKS

COMMENTS