

HLA-DQA1 (https://www.ghpbase.org/search-criteria?/and+Gene Ghpbase=~HLA-DQA1~#ghpbase-summary-title)	Ghpbase Gene	GP0000478	GephID
Published	Entry Status	Martin	Main curator

Trait Category	Trait	Trait State in Taxon A	Trait State in Taxon B	Ancestral State	Taxonomic Status
Physiology (#https://www.gephebase.org/search-criteria?/and+Trait+Category=~Physiology ^#gephebase-summary-title)					
Histocompatibility (#https://www.gephebase.org/search-criteria?/and+Trait=~Histocompatibility ^#gephebase-summary-title)					
Primates (with focus on baboon intraspecific variation)					
Primates (with focus on baboon intraspecific variation)					
Data not curated					
Intraspecific (#https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=~Intraspecific ^#gephebase-summary-title)					

Taxon A		Taxon B	
Papio	Latin Name	Papio	Latin Name
(https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=~Papio~#gephebase-summary-title)		(https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=~Papio~#gephebase-summary-title)	
	Common Name		Common Name
baboons		baboons	
	Synonyms		Synonyms
baboons; Papio Erxleben, 1777		baboons; Papio Erxleben, 1777	
	Rank		Rank
genus		genus	
	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; Cercopithecoidea; Cercopithecidae; Cercopithecinae		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; Cercopithecoidea; Cercopithecidae; Cercopithecinae	
	Parent		Parent
Cercopithecinae () - (Rank: subfamily)		Cercopithecinae () - (Rank: subfamily)	
(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9528)		(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9528)	
	NCBI Taxonomy ID		NCBI Taxonomy ID
9554		9554	
(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9554)		(https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9554)	
	is Taxon A an Intraspecies?		is Taxon B an Intraspecies?
No		No	

	Generic Gene Name	UniProtKB Homo sapiens
HLA-DQA1		P01909 (http://www.uniprot.org/uniprot/P01909)
	Synonyms	GenebankID or UniProtKB
DQA1; DQ-A1; CELIAC1; HLA-DQA		CUQ94670 (https://www.ncbi.nlm.nih.gov/nuccore/CUQ94670)
	String	
-		
	Sequence Similarities	
Belongs to the MHC class II family.		
	GO - Molecular Function	
GO:0032395 : MHC class II receptor activity (https://www.ebi.ac.uk/QuickGO/term/GO:0032395)		
GO:0042605 : peptide antigen binding (https://www.ebi.ac.uk/QuickGO/term/GO:0042605)		
	GO - Biological Process	
GO:0006955 : immune response (https://www.ebi.ac.uk/QuickGO/term/GO:0006955)		
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>)
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:0016020 : membrane (<https://www.ebi.ac.uk/QuickGO/term/GO:0016020>)
GO:0005887 : integral component of plasma membrane
(<https://www.ebi.ac.uk/QuickGO/term/GO:0005887>)
GO:0030669 : clathrin-coated endocytic vesicle membrane
(<https://www.ebi.ac.uk/QuickGO/term/GO:0030669>)
GO:0010008 : endosome membrane
(<https://www.ebi.ac.uk/QuickGO/term/GO:0010008>)
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>)

	Presumptive Null
Unknown (https://www.gephebase.org/search-criteria?/and+Presumptive Null=~Unknown~#gephebase-summary-title)	
	Molecular Type
Cis-regulatory (https://www.gephebase.org/search-criteria?/and+Molecular Type=~Cis-regulatory~#gephebase-summary-title)	
	Aberration Type
Unknown (https://www.gephebase.org/search-criteria?/and+Aberration Type=~Unknown~#gephebase-summary-title)	
	Molecular Details of the Mutation
Promoter variation	
	Experimental Evidence
Candidate Gene (https://www.gephebase.org/search-criteria?/and+Experimental Evidence=~Candidate Gene~#gephebase-summary-title)	
	Main Reference
Ancient polymorphism and functional variation in the primate MHC-DQA1 5' cis-regulatory region. (2006) (https://pubmed.ncbi.nlm.nih.gov/17053068)	
	Authors
Loisel DA; Rockman MV; Wray GA; Altmann J; Alberts SC	
	Abstract
Precise regulation of MHC gene expression is critical to vertebrate immune surveillance and response. Polymorphisms in the 5' proximal promoter region of the human class II gene HLA-DQA1 have been shown to influence its transcriptional regulation and may contribute to the pathogenesis of autoimmune diseases. We investigated the evolutionary history of this cis-regulatory region by sequencing the DQA1 5' proximal promoter region in eight nonhuman primate species. We observed unexpectedly high levels of sequence variation and multiple strong signatures of balancing selection in this region. Specifically, the considerable DQA1 promoter region diversity was characterized by abundant shared (or trans-species) polymorphism and a pronounced lack of fixed differences between species. The majority of transcription factor binding sites in the DQA1 promoter region were polymorphic within species, and these binding site polymorphisms were commonly shared among multiple species despite evidence for negative selection eliminating a significant fraction of binding site mutations. We assessed the functional consequences of intraspecific promoter region diversity using a cell line-based reporter assay and detected significant differences among baboon DQA1 promoter haplotypes in their ability to drive transcription in vitro. The functional differentiation of baboon promoter haplotypes, together with the significant deviations from neutral sequence evolution, suggests a role for balancing selection in the evolution of DQA1 transcriptional regulation in primates.	
	Additional References

RELATED GEPHE

	Related Genes
No matches found.	
	Related Haplotypes
No matches found.	

EXTERNAL LINKS

COMMENTS

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