

GEPHE SUMMARY

SIGLEC17P (pseudogene) (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^SIGLEC17P (pseudogene)^#gephebase-summary-title)	Gephebase Gene GP00001053	GephelD Main curator
Published	Entry Status Martin	

PHENOTYPIC CHANGE

Trait Category			
Physiology (https://www.gephebase.org/search-criteria?/and+Trait Category=^Physiology^#gephebase-summary-title)	Trait		
Pathogen resistance (https://www.gephebase.org/search-criteria?/and+Trait=^Pathogen resistance^#gephebase-summary-title)	Trait State in Taxon A		
Other Primates	Trait State in Taxon B		
Homo sapiens	Ancestral State		
Taxon A	Taxonomic Status		
Interspecific (https://www.gephebase.org/search-criteria?/and+Taxonomic Status=^Interspecific^#gephebase-summary-title)			
Taxon A	Latin Name	Taxon B	Latin Name
Primates (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Primates^#gephebase-summary-title)		Homo sapiens (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Homo+sapiens^#gephebase-summary-title)	
-	Common Name		Common Name
Primate; Primates Linnaeus, 1758	Synonyms	human	Synonyms
order	Rank	human; man; Homo sapiens Linnaeus, 1758; Homo sapiens; Homo sapiens; Homo sapien; Homo sapiens; Homo sapien; Homo sapien; Homo sapience; Homo sapiense; Homo sapients; Homo sapines; Homo spaiens; Homo spiens; Homo sapiens	Rank
cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria; Euarchontoglires	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	Lineage
Euarchontoglires () - (Rank: superorder) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 314146)	Parent	Homo () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9605)	Parent
9443 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9443)	NCBI Taxonomy ID		NCBI Taxonomy ID
No	is Taxon A an Infraspecies?	9606 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9606)	is Taxon B an Infraspecies?
		No	

GENOTYPIC CHANGE

SIGLEC13	Generic Gene Name Q64JA4 (http://www.uniprot.org/uniprot/Q64JA4)	UniProtKB Pan troglodytes
SIGLEC-13	Synonyms 0	GenebankID or UniProtKB
9598.ENSPTRP00000054310 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9598.ENSPTRP00000054310)	String	
Belongs to the immunoglobulin superfamily. SIGLEC (sialic acid binding Ig-like lectin) family.	Sequence Similarities	
GO - Molecular Function GO:0030246 : carbohydrate binding (https://www.ebi.ac.uk/QuickGO/term/GO:0030246)		
GO - Biological Process GO:0007155 : cell adhesion (https://www.ebi.ac.uk/QuickGO/term/GO:0007155)		

GO - Cellular Component

GO:0016021 : integral component of membrane

(<https://www.ebi.ac.uk/QuickGO/term/GO:0016021>)

Presumptive Null

Yes (<https://www.gephebase.org/search-criteria?/and+Presumptive+Null=%Yes%#gephebase-summary-title>)

Molecular Type

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

Aberration Type

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

Deletion Size

1-9 bp

Molecular Details of the Mutation

1bp deletion resulting in frame-shift (pseudogenization)

Experimental Evidence

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

Main Reference

Specific inactivation of two immunomodulatory SIGLEC genes during human evolution. (2012) (<https://pubmed.ncbi.nlm.nih.gov/22665810>)

Authors

Wang X; Mitra N; Secundino I; Banda K; Cruz P; Padler-Karavani V; Verhagen A; Reid C; Lari M; Rizzi E; Balsamo C; Corti G; De Bellis G; Longo L; Beggs W; Caramelli D; Tishkoff SA; Hayakawa T; Green ED; Mullikin JC; Nizet V; Bui J; Varki A

Abstract

Sialic acid-recognizing Ig-like lectins (Siglecs) are signaling receptors that modulate immune responses, and are targeted for interactions by certain pathogens. We describe two primate Siglecs that were rendered nonfunctional by single genetic events during hominin evolution after our common ancestor with the chimpanzee. SIGLEC13 was deleted by an Alu-mediated recombination event, and a single base pair deletion disrupted the ORF of SIGLEC17. Siglec-13 is expressed on chimpanzee monocytes, innate immune cells that react to bacteria. The human SIGLEC17P pseudogene mRNA is still expressed at high levels in human natural killer cells, which bridge innate and adaptive immune responses. As both resulting pseudogenes are homozygous in all human populations, we resurrected the originally encoded proteins and examined their functions. Chimpanzee Siglec-13 and the resurrected human Siglec-17 recruit a signaling adapter and bind sialic acids. Expression of either Siglec in innate immune cells alters inflammatory cytokine secretion in response to Toll-like receptor-4 stimulation. Both Siglecs can also be engaged by two potentially lethal sialylated bacterial pathogens of newborns and infants, agents with a potential impact on reproductive fitness. Neanderthal and Denisovan genomes show human-like sequences at both loci, corroborating estimates that the initial pseudogenization events occurred in the common ancestral population of these hominins. Both loci also show limited polymorphic diversity, suggesting selection forces predating the origin of modern humans. Taken together, these data suggest that genetic elimination of Siglec-13 and/or Siglec-17 represents signatures of infectious and/or other inflammatory selective processes contributing to population restrictions during hominin origins.

Additional References

RELATED GEPHE

Related Genes

13 (ATP2B4, CCL3L1, Duffy, Glucose-6-phosphate dehydrogenase (G6PD), Glycophorin GYPA-GYPB-GYPE cluster, hemoglobin; HBB, HLA-DRB1, Human Leukocyte Antigen-B (HLA-B), MARVELD3, Mitochondrial antiviral signaling (MAVS), SIGLEC13, TRIM5alpha, TRIM5alpha-CypA chimeric gene) (<https://www.gephebase.org/search-criteria?/or+Taxon+ID=%9443%and+Trait=Pathogen+resistance/or+Taxon+ID=%9606%and+Trait=Pathogen+resistance/and+groupHaplotypes=true#gephebase-summary-title>)

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS