

GEPHE SUMMARY

SIGLEC13 (https://www.gephebase.org/search-criteria?/and+Gene+Gephebase=^SIGLEC13^#gephebase-summary-title)	Gephebase Gene	GP00001052	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 (https://www.gephebase.org/search-criteria?/and+Trait=^Pathogen+resistance^#gephebase-summary-title)	Trait		
Other Primates	Trait State in Taxon A		
Homo sapiens	Trait State in Taxon B		
Data not curated	Ancestral State		
Interspecific (https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=^Interspecific^#gephebase-summary-title)	Taxonomic Status		
	Taxon A	Taxon B	
Primates (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Primates^#gephebase-summary-title)	Latin Name	Homo sapiens (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Homo+sapiens^#gephebase-summary-title)	Latin Name
-	Common Name	human	Common Name
Primata; Primates Linnaeus, 1758	Synonyms	human; man; Homo sapiens Linnaeus, 1758; Home sapiens; Homo sampiens; Homo sapeins; Homo sapien; Homo sapians; Homo sapien; Homo sapience; Homo sapiense; Homo sapients; Homo sapines; Homo spaiens; Homo spiens; Humo sapiens	Synonyms
order	Rank	species	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	9606 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9606)	NCBI Taxonomy ID
No	is Taxon A an Intraspecies?	No	is Taxon B an Intraspecies?

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:0030246 : carbohydrate binding (https://www.ebi.ac.uk/QuickGO/term/GO:0030246)	GO - Molecular Function		
GO:0007155 : cell adhesion (https://www.ebi.ac.uk/QuickGO/term/GO:0007155)	GO - Biological Process		

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

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

Presumptive Null

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

Molecular Type

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

Aberration Type

Gene deletion by Alu-mediated recombination

Molecular Details of the Mutation

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

Experimental Evidence

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

Main Reference

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

Authors

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

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), SIGLEC17P (pseudogene), 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 Genes

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS