

## GEPHE SUMMARY

		Gephebase Gene		GepheID
SIGLEC17P (pseudogene) ( <a href="https://www.gephebase.org/search-criteria?/and+Gene">https://www.gephebase.org/search-criteria?/and+Gene</a> )			GP00001053	
Gephebase="SIGLEC17P (pseudogene)"#gephebase-summary-title)				Main curator
Published		Entry Status	Martin	

## PHENOTYPIC CHANGE

		Trait Category		
Physiology ( <a href="https://www.gephebase.org/search-criteria?/and+Trait">https://www.gephebase.org/search-criteria?/and+Trait</a> )				
Category="Physiology"#gephebase-summary-title)		Trait		
Pathogen resistance ( <a "="" href="https://www.gephebase.org/search-criteria?/and+Trait=">https://www.gephebase.org/search-criteria?/and+Trait="</a> )				
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 ( <a href="https://www.gephebase.org/search-criteria?/and+Taxonomic">https://www.gephebase.org/search-criteria?/and+Taxonomic</a> )				
Status="Interspecific"#gephebase-summary-title)				
Taxon A		Taxon B		
	Latin Name		Latin Name	
Primates		Homo sapiens		
( <a href="https://www.gephebase.org/search-criteria?/and+Taxon">https://www.gephebase.org/search-criteria?/and+Taxon</a> and		( <a href="https://www.gephebase.org/search-criteria?/and+Taxon">https://www.gephebase.org/search-criteria?/and+Taxon</a> and		
Synonyms="Primates"#gephebase-summary-title)		Synonyms="Homo		
-	Common Name	sapiens"#gephebase-summary-title)		Common Name
		human		
	Synonyms			Synonyms
Primata; Primates Linnaeus, 1758		human; man; Homo sapiens Linnaeus, 1758; Home sapiens; Homo sampiens; Homo sapeins;		
	Rank	Homo sapien; Homo sapians; Homo sapien; Homo sapience; Homo sapiense; Homo		
order		sapients; Homo sapines; Homo spaiens; Homo spiens; Humo sapiens		Rank
	Lineage	species		
cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia;				Lineage
Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii;		cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia;		
Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria;		Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii;		
Euarchontoglires		Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria;		
	Parent	Euarchontoglires; Primates; Haplorrhini; Simiiformes; Catarrhini; Hominoidea; Hominidae;		
Euarchontoglires () - (Rank: superorder)		Homininae; Homo		Parent
( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=314146">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=314146</a> )				
	NCBI Taxonomy ID	Homo () - (Rank: genus)		
9443		( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9605">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9605</a> )		
( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9443">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9443</a> )				NCBI Taxonomy ID
	is Taxon A an Intraspecies?	9606		
No		( <a href="https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9606">https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9606</a> )		is Taxon B an Intraspecies?
		No		

## GENOTYPIC CHANGE

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

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

Presumptive Null

Coding ([https://www.gephebase.org/search-criteria?/and+Molecular Type=~Coding^#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Molecular+Type=~Coding^#gephebase-summary-title))

Molecular Type

Deletion ([https://www.gephebase.org/search-criteria?/and+Aberration Type=~Deletion^#gephebase-summary-title](https://www.gephebase.org/search-criteria?/and+Aberration+Type=~Deletion^#gephebase-summary-title))

Aberration Type

1-9 bp

Deletion Size

1bp deletion resulting in frame-shift (pseudogenization)

Molecular Details of the Mutation

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

No matches found.

Related Haplotypes

## EXTERNAL LINKS

## COMMENTS