GEPHE SUMMARY

Gephebase Gene
SIGLEC17P (pseudogene) (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^SIGLEC17P (pseudogene)^#gephebase-summary-title)

GepheID
GP00001053

Entry Status
Published

Main curator
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
Primates (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Primates^#gephebase-summary-title)

Common Name
-

Synonyms
Primata; Primates Linnaeus, 1758

Rank
order

Lineage
cellular organisms; Eukaryota; Opisthokonta; Metazoa; Eumetazoa; Bilateria; Deuterostomia; Chordata; Craniata; Vertebrata; Gnathostomata; Teleostomi; Euteleostomi; Sarcopterygii; Dipnotetrapodomorpha; Tetrapoda; Amniota; Mammalia; Theria; Eutheria; Boreoeutheria; Euarchontoglires

Parent
Euarchontoglires () - (Rank: superorder) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 314146)

NCBI Taxonomy ID
9443 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9443)

is Taxon A an Infraspecies?
No

Taxon B

Latin Name
Homo sapiens (https://www.gephebase.org/search-criteria?/and+Taxon and Synonyms=^Homo sapiens^#gephebase-summary-title)

Common Name
human

Synonyms
human; man; Homo sapiens Linnaeus, 1758; Home sapiens; Homo sampiens; Homo sapeins; Homo sapian; Homo sapians; Homo sapien; Homo sapience; Homo sapiense; Homo sapients; Homo sapines; Homo spaiens; Homo spiens; Humo sapiens

Rank
species

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

Parent
Homo () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9605)

NCBI Taxonomy ID
9606 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id= 9606)

is Taxon B an Infraspecies?
No

GENOTYPIC CHANGE

Generic Gene Name
SIGLEC13

Synonyms
SIGLEC-13

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 (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)

UniProtKB Pan troglodytes
Q64JA4 (http://www.uniprot.org/uniprot/Q64JA4)

GenebankID or UniProtKB
()

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