GEPHE SUMMARY

Gephebase Gene

CCL3L1 (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^CCL3L1^#gephebase-summary-title)

GepheID

GP00000177

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 (HIV) (https://www.gephebase.org/search-criteria?/and+Trait=^Pathogen resistance (HIV)^#gephebase-summary-title)

Trait State in Taxon A

Homo sapiens

Trait State in Taxon B

Homo sapiens

Ancestral State

Taxon A

Taxonomic Status

Intraspecific (https://www.gephebase.org/search-criteria?/and+Taxonomic Status=^Intraspecific^#gephebase-summary-title)

Taxon A

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 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

CCL3L1

Synonyms

LD78; 464.2; SCYA3L; G0S19-2; SCYA3L1; D17S1718; LD78BETA

String

9606.ENSP00000408984 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier=9606.ENSP00000408984)

Sequence Similarities

Belongs to the intercrine beta (chemokine CC) family.

GO - Molecular Function

GO:0048020 : CCR chemokine receptor binding (https://www.ebi.ac.uk/QuickGO/term/GO:0048020)
GO:0008009 : chemokine activity (https://www.ebi.ac.uk/QuickGO/term/GO:0008009)

GO - Biological Process

UniProtKB Homo sapiens

P16619 (http://www.uniprot.org/uniprot/P16619)

GenebankID or UniProtKB

X52149 (https://www.ncbi.nlm.nih.gov/nuccore/X52149)

GO:0007186 : G protein-coupled receptor signaling pathway (https://www.ebi.ac.uk/QuickGO/term/GO:0007186)
GO:0070374 : positive regulation of ERK1 and ERK2 cascade (https://www.ebi.ac.uk/QuickGO/term/GO:0070374)
GO:0008285 : negative regulation of cell proliferation (https://www.ebi.ac.uk/QuickGO/term/GO:0008285)
GO:0006954 : inflammatory response (https://www.ebi.ac.uk/QuickGO/term/GO:0006954)
GO:0071346 : cellular response to interferon-gamma (https://www.ebi.ac.uk/QuickGO/term/GO:0071346)
GO:0071347 : cellular response to interleukin-1 (https://www.ebi.ac.uk/QuickGO/term/GO:0071347)
GO:0071356 : cellular response to tumor necrosis factor (https://www.ebi.ac.uk/QuickGO/term/GO:0071356)
GO:0070098 : chemokine-mediated signaling pathway (https://www.ebi.ac.uk/QuickGO/term/GO:0070098)
GO:0019221 : cytokine-mediated signaling pathway (https://www.ebi.ac.uk/QuickGO/term/GO:0019221)
GO:0048247 : lymphocyte chemotaxis (https://www.ebi.ac.uk/QuickGO/term/GO:0048247)
GO:0002548 : monocyte chemotaxis (https://www.ebi.ac.uk/QuickGO/term/GO:0002548)
GO:0030593 : neutrophil chemotaxis (https://www.ebi.ac.uk/QuickGO/term/GO:0030593)
GO:0043547 : positive regulation of GTPase activity (https://www.ebi.ac.uk/QuickGO/term/GO:0043547)
GO:0050729 : positive regulation of inflammatory response (https://www.ebi.ac.uk/QuickGO/term/GO:0050729)

GO - Cellular Component

GO:0005576 : extracellular region (https://www.ebi.ac.uk/QuickGO/term/GO:0005576)
GO:0005615 : extracellular space (https://www.ebi.ac.uk/QuickGO/term/GO:0005615)

Presumptive Null

No (https://www.gephebase.org/search-criteria?/and+Presumptive Null=^No^#gephebase-summary-title)

Molecular Type

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

Aberration Type

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

Indel Size

unknown

Molecular Details of the Mutation

Copy number Variant

Experimental Evidence

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

Main Reference

The influence of CCL3L1 gene-containing segmental duplications on HIV-1/AIDS susceptibility. (2005) (https://pubmed.ncbi.nlm.nih.gov/15637236)

Authors

Gonzalez E; Kulkarni H; Bolivar H; Mangano A; Sanchez R; Catano G; Nibbs RJ; Freedman BI; Quinones MP; Bamshad MJ; Murthy KK; Rovin BH; Bradley W; Clark RA; Anderson SA; O'connell RJ; Agan BK; Ahuja SS; Bologna R; Sen L; Dolan MJ; Ahuja SK

Abstract

Segmental duplications in the human genome are selectively enriched for genes involved in immunity, although the phenotypic consequences for host defense are unknown. We show that there are significant interindividual and interpopulation differences in the copy number of a segmental duplication encompassing the gene encoding CCL3L1 (MIP-1alphaP), a potent human immunodeficiency virus-1 (HIV-1)-suppressive chemokine and ligand for the HIV coreceptor CCR5. Possession of a CCL3L1 copy number lower than the population average is associated with markedly enhanced HIV/acquired immunodeficiency syndrome (AIDS) susceptibility. This susceptibility is even greater in individuals who also possess disease-accelerating CCR5 genotypes. This relationship between CCL3L1 dose and altered HIV/AIDS susceptibility points to a central role for CCL3L1 in HIV/AIDS pathogenesis and indicates that differences in the dose of immune response genes may constitute a genetic basis for variable responses to infectious diseases.

Additional References

RELATED GEPHE

Related Genes

10 (ATP2B4, Duffy, Glucose-6-phosphate dehydrogenase (G6PD), Glycophorin GYPA-GYPB-GYPE cluster, hemoglobin; HBB, HLA-DRB1, Human Leukocyte Antigen-B (HLA-B), MARVELD3, SIGLEC13, SIGLEC17P (pseudogene)) (https://www.gephebase.org/search-criteria?/or+Taxon ID=^9606^/and+Trait=Pathogen resistance/and+groupHaplotypes=true#gephebase-summary-title)

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS