

CCL3L1 (https://www.gephebase.org/search-criteria?/and+GeneGephebase=~CCL3L1*#gephebase-summary-title)	Gephebase Gene	GP00000177	GepheID
Published	Entry Status	Martin	Main curator

Physiology (https://www.gephebase.org/search-criteria?/and+Trait+Category=^Physiology^#gephebase-summary-title)		Trait Category	
Pathogen resistance (HIV) (https://www.gephebase.org/search-criteria?/and+Trait=^Pathogen resistance (HIV)^#gephebase-summary-title)		Trait	
Homo sapiens		Trait State in Taxon A	
Homo sapiens		Trait State in Taxon B	
Taxon A		Ancestral State	
Intraspecific (https://www.gephebase.org/search-criteria?/and+Taxonomic+Status=^Intraspecific^#gephebase-summary-title)		Taxonomic Status	
Taxon A		Taxon B	
Homo sapiens (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Homo sapiens^#gephebase-summary-title)	Latin Name	Homo sapiens (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Homo sapiens^#gephebase-summary-title)	Latin Name
human	Common Name	human	Common Name
human; man; Homo sapiens Linnaeus, 1758; Home sapiens; Homo sampiens; Homo sapeins; Homo sapien; Homo sapians; Homo sapien; Homo sapience; Homo sapience; Homo sapients; Homo sapines; Homo spaiens; Homo spiens; Humo sapiens	Synonyms	human; man; Homo sapiens Linnaeus, 1758; Home sapiens; Homo sampiens; Homo sapeins; Homo sapien; Homo sapians; Homo sapien; Homo sapience; Homo sapience; Homo sapients; Homo sapines; Homo spaiens; Homo spiens; Humo sapiens	Synonyms
species	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; Primates; Haplorrhini; Simiiformes; Catarrhini; Hominoidea; Hominidae; Homininae; Homo	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
Homo () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9605)	Parent	Homo () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9605)	Parent
9606 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9606)	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?

CCL3L1	Generic Gene Name	P16619 (http://www.uniprot.org/uniprot/P16619)	UniProtKB Homo sapiens
LD78; 464.2; SCYA3L; G0S19-2; SCYA3L1; D17S1718; LD78BETA	Synonyms	X52149 (https://www.ncbi.nlm.nih.gov/nucore/X52149)	GenebankID or UniProtKB
9606.ENSP00000408984 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSP00000408984)	String		
Belongs to the intercrine beta (chemokine CC) family.	Sequence Similarities		
GO:0048020 : CCR chemokine receptor binding (https://www.ebi.ac.uk/QuickGO/term/GO:0048020)	GO - Molecular Function		
GO:0008009 : chemokine activity (https://www.ebi.ac.uk/QuickGO/term/GO:0008009)	GO - Biological Process		

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](https://www.gephebase.org/search-criteria?/and+Presumptive Null=))

Molecular Type

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

Aberration Type

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

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](https://www.gephebase.org/search-criteria?/and+Experimental Evidence=))

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](https://www.gephebase.org/search-criteria?/or+Taxon ID=))

Related Haplotypes

No matches found.

EXTERNAL LINKS

COMMENTS

