GEPHE SUMMARY

Gephebase Gene

Human Leukocyte Antigen-B (HLA-B) (https://www.gephebase.org/search-criteria?/and+Gene Gephebase=^Human Leukocyte Antigen-B (HLA-B)^#gephebase-summary-title)

Entry Status

Published

GephelD

GP00000492

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

Trait State in Taxon A

Homo sapiens

Trait State in Taxon B

Homo sapiens; HIV controllers

Ancestral State

Data not curated

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

HLA-B

Synonyms

AS; HLAB; B-4901

String

9606.ENSP00000399168 (http://string-db.org/newstring_cgi/show_network_section.pl?identifier= 9606.ENSP00000399168)

Sequence Similarities

Belongs to the MHC class I family.

GO - Molecular Function

GO:0005102 : signaling receptor binding (https://www.ebi.ac.uk/QuickGO/term/GO:0005102)
GO:0042605 : peptide antigen binding

UniProtKB Homo sapiens

P01889 (http://www.uniprot.org/uniprot/P01889)

GenebankID or UniProtKB

X64454 (https://www.ncbi.nlm.nih.gov/nuccore/X64454)

(https://www.ebi.ac.uk/QuickGO/term/GO:0042605)

GO - Biological Process

GO:0006955 : immune response (https://www.ebi.ac.uk/QuickGO/term/GO:0006955)
GO:0043312 : neutrophil degranulation
(https://www.ebi.ac.uk/QuickGO/term/GO:0043312)
GO:0060333 : interferon-gamma-mediated signaling pathway
(https://www.ebi.ac.uk/QuickGO/term/GO:0060333)
GO:0002479 : antigen processing and presentation of exogenous peptide antigen via MHC class I, TAP-dependent (https://www.ebi.ac.uk/QuickGO/term/GO:0002479)
GO:0002480 : antigen processing and presentation of exogenous peptide antigen via MHC class I, TAP-independent (https://www.ebi.ac.uk/QuickGO/term/GO:0002480)
GO:0002474 : antigen processing and presentation of peptide antigen via MHC class I
(https://www.ebi.ac.uk/QuickGO/term/GO:0002474)
GO:0042270 : protection from natural killer cell mediated cytotoxicity
(https://www.ebi.ac.uk/QuickGO/term/GO:0042270)
GO:2001198 : regulation of dendritic cell differentiation
(https://www.ebi.ac.uk/QuickGO/term/GO:2001198)
GO:0050776 : regulation of immune response
(https://www.ebi.ac.uk/QuickGO/term/GO:0050776)
GO:0032655 : regulation of interleukin-12 production
(https://www.ebi.ac.uk/QuickGO/term/GO:0032655)
GO:0032675 : regulation of interleukin-6 production
(https://www.ebi.ac.uk/QuickGO/term/GO:0032675)
GO:0002667 : regulation of T cell anergy
(https://www.ebi.ac.uk/QuickGO/term/GO:0002667)
GO:0060337 : type I interferon signaling pathway
(https://www.ebi.ac.uk/QuickGO/term/GO:0060337)
GO:0016032 : viral process (https://www.ebi.ac.uk/QuickGO/term/GO:0016032)

GO - Cellular Component

GO:0005886 : plasma membrane (https://www.ebi.ac.uk/QuickGO/term/GO:0005886)
GO:0000139 : Golgi membrane (https://www.ebi.ac.uk/QuickGO/term/GO:0000139)
GO:0016020 : membrane (https://www.ebi.ac.uk/QuickGO/term/GO:0016020)
GO:0070062 : extracellular exosome (https://www.ebi.ac.uk/QuickGO/term/GO:0070062)
GO:0005887 : integral component of plasma membrane
(https://www.ebi.ac.uk/QuickGO/term/GO:0005887)
GO:0005794 : Golgi apparatus (https://www.ebi.ac.uk/QuickGO/term/GO:0005794)
GO:0005783 : endoplasmic reticulum
(https://www.ebi.ac.uk/QuickGO/term/GO:0005783)
GO:0009986 : cell surface (https://www.ebi.ac.uk/QuickGO/term/GO:0009986)
GO:0031901 : early endosome membrane
(https://www.ebi.ac.uk/QuickGO/term/GO:0031901)
GO:0055038 : recycling endosome membrane
(https://www.ebi.ac.uk/QuickGO/term/GO:0055038)
GO:0012507 : ER to Golgi transport vesicle membrane
(https://www.ebi.ac.uk/QuickGO/term/GO:0012507)
GO:0071556 : integral component of lumenal side of endoplasmic reticulum membrane
(https://www.ebi.ac.uk/QuickGO/term/GO:0071556)
GO:0042612 : MHC class I protein complex
(https://www.ebi.ac.uk/QuickGO/term/GO:0042612)
GO:0030670 : phagocytic vesicle membrane
(https://www.ebi.ac.uk/QuickGO/term/GO:0030670)
GO:0030667 : secretory granule membrane
(https://www.ebi.ac.uk/QuickGO/term/GO:0030667)

Mutation #1

Presumptive Null

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

Molecular Type

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

Aberration Type

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

SNP Coding Change

Nonsynonymous

Molecular Details of the Mutation

Arg62Gly

Experimental Evidence

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

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	Arg	Gly	62

Main Reference

The major genetic determinants of HIV-1 control affect HLA class I peptide presentation. (2010) (https://pubmed.ncbi.nlm.nih.gov/21051598)

Authors

; Pereyra F; Jia X; McLaren PJ; Telenti A; de Bakker PI; Walker BD; Ripke S; Brumme CJ; Pulit SL; Carrington M; Kadie CM; Carlson JM; Heckerman D; Graham RR; Plenge RM; Deeks SG; Gianniny L; Crawford G; Sullivan J; Gonzalez E; Davies L; Camargo A; Moore JM; Beattie N; Gupta S; Crenshaw A; Burtt NP; Guiducci C; Gupta N; Gao X; Qi Y; Yuki Y; Piechocka-Trocha A; Cutrell E; Rosenberg R; Moss KL; Lemay P; O'Leary J; Schaefer T; Verma P; Toth I; Block B; Baker B; Rothchild A; Lian J; Proudfoot J; Alvino DM; Vine S; Addo MM; Allen TM; Altfeld M; Henn MR; Le Gall S; Streeck H; Haas DW; Kuritzkes DR; Robbins GK; Shafer RW; Gulick RM; Shikuma CM; Haubrich R; Riddler S; Sax PE; Daar ES; Ribaudo HJ; Agan B; Agarwal S; Ahern RL; Allen BL; Altidor S; Altschuler EL; Ambardar S; Anastos K; Anderson B; Anderson V; Andrady U; Antoniskis D; Bangsberg D; Barbaro D; Barrie W; Bartczak J; Barton S; Basden P; Basgoz N; Bazner S; Bellos NC; Benson AM; Berger J; Bernard NF; Bernard AM; Birch C; Bodner SJ; Bolan RK; Boudreaux ET; Bradley M; Braun JF; Brndjar JE; Brown SJ; Brown K; Brown ST; Burack J; Bush LM; Cafaro V; Campbell O; Campbell J; Carlson RH; Carmichael JK; Casey KK; Cavacuiti C; Celestin G; Chambers ST; Chez N; Chirch LM; Cimoch PJ; Cohen D; Cohn LE; Conway B; Cooper DA; Cornelson B; Cox DT; Cristofano MV; Cuchural G; Czartoski JL; Dahman JM; Daly JS; Davis BT; Davis K; Davod SM; DeJesus E; Dietz CA; Dunham E; Dunn ME; Ellerin TB; Eron JJ; Fangman JJ; Farel CE; Ferlazzo H; Fidler S; Fleenor-Ford A; Frankel R; Freedberg KA; French NK; Fuchs JD; Fuller JD; Gaberman J; Gallant JE; Gandhi RT; Garcia E; Garmon D; Gathe JC; Gaultier CR; Gebre W; Gilman FD; Gilson I; Goepfert PA; Gottlieb MS; Goulston C; Groger RK; Gurley TD; Haber S; Hardwicke R; Hardy WD; Harrigan PR; Hawkins TN; Heath S; Hecht FM; Henry WK; Hladek M; Hoffman RP; Horton JM; Hsu RK; Huhn GD; Hunt P; Hupert MJ; Illeman ML; Jaeger H; Jellinger RM; John M; Johnson JA; Johnson KL; Johnson H; Johnson K; Joly J; Jordan WC; Kauffman CA; Khanlou H; Killian RK; Kim AY; Kim DD; Kinder CA; Kirchner JT; Kogelman L; Kojic EM; Korthuis PT; Kurisu W; Kwon DS; LaMar M; Lampiris H; Lanzafame M; Lederman MM; Lee DM; Lee JM; Lee MJ; Lee ET; Lemoine J; Levy JA; Llibre JM; Liguori MA; Little SJ; Liu AY; Lopez AJ; Loutfy MR; Loy D; Mohammed DY; Man A; Mansour MK; Marconi VC; Markowitz M; Marques R; Martin JN; Martin HL; Mayer KH; McElrath MJ; McGhee TA; McGovern BH; McGowan K; McIntyre D; Mcleod GX; Menezes P; Mesa G; Metroka CE; Meyer-Olson D; Miller AO; Montgomery K; Mounzer KC; Nagami EH; Nagin I; Nahass RG; Nelson MO; Nielsen C; Norene DL; O'Connor DH; Ojikutu BO; Okulicz J; Oladehin OO; Oldfield EC; Olender SA; Ostrowski M; Owen WF; Pae E; Parsonnet J; Pavlatos AM; Perlmutter AM; Pierce MN; Pincus JM; Pisani L; Price LJ; Proia L; Prokesch RC; Pujet HC; Ramgopal M; Rathod A; Rausch M; Ravishankar J; Rhame FS; Richards CS; Richman DD; Rodes B; Rodriguez M; Rose RC; Rosenberg ES; Rosenthal D; Ross PE; Rubin DS; Rumbaugh E; Saenz L; Salvaggio MR; Sanchez WC; Sanjana VM; Santiago S; Schmidt W; Schuitemaker H; Sestak PM; Shalit P; Shay W; Shirvani VN; Silebi VI; Sizemore JM; Skolnik PR; Sokol-Anderson M; Sosman JM; Stabile P; Stapleton JT; Starrett S; Stein F; Stellbrink HJ; Sterman FL; Stone VE; Stone DR; Tambussi G; Taplitz RA; Tedaldi EM; Telenti A; Theisen W; Torres R; Tosiello L; Tremblay C; Tribble MA; Trinh PD; Tsao A; Ueda P; Vaccaro A; Valadas E; Vanig TJ; Vecino I; Vega VM; Veikley W; Wade BH; Walworth C; Wanidworanun C; Ward DJ; Warner DA; Weber RD; Webster D; Weis S; Wheeler DA; White DJ; Wilkins E; Winston A; Wlodaver CG; van't Wout A; Wright DP; Yang OO; Yurdin DL; Zabukovic BW; Zachary KC; Zeeman B; Zhao M

Abstract

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Additional References

Mutation #2

Presumptive Null

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

Molecular Type

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

Aberration Type

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

SNP Coding Change

Nonsynonymous

Molecular Details of the Mutation

Asn63Glu

Experimental Evidence

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

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	Asn	Glu	63

Main Reference

The major genetic determinants of HIV-1 control affect HLA class I peptide presentation. (2010) (https://pubmed.ncbi.nlm.nih.gov/21051598)

Authors

; Pereyra F; Jia X; McLaren PJ; Telenti A; de Bakker PI; Walker BD; Ripke S; Brumme CJ; Pulit SL; Carrington M; Kadie CM; Carlson JM; Heckerman D; Graham RR; Plenge RM; Deeks SG; Gianniny L; Crawford G; Sullivan J; Gonzalez E; Davies L; Camargo A; Moore JM; Beattie N; Gupta S; Crenshaw A; Burtt NP; Guiducci C; Gupta N; Gao X; Qi Y; Yuki Y; Piechocka-Trocha A; Cutrell E; Rosenberg R; Moss KL; Lemay P; O'Leary J; Schaefer T; Verma P; Toth I; Block B; Baker B; Rothchild A; Lian J; Proudfoot J; Alvino DM; Vine S; Addo MM; Allen TM; Altfeld M; Henn MR; Le Gall S; Streeck H; Haas DW; Kuritzkes DR; Robbins GK; Shafer RW; Gulick RM; Shikuma CM; Haubrich R; Riddler S; Sax PE; Daar ES; Ribaudo HJ; Agan B; Agarwal S; Ahern RL; Allen BL; Altidor S; Altschuler EL; Ambardar S; Anastos K; Anderson B; Anderson V; Andrady U; Antoniskis D; Bangsberg D; Barbaro D; Barrie W; Bartczak J; Barton S; Basden P; Basgoz N; Bazner S; Bellos NC; Benson AM; Berger J; Bernard NF; Bernard AM; Birch C; Bodner SJ; Bolan RK; Boudreaux ET; Bradley M; Braun JF; Brndjar JE; Brown SJ; Brown K; Brown ST; Burack J; Bush LM; Cafaro V; Campbell O; Campbell J; Carlson RH; Carmichael JK; Casey KK; Cavacuiti C; Celestin G; Chambers ST; Chez N; Chirch LM; Cimoch PJ; Cohen D; Cohn LE; Conway B; Cooper DA; Cornelson B; Cox DT; Cristofano MV; Cuchural G; Czartoski JL; Dahman JM; Daly JS; Davis BT; Davis K; Davod SM; DeJesus E; Dietz CA; Dunham E; Dunn ME; Ellerin TB; Eron JJ; Fangman JJ; Farel CE; Ferlazzo H; Fidler S; Fleenor-Ford A; Frankel R; Freedberg KA; French NK; Fuchs JD; Fuller JD; Gaberman J; Gallant JE; Gandhi RT; Garcia E; Garmon D; Gathe JC; Gaultier CR; Gebre W; Gilman FD; Gilson I; Goepfert PA; Gottlieb MS; Goulston C; Groger RK; Gurley TD; Haber S; Hardwicke R; Hardy WD; Harrigan PR; Hawkins TN; Heath S; Hecht FM; Henry WK; Hladek M; Hoffman RP; Horton JM; Hsu RK; Huhn GD; Hunt P; Hupert MJ; Illeman ML; Jaeger H; Jellinger RM; John M; Johnson JA; Johnson KL; Johnson H; Johnson K; Joly J; Jordan WC; Kauffman CA; Khanlou H; Killian RK; Kim AY; Kim DD; Kinder CA; Kirchner JT; Kogelman L; Kojic EM; Korthuis PT; Kurisu W; Kwon DS; LaMar M; Lampiris H; Lanzafame M; Lederman MM; Lee DM; Lee JM; Lee MJ; Lee ET; Lemoine J; Levy JA; Llibre JM; Liguori MA; Little SJ; Liu AY; Lopez AJ; Loutfy MR; Loy D; Mohammed DY; Man A; Mansour MK; Marconi VC; Markowitz M; Marques R; Martin JN; Martin HL; Mayer KH; McElrath MJ; McGhee TA; McGovern BH; McGowan K; McIntyre D; Mcleod GX; Menezes P; Mesa G; Metroka CE; Meyer-Olson D; Miller AO; Montgomery K; Mounzer KC; Nagami EH; Nagin I; Nahass RG; Nelson MO; Nielsen C; Norene DL; O'Connor DH; Ojikutu BO; Okulicz J; Oladehin OO; Oldfield EC; Olender SA; Ostrowski M; Owen WF; Pae E; Parsonnet J; Pavlatos AM; Perlmutter AM; Pierce MN; Pincus JM; Pisani L; Price LJ; Proia L; Prokesch RC; Pujet HC; Ramgopal M; Rathod A; Rausch M; Ravishankar J; Rhame FS; Richards CS; Richman DD; Rodes B; Rodriguez M; Rose RC; Rosenberg ES; Rosenthal D; Ross PE; Rubin DS; Rumbaugh E; Saenz L; Salvaggio MR; Sanchez WC; Sanjana VM; Santiago S; Schmidt W; Schuitemaker H; Sestak PM; Shalit P; Shay W; Shirvani VN; Silebi VI; Sizemore JM; Skolnik PR; Sokol-Anderson M; Sosman JM; Stabile P; Stapleton JT; Starrett S; Stein F; Stellbrink HJ; Sterman FL; Stone VE; Stone DR; Tambussi G; Taplitz RA; Tedaldi EM; Telenti A; Theisen W; Torres R;

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Additional References

Mutation #3

Presumptive Null

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

Molecular Type

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

Aberration Type

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

SNP Coding Change

Nonsynonymous

Molecular Details of the Mutation

Tyr/Asn 67 Met

Experimental Evidence

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

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	-	Met	67

Main Reference

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Authors

; Pereyra F; Jia X; McLaren PJ; Telenti A; de Bakker PI; Walker BD; Ripke S; Brumme CJ; Pulit SL; Carrington M; Kadie CM; Carlson JM; Heckerman D; Graham RR; Plenge RM; Deeks SG; Gianniny L; Crawford G; Sullivan J; Gonzalez E; Davies L; Camargo A; Moore JM; Beattie N; Gupta S; Crenshaw A; Burtt NP; Guiducci C; Gupta N; Gao X; Qi Y; Yuki Y; Piechocka-Trocha A; Cutrell E; Rosenberg R; Moss KL; Lemay P; O'Leary J; Schaefer T; Verma P; Toth I; Block B; Baker B; Rothchild A; Lian J; Proudfoot J; Alvino DM; Vine S; Addo MM; Allen TM; Altfeld M; Henn MR; Le Gall S; Streeck H; Haas DW; Kuritzkes DR; Robbins GK; Shafer RW; Gulick RM; Shikuma CM; Haubrich R; Riddler S; Sax PE; Daar ES; Ribaudo HJ; Agan B; Agarwal S; Ahern RL; Allen BL; Altidor S; Altschuler EL; Ambardar S; Anastos K; Anderson B; Anderson V; Andrady U; Antoniskis D; Bangsberg D; Barbaro D; Barrie W; Bartczak J; Barton S; Basden P; Basgoz N; Bazner S; Bellos NC; Benson AM; Berger J; Bernard NF; Bernard AM; Birch C; Bodner SJ; Bolan RK; Boudreaux ET; Bradley M; Braun JF; Brndjar JE; Brown SJ; Brown K; Brown ST; Burack J; Bush LM; Cafaro V; Campbell O; Campbell J; Carlson RH; Carmichael JK; Casey KK; Cavacuiti C; Celestin G; Chambers ST; Chez N; Chirch LM; Cimoch PJ; Cohen D; Cohn LE; Conway B; Cooper DA; Cornelson B; Cox DT; Cristofano MV; Cuchural G; Czartoski JL; Dahman JM; Daly JS; Davis BT; Davis K; Davod SM; DeJesus E; Dietz CA; Dunham E; Dunn ME; Ellerin TB; Eron JJ; Fangman JJ; Farel CE; Ferlazzo H; Fidler S; Fleenor-Ford A; Frankel R; Freedberg KA; French NK; Fuchs JD; Fuller JD; Gaberman J; Gallant JE; Gandhi RT; Garcia E; Garmon D; Gathe JC; Gaultier CR; Gebre W; Gilman FD; Gilson I; Goepfert PA; Gottlieb MS; Goulston C; Groger RK; Gurley TD; Haber S; Hardwicke R; Hardy WD; Harrigan PR; Hawkins TN; Heath S; Hecht FM; Henry WK; Hladek M; Hoffman RP; Horton JM; Hsu RK; Huhn GD; Hunt P; Hupert MJ; Illeman ML; Jaeger H; Jellinger RM; John M; Johnson JA; Johnson KL; Johnson H; Johnson K; Joly J; Jordan WC; Kauffman CA; Khanlou H; Killian RK; Kim AY; Kim DD; Kinder CA; Kirchner JT; Kogelman L; Kojic EM; Korthuis PT; Kurisu W; Kwon DS; LaMar M; Lampiris H; Lanzafame M; Lederman MM; Lee DM; Lee JM; Lee MJ; Lee ET; Lemoine J; Levy JA; Llibre JM; Liguori MA; Little SJ; Liu AY; Lopez AJ; Loutfy MR; Loy D; Mohammed DY; Man A; Mansour MK; Marconi VC; Markowitz M; Marques R; Martin JN; Martin HL; Mayer KH; McElrath MJ; McGhee TA; McGovern BH; McGowan K; McIntyre D; Mcleod GX; Menezes P; Mesa G; Metroka CE; Meyer-Olson D; Miller AO; Montgomery K; Mounzer KC; Nagami EH; Nagin I; Nahass RG; Nelson MO; Nielsen C; Norene DL; O'Connor DH; Ojikutu BO; Okulicz J; Oladehin OO; Oldfield EC; Olender SA; Ostrowski M; Owen WF; Pae E; Parsonnet J; Pavlatos AM; Perlmutter AM; Pierce MN; Pincus JM; Pisani L; Price LJ; Proia L; Prokesch RC; Pujet HC; Ramgopal M; Rathod A; Rausch M; Ravishankar J; Rhame FS; Richards CS; Richman DD; Rodes B; Rodriguez M; Rose RC; Rosenberg ES; Rosenthal D; Ross PE; Rubin DS; Rumbaugh E; Saenz L; Salvaggio MR; Sanchez WC; Sanjana VM; Santiago S; Schmidt W; Schuitemaker H; Sestak PM; Shalit P; Shay W; Shirvani VN; Silebi VI; Sizemore JM; Skolnik PR; Sokol-Anderson M; Sosman JM; Stabile P; Stapleton JT; Starrett S; Stein F; Stellbrink HJ; Sterman FL; Stone VE; Stone DR; Tambussi G; Taplitz RA; Tedaldi EM; Telenti A; Theisen W; Torres R; Tosiello L; Tremblay C; Tribble MA; Trinh PD; Tsao A; Ueda P; Vaccaro A; Valadas E; Vanig TJ; Vecino I; Vega VM; Veikley W; Wade BH; Walworth C; Wanidworanun C; Ward DJ; Warner DA; Weber RD; Webster D; Weis S; Wheeler DA; White DJ; Wilkins E; Winston A; Wlodaver CG; van't Wout A; Wright DP; Yang OO; Yurdin DL; Zabukovic BW; Zachary KC; Zeeman B; Zhao M

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Additional References

Mutation #4

Presumptive Null

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

Molecular Type

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

Aberration Type

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

SNP Coding Change

Nonsynonymous

Molecular Details of the Mutation

Lys/Arg 70 Ser

Experimental Evidence

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

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	-	Ser	70

Main Reference

The major genetic determinants of HIV-1 control affect HLA class I peptide presentation. (2010) (https://pubmed.ncbi.nlm.nih.gov/21051598)

Authors

; Pereyra F; Jia X; McLaren PJ; Telenti A; de Bakker PI; Walker BD; Ripke S; Brumme CJ; Pulit SL; Carrington M; Kadie CM; Carlson JM; Heckerman D; Graham RR; Plenge RM; Deeks SG; Gianniny L; Crawford G; Sullivan J; Gonzalez E; Davies L; Camargo A; Moore JM; Beattie N; Gupta S; Crenshaw A; Burtt NP; Guiducci C; Gupta N; Gao X; Qi Y; Yuki Y; Piechocka-Trocha A; Cutrell E; Rosenberg R; Moss KL; Lemay P; O'Leary J; Schaefer T; Verma P; Toth I; Block B; Baker B; Rothchild A; Lian J; Proudfoot J; Alvino DM; Vine S; Addo MM; Allen TM; Altfeld M; Henn MR; Le Gall S; Streeck H; Haas DW; Kuritzkes DR; Robbins GK; Shafer RW; Gulick RM; Shikuma CM; Haubrich R; Riddler S; Sax PE; Daar ES; Ribaudo HJ; Agan B; Agarwal S; Ahern RL; Allen BL; Altidor S; Altschuler EL; Ambardar S; Anastos K; Anderson B; Anderson V; Andrady U; Antoniskis D; Bangsberg D; Barbaro D; Barrie W; Bartczak J; Barton S; Basden P; Basgoz N; Bazner S; Bellos NC; Benson AM; Berger J; Bernard NF; Bernard AM; Birch C; Bodner SJ; Bolan RK; Boudreaux ET; Bradley M; Braun JF; Brndjar JE; Brown SJ; Brown K; Brown ST; Burack J; Bush LM; Cafaro V; Campbell O; Campbell J; Carlson RH; Carmichael JK; Casey KK; Cavacuiti C; Celestin G; Chambers ST; Chez N; Chirch LM; Cimoch PJ; Cohen D; Cohn LE; Conway B; Cooper DA; Cornelson B; Cox DT; Cristofano MV; Cuchural G; Czartoski JL; Dahman JM; Daly JS; Davis BT; Davis K; Davod SM; DeJesus E; Dietz CA; Dunham E; Dunn ME; Ellerin TB; Eron JJ; Fangman JJ; Farel CE; Ferlazzo H; Fidler S; Fleenor-Ford A; Frankel R; Freedberg KA; French NK; Fuchs JD; Fuller JD; Gaberman J; Gallant JE; Gandhi RT; Garcia E; Garmon D; Gathe JC; Gaultier CR; Gebre W; Gilman FD; Gilson I; Goepfert PA; Gottlieb MS; Goulston C; Groger RK; Gurley TD; Haber S; Hardwicke R; Hardy WD; Harrigan PR; Hawkins TN; Heath S; Hecht FM; Henry WK; Hladek M; Hoffman RP; Horton JM; Hsu RK; Huhn GD; Hunt P; Hupert MJ; Illeman ML; Jaeger H; Jellinger RM; John M; Johnson JA; Johnson KL; Johnson H; Johnson K; Joly J; Jordan WC; Kauffman CA; Khanlou H; Killian RK; Kim AY; Kim DD; Kinder CA; Kirchner JT; Kogelman L; Kojic EM; Korthuis PT; Kurisu W; Kwon DS; LaMar M; Lampiris H; Lanzafame M; Lederman MM; Lee DM; Lee JM; Lee MJ; Lee ET; Lemoine J; Levy JA; Llibre JM; Liguori MA; Little SJ; Liu AY; Lopez AJ; Loutfy MR; Loy D; Mohammed DY; Man A; Mansour MK; Marconi VC; Markowitz M; Marques R; Martin JN; Martin HL; Mayer KH; McElrath MJ; McGhee TA; McGovern BH; McGowan K; McIntyre D; Mcleod GX; Menezes P; Mesa G; Metroka CE; Meyer-Olson D; Miller AO; Montgomery K; Mounzer KC; Nagami EH; Nagin I; Nahass RG; Nelson MO; Nielsen C; Norene DL; O'Connor DH; Ojikutu BO; Okulicz J; Oladehin OO; Oldfield EC; Olender SA; Ostrowski M; Owen WF; Pae E; Parsonnet J; Pavlatos AM; Perlmutter AM; Pierce MN; Pincus JM; Pisani L; Price LJ; Proia L; Prokesch RC; Pujet HC; Ramgopal M; Rathod A; Rausch M; Ravishankar J; Rhame FS; Richards CS; Richman DD; Rodes B; Rodriguez M; Rose RC; Rosenberg ES; Rosenthal D; Ross PE; Rubin DS; Rumbaugh E; Saenz L; Salvaggio MR; Sanchez WC; Sanjana VM; Santiago S; Schmidt W; Schuitemaker H; Sestak PM; Shalit P; Shay W; Shirvani VN; Silebi VI; Sizemore JM; Skolnik PR; Sokol-Anderson M; Sosman JM; Stabile P; Stapleton JT; Starrett S; Stein F; Stellbrink HJ; Sterman FL; Stone VE; Stone DR; Tambussi G; Taplitz RA; Tedaldi EM; Telenti A; Theisen W; Torres R; Tosiello L; Tremblay C; Tribble MA; Trinh PD; Tsao A; Ueda P; Vaccaro A; Valadas E; Vanig TJ; Vecino I; Vega VM; Veikley W; Wade BH; Walworth C; Wanidworanun C; Ward DJ; Warner DA; Weber RD; Webster D; Weis S; Wheeler DA; White DJ; Wilkins E; Winston A; Wlodaver CG; van't Wout A; Wright DP; Yang OO; Yurdin DL; Zabukovic BW; Zachary KC; Zeeman B; Zhao M

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Additional References

Mutation #5

Presumptive Null

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

Molecular Type

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

Aberration Type

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

SNP Coding Change

Nonsynonymous

Molecular Details of the Mutation

Ser/Thr/Arg 97 Val

Experimental Evidence

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

	Taxon A	Taxon B	Position
Codon	-	-	-
Amino-acid	-	Val	97

Main Reference

The major genetic determinants of HIV-1 control affect HLA class I peptide presentation. (2010) (https://pubmed.ncbi.nlm.nih.gov/21051598)

Authors

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Additional References

RELATED GEPHE

Related Genes

10 (ATP2B4, CCL3L1, Duffy, Glucose-6-phosphate dehydrogenase (G6PD), Glycophorin GYPA-GYPB-GYPE cluster, hemoglobin; HBB, HLA-DRB1, 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

Needs curation @SeveralMutationsWithEffect