

GEPHE SUMMARY

	Gephebase Gene		GepheID
Human Leukocyte Antigen-B (HLA-B) (https://www.gephebase.org/search-criteria?/and+Gene=Gephebase+^Human+Leukocyte+Antigen-B+(HLA-B)^#gephebase-summary-title)		GP00000492	
	Entry Status	Martin	Main curator
Published			

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		Taxon B
	Latin Name		Latin Name
Homo sapiens (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Homo+sapiens^#gephebase-summary-title)		Homo sapiens (https://www.gephebase.org/search-criteria?/and+Taxon+and+Synonyms=^Homo+sapiens^#gephebase-summary-title)	
	Common Name		Common Name
human		human	
	Synonyms		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		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		Rank
species		species	
	Lineage		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		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		Parent
Homo () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9605)		Homo () - (Rank: genus) (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9605)	
	NCBI Taxonomy ID		NCBI Taxonomy ID
9606 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9606)		9606 (https://www.ncbi.nlm.nih.gov/Taxonomy/Browser/wwwtax.cgi?id=9606)	
	is Taxon A an Intraspecies?		is Taxon B an Intraspecies?
No		No	

GENOTYPIC CHANGE

	Generic Gene Name		UniProtKB Homo sapiens
HLA-B		P01889 (http://www.uniprot.org/uniprot/P01889)	
	Synonyms		GenebankID or UniProtKB
AS; HLAB; B-4901		X64454 (https://www.ncbi.nlm.nih.gov/nuccore/X64454)	
	String		
9606.ENSPO0000399168 (http://string-db.org/newstring.cgi/show_network_section.pl?identifier=9606.ENSPO0000399168)			
	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			

GO - Biological Process

GO - Cellular Component

(<https://www.ebi.ac.uk/QuickGO/term/GO:0030667>)

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; Burt 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; Ribaud 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; Bartzak 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; Ellerlin 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; Pujat 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](https://www.gephebase.org/search-criteria?/and+Presumptive Null=))

Molecular Type

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

Aberration Type

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

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

Taxon A		Taxon B		Position
Codon		-		-
Amino-acid		Asn		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; Burt 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; Ribaud 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; Bartzak 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; Ellerlin 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; Pujat 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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Abstract

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Additional References

Mutation #3

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

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

Aberration Type

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

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

	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; Burt 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; Ribaud 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; Cornelison 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; Ellerlin 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; Pujat HC; Ramgopal N; 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](https://www.gephebase.org/search-criteria?/and+Presumptive Null=))

Molecular Type

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

SNP (https://www.gephebase.org/search-criteria?/and+Aberration Type=^SNP^#gephebase-summary-title) Nonsynonymous Lys/Arg 70 Ser Association Mapping (https://www.gephebase.org/search-criteria?/and+Experimental Evidence=^Association Mapping^#gephebase-summary-title)	Aberration Type
	SNP Coding Change
	Molecular Details of the Mutation
	Experimental Evidence
	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; Burt 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; Ribaldo 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; Pujat 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 #5	
No (https://www.gephebase.org/search-criteria?/and+Presumptive Null=^No^#gephebase-summary-title)	Presumptive Null
Coding (https://www.gephebase.org/search-criteria?/and+Molecular Type=^Coding^#gephebase-summary-title)	Molecular Type
SNP (https://www.gephebase.org/search-criteria?/and+Aberration Type=^SNP^#gephebase-summary-title)	Aberration Type
Nonsynonymous	SNP Coding Change
Ser/Thr/Arg 97 Val	Molecular Details of the Mutation
Association Mapping (https://www.gephebase.org/search-criteria?/and+Experimental Evidence=^Association Mapping^#gephebase-summary-title)	Experimental Evidence

; 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; Burt 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; Ribaud 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; Ellerlin 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; Libre 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; Pujat 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

RELATED GEPHE

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>)

No matches found.

Related Genes

Related Haplotypes

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COMMENTS

Needs curation @SeveralMutationsWithEffect