BCR (Breakpoint Cluster Region Protein) is one of a number of genes affected by aberrant somatic hypermutation in B-cell lymphomas, which complicates the interpretation of mutations at this locus.

Relevance tier by entity

Entity	Tier	Description
BL	2-a	aSHM target; Although recurrent, the relevance of mutations in BL is tenuous
DLBCL	1-a	aSHM target and high-confidence DLBCL gene

Mutation incidence in large patient cohorts (GAMBL reanalysis)

Entity	source	frequency (%)
BL	GAMBL genomes+capture	6.00
BL	Thomas cohort	6.40
BL	Panea cohort	10.90
DLBCL	GAMBL genomes	4.97
DLBCL	Schmitz cohort	6.38
DLBCL	Reddy cohort	3.90
DLBCL	Chapuy cohort	4.27

Mutation pattern and selective pressure estimates

Entity	aSHM	Significant selection	dN/dS (missense)	dN/dS (nonsense)
BL	Yes	No	2.733	2.641
DLBCL	Yes	No	1.249	3.642
FL	Yes	No	7.439	0.000

aSHM regions

chr_name	hg19_start	hg19_end	region	regulatory_comment
chr22	23522060	23528313	TSS	NA

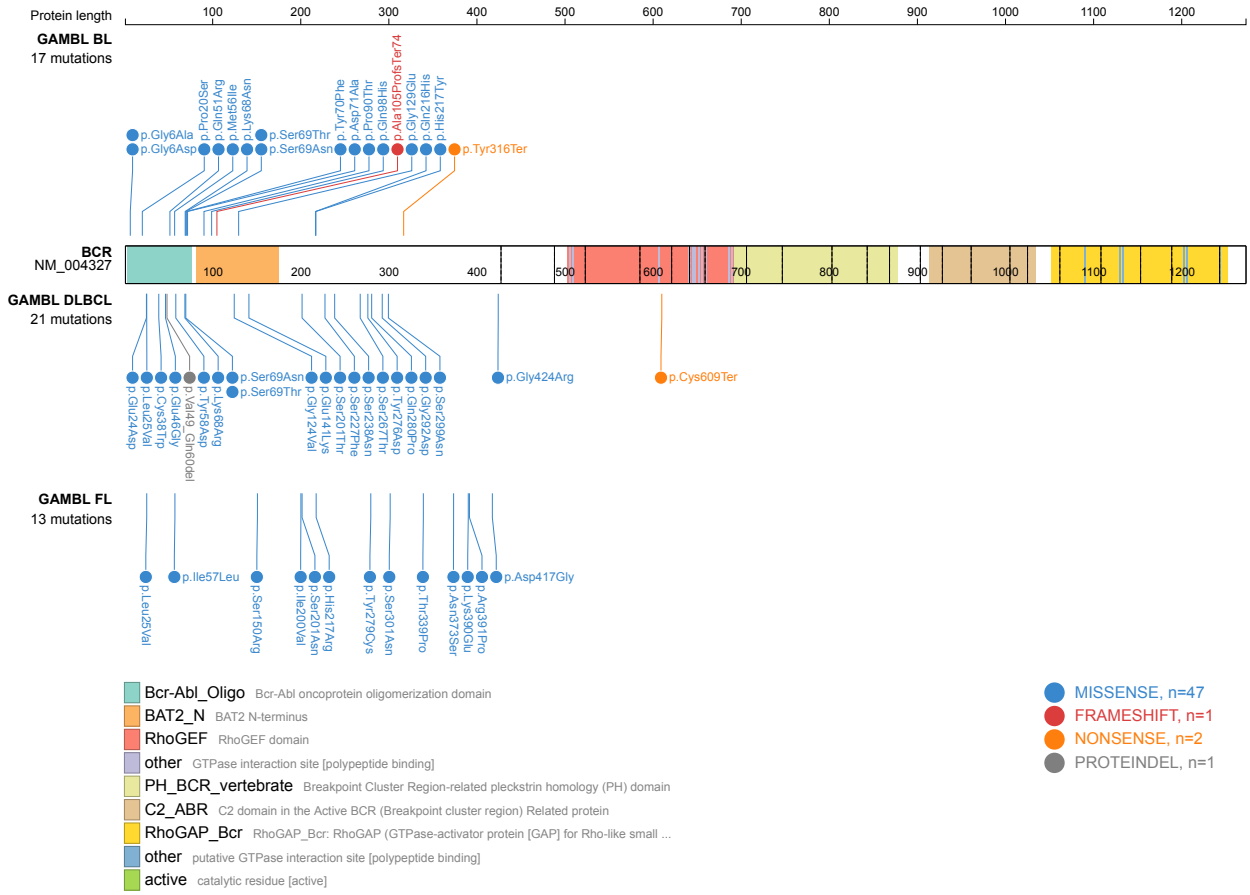
> [!NOTE] First described in BL in 2019 by Panea RI

BCR Hotspots

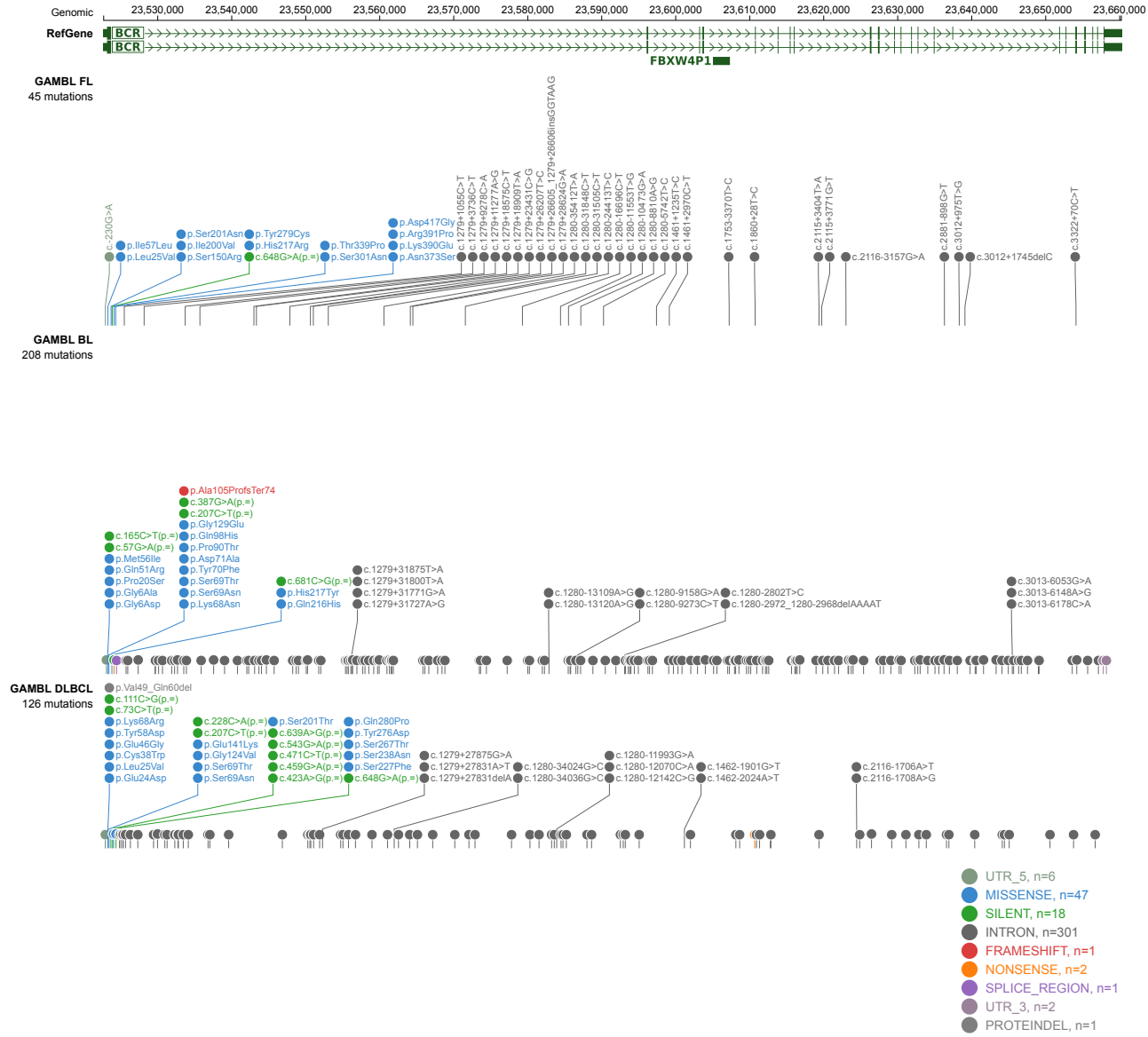
Chromosome	Coordinate (hg19)	ref>alt	HGVSp
chr22	23523350	A>G	K68R

Chromosome	Coordinate (hg19)	ref>alt	HGVSp
chr22	23523351	G>T	K68N
chr22	23523353	G>A	S69N
chr22	23523353	G>C	S69T
chr22	23523356	A>T	Y70F
chr22	23523359	A>C	D71A
chr22	23523374	G>A	G76D

View coding variants in ProteinPaint hg19 or hg38



View all variants in GenomePaint hg19 or hg38


Genomic
23,530,000
23,540,000
23,550,000
23,560,000
23,570,000
23,580,000
23,590,000
23,600,000
23,610,000
23,620,000
23,630,000
23,640,000
23,650,000
23,660,000
RefGene
BCR
BCR
FBXW4P1
GAMBL FL
45 mutations
c.-230G>A
p.Ile57Leu
p.Leu25Val
p.Ser201Asn
p.Ile200Val
p.Ser150Arg
p.Tyr279Cys
p.His217Arg
c.648G>A(p.=)
p.Thr339Pro
p.Ser301Asn
p.Asp417Gly
p.Arg391Pro
p.Lys390Glu
p.Asn373Ser
c.1279+1055C>T
c.1279+1733C>T
c.1279+9278C>A
c.1279+11277A>G
c.1279+18575C>T
c.1279+18909T>A
c.1279+23431C>G
c.1279+26207T>C
c.1279+26606insGGTAAG
c.1279+26605_1279+26606insGGTAAG
c.1279+28624G>A
c.1280-3541121>A
c.1280-31848C>T
c.1280-31855>T
c.1280-24413T>C
c.1280-16696C>T
c.1280-11553T>G
c.1280-10473G>A
c.1280-8810A>G
c.1280-5742T>C
c.1461+12357>C
c.1461+2970C>T
c.1753-3370T>C
c.1860+28T>C
c.2115-3404T>A
c.2115-3771G>T
c.2116-3157G>A
c.2881-898G>T
c.3012+975T>G
c.3012+1745delC
c.3322+70C>T
GAMBL BL
208 mutations
p.Ala105ProfsTer74
c.387G>A(p.=)
c.207C>T(p.=)
p.Gly129Glu
c.165C>T(p.=)
p.Gln98His
c.57G>A(p.=)
p.Pro90Thr
p.Met56Ile
p.Asp71Ala
p.Gln51Arg
p.Tyr70Phe
p.Pro20Ser
p.Ser69Thr
p.Gly6Ala
p.Ser69Asn
p.Gly6Asp
p.Lys68Asn
c.681C>G(p.=)
p.His217Tyr
p.Gln216His
c.1279+31875T>A
c.1279+31800T>A
c.1279+31771G>A
c.1279+31727A>G
c.1280-13109A>G
c.1280-13120A>G
c.1280-9158G>A
c.1280-9273C>T
c.1280-2802T>C
c.1280-2972_1280-2968delAAAAT
c.3013-6053G>A
c.3013-6148A>G
c.3013-6178C>A
GAMBL DLBCL
126 mutations
p.Val49_Gln60del
c.111C>G(p.=)
c.73C>T(p.=)
p.Lys68Arg
p.Tyr58Asp
p.Glu46Gly
p.Cys38Trp
p.Leu25Val
p.Glu24Asp
c.228C>A(p.=)
c.207C>T(p.=)
p.Glu141Lys
p.Gly124Val
p.Ser69Thr
p.Ser69Asn
p.Ser201Thr
c.639A>G(p.=)
c.543G>A(p.=)
c.471C>T(p.=)
c.459G>A(p.=)
c.423A>G(p.=)
p.Gln280Pro
p.Tyr276Asp
p.Ser267Thr
p.Ser238Asn
p.Ser227Phe
c.648G>A(p.=)
c.1279+27875G>A
c.1279+27831A>T
c.1279+27831delA
c.1280-11993G>A
c.1280-34024G>C
c.1280-34036G>C
c.1280-12070C>A
c.1280-12142C>G
c.1462-1901G>T
c.1462-2024A>T
c.2116-1706A>T
c.2116-1708A>G
UTR_5, n=6
MISSENSE, n=47
SILENT, n=18
INTRON, n=301
FRAMESHIFT, n=1
NONSENSE, n=2
SPLICE_REGION, n=1
UTR_3, n=2
PROTEINDEL, n=1