HNRNPU

Overview

HNRNPU 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	1-a	aSHM target and high-confidence BL gene
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	5.77
BL	Thomas cohort	6.40
BL	Panea cohort	8.90
DLBCL	GAMBL genomes	2.49
DLBCL	Schmitz cohort	3.40
DLBCL	Reddy cohort	2.40
DLBCL	Chapuy cohort	3.42

Mutation pattern and selective pressure estimates

Entity	aSHM	Significant selection	dN/dS (missense)	dN/dS (nonsense)
BL	Yes	Yes	1.081	27.052
DLBCL	Yes	No	1.712	0.000
FL	Yes	No	4.865	31.056

aSHM regions

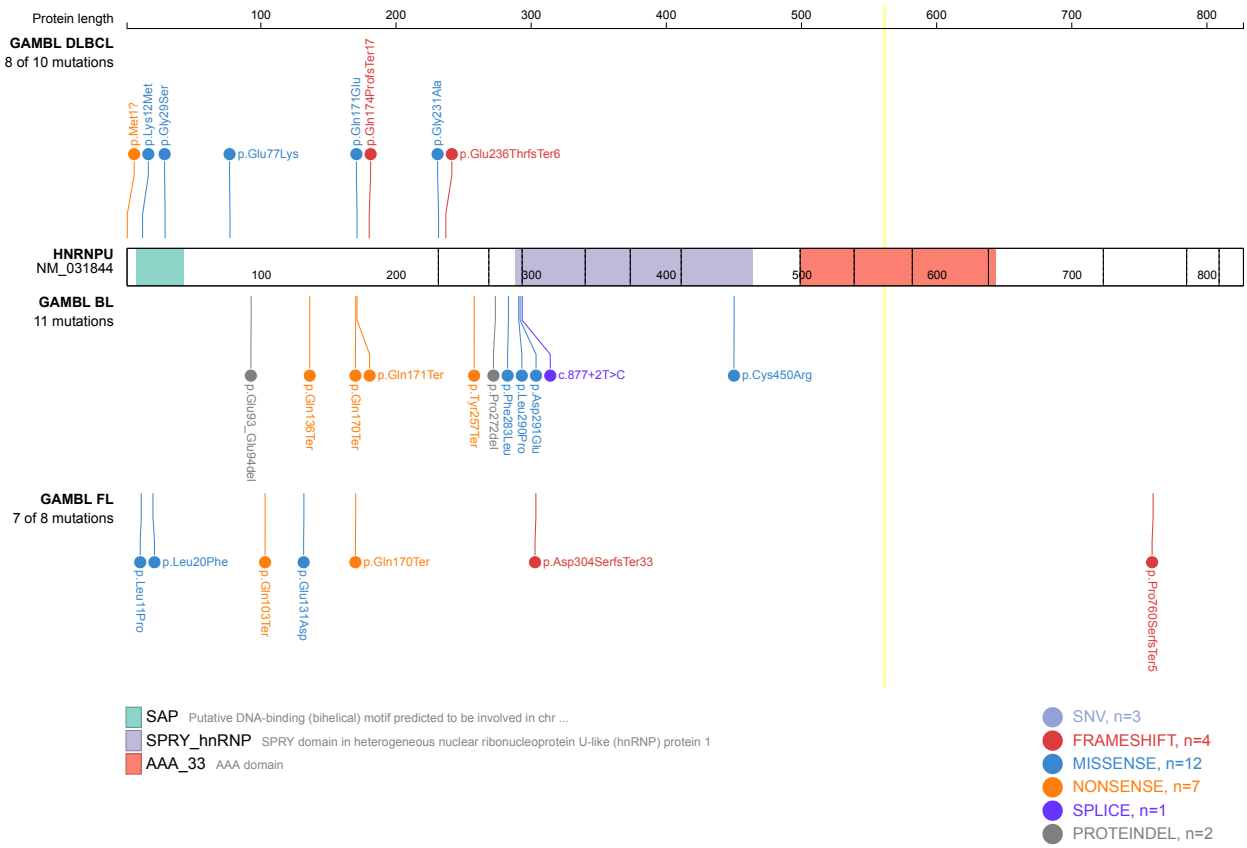
chr_name	hg19_start	hg19_end	region	regulatory_comment
chr1	245023502	245029083	TSS	NA

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

HNRNPU Hotspots

Chromosome	Coordinate (hg19)	ref>alt	HGVSp
chr1	245027102	G>A	Q170*
chr1	245027099	G>C	Q171E
chr1	245027099	G>A	Q171*

View coding variants in ProteinPaint hg19 or hg38



View all variants in GenomePaint hg19 or hg38


Genomic
245,014,000
245,016,000
245,018,000
245,020,000
245,022,000
245,024,000
245,026,000
RefGene
HNRNPU
HNRNPU
SNORA100
GAMBL FL
13 mutations
c.*54C>T
p.Pro760SerfsTer5
c.1744-5T>A
p.Asp304SerfsTer33
c.877+127A>C
c.691+17A>G
p.Gln170Ter
c.432G>A(p.=)
p.Glu131Asp
p.Gln103Ter
p.Leu20Phe
p.Leu11Pro
A>G
GAMBL DLBCL
19 mutations
A>G
T>C
c.*250T>A
c.1494+60G>A
c.878-932G>A
c.877+991C>T
p.Glu236ThrfsTer6
p.Gly231Ala
p.Gln174ProfsTer17
p.Gln171Glu
c.316C>T(p.=)
p.Glu77Lys
p.Gly29Ser
p.Lys12Met
c.31C>T(p.=)
c.51A>G(p.=)
c.-10G>A
p.Met1?
c.-110A>G
GAMBL BL
34 mutations
c.*881_*883delAGT
c.*811T>G
c.2353-69C>T
c.1495-10_1495-6delTTTTC
c.1495-98A>G
p.Cys450Arg
c.1230+224C>A
c.1018-151A>G
c.1017+108C>G
c.878-564G>A
c.877+743_877+744insAAA
c.877+376G>A
c.877+94A>G
c.877+105T>A
c.877+2T>C
c.877+3A>C
c.877+25T>G
p.Leu290Pro
p.Asp291Glu
p.Phe283Leu
p.Pro272del
c.822A>T(p.=)
c.803+24A>G
p.Tyr257Ter
c.692-119C>G
c.692-373C>T
c.691+349T>G
p.Gln170Ter
p.Gln171Ter
c.435C>T(p.=)
p.Gln136Ter
c.315G>A(p.=)
p.Glu93_Glu94del
c.-32G>T
UTR_3, n=4
FRAMESHIFT, n=4
SPLICE_REGION, n=3
INTRON, n=20
NONSENSE, n=7
SILENT, n=7
MISSENSE, n=12
SNV, n=3
UTR_5, n=3
SPLICE, n=1
PROTEINDEL, n=2