

<i>SAMD9L</i> variants (cDNA) NM_152703	<i>SAMD9L</i> variants (protein)	VAF in DNA from Fibroblasts	VAF in DNA from Blood	VAF in DNA Bone Marrow
c.2915T>C	p.Ile973Thr	54% (102 out of 190 reads)	41% (67 out of 164 reads)	47% (95 out of 216 reads)
c.3229C>T	p.Arg1077*	Not detected (0 out of 183 reads)	6% (8 out of 136 reads)	4% (8 out of 212 reads)
c.3456_3458del	p.Leu1153del	Not detected (0 out of 208 reads)	26% (41 out of 160 reads)	31% (68 out of 217 reads)

Table e-2: Distribution of *SAMD9L* variants in different tissues. VAF= Variant allele frequency