Supplemental Table 1

Gene name	Location	Variant classification	HGVSc	HGVSp	Consequence	CONSTITUTIONAL DNA			TUMOR DNA			TUMOR RNA
						Copy number	VAF (%)	Depth	Copy number	VAF (%)	Depth	VAF (%)
PMS2	Chr7	PV / SNV	NM_000088.4: c.695G>T	NP_000526.2: p.(Gly232Val)	Missense variant	**2**	**55**	**84**	.	50	147	68
NF1	Chr17	PV / insertion	NM_000267.3: c.2033dup	NP_000258.1: p.(Ile679Aspfs*21)	Frameshift variant	**2**	**20**	**119**	.	47	215	0
PMS2	Chr7	PV / SNV	NM_000535.7: c.2275+1G>A	.	Splice donor variant	**2**	**45**	**62**	.	29	139	0
TP53	Chr17	PV / SNV	NM_000546.6: c.817C>T	NP_000537.3: p.(Arg273Cys)	Missense variant	.	.	.	2	48	223	77
FANCA	Chr16	PV / SNV	NM_000135.4: c.3624C>T	NP_000126.2: p.(Ser1208=)	Splice region & synonymous variant	.	.	.	2	47	143	0
TGFBR2	Chr3	PV / SNV	NM_003242.6: c.1336G>A	NP_001020018.1: p.(Asp446Asn)	Missense variant	.	.	.	2	46	215	15
SETD2	Chr3	PV / insertion	NM_001349370.3: c.4087dup	NP_001336299.1: p.(Arg1363Lysfs*8)	Frameshift variant	.	.	.	2	45	244	22
SMO	Chr7	PV / SNV	NM_005631.5: c.1965G>A	NP_005622.1: p.(Trp655*)	Nonsense	.	.	.	2	44	224	42
KDM6A	ChrX	PV / SNV	NM_001291415.2: c.4207C>T	NP_001278344.1: p.(Arg1403*)	Nonsense	.	.	.	2.1	41	241	0
POLE	Chr12	PV / SNV	NM_006231.4 c.857C>G	NP_006222.2: p.(Pro286Arg)	Missense variant	.	.	.	2	41	115	58
ATR	Chr3	PV / SNV	NM_001184.4: c.7597C>T	NP_001175.2: p.(Arg2533*)	Nonsense	.	.	.	2	38	222	31
NF1	Chr17	PV / SNV	NM_000267.3: c.532G>T	NP_000258.1: p.(Glu178*)	Nonsense	.	.	.	2	37	141	50
APC	Chr5	PV / SNV	NM_000038.6: c.2626C>T	NP_000029.2: p.(Arg876*)	Nonsense	.	.	.	2	34	307	50
TP53	Chr17	PV / SNV	NM_001126112.3: c.586C>T	NP_001119584.1: p.(Arg196*)	Nonsense	.	.	.	2	30	253	0
PPP2R1A	Chr19	PV / SNV	NM_001363656.2: c.7C>T	NP_001350585.1: p.(Arg3Trp)	Missense variant	.	.	.	2	25	213	28
ARID1B	Chr6	PV / SNV	NM_001346813.1: c.2455C>T	NP_001333742.1: p.(Gln819*)	Nonsense	.	.	.	2	14	238	16

Abbreviation : VAF : variant allele frequency, PV : pathogenic variant, SVN : small nucleotide variant