

## ION REPORTER ANALYSIS RESULT AND IGV SNAPSHOT

Locus	Oncomi...	Oncomine G...	Genes	Amino Acid ...	Genot...	Type	Allele Ratio	Coverage
chr17:41246153	Deleterious	Loss-of-function	BRCA1	p.Tyr465Ter	A/C	SNV	A=0.5479, C=0.4521	2568
chr17:41245466			BRCA1	p.Ser694=	G/A	SNV	G=0.5164, A=0.4836	3999
chr17:41245237			BRCA1	p.Leu771=	A/G	SNV	A=0.5276, G=0.4724	2208
chr17:41244936			BRCA1	p.Pro871Leu	G/A	SNV	G=0.495, A=0.505	3998
chr17:41244435			BRCA1	p.Glu1038Gly	T/C	SNV	T=0.5188, C=0.4813	4000
chr17:41244000			BRCA1	p.Lys1183Arg	T/C	SNV	T=0.5053, C=0.4947	3998
chr17:41234470			BRCA1	p.Ser1436=	A/G	SNV	A=0.5241, G=0.4759	1595
chr17:41231516			BRCA1	p.?	C/T	SNV	C=0.5116, T=0.4884	2758
chr17:41223094			BRCA1	p.Ser1613Gly	T/C	SNV	T=0.5177, C=0.4823	3622
chr17:41219804			BRCA1	p.?	T/C	SNV	T=0.5171, C=0.4829	3185

## METHOD AND SEQUENCING METRICS BY NGS ANALYSIS

- Genomic DNA was extracted from EDTA peripheral blood sample, and quantified using Qubit Fluorometer. Approximately 20 ng DNA is used for target amplification. The amplified DNA is subjected to adapter ligation and Ion Xpress™ Barcode generation for specific library preparation. The generated high quality library is subjected to NGS on the ION S5 sequencing platform
- Ion torrent S5 sequencing produced 7,22,854 total reads in this sample, the mean read length was 113 bp. The average base coverage depth was 3252x, with a mean percentage of reads on target was 99.23%, and a uniformity of base coverage was 99.55%