Resequencing Report

Sample: LP9006283-DNA_A01

Report Date: 02/13/2015 18:48:11 (UTC)

Illumina_WGS_Gel, Version v3.0.8-39-gf08c608

Sample Information

Sample ID:	LP9006283-DNA_A01
Sample Name:	LP9006283-DNA_A01
Total PF Reads:	844,844,612
Percent Q30 Bases:	82.7%

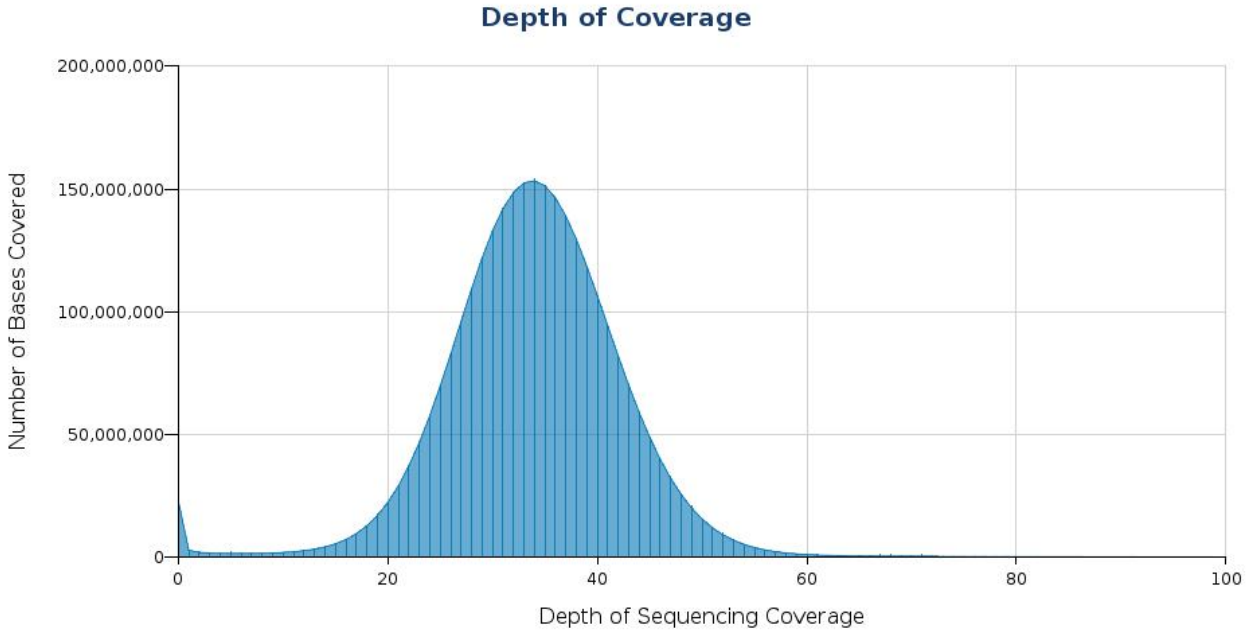
Read Level Statistics

Read	Total Aligned Reads	Percent Aligned Reads
1	408,459,374	96.7%
2	398,466,538	94.3%

Base Level Statistics

Read	Total Aligned Bases	Percent Aligned Bases	Mismatch Rate
1	60,187,932,343	95.0%	1.17%
2	57,407,403,872	90.6%	2.06%

Coverage Histogram (mean coverage 37.9)



Small Variants Summary

	SNVs	Insertions	Deletions
Total Passing	3,596,143	310,194	314,328
Percent Found in dbSNP	98.8%	79.4%	82.6%
Het/Hom Ratio	1.6	1.8	2.5
Ts/Tv Ratio	2.1	-	-

Variants by Sequence Context

	SNVs	Insertions	Deletions
In Genes	1,668,133	149,499	151,540
In Exons	50,471	3,222	3,062
In Coding Regions	22,161	262	302
In UTR Regions	28,310	2,960	2,760
In Splice Site Regions	3,190	329	282
In Mature microRNA	77	9	19

Genes include exons, introns and UTR regions. Exons include coding and UTR regions. UTR regions include 5' and 3' UTR regions. Splice site regions include regions annotated as splice acceptor, splice donor, splice site or splice region.

Variants by Consequence

	SNVs	Insertions	Deletions
Frameshifts	-	109	98
Non-synonymous	10,528	146	190
Synonymous	11,544	-	-
Stop Gained	70	0	0
Stop Lost	19	0	0

Variation consequences are calculated following the guidelines at http://uswest.ensembl.org/info/genome/variation/predicted_data.html#consequences

Structural Variants Summary

Variant Type	Total	In Genes
CNV	77	42
SV Insertions	1,985	971
SV Deletions	4,624	2,303
SV Tandem Duplications	97	49
SV Inversions	350	253

Fragment Length Summary

Fragment Length Median	Minimum	Maximum	Standard Deviation
481 bp	138 bp	877 bp	122 bp

Note: The minimum and maximum are calculated from values within approximately three standard deviations (excluding the lower and upper 0.15% of the data) to account for potential outliers.

Duplicate Information

Percent Duplicate Paired Reads
15.0%

Analysis Details

Settings

Setting Name	Value
Reference Genome	Homo sapiens (Ensembl GRCh37)
Annotation Source	Ensembl
Flag PCR Duplicates	True
SV Caller	Manta
CNV Caller	Canvas

Software Versions

Software	Version
Illumina_WGS_Gel (BaseSpace Workflow)	v3.0.8-39-gf08c608
Isis (Analysis Software)	2.5.55.16
SAMtools	0.1.19-isis-1.0.3
Isaac (Aligner)	
Isaac Variant Caller	starka-2.1.4.2
Manta (SV Caller)	0.23.1
Canvas (CNV Caller)	1.1.0.5
IONA (Annotation Service)	1.0.10.37

Data collections

Data Collection	Version
Annotation Dataset	72.5