Resequencing Report

Sample: LP9006281-DNA_A01

Report Date: 02/08/2015 02:18:16 (UTC)

Illumina_WGS_Gel, Version v3.0.8-39-gf08c608

Sample Information

Sample ID:	LP9006281-DNA_A01
Sample Name:	LP9006281-DNA_A01
Total PF Reads:	803,263,602
Percent Q30 Bases:	81.3%

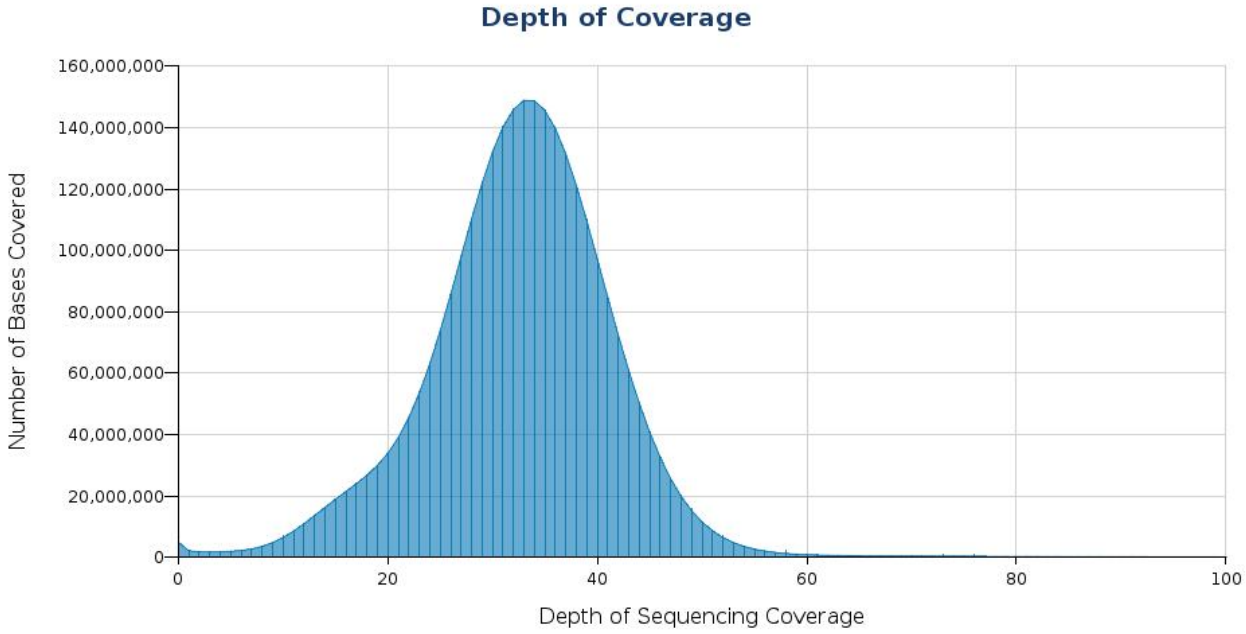
Read Level Statistics

Read	Total Aligned Reads	Percent Aligned Reads
1	384,370,424	95.7%
2	371,815,380	92.6%

Base Level Statistics

Read	Total Aligned Bases	Percent Aligned Bases	Mismatch Rate
1	56,123,161,008	93.2%	1.50%
2	53,555,957,141	88.9%	2.18%

Coverage Histogram (mean coverage 35.3)



Small Variants Summary

	SNVs	Insertions	Deletions
Total Passing	3,561,824	302,570	307,924
Percent Found in dbSNP	98.5%	79.8%	83.0%
Het/Hom Ratio	1.5	1.7	2.3
Ts/Tv Ratio	2.1	-	-

Variants by Sequence Context

	SNVs	Insertions	Deletions
In Genes	1,655,379	145,875	148,701
In Exons	50,425	3,251	3,040
In Coding Regions	22,110	277	290
In UTR Regions	28,315	2,974	2,750
In Splice Site Regions	3,173	345	274
In Mature microRNA	78	9	21

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	-	121	94
Non-synonymous	10,440	149	181
Synonymous	11,583	-	-
Stop Gained	64	1	0
Stop Lost	23	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	82	53
SV Insertions	1,797	865
SV Deletions	4,402	2,217
SV Tandem Duplications	96	46
SV Inversions	298	229

Fragment Length Summary

Fragment Length Median	Minimum	Maximum	Standard Deviation
475 bp	130 bp	862 bp	123 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
11.8%

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