Resequencing Report

Sample: LP9006282-DNA_A01

Report Date: 02/08/2015 07:51:49 (UTC)

Illumina_WGS_Gel, Version v3.0.8-39-gf08c608

Sample Information

Sample ID:	LP9006282-DNA_A01
Sample Name:	LP9006282-DNA_A01
Total PF Reads:	810,479,300
Percent Q30 Bases:	81.5%

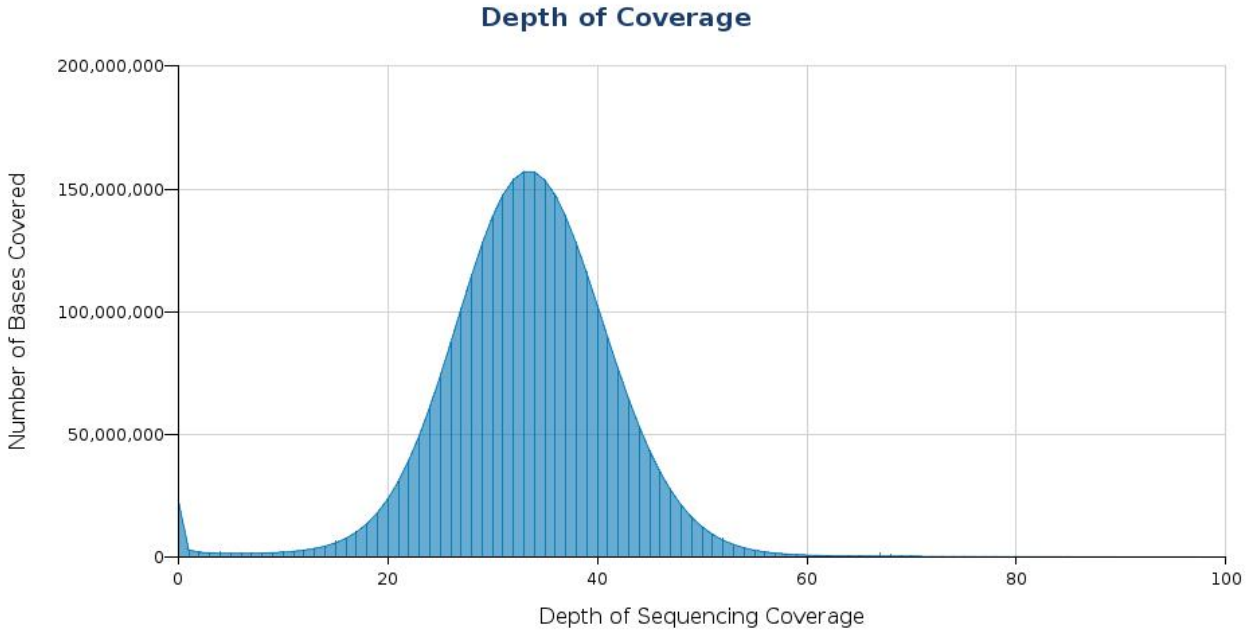
Read Level Statistics

Read	Total Aligned Reads	Percent Aligned Reads
1	389,989,560	96.2%
2	378,431,220	93.4%

Base Level Statistics

Read	Total Aligned Bases	Percent Aligned Bases	Mismatch Rate
1	57,316,632,829	94.3%	1.24%
2	54,163,648,905	89.1%	2.26%

Coverage Histogram (mean coverage 35.9)



Small Variants Summary

	SNVs	Insertions	Deletions
Total Passing	3,588,500	308,466	312,216
Percent Found in dbSNP	98.9%	79.8%	83.0%
Het/Hom Ratio	1.6	1.8	2.5
Ts/Tv Ratio	2.1	-	-

Variants by Sequence Context

	SNVs	Insertions	Deletions
In Genes	1,673,329	149,133	150,887
In Exons	50,380	3,234	3,097
In Coding Regions	22,009	255	278
In UTR Regions	28,371	2,979	2,819
In Splice Site Regions	3,234	341	320
In Mature microRNA	76	6	26

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	-	102	89
Non-synonymous	10,405	147	179
Synonymous	11,511	-	-
Stop Gained	72	0	0
Stop Lost	20	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	78	43
SV Insertions	1,875	898
SV Deletions	4,538	2,320
SV Tandem Duplications	109	52
SV Inversions	367	278

Fragment Length Summary

Fragment Length Median	Minimum	Maximum	Standard Deviation
475 bp	131 bp	851 bp	121 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
11.6%

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