DRAGEN Enrichment Sequencing Report

Sample: W9YAF-B-D-GE-SL-S69-L004

Analysis Name: 06_Feb_24_LC-189_BRCA_1_2_GE
Report Date: 2024-02-06T11:10:16

DRAGEN Enrichment, Version 3.9.5
For Research Use Only. Not for use in diagnostic procedures.

Sample Information

Sample ID	Total PF Reads	Unique PF Reads	Percent Q30 Bases	Mean Target Coverage Depth	Percent Autosome Callability
W9YAF-B-D-GE-SL-S69-L004	2,818,674	2,177,174	94.13%	268.9	96.67%

Note: Percent autosome callability is the percent of non-N reference positions in the autosomes with a PASSing genotype call. The callability value for autosomes and allosomes can be found in the *.summary.csv output file under "Percent callability".

Enrichment Summary

Target Manifest	Total Length of Targeted Reference	Number of Targeted Regions	Padding Size
GERMLINE_Panel_163_GRCh37_13_Mar_23_used.bed	560,897 bp	2,588	150 bp

Note: All enrichment values are calculated without padding (sequence immediately upstream and downstream) unless otherwise stated. If any targeted region overlaps another region, the region positions will be adjusted to remove overlaps (check manifest file in the output directory for differences).

The total length of the targeted reference excludes no-call positions (Ns) in the reference sequence and might result in a smaller value than expected from the targeted regions coordinates alone.

Read Level Enrichment

Total Aligned Reads	Percent Aligned Reads	Targeted Aligned Reads	Read Enrichment	Padded Target Aligned Reads	Padded Read Enrichment
2,163,164	99.36%	1,402,453	64.83%	1,525,912	70.54%

Note: Reads flagged as duplicates are excluded from the read level metrics.

Base Level Enrichment

Total Aligned Bases	Percent Aligned Bases	Targeted Aligned Bases	Base Enrichment	Padded Target Aligned Bases	Padded Base Enrichment
309,611,335	99.32%	150,827,535	48.72%	213,716,668	69.03%

Note: Reads flagged as duplicates are excluded from the base level metrics.

Small Variants Summary

	SNVs	Insertions	Deletions
Total Passing	309	12	30
Het/Hom Ratio	2.121	2.000	4.000
Ts/Tv Ratio	2.810	-	-

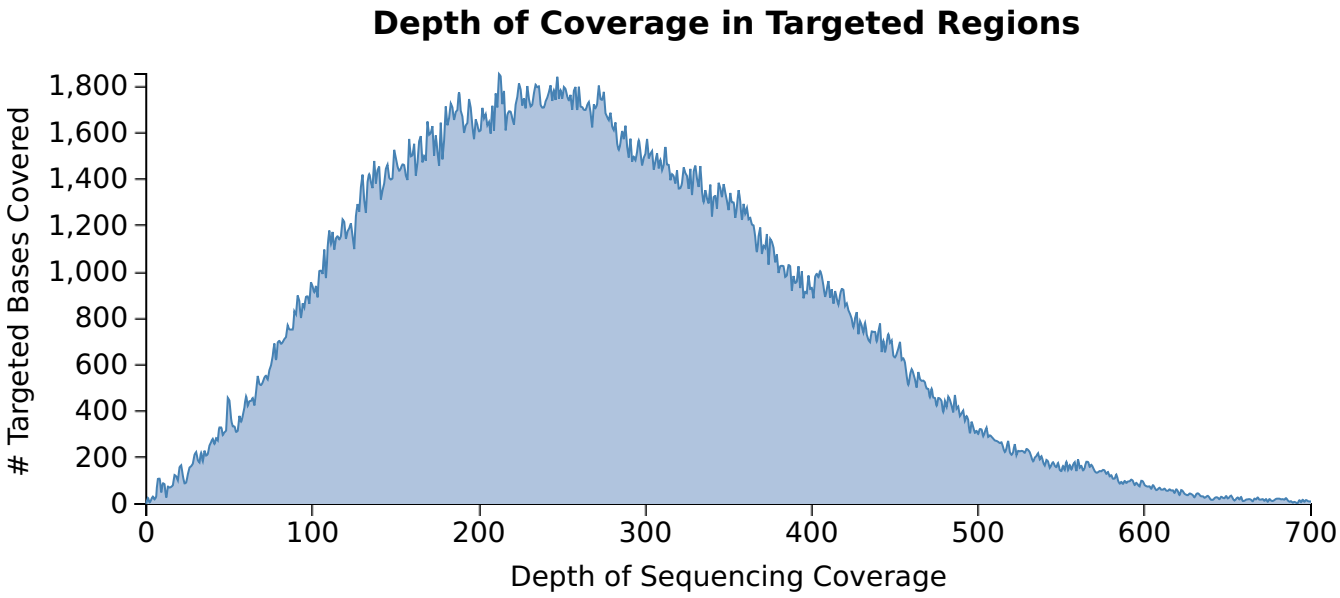
Structural Variants Summary

Variant Type	Total
SV Insertions	0
SV Deletions	0
SV Tandem Duplications	0
SV Breakends	0

Coverage Summary

Mean Target Coverage Depth	Uniformity of Coverage (Pct > 0.2*mean)
268.9	98.30%

Note: Uniformity of coverage is the percentage of targeted base positions in which the read depth is greater than 0.2 times the mean region target coverage depth.



Depth of Coverage	Number of Targeted Bases Covered at Indicated Depth of Coverage	Number of Targeted Bases Covered at or Above Indicated Depth of Coverage	Target Coverage at or Above Indicated Depth of Coverage
1X	26	560,884	100.00%
10X	89	560,480	99.93%
20X	158	559,631	99.77%
30X	224	558,168	99.51%
50X	446	552,858	98.57%
100X	938	522,415	93.14%
200X	1,607	381,934	68.09%

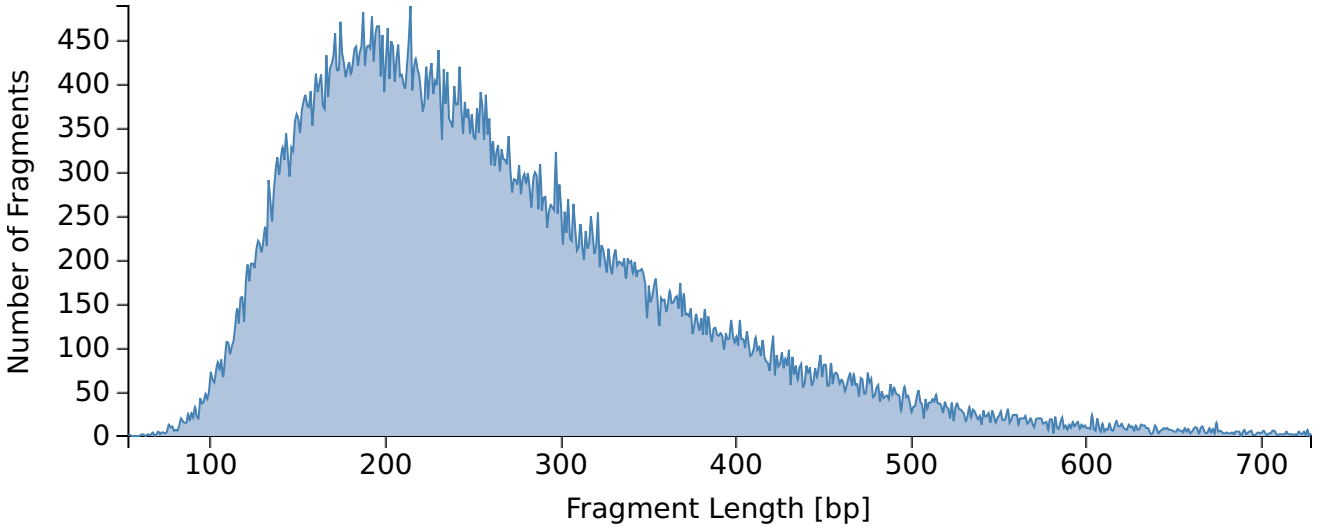
Note: Reads flagged as duplicates are excluded from the coverage statistics. The target coverage percentage is calculated for non-N reference positions in the targeted regions. Read cycles with no-calls (Ns) do not add to the target coverage.

Fragment Length Summary

Fragment Length Median	Minimum	Maximum	Standard Deviation
238 bp	53 bp	728 bp	98 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.

Fragment Length Distribution



Duplicate Information

Percent Duplicate Aligned Reads
22.87%

Analysis Details

Settings

Setting Name	Value
Reference Genome	Homo sapiens (UCSC hg19-altaware)
Targeted Regions	GERMLINE_Panel_163_GRCh37_13_Mar_23_used.bed
Base Padding	150

Software Versions

Software	Version
DRAGEN Enrichment (BaseSpace Workflow)	3.9.5
DRAGEN Enrichment Workflow	0.15.2-O0014dragen-3.9.5-pyparsing-fix
Bam Metrics	v0.0.22
Hash Table Build (Aligner)	01.003.044.3.5.3-38-gefdaeaff-hv-8