QC Metrics Report - Trusight Oncology 500 DNA library only

Accession # 39-22 (S S)

Level 1A: Read and coverage metrics across TruSight Oncology 500 target space. Reference average (Low - High (Average/median)) is derived from samples used in TruSight Oncology 500 validation.

Metric	Value	Reference Average (Low - High (Average/Median))
Total reads	71,759,460	- - - (-)
% mapped reads	96.43%	- - - (-)
on target reads	56,554,049	- - - (-)
% on target reads	81.73%	- - - (-)
Unique on target reads*	20,461,034	- - - (-)
% unique on target reads*	36.18%	- - - (-)
% positions > 50x unique	97.77%	- - - (-)
% positions > 100x unique	94.95%	- - - (-)
% positions > 250x unique	70.59%	- - - (-)
Average Unique Coverage	348.0	- - - (-)

*Unique on target reads calculated based on total number of UMI families.

Level 1B: Expanded DNA QC metrics generated by Illumina TruSight Oncology 500 secondary analysis software.

[DNA Library QC Metrics - Contamination Report]

Metric	Value	Recommended Threshold
CONTAMINATION_SCORE (NA)	384	0 - 3106
CONTAMINATION_P_VALUE (NA)	1.000	0 - 0.049

*The combination of both the Contamination Score and Contamination p-value should be used to evaluate whether or not a sample contains foreign DNA. Therefore, the recommended guideline quality threshold to indicate the potential presence of foreign DNA is a Contamination Score > 3106 AND a Contamination p-value > 0.05.

[DNA Library QC Metrics for Small Variant Calling and TMB]

Metric	Value	Recommended Threshold
MEDIAN_INSERT_SIZE (bp)	81	>= 70
MEDIAN_EXON_COVERAGE (Count)	340	>= 150
PCT_EXON_50X (%)	98.3	90 - 100%

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[DNA Library QC Metrics for MSI]

Metric	Value	Recommended Threshold
USABLE_MSI_SITES (Count)	115	>= 40

[DNA Library QC Metrics for CNV]

Metric	Value	Recommended Threshold
COVERAGE_MAD (Count)	0.129	0 - 0.210
MEDIAN_BIN_COUNT_CNV_TARGET (Count)	5.800	>= 1.0

[DNA Expanded Metrics]

Metric	Value	Recommended Threshold
TOTAL_PF_READS (Count)	70,470,924	NA
MEAN_FAMILY_SIZE (Count)	2.8	NA
MEDIAN_TARGET_COVERAGE (Count)	345.0	NA
PCT_CHIMERIC_READS (%)	1.9	NA
PCT_EXON_100X (%)	95.6	NA
PCT_READ_ENRICHMENT (%)	81.0	NA
PCT_USABLE_UMI_READS (%)	99.9	NA
MEAN_TARGET_COVERAGE (Count)	348.2	NA
PCT_ALIGNED_READS (%)	96.4	NA
PCT_CONTAMINATION_EST (%)	0.9	NA
PCT_PF_UQ_READS (%)	100.0	NA
PCT_TARGET_0.4X_MEAN (%)	90.7	NA
PCT_TARGET_100X (%)	94.9	NA
PCT_TARGET_250X (%)	70.6	NA

Level 2: Exon Coverage Metrics
100x coverage for > 50% of positions was not achieved for the targeted exon regions listed below:

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Gene	Transcript acc #	Exon*	% position >=100x	% position >=250x
BRD4	NM_058243.2	19	37.3%	0.8%
CUX1	NM_181552.3	24	26.3%	0.0%
GATA6	NM_005257.4	2	24.9%	0.0%
GATA6	NM_005257.4	3	48.8%	0.0%
POLE	NM_006231.2	1	0.0%	0.0%
ABL2	NM_007314.3	1	46.2%	0.0%
FANCA	NM_000135.2	1	0.0%	0.0%
GID4	NM_024052.4	1	38.3%	0.0%
IGF2	NM_000612.4	4	35.0%	0.0%
MAP3K4	NM_005922.2	1	40.0%	0.0%
NRG1	NM_013956.3	1	35.9%	0.0%
TCF3	NM_003200.3	15	43.2%	0.0%
TCF3	NM_003200.3	12	0.0%	0.0%
TCF3	NM_003200.3	11	44.9%	0.0%
TCF3	NM_003200.3	10	22.0%	0.0%
TCF3	NM_003200.3	7	43.4%	0.0%
DCUN1D1	NM_020640.2	1	0.0%	0.0%
GNA11	NM_002067.2	1	0.0%	0.0%
SDHA	NM_004168.2	1	0.0%	0.0%
FANCE	NM_021922.2	1	26.3%	0.0%
NTRK1	NM_002529.3	1	33.0%	0.0%
TERT	TERT_Promoter	0'	5.4%	0.0%
GPR124	NM_032777.9	1	29.0%	0.0%
ANKRD11	NM_013275.5	13	18.0%	0.0%
DIS3	NM_014953.3	1	41.1%	30.3%
GPS2	NM_004489.4	2	25.8%	0.0%
TFE3	NM_006521.4	1	17.6%	0.0%
MED12	NM_005120.2	17	27.8%	0.0%
DNMT1	NM_001130823.1	5	0.0%	0.0%
INSR	NM_000208.2	1	0.0%	0.0%
TGFBR1	NM_004612.2	1	0.0%	0.0%
DNMT3A	NM_022552.4	2	25.3%	0.0%

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PTCH1	NM_000264.3	1	32.8%	18.6%
SH2B3	NM_005475.2	2	34.0%	0.0%
ARAF	NM_001654.4	16	34.8%	0.0%
CCNE1	NM_001238.2	2	0.0%	0.0%
IRF4	NM_002460.3	2	37.4%	0.0%
PTEN	NM_000314.4	9	49.7%	0.5%
DOT1L	NM_032482.2	13	43.9%	0.0%
ARID1A	NM_006015.4	1	12.3%	0.0%
ARID1B	NM_017519.2	1	49.6%	11.5%
FGF19	NM_005117.2	1	0.0%	0.0%
PTPRS	NM_002850.3	18	27.9%	0.0%
PTPRS	NM_002850.3	16	23.3%	0.0%
PTPRS	NM_002850.3	15	0.0%	0.0%
PTPRS	NM_002850.3	13	17.6%	0.0%
FGF2	NM_002006.4	1	48.7%	0.0%
KMT2A	NM_001197104.1	1	32.2%	0.0%
PTPRT	NM_007050.5	1	0.0%	0.0%
TOP1	NM_003286.2	1	0.0%	0.0%
EGFR	NM_005228.3	1	0.0%	0.0%
PAX7	NM_001135254.1	7	40.8%	0.0%
FGF3	NM_005247.2	1	0.0%	0.0%
RAB35	NM_006861.6	1	0.0%	0.0%
CDH1	NM_004360.3	1	0.0%	0.0%
FGF4	NM_002007.2	2	0.0%	0.0%
FGF4	NM_002007.2	1	0.0%	0.0%
KDM5C	NM_001146702.1	24	0.0%	0.0%
KDM5C	NM_001146702.1	23	19.0%	0.0%
KDM5C	NM_001146702.1	21	49.1%	0.0%
KDM5C	NM_001146702.1	20	0.0%	0.0%
KDM5C	NM_001146702.1	12	43.4%	0.0%
SMARCD1	NM_003076.4	1	36.1%	0.0%
ATRX	NM_000489.3	1	30.4%	0.0%

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KDM6A	NM_021140.2	1	37.2%	0.0%
SMC1A	NM_006306.3	23	0.0%	0.0%
SMC1A	NM_006306.3	20	16.9%	0.0%
FGF8	NM_033164.3	3	0.0%	0.0%
MST1	NM_020998.3	14	22.2%	0.0%
MST1	NM_020998.3	7	0.0%	0.0%
TSC2	NM_000548.3	32	0.0%	0.0%
PDK1	NM_002610.4	1	40.2%	11.6%
SMO	NM_005631.4	1	14.5%	0.0%
EPHA5	NM_004439.5	1	34.2%	0.0%
PDPK1	NM_002613.4	2	34.5%	22.7%
FGFR3	NM_000142.4	2	0.0%	0.0%
CEBPA	NM_004364.3	1	16.3%	0.0%
RANBP2	NM_006267.4	11	11.7%	0.0%
RANBP2	NM_006267.4	14	48.9%	0.0%
CENPA	NM_001809.3	1	0.0%	0.0%
KMT2B	NM_014727.1	1	0.0%	0.0%
KMT2B	NM_014727.1	16	28.7%	0.0%
MYCN	NM_005378.4	2	42.5%	0.0%
WT1	NM_024426.4	3	3.8%	0.0%
WT1	NM_024426.4	1	32.0%	0.0%
BBC3	NM_014417.4	2	0.0%	0.0%
ERCC2	NM_000400.3	10	37.2%	0.0%
FLT1	NM_002019.4	1	29.9%	0.0%
RB1	NM_000321.2	1	32.9%	0.0%
FLT3	NM_004119.2	1	0.0%	0.0%
RBM10	NM_005676.4	9	0.0%	0.0%
RBM10	NM_005676.4	10	46.0%	0.0%
RBM10	NM_005676.4	11	0.0%	0.0%
RBM10	NM_005676.4	12	0.0%	0.0%
RBM10	NM_005676.4	13	0.5%	0.0%
RBM10	NM_005676.4	17	47.0%	0.0%

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RBM10	NM_005676.4	18	39.2%	0.0%
RBM10	NM_005676.4	19	0.0%	0.0%
RBM10	NM_005676.4	20	10.9%	0.0%
RBM10	NM_005676.4	22	49.1%	0.0%
RBM10	NM_005676.4	4	0.0%	0.0%
FLT4	NM_182925.4	20	0.0%	0.0%
FLT4	NM_182925.4	19	44.4%	0.0%
FLT4	NM_182925.4	1	0.0%	0.0%
LAMP1	NM_005561.3	1	4.7%	0.0%
PIK3CD	NM_005026.3	6	48.6%	0.0%
PIK3CD	NM_005026.3	12	0.0%	0.0%
PIK3CD	NM_005026.3	16	18.4%	0.0%
PIK3CD	NM_005026.3	17	34.6%	0.0%
RECQL4	NM_004260.3	12	24.0%	0.0%
RECQL4	NM_004260.3	3	0.0%	0.0%
RECQL4	NM_004260.3	2	0.0%	0.0%
SRC	NM_198291.2	4	37.9%	0.0%
CIC	NM_015125.3	13	48.8%	0.0%
FOXL2	NM_023067.3	1	25.4%	0.0%
RET	NM_020975.4	1	5.3%	0.0%
FOXO1	NM_002015.3	1	36.0%	7.6%
PIK3R2	NM_005027.3	6	0.0%	0.0%
NF1	NM_001042492.2	1	0.0%	0.0%
BCR	NM_004327.3	18	0.0%	0.0%
BCR	NM_004327.3	20	8.0%	0.0%
CSF3R	NM_156039.3	7	42.8%	0.0%
LZTR1	NM_006767.3	14	34.3%	0.0%
STAT5A	NM_003152.3	7	0.0%	0.0%
STAT5A	NM_003152.3	8	0.0%	0.0%
STAT5A	NM_003152.3	9	0.0%	0.0%
MAGI2	NM_012301.3	22	30.1%	0.2%
STAT5B	NM_012448.3	8	0.0%	0.0%

STAT5B	NM_012448.3	7	0.0%	0.0%
STAT5B	NM_012448.3	6	0.0%	0.0%
MALT1	NM_006785.3	1	0.0%	0.0%
NKX2-1	NM_003317.3	2	40.9%	0.0%
GATA1	NM_002049.3	2	16.6%	0.0%
GATA1	NM_002049.3	6	43.2%	0.0%
NKX3-1	NM_006167.3	1	0.0%	0.0%
MAP2K2	NM_030662.3	6	38.3%	0.0%
RPS6KA4	NM_003942.2	16	21.0%	0.0%
RPS6KA4	NM_003942.2	17	28.9%	0.0%
IDH2	NM_002168.2	1	0.0%	0.0%
MAP2K4	NM_003010.3	1	0.0%	0.0%
SUZ12	NM_015355.2	6	43.8%	0.0%
GATA4	NM_002052.3	2	0.0%	0.0%
MAP3K1	NM_005921.1	1	10.7%	0.0%
NOTCH3	NM_000435.2	24	22.7%	0.0%
NOTCH3	NM_000435.2	1	0.0%	0.0%
POLD1	NM_002691.3	6	41.3%	0.0%

Level 3: Clinically Actionable Genomic Position Coverage Metrics
100x coverage was not achieved for the clinically actionable positions listed below:

Gene	Variant	Disease	Depth
SRSF2	p.P95_R102del	Acute monoblastic and monocytic leukemia | Acute myeloid leukemia | Acute myeloid leukemia, disease | Chronic myelomonocytic leukemia | Evans syndrome | Leukemia | Malignant mastocytosis | Myelodysplastic syndrome | Myelodysplastic/myel oproliferative disease | Myelofibrosis | Myeloproliferative disorder | Myeloproliferative neoplasm | Refractory anemia with excess blasts-1 | Refractory anemia with excess blasts-2 | Thrombocytosis	83
CEBPA	p.N356_C357delinsLAM G	Acute myeloid leukemia	51
CEBPA	p.N356_C357delinsLGL GQG	Acute myeloid leukemia	51
CEBPA	p.L220Pfs*101	Acute myeloid leukemia	57
CEBPA	p.P189_P189del	Acute myeloid leukemia	12

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CEBPA	p.P187Rfs*131	Acute myeloid leukemia	12
CEBPA	p.Y147Pfs*24	Acute myeloid leukemia	37
CEBPA	p.D107*	Acute myeloid leukemia	12
CEBPA	p.G96Afs*56	Acute myeloid leukemia	13
CEBPA	p.K92Wfs*65	Acute myeloid leukemia	28
CEBPA	p.Q83Pfs*25	Acute myeloid leukemia	74
CEBPA	p.P49Rfs*111	Acute myeloid leukemia	46
CEBPA	p.A37Gfs*123	Acute myeloid leukemia	5
CEBPA	p.H24Pfs*84	Acute myeloid leukemia	64
CEBPA	p.H24Afs*84	Acute myeloid leukemia	77
CEBPA	p.P23Rfs*137	Acute myeloid leukemia	77
CEBPA	p.S21Rfs*139	Acute myeloid leukemia	87
DNMT3A	chr2:g.25469487A>C	Acute myeloid leukemia	84
DNMT3A	chr2:g.25469488C>T	Acute myeloid leukemia	88
DNMT3A	p.W297R	Acute myeloid leukemia	87
GATA2	p.A318P	Acute myeloid leukemia	92
GATA2	p.A318T	Acute myeloid leukemia	92
GATA2	p.G151Rfs*34	Acute myeloid leukemia	82
RUNX1	p.P425Mfs	Acute myeloid leukemia	92
SH2B3	p.P97Rfs*22	Acute myeloid leukemia	38
GATA2	p.G320D	Acute myeloid leukemia with inv(3)(q21q26.2) or t(3;3)(q21;q26.2); RPN1-EVI1 | Myelodysplastic syndrome	92
CEBPA	p.D80Rfs*28	Acute myeloid leukemia with myelodysplasia-relat ed changes	87
CEBPA	p.G53Lfs*52	Acute myeloid leukemia | Acute myeloid leukemia, disease	74
SRSF2	p.P95_S101del	Acute myeloid leukemia | Acute myeloid leukemia, disease	86
RUNX1	p.P425L	Acute myeloid leukemia | Acute myeloid leukemia, disease | Anemia | Diffuse non-Hodgkin's lymphoma | Generalized enlarged lymph nodes | Myelodysplastic syndrome	84

CEBPA	p.H24Afs*84	Acute myeloid leukemia | Acute myeloid leukemia, disease | Chronic myelomonocytic leukemia | Myelodysplastic syndrome	77
SRSF2	p.P95_R102del	Acute myeloid leukemia | Chronic myelomonocytic leukemia | Leukemia | Myelodysplastic syndrome | Myelofibrosis | Myeloid leukemia (category) | Myeloproliferative neoplasm | Pancytopenia	83
NOTCH1	p.A2331T	Acute myeloid leukemia | Myelodysplastic syndrome	94
RUNX1	p.L58_G59insF*	Acute myeloid leukemia | Myelodysplastic syndrome	85
CEBPA	p.E59*	Acute myeloid leukemia | Myelodysplastic syndrome (clinical)	90
SH2B3	p.E208Q	Acute myeloid leukemia | Myelofibrosis	68
NF1	chr17:g.29559090G>A	Acute myeloid leukemia | Refractory anemia with excess blasts-2	54
CEBPA	p.P198Afs*123	Acute myeloid leukemia, disease	25
CEBPA	chr19:g.33793002_337 93003insA	Acute myeloid leukemia, disease	12
CEBPA	p.Q83Sfs*77	Acute myeloid leukemia, disease	69
CEBPA	p.A40Rfs*68	Acute myeloid leukemia, disease	7
DNMT3A	chr2:g.25469487A>T	Acute myeloid leukemia, disease	84
GATA2	p.A318V	Acute myeloid leukemia, disease	94
WT1	p.W102*	Acute myeloid leukemia, disease	20
ARID1A	p.Q321Rfs*42	Adenocarcinoma of stomach	19
CEBPA	p.H205Pfs*115	Anemia	47
CEBPA	p.P204Rfs*115	Anemia	43
CEBPA	p.L201P	Anemia	40
CEBPA	p.H200Pfs*121	Anemia	38
CEBPA	p.A199P	Anemia	30
CEBPA	p.P198Afs*123	Anemia	25
CEBPA	p.P197Q	Anemia	24
CEBPA	p.P197Sfs*124	Anemia	21
CEBPA	p.D80Gfs*28	Anemia	85

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FGFR3	p.E135K	Carcinoma of colon	92
TSC2	p.Q1192*	Carcinoma of thyroid	91
PTEN	p.W111*	Carcinoma of uterus | Glioblastoma | Glioblastoma multiforme | Neoplasm of endometrium	89
DNMT3A	chr2:g.25470620T>C	Essential thrombocythemia	63
SH2B3	p.E153Rfs*20	Essential thrombocythemia	54
PIK3CD	c.195C>T	Giant cell glioblastoma	79
PTCH1	chr9:g.98270646_9827 0647insGCC	Glioblastoma multiforme of brain | Malignant glioma of brain | Oligodendroglioma of brain	4
CREBBP	p.V1942G	Malignant melanoma	38
ARID1A	p.N1571I	Malignant melanoma, metastatic	62
PTCH1	chr9:g.98270573_9827 0573delG	Medulloblastoma	43
VHL	p.G114C	Metastatic renal cell carcinoma	79
CEBPA	p.P204Rfs*114	Myelodysplastic syndrome	45
CEBPA	p.Q83Sfs*77	Myelodysplastic syndrome	69
CUX1	p.A1329Rfs*155	Myelodysplastic syndrome	34
GATA2	p.W10C	Myelodysplastic syndrome	39
RUNX1	p.W477*	Myelodysplastic syndrome	53
RUNX1	p.E473Gfs	Myelodysplastic syndrome	70
RUNX1	p.M418_V419delinsRG	Myelodysplastic syndrome	80
RUNX1	p.A66Gfs*69	Myelodysplastic syndrome	64
CEBPA	p.L52Sfs*103	Myelodysplastic syndrome (clinical)	80
GATA2	p.Q20*	Myelodysplastic syndrome (clinical)	75
CUX1	p.G1023Afs*37	Myelofibrosis	56
CUX1	p.P1375S	Pancytopenia	66
CEBPA	p.N356H	Polycythemia vera	62
RUNX1	p.E473Gfs	Refractory anemia with excess blasts I	70
IKZF1	p.V197I	Refractory anemia with excess blasts-2	88
POLD1	chr19:g.50919866_509 19866delG	Secondary malignant neoplasm of intestinal tract	60

NOTCH1	p.V1671I	Thrombocytopenic disorder	39
ARID1A	p.Y308Lfs*92	Uncertain diagnosis	44
CEBPA	p.E167Gfs*3	Uncertain diagnosis	63
CEBPA	p.P39Rfs*71	Uncertain diagnosis	7

Notes: a) This QC report is generated for DNA sequencing only. b) Run Metrics are not generated as analysis started from FASTQ files. c) Clinically actionable positions are identified based on variants reported as clinically relevant in patients tumor type or any other tumor type in cancer patients analyzed by PierianDx.