Seraseq® ctDNA v4 Reference Materials

Accelerate liquid biopsy assay validation and clinical implementation with a comprehensive reference material

INTRODUCTION

As liquid biopsy assays gain acceptance for a growing variety of clinical applications, an acute need for highly multiplexed reference materials bearing a broad range of clinically relevant variant types has arisen. But searching for useful remnant patient specimens is prohibitively time-consuming, and singleplex cell lines are inefficient and add unnecessary expense.

Seraseq ctDNA v4 reference materials are purpose-built to accelerate the thorough validation and clinical implementation of NGS-based ctDNA assays. Seraseq ctDNA v4 contains an unprecedented 93 variants including SNPs, indels, translocations and CNVs. Seraseq ctDNA v4 is manufactured with a new fragmentation process which results lower background than previous Seraseq ctDNA offerings to support greater confidence at low variant allele frequencies. These orthogonally validated truth-set materials can trim weeks or even months off your assay validation timeline with the most complete coverage of actionable variant types in a single reference sample. With the input of recognized clinical genomics experts, the Seraseq ctDNA v4 Reference Materials focus on clinically relevant variants that provide actionable feedback about patient samples.

HIGHLIGHTS

Diverse set of 93 variants including CNVs and translocations

Improved manufacturing to reduce background noise

Consistently performing with batch-specific yield information provided

FEATURES AND BENEFITS

- 93 unique multiplexed variants in 71 genes, covering 43 SNVs, 19 deletions, 5 insertions, 4 INDELs, 12 CNVs, and 10 translocations for the broadest coverage of clinically relevant variant types in a single reference material
- Four different variant allele frequencies (VAF) – WT (0%), 0.1%, 0.5%, and 5% - challenge your limit-of-detection or match typical AF of solid tumor NGS assays
- 83 variants are FDA drug targets or referenced in NCCN guidelines to guide treatment
- Variants precisely quantitated with digital PCR and orthogonally validated by NGS
- Blended with well-characterized GM24385 human genomic DNA as background wild-type material
- Available as mutation mix (DNA in 0.1x TE) and reference material (extractable plasma) formats
- Manufactured in cGMP-compliant, ISO 13485 certified facilities

ORDERING INFORMATION

Material #	Product	Concentration*	Fill Volume	Total Mass
0710-3097	Seraseq ctDNA Mutation Mix v4 AF0.1%	10 ng/µL	25 µL	250 ng
0710-3099	Seraseq ctDNA Mutation Mix v4 AF0.5%	10 ng/µL	25 µL	250 ng
0710-3100	Seraseq ctDNA Mutation Mix v4 AF5%	10 ng/µL	25 µL	250 ng
0710-3101	Seraseq ctDNA Mutation Mix v4 WT	10 ng/µL	25 µL	250 ng

* Concentration targets are based on the Qubit dsDNA BR Assay.

Material #	Product	Concentration*	Fill Volume	Total Mass
0710-3992	Seraseq ctDNA Reference Material v4 AF0.1%	25 ng/mL	5 mL	125 ng
0710-3991	Seraseq ctDNA Reference Material v4 AF0.5%	25 ng/mL	5 mL	125 ng
0710-3990	Seraseq ctDNA Reference Material v4 AF5%	25 ng/mL	5 mL	125 ng
0710-3989	Seraseq ctDNA Reference Material v4 WT	25 ng/mL	5 mL	125 ng

Concentration targets are based on average of 3x1mL extractions using QIAGEN QIAamp® Circulating Nucleic Acid Kit with carrier RNA followed by measurement with the Qubit dsDNA BR Assay.

MUTATIONS PRESENT IN THE SERASEQ® ctDNA v4 PRODUCTS (0.1%, 0.5%, 5%)

Gene	Nucleotide Change	Transcript	Variant Type	COSMIC ID
AKT1	c.49G>A	NM_005163.2	SNV	COSM33765
AR	c.2623C>T	NM_000044.6	SNV	COSM238555
ATM	c.1058_1059del	NM_000051.4	Deletion	COSM21924
BRAF	c.1799T>A	NM_004333.6	SNV	COSM476
BRCA1	c.1961del	NM_007294.4	Deletion	COSM219054
BRCA2	c.7934del	NM_000059.4	Deletion	COSM1738241
CDKN2A	c.9_32dup	NM_000077.5	Insertion	COSM13442
CHEK1	c.676del	NM_001114122.3	Deletion	COSM1352376*
CHEK2	c.1116_1117delinsGT	NM_007194.4	INDEL	COSM384945**
EGFR	c.2235_2249del	NM_005228.5	Deletion	COSM6223
EGFR	c.2303G>T	NM_005228.5	SNV	COSM6241
EGFR	c.2310_2311insGGT	NM_005228.5	Insertion	COSM12378
EGFR	c.2369C>T	NM_005228.5	SNV	COSM6240
EGFR	c.2389T>A	NM_005228.5	SNV	COSM6493937
EGFR	c.2573T>G	NM_005228.5	SNV	COSM6224
ERBB2	c.2313_2324dup	NM_004448.4	Insertion	COSM20959
ESR1	c.1613A>G	NM_000125.4	SNV	COSM94250
FGFR3	c.746C>G	NM_000142.5	SNV	COSM715
HRAS	c.182A>G	NM_005343.4	SNV	COSM499
HRAS	c.37G>C	NM_005343.4	SNV	COSM486
IDH1	c.394C>T	NM_005896.4	SNV	COSM28747
IDH2	c.419G>A	NM_002168.4	SNV	COSM41590
IDH2	c.515G>A	NM_002168.4	SNV	COSM33733
KIT	c.2361+67_2361+72delTTTTTT	NM_000222.3	Deletion	N/A
KIT	c.2447A>T	NM_000222.3	SNV	COSM1314
KRAS	c.183A>C	NM_004985.5	SNV	COSM554
KRAS	c.34G>T	NM_004985.5	SNV	COSM516
KRAS	c.35G>A	NM_004985.5	SNV	COSM521

MUTATIONS PRESENT IN THE SERASEQ® ctDNA v4 PRODUCTS (0.1%, 0.5%, 5%) continued

Gene	Nucleotide Change	Transcript	Variant Type	COSMIC ID
MAP2K1	c.370C>T	NM_002755.4	SNV	COSM235614
MAP4K3	c.246-2475_246-2470delTTTTTT	NM_003618.4	NM_003618.4	N/A
MAP4K3	c.998-35_998-30delAAAAAA	NM_003618.4	Deletion	N/A
MET	c.3082+1del	NM_001127500.3	Deletion	COSM6947926
MLH1	c.232_243delinsATGTAAGG	NM_000249.4	INDEL	N/A
MSH2	c.1662-12_1677del	NM_000251.3	Deletion	N/A
MSH2	c.942+20_942+29delAAAAAAAAAA	NM_000251.3	Deletion	N/A
MSH6	c.2056_2060delinsCTTCTACCTCAAAAA	NM_000179.3	INDEL	N/A
MTOR	c.6644C>A	NM_004958.4	SNV	COSM20417
NF1	c.3738_3747del	NM_001042492.3	Deletion	COSM510741
NRAS	c.182A>G	NM_002524.5	SNV	COSM584
NTRK1	c.1783G>A	NM_002529.4	SNV	COSM9113104
NTRK2	c.1915G>A	NM_006180.6	SNV	N/A
NTRK3	c.1867G>A	NM_001012338.3	SNV	COSM6951362
PALB2	c.839del	NM_024675.4	Deletion	COSM1376815
PDGFRA	c.2525A>T	NM_006206.6	SNV	COSM736
PIK3CA	c.1633G>A	NM_006218.4	SNV	COSM763
PIK3CA	c.3140A>G	NM_006218.4	SNV	COSM775
PIK3CA	c.3203dup	NM_006218.4	Insertion	COSM249879
PIK3R1	c.1727_1729del	NM_181523.3	Deletion	COSM35737
PMS2	c.861_864del	NM_000535.7	Deletion	COSM5547641
PTCH1	c.2307_2308delinsTT	NM_000264.5	INDEL	COSM17587
PTEN	c.800del	NM_000314.8	Deletion	COSM5809
PTEN	c.741dup	NM_000314.8	Insertion	COSM4986
RAD51C	c.242C>A	NM_058216.3	SNV	N/A
RAD51C	c.338dup	NM_058216.3	SNV	N/A
RAD51D	c.271A>T	NM_002878.4	SNV	N/A

MUTATIONS PRESENT IN THE SERASEQ® ctDNA v4 PRODUCTS (0.1%, 0.5%, 5%) continued

Gene	Nucleotide Change	Transcript	Variant Type	COSMIC ID
RAD51D	c.392dup	NM_002878.4	SNV	N/A
RAF1	c.770C>T	NM_002880.4	SNV	COSM181063
RB1	c.751C>T	NM_000321.3	SNV	COSM878
RET	c.2753T>C	NM_020975.6	SNV	COSM965
SLC7A8	c.-231_-224delTTTTTTTT	NM_012244.4	Deletion	N/A
SMARCB1	c.118C>T	NM_003073.5	SNV	COSM1002
STK11	c.734+1G>T	NM_000455.5	SNV	COSM51523
TERT	c.-124C>T	NM_198253.3	SNV	N/A
TERT	c.-146C>T	NM_198253.3	SNV	N/A
TP53	c.723del	NM_000546.6	Deletion	COSM6530
TP53	c.743G>A	NM_000546.6	SNV	COSM10662
TP53	c.818G>A	NM_000546.6	SNV	COSM10660
TSC1	c.1263+1G>T	NM_000368.5	SNV	COSM1738312
TSC2	c.2640-1G>A	NM_000548.5	SNV	COSM3361675
VHL	c.481C>T	NM_000551.4	SNV	COSM17612
ZNF2	c.*1525_*1530delTTTTTT	NM_021088.4	Deletion	N/A
CD74::NRG1	Intron 6::Intron 5	NM_001025159.3::NM_013964.5	Translocation	N/A
CD74::ROS1	Intron 6::Intron 34	NM_001025159.3::NM_001378902.1	Translocation	N/A
COL1A1::PDGFB	Intron 25::Intron 1	NM_000088.3::NM_002608.3	Translocation	N/A
EML4::ALK	Intron 13::Intron 19	NM_019063.5::NM_004304.5	Translocation	N/A
ETV6::NTRK3	Intron 5::Intron 14	NM_001987.5::NM_002530.4	Translocation	N/A
FGFR2::BICC1	Intron 17::Intron 2	NM_000141.5::NM_001080512.3	Translocation	N/A
FGFR3::TACC3	Exon 18::Intron 7	NM_000142.5::NM_006342.3	Translocation	N/A
NCOA4::RET	Intron 7::Intron 11	NM_001145263.2::NM_020975.6	Translocation	N/A
PML::NTRK2	Intron 2::Intron 12	NM_002675.4::NM_006180.6	Translocation	N/A
TPM3::NTRK1	Intron 7::Intron 9	NM_153649.4::NM_002529.4	Translocation	N/A
AKT2	Amplification	19:40736224_40791252	CNV	N/A

MUTATIONS PRESENT IN THE SERASEQ® ctDNA v4 PRODUCTS (0.1%, 0.5%, 5%) continued

Gene	Nucleotide Change	Transcript	Variant Type	COSMIC ID
CCND1	Amplification	NM_053056.3:11:69455924_69469242	CNV	N/A
CCNE1	Amplification	NM_001238.4:19:30302898_30315219	CNV	N/A
CDK4	Amplification	NM_000075.4:58141510_58146093	CNV	N/A
ERBB2	Amplification	NM_004448.4:17:37844347_37884911	CNV	N/A
FGF19	Amplification	NM_005117.3:11:69513006_69518790	CNV	N/A
FGF3	Amplification	NM_005247.4:11:69624736_69634184	CNV	N/A
FGF4	Amplification	NM_002007.4:69587797_69590109	CNV	N/A
FGFR1	Amplification	NM_023110.3:8:38268661_38326153	CNV	N/A
MET***	Amplification	NM_001127500.3:7:116312250_116438431	CNV	N/A
MYC	Amplification	NM_002467.6:8:128747680_128755197	CNV	N/A
MYCN	Amplification	NM_005378.6:2:16080672_16087126	CNV	N/A

** COSMIC uses transcript ENST00000328354

*** MET gene is covered using overlapping DNA constructs. The overlapping regions are expected to show higher copy number levels than the rest of the gene.

NOTE: Above list does not include variants present in the GM24385 background. Indels are defined as deletion/insertions less than 10 base pairs.

MKT-01055-Rev. 03

seracare

seracare.com • 508.244.6400 • 800.676.1881 Toll Free • 508.634.3334 Fax