Seraseq® Inherited Cancer DNA Mix v2

Assay validation and daily-run QC material for cancer-predisposition testing

INTRODUCTION

Germline testing for inherited cancers plays a critical role in identifying individuals at an elevated risk for developing cancer due to genetic predispositions. Unlike somatic mutations, which are acquired throughout a person's life, germline mutations are inherited genetic alterations present from birth and found in every cell of the body. These mutations are often passed down through generations and can significantly increase the likelihood of certain cancers developing, such as breast, ovarian, colorectal, and prostate cancers, among others.

With advancements in next-generation sequencing (NGS) and more comprehensive testing panels, clinical labs can now screen for a wide array of mutations in genes known to be associated with cancer susceptibility. Key genes, including BRCA1, BRCA2, MLH1, and others, are routinely evaluated in individuals with a family history of cancer, providing vital information that can inform personalized risk management, early intervention strategies, and tailored treatment plans.

Germline testing for inherited cancer syndromes has become an essential component of clinical practice, guided by testing guidelines and quality standards from regulatory bodies like the College of American Pathologists (CAP) and the Clinical Laboratory Improvement Amendments (CLIA). Accurate detection of both common and rare genetic variants, including challenging-to-detect mutations, is crucial for delivering reliable results. Comprehensive reference materials like SeraCare's Seraseq Inherited Cancer DNA Mix v2 support clinical labs in their efforts to develop, validate, and routinely assess the performance of their assays, ensuring high-quality, accurate, and clinically impactful genetic testing for inherited cancers.

FEATURES AND BENEFITS

- **Comprehensive Variant Coverage:** Screen confidently for 61 clinically relevant variants across 56 cancer-associated genes. This enhanced coverage offers 2.5 times more mutations than our previous version, ensuring even rare and challenging variants are accounted for.
- **Unmatched Complexity:** Incorporates difficult-to-detect mutations, such as the MSH2 Boland inversion, PMS2 pseudogene homologous variant, and EPCAM 3' large deletion, ensuring robust testing capabilities.
- **High Variant Diversity:** Includes deletions, duplications, insertions, INDELs, SNVs, and inversions as well as a variety of complex variant types such as homopolymer variants, high-GC region variants, microsatellite variants, whole exon deletion, and deep splice site variants.
- **Single-Vial Convenience:** Simplify workflows with a highly multiplexed reference material packaged in a single vial, reducing costs and saving time while boosting QC consistency.
- **Regulatory Compliance Support:** Designed to support compliance with regulatory standards like CAP, CLIA ensures your lab meets performance, quality, and reporting requirements effortlessly.
- **Sustainable source:** Manufactured in GMP-compliant, ISO 13485-certified facilities, the Seraseq Inherited Cancer DNA Mix v2 offers lot-to-lot consistency and an uninterrupted supply chain.

HIGHLIGHTS

High-quality third-party QC material in mutation mix format to develop, validate, monitor, and troubleshoot your assay.

Convenient highly multiplexed material in a single vial format to save time, cost, and increase QC consistency.

Ensure lot-to-lot consistency and a reliable supply with materials manufactured in GMP-compliant and ISO 13485-certified facilities.

ORDERING INFORMATION

Material #	Product	Concentration*	Fill Size	Total Mass
0730-0069	Seraseq® Inherited Cancer DNA Mix v2	25 ng/µL	1 vial x 20µL	500 ng

*Reported concentration values are based on the Qubit dsDNA BR Assay.

Not for In Vitro Diagnostic Use. Research Use Only.

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

Gene	Nucleotide Change	Variant Type	Variant Size (bp)	Inherited Disorder
APC	c.4348C>T	SNV	1	Familial multiple polyposis syndrome; Familial adenomatous polyposis 1; APC-Associated Polyposis Disorders; Hereditary cancer-predisposing syndrome
	c.4666dup	Duplication	1	Familial multiple polyposis syndrome; Familial adenomatous polyposis 1; APC-Associated Polyposis Disorders; Hereditary cancer-predisposing syndrome
ATM	c.640del	Deletion	1	Ataxia-telangiectasia syndrome; Hereditary cancer-predisposing syndrome
AXIN2	c.1994dup	Duplication	1	Oligodontia-cancer predisposition syndrome; Hereditary cancer-predisposing syndrome
BAP1	c.1433_1449dup	Duplication	17	BAP1-related tumor predisposition syndrome
BARD1	c.1600_1634delinsGCG	Indel	35	Familial breast cancer
BMPR1A	c.1131_1153dup	Duplication	23	Juvenile polyposis syndrome
BRCA1	c.3481_3491del	Deletion	11	Hereditary breast and ovarian cancer predisposing syndrome
BRCA2	c.1813dup	Duplication	1	Hereditary breast and ovarian cancer predisposing syndrome
	c.8954-8_9136del	Deletion	284	Hereditary breast and ovarian cancer predisposing syndrome
	c.9342_9343insAluY	Insertion	329	HBOC syndrome, Breast, fallopian tube, melanoma, ovarian, pancreatic, peritoneal, prostate, fanconi anemia, complementation group D1
BRIP1	c.2392C>T	SNV	1	Hereditary breast and ovarian cancer predisposing syndrome, Fanconi anemia
CDH1	c.2037_2061dup	Duplication	25	Hereditary diffuse gastric adenocarcinoma
CDK4	c.70C>T	SNV	1	Familial melanoma
CDKN1B	c.59_77dup	Duplication	19	Endocrine neoplasia; MEN Type 4; Gastrinoma; GEP; nonendocrine parathyroid, pituitary
CDKN2A	c.9_32dup	Insertion	24	Familial Atypical Multiple Mole Melanoma syndrome (FAMMM-PC); Cutaneous melanoma; Melanoma and neural system tumor syndrome; Melanoma-pancreatic cancer syndrome
CHEK2	c.1100del	Deletion	1	Astrocytoma; Nephroblastoma; CHEK2-Related Cancer Susceptibility
DICER1	c.682_724dup	Duplication	43	DICER1 syndrome
EPCAM	c.859-1462_*1999del	Deletion	4,909	Lynch syndrome
FH	c.37_92del	Deletion	56	Hereditary uterine fibroids, leiomyomatosis and renal cell cancer (HLRCC)
FLCN	c.1285dup	Duplication	1	Birt-Hogg-Dube Syndrome; Familial spontaneous pneumothorax; Multiple fibrofolliculomas

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

Gene	Nucleotide Change	Variant Type	Variant Size (bp)	Inherited Disorder
HOXB13	c.844_845del	Deletion	2	Hereditary prostate cancer, Gastric cancer
KIT	c.1676T>C	SNV	1	Familial Gastrointestinal Stromal Tumor (GIST); Melanoma; Thymoma
MAX	c.183_195del	Deletion	13	Hereditary pheochromocytoma-paraganglioma
MEN1	c.1382_1404dup	Duplication	23	Multiple endocrine neoplasia, type 1
MET	c.3281A>G	SNV	1	Hereditary Papillary Renal Carcinoma (HPRCC)
MITF	c.773_785dup	Duplication	13	Genetic deafness (autosomal dominant inheritance)
MLH1	c.1852_1854del	Deletion	3	Lynch syndrome
MSH2	c.942+3A>T	SNV	1	Lynch syndrome
MSH2 Boland Inversion 3'	c.-125-9509096_1277-3165inv	Inversion	9,548,413	Lynch syndrome
MSH6	c.3261dup	Duplication	1	Lynch Syndrome; Hereditary nonpolyposis colon cancer
MUTYH	c.549_576del	Deletion	28	Familial adenomatous polyposis 2
NBN	c.667_668insTTTATATTTTTA TTATATAAAATATAATAAAGAAA ACATTTATATTTTATAATATAAAA ATATTATATTATATATAAAATA TAATAAAGAAAACATTATATA	Insertion	102	Microcephaly, normal intelligence and immunodeficiency; Seemanova Syndrome II; Nijmegen breakage syndrome
NF1	c.2410-110_2850+65delinsAAAA	Indel	616	Neurofibromatosis type 1
	c.4600C>T	SNV	1	Neurofibromatosis type 2
NTHL1	c.417_436dup	Duplication	20	Hereditary cancer-predisposing syndrome
PALB2	c.3114-1_3201+2del	Deletion	91	Lynch syndrome/HNPCC, Brain, colorectal, endometrial, ovarian, pancreas, prostate, renal pelvis and/or urea, stomach, and others, CMMRD
PDGFRA	c.2537A>T	SNV	1	GIST-plus syndrome
PMS2	c.2243_2246del	Deletion	4	Lynch syndrome
	c.861_864del	Deletion	4	Lynch syndrome
POLD1	c.1433G>A	SNV	1	Familial Colorectal Cancer susceptibility (CRC)
POLE	c.1270C>G	SNV	1	Familial Colorectal Cancer susceptibility (CRC); Cystic fibrosis-gastritis-megaloblastic anemia syndrome
PTCH1	c.202-16_227del	Deletion	42	Gorlin syndrome
PTEN	c.750_751del	Deletion	2	PTEN hamartoma tumor syndrome; Cowden syndrome
RAD50	c.326_329del	Deletion	4	Hereditary cancer-predisposing syndrome; Nijmegen breakage syndrome, Breast carcinoma
RAD51C	c.706-2A>G	SNV	1	Hereditary nonpolyposis colon cancer
RAD51D	c.694_715delinsTGAGAGC TGAAGACCCTGGCCT	Indel	22	MEN2, Medullary thyroid carcinoma, parathyroid adenoma or hyperplasia, pheochromocytoma

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

Gene	Nucleotide Change	Variant Type	Variant Size (bp)	Inherited Disorder
RET	c.2753T>C	SNV	1	Multiple endocrine neoplasia, type 2a; Medullary thyroid carcinoma
SDHA	c.1785delinsCTTCTGGCGC GCATGCCAGG	Indel	20	Hereditary cancer-predisposing syndrome
SDHAF2	c.267del	Deletion	1	Hereditary pheochromocytoma-paraganglioma; Hereditary cancer-predisposing syndrome
SDHB	c.42_43insCACTCTCCTTG AGGCGCCGGTTGCCG	Insertion	26	Hereditary pheochromocytoma-paraganglioma
SDHC	c.250_251del	Deletion	2	Paragangliomas 3; Gastrointestinal stromal tumor
SDHD	c.383_386dup	Duplication	4	Carney-Stratakis syndrome; Paragangliomas with sensorineural hearing loss; Pheochromocytoma; Cowden syndrome 3
SMAD4	c.1349_1376dup	Duplication	28	Juvenile polyposis syndrome; Familial thoracic aortic aneurysm and aortic dissection; Hereditary cancer-predisposing syndrome
SMARCA4	c.917_941del	Deletion	25	Rhabdoid tumor predisposition syndrome 2
STK11	c.291-10_922del	Deletion	4,580	Peutz-Jeghers syndrome
TMEM127	c.24_48del	Deletion	25	Hereditary pheochromocytoma-paraganglioma
TP53	c.524G>A	SNV	1	Li-Fraumeni syndrome
TSC1	c.850_881delinsGCTTTCCT CATCGTT	Indel	32	Tuberous sclerosis 1
TSC2	c.2640-1G>A	SNV	1	Tuberous sclerosis 2
VHL	c.481C>T	SNV	1	Von Hippel-Lindau syndrome; Familial infantile myasthenia

NOTE: Above list does not include variants present in the GM24385 background. Target variant allele frequency is 50%.

ABOUT US

LGC Clinical Diagnostics offers a comprehensive portfolio of reference materials for oncology, reproductive health, and other genomic applications, designed and manufactured to meet the precision demanded by NGS and other molecular assays. The portfolio includes high quality ground-truth RNA, ctDNA and genomic DNA-based reference materials that are NGS platform agnostic for tumor profiling, immuno-oncology, liquid biopsy, NIPT, pharmacogenomics, inherited diseases and many other assays.

For more information visit seracare.com.

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