PRODUCT SHEET

Seraseq® CNV Copy Loss DNA Mix

Comprehensive reference material for NGS-based CNV gene copy loss assay development, validation, and routine QC use

Modern methods for cancer screening in patient diagnosis, prognosis, and treatment often include detection of genetic variants at the DNA or RNA level. Among those lesions, copy number variants (CNVs), which can consist of either gains through gene amplification or reduction through gene loss, play major roles as oncogenic biomarkers. Copy losses in the TP53 and PTEN genes represent classic loss of heterozygosity (LOH) oncogenic models. Furthermore, recent research has demonstrated that deletion of the gene encoding methyl thioadenosine phosphorylase (MTAP) plays a role in enhancing therapeutic efficacy of drugs in the PRMT5 and MAT2A inhibitor class.

Accurate detection of pathogenic gene copy losses in targeted next-generation sequencing (NGS) assays can be challenging due to complex analysis of associated aneuploidy, partial deletions, and other genomic rearrangements. Therefore, there is a critical need for comprehensive reference materials carrying multiple copy loss CNVs in clinically-relevant genes, but availability of such materials has been scant.

To support clinical laboratories in developing, characterizing, validating, and routinely analyzing assays that include gene loss CNVs, LGC has developed a novel reference material in which multiple genes have sustained either partial or complete deletions.

The Seraseq® CNV Copy Loss DNA Mix is a reference material derived from a tumor cell line with well-characterized, multiple gene copy losses, including MTAP, TP53, and PTEN (Table 1). Available in purified genomic DNA format, it is suitable for NGS assays that include detection of gene copy loss CNVs.

Highlights

- High quality reference material containing gene-level deletions including MTAP, TP53, and PTEN.
- Available in mutation mix format to develop, validate, monitor, and troubleshoot your assay.
- Manufactured under cGMP compliance in ISO 13485 certified facilities.

FEATURES AND BENEFITS

- 7 clinically relevant CNV gene copy losses associated with tumors
- A single reference material carrying partial or complete gene copy losses
- Copy numbers quantified using sensitive dPCR assays and/or whole genome, long read sequencing
- Manufactured in cGMP-compliant, ISO 13485-certified facilities

ORDERING INFORMATION

Product	Material	Conc.	Fill Volume	Total Mass
Seraseq® CNV Copy Loss DNA Mix	0700-4109	10 ng/µL	20 µL	200 ng

**Concentration as determined by Qiagen Qubit dsDNA BR Assay. See the Technical Product Report for more details.*

To place an order, please contact us at +1 508.244.6400 and +1 800.676.1881 or email CDx-CustomerService@lgcgroup.com.

Table 1

Gene	Number of full-length gene copies
CDKN2A	0
CDKN2B	0
CYP2D6	1
MAP2K4	1
MINPP1	1
MTAP	0
PTEN	0
TP53	0

NOTE: The above list does not include variants present in the cell line background.

VISUALIZATION OF GENE-LEVEL DELETIONS

This product contains partial or complete gene copy losses native to a cell line background. Screenshots (Figures 1 and 2) derived from long-read NGS, display the read alignments of the MTAP and TP53 genes.

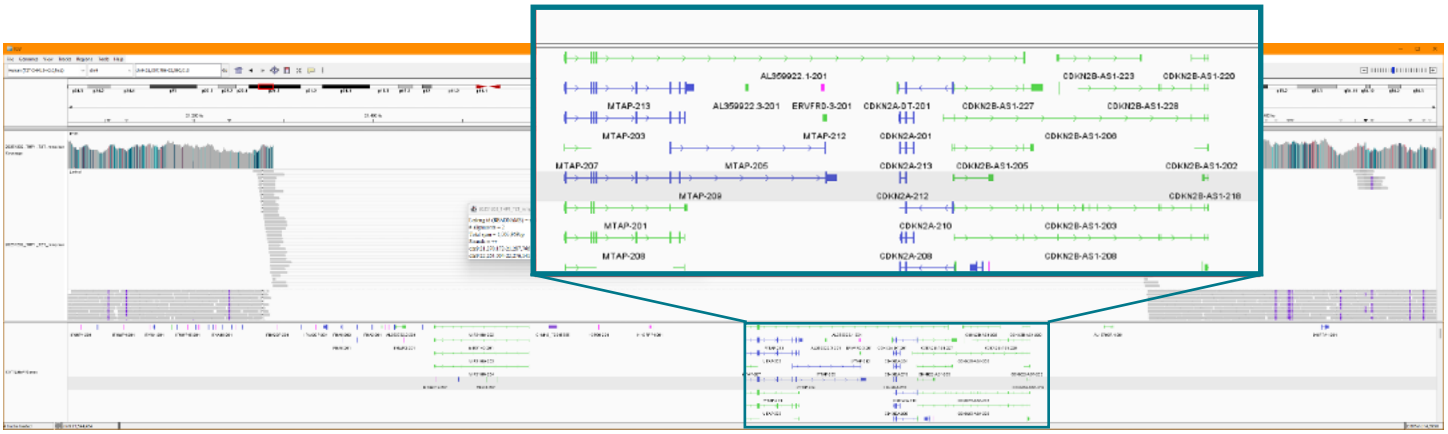

Figure 1. Visualization of CDKN2A, CDKN2B, and MTAP complete deletions. The region of chromosome 9 that contains MTAP, CDKN2A, and CDKN2B is completely deleted and surrounded by regions that show loss of heterozygosity (LOH) based on the presence of only homozygous SNPs (the vertical colored lines on a gray background). Magnified section shows the region containing MTAP, CDKN2A, and CDKN2B.

Figure 2. Visualization of PTEN partial deletion. The entire chromosome 10 that contains PTEN is present at 1 copy. Part of the remaining PTEN contains a larger deletion—some PTEN remains. The 3' untranslated region of the version with the deletion has several additional mutations.

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