

PRESS RELEASE

MRC Holland Announces digitalMLPA – A Revolution in Copy Number Variation Detection

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digitalMLPA allows for the simultaneous copy number determination of up to 1000 different DNA sequences in one simple reaction.

MRC Holland announces the launch of its SALSA® digitalMLPA technology, a novel technique that enables the simultaneous quantification of hundreds of genomic targets. Alterations in the number of gene copies (known as copy number variations, or CNVs) play a causal role in a wide range genetic disorders, but sequencing techniques often fail to adequately detect these changes, especially in complex genomic regions. MRC Holland has pioneered the way in accurate CNV detection with their well-known industry standard SALSA® MLPA® assays. With the arrival of digitalMLPA, the scale on which reliable, robust, and low-cost copy number variation determination can be applied has increased >10 fold: with its capability to quantify up to 1000 targets per assay digitalMLPA streamlines CNV detection.

digitalMLPA combines MRC Holland's proprietary MLPA technology with the power of next generation sequencing (NGS) for data generation. Data generated from digitalMLPA assays is analyzed using free MRC Holland-developed software, Coffalyser digitalMLPA. digitalMLPA has several advantages over other methods used in large-scale genomic profiling, as it can reliably determine the copy number of sequences that differ by single nucleotide and requires only 20 ng of sample DNA. With digitalMLPA, only information about DNA regions specifically targeted by digitalMLPA probes is uncovered. This is because digitalMLPA probes are amplified, not sample DNA, thus limiting incidental findings. In addition, digitalMLPA probes are amplified by a universal primer pair, eliminating amplification bias, which in turn reduces probe read depth variability and the read depth coverage needed. A large number of reaction control probes and sample identification probes are also included in every probemix, ensuring that a high level of quality is met in every reaction. Up to 192 (soon to be 384) digitalMLPA samples can be analyzed together, with no clean up or quantification needed prior to loading, and – if desired – samples can be combined with other NGS sequencing libraries on a single lane, saving both time and money.

The first digitalMLPA assay on the market is D001 Hereditary Cancer Panel 1. This panel offers 566 probes targeting 28 genes associated with various hereditary cancers, including breast, ovarian, colorectal, gastric, prostate, pancreatic, endometrial and melanoma. D001 Hereditary Cancer Panel 1 is the perfect complement to existing NGS-based testing, as it improves the detection of CNVs in critical cancer genes, including complicated ones like *PMS2* and *STK11*. MRC Holland's digitalMLPA technology is rapidly expanding with the impending availability of additional assays on the market.

MRC Holland has been providing reliable genetic tests for over 15 years, and this latest development provides yet another leap forward in the field of copy number variation determination from the company that remains the standard in the field.

digitalMLPA products are for research use only, not for use in diagnostic purposes unless specifically stated otherwise.

About MRC Holland

MRC Holland specialises in the production of assays for gene copy number determination and gene variant detection. MRC Holland is best known as the producer of Multiplex Ligation-dependent Probe Amplification (MLPA®) assays. MLPA continues to be the gold standard in the field of gene copy number detection, and MRC Holland's >350 different MLPA assays are used on millions of samples every year. It is MRC Holland's mission to manufacture and distribute affordable and reliable genetic tests worldwide. The company's products are easy to use and implement into research and diagnostic settings, as they run on standard laboratory equipment, use standard platforms and free MRC Holland-developed software for analysis. To learn more, visit www.mrcholland.com.

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