

Product Description SALSA[®] Artificial Duplication DNA SD024-S01

Catalogue number: SD024

Artificial duplication DNA SD024: Contains 100 µl of SALSA SD024 Artificial duplication DNA at a concentration of 10 ng/µl, which is sufficient for 20 MLPA reactions. MLPA reactions used as an artificial positive control should be performed with 5 µl of Artificial duplication DNA. This Artificial duplication DNA is to be used with the following SALSA MLPA probemixes P002-D1 BRCA1, P045-B3/C1/D1 BRCA2/CHEK2, P077-A3/B1 BRCA2 Confirmation, P087-C1/D1 BRCA1 Confirmation and P090-A4/B1/C1 BRCA2.

Intended use: This SD024 DNA can be used as an artificial positive control DNA for the MLPA probemix versions as specified above and in Table 1. These probemix versions are used to detect copy number changes associated with hereditary predisposition for breast cancer. Inclusion of one reaction on SD024 DNA in an MLPA experiment can be of use in the implementation and validation of the MLPA technique.

This product is for research use only (RUO).

Product Description: MRC-Holland is unable to provide positive human DNA samples (for deletion or duplication). As an alternative we have prepared a mixture of female genomic DNA from healthy individuals and a carefully titrated amount of plasmid that contains the target sequences recognised by several probes present in the above mentioned MLPA probemixes.

The plasmid included in SD024 DNA contains partial sequences of the BRCA1 and BRCA2 genes. These sequences will be detected by a number of MLPA probes that are present in the aforementioned probemix versions (for details, see Table 1). The amount of plasmid present is one copy of plasmid / two haploid genome copies. The use of SD024 in MLPA reactions performed with the aforementioned probemixes will therefore show a duplication of several sequences.

The plasmid included in the SD024 DNA also contains a partial sequence of the CHEK2 gene. This sequence will be detected by a mutation-specific MLPA probe that is present in the P045-B3/C1/D1 probemix versions (for details, see Table 1) and will therefore generate a signal for this probe. The amount of plasmid present in SD024 is one copy of plasmid / two haploid genome copies, mimicking the signal obtained on patient samples that contain a heterozygous mutation.

Both the MLPA reaction and the analysis of results should be performed according to the instructions in the MLPA[®] General Protocol. Coffalyser.Net software must be used for analysis of MLPA experiments. This software is available free of charge on www.mlpa.com.

Storage Upon arrival, Binning DNA must be stored between -25 °C and -15 °C. When stored at recommended conditions, this product is stable for at least one year after shipment. The expiry date is mentioned on the label of the vial.

More information: www.mlpa.com; www.mlpa.eu

| | |
|-------------------------------------------------------------------------------------|---------------------------------------------------------------------------------------------------------------------------------------------------|
|  | MRC-Holland bv; Willem Schoutenstraat 1 1057 DL, Amsterdam, The Netherlands |
| E-mail | info@mlpa.com (information & technical questions); order@mlpa.com (orders) |
| Phone | +31 888 657 200 |

Table 1. Probe targets duplicated in SD024-S01 Artificial duplication DNA

The following probes will appear to have a heterozygous duplication (probe ratios between 1.3 and 1.7) in MLPA reactions on SD024 DNA when using normal DNA from healthy individuals for reference reactions.

| Product | Gene/Exon | Probe length | Probe ID | Present in probemix version | Remarks |
|-----------------|--------------------|--------------|--------------|-----------------------------|-------------------------|
| P002 | BRCA1/exon 6 (5) | 374 nt | 20032-L27342 | D1 | |
| | BRCA1/exon 16 (15) | 160 nt | 20022-L27333 | D1 | |
| | BRCA1/exon 22 (21) | 412 nt | 00785-L23318 | D1 | |
| P045 | BRCA2/exon 4 | 197 nt | 01600-L04671 | B3 | |
| | BRCA2/exon 4 | 202 nt | 01600-L23751 | C1, D1 | |
| | BRCA2/exon 8 | 454 nt | 20632-L28323 | C1, D1 | |
| | BRCA2/exon 11 | 142 nt | 18385-L23778 | C1, D1 | |
| | BRCA2/exon 13 | 301 nt | 02280-L01771 | B3 | |
| | BRCA2/exon 13 | 313 nt | 02280-L28326 | C1, D1 | |
| | BRCA2/exon 21 | 409 nt | 02069-L01970 | B3 | |
| | BRCA2/exon 21 | 373 nt | 20629-L28321 | C1, D1 | |
| | CHEK2/exon 11 * | 495 nt | 01772-L15680 | B3 | CHEK2-1100delC mutation |
| CHEK2/exon 11 * | 490 nt | 01772-L01336 | C1, D1 | CHEK2-1100delC mutation | |
| P077 | BRCA2/exon 8 | 391 nt | 12319-L13312 | A3 | |
| | BRCA2/exon 11 | 196 nt | 12296-L13289 | A3, B1 | |
| | BRCA2/exon 18 | 274 nt | 12307-L13300 | A3, B1 | |
| P087 | BRCA1/exon 15 (14) | 329 nt | 02821-L02250 | C1 | |
| | BRCA1/exon 18 (17) | 185 nt | 03398-L02254 | C1, D1 | |
| | BRCA1/exon 23 (22) | 416 nt | 02830-L02259 | C1 | |
| | BRCA1/exon 15 (14) | 209 nt | 21956-L30984 | D1 | |
| P090 | BRCA2/exon 4 | 197 nt | 01600-L04671 | A4 | |
| | BRCA2/exon 4 | 202 nt | 01600-L23751 | B1, C1 | |
| | BRCA2/exon 8 | 454 nt | 20632-L28323 | B1, C1 | |
| | BRCA2/exon 11 | 142 nt | 18385-L23778 | B1, C1 | |
| | BRCA2/exon 13 | 301 nt | 02280-L01771 | A4 | |
| | BRCA2/exon 13 | 313 nt | 02280-L28326 | B1, C1 | |
| | BRCA2/exon 21 | 409 nt | 02069-L01970 | A4 | |
| | BRCA2/exon 21 | 373 nt | 20629-L28321 | B1, C1 | |

* Contrary to the other probes, this probe shows a mutation signal and not a duplicated signal in SD reaction.

Note: Exon numbering used here may differ from literature! Please notify us of any mistakes: info@mlpa.com.

BRCA1 exon numbering is based on **NM_007294.3**. Please note that the BRCA1 exon numbering in the BRCA1 LRG sequence and in the NCBI NG 005905.2 reference sequence is different, the LRG exon numbering is indicated between brackets. For more detailed information, please consult the product descriptions of P002-D1 and P087-C1/D1 BRCA1 Confirmation SALSA MLPA probemixes.

Please note that SD024 DNA consists of female genomic DNA and a plasmid that contains the target sequences detected by the above mentioned probes and the sequence of the 105 nt chromosome Y specific control fragment. The amount of plasmid in this Artificial duplication DNA (relative to the genomic DNA) results in a relative probe signal for the 105 nt probe on this female DNA which is similar to the relative probe signal obtained on male DNA samples. As a result, the 100 and 105 nt control fragments indicate the presence of two copies chromosome X and one copy chromosome Y.

Implemented Changes – compared to the previous SD024 product description versions

Version 12 – 28 May 2019 (11)

- Information about D1 version of P045 BRCA2/CHEK2 MLPA probemix included on page 1 and 2 (Table 1)

Version 11 – 26 March 2019 (11)

- Information about C1 version of P090 BRCA2 MLPA probemix included on page 1 and 2 (Table 1)
- Removed Table 1 footnote about the height of 105 nt Y probe.

Version 10 – 04 June 2018 (11)

- Information about D1 version of P087 BRCA1 MLPA probemix included on page 1 and 2 (Table 1), and name change of probemix P087 BRCA1 to BRCA1 Confirmation.

Version 09 – 29 May 2018 (11)

- Minor textual and layout changes on page 1.
- Table 1 adjusted on page 2.

Version 08 – 3 October 2017 (11)

- Minor textual and layout changes.
- Information about other Artificial duplication DNA samples removed from page 1.

Version 07 (11) – 27 September 2017

- Information about B1 version of P077 BRCA2 MLPA probemix included on page 1 and 2 (Table 1).
- Updated information about Other SALSA® MLPA® Artificial duplication DNA samples on page 1.
- Minor textual changes.

Version 06 (11) – 23 August 2016

- Information about new versions of P090 BRCA1 and P045 BRCA2/CHEK2 MLPA probemixes included.
- Information on mutation-specific probe for CHEK2 gene of P045-B3/C1 probemix added on page 1.
- Updated information about Other SALSA® MLPA® Artificial duplication DNA samples on page 1.
- Various minor textual changes.

Version 05 – 12 May 2016 (10)

- Lot removed throughout document.
- Updated information about Other SALSA® MLPA® Artificial duplication DNA samples on page 1.
- Table 1 adjusted.
- Various minor textual changes.
- Various minor layout changes.

Version 04 (07)

- Product description adapted to a new lot.
- Information about old versions of MLPA probemixes removed: P002-C1 and P077-A2.
- Table 1: Note about BRCA1/exon 22 probe in P002-D1 removed, note added about BRCA1 exon numbering and minor textual changes.
- Various minor textual changes on page 1.
- Various minor layout changes.

Version 03 (04)

- Information about new version of P002 BRCA1 MLPA probemix included and information about old version removed in text on page 1 and table 1.
- Updated information about Other SALSA® MLPA® Artificial Duplication DNA samples on page 1.
- Updated contact details on page 1.
- Note added to clarify that exon numbering may differ from literature to table 1.
- Minor textual changes on page 1, table 1 and in header.

Version 02 (02)

- Version number added to MLPA probemixes for which SD can be used
- Changes in title: name and lot number of SD
- Minor textual changes

Version 01 (01)

- Not applicable, new document.