

## Product Description SALSA<sup>®</sup> Reference Selection DNA SD082-S01

## Version S01.

Catalogue number: SD082: SALSA<sup>®</sup> Reference Selection DNA, 6 reactions

To be used with the following SALSA MLPA probemix: P021-B1 SMA, in combination with a SALSA<sup>®</sup> MLPA<sup>®</sup> reagent kit, available for various number of reactions. MLPA reagent kits are either provided with FAM or Cy5.0 dye-labelled PCR primer, suitable for Applied Biosystems and Beckman capillary sequencers, respectively (see www.mlpa.com).

**Certificate of Analysis:** Information regarding storage conditions, quality tests, and a sample electropherogram from the current sales lot is available at www.mlpa.com.

**Precautions and warnings:** For professional use only. Always consult the most recent product description AND the corresponding probemix product description AND the MLPA General Protocol or the MS-MLPA General Protocol before use: www.mlpa.com. It is the responsibility of the user to be aware of the latest scientific knowledge of the application before drawing any conclusions from findings generated with this product. Reference Selection DNA should never be used as a reference sample in the MLPA data analysis.

**Intended use:** This SD082 DNA can be used to select suitable reference DNA samples for the MLPA probemix version as specified above and in Table 1. We recommend the use of this SD082 Reference Selection DNA only for initial experiments on DNA samples from healthy individuals with the intention to select suitable reference DNA samples. Reference Selection DNA should never be used as a reference sample in the MLPA data analysis.

Reference DNA samples for use in MLPA experiments should preferably be derived from the same type of tissue, and be purified by the same method, as the DNA samples to be tested. For certain applications, the selection of suitable reference DNA samples is complicated.

For SALSA MLPA Probemix P021-B1, the percentage of all samples that are suitable as reference sample is population dependent. Based on analysis of cell lines from the 1000 genome collection, we estimate that samples with two copies *SMN1*, two copies *SMN2*, two copies of the NAIP gene and absence of a SMN2 $\Delta$ 7-8 copy are expected in approximately 35% of Caucasian samples; 50% of Asian samples and 15-20% of African samples.

**Experimental set up:** MLPA reactions for reference selection purposes should be performed with 5  $\mu$ l of SD082 Reference Selection DNA. Include three reactions with SALSA Reference Selection DNA SD082 in the initial MLPA experiment to select suitable reference DNA samples that have probe ratios between 0.80 and 1.20 for each probe in the probemix.

**Data analysis:** Coffalyser.Net software must be used for analysis of MLPA experiments. This software is available free of charge on www.mlpa.com.

**Reference Selection DNA content:** SD082 Reference Selection DNA is human genomic DNA purified from a selected cell line. This cell line has two copies for the *SMN1* and *SMN2* genes, no copies of the SMN2 $\Delta$ 7-8 variant and two copies of the *NAIP* gene. The P021-B1 probemix contains four probes that are specific for either *SMN1* or *SMN2* and that detect 2 copies of that gene in SD082 DNA. The P021-B1 probemix contains 17 probes that detect a sequence which is present in both *SMN1* and *SMN2* and which detect four copies in SD082.

**Storage and stability:** Upon arrival, Reference Selection DNA must be stored between -25 °C and -15 °C, in the original packaging. When stored under the recommended conditions, a shelf life of at least 1 year is guaranteed, also after opening. The expiry date is mentioned on the label of the vial.

| Probemix | Gene/Exon         | Probe  | Probe ID     | Present in       | Copy number |
|----------|-------------------|--------|--------------|------------------|-------------|
|          |                   | length |              | probemix version |             |
| P021     | Reference         | 175 nt | 00808-L00638 | B1               | 2           |
|          | SMN1/SMN2/1       | 184 nt | 21519-L30024 | B1               | 4           |
|          | SMN1/SMN2/intr. 6 | 193 nt | 22121-L31133 | B1               | 4           |
|          | SMN1/SMN2/5       | 199 nt | 21518-L30023 | B1               | 4           |
|          | Reference         | 211 nt | 18964-L24756 | B1               | 2           |
|          | SMN1/SMN2/2a      | 221 nt | 21517-L30022 | B1               | 4           |
|          | SMN1/SMN2/intr. 7 | 229 nt | 22122-L31134 | B1               | 4           |
|          | NAIP/5            | 238 nt | 01259-L00811 | B1               | 2           |
|          | Reference         | 247 nt | 19086-L24973 | B1               | 2           |
|          | Reference         | 256 nt | 19625-L26284 | B1               | 2           |
|          | SMN1/SMN2/intr. 7 | 265 nt | 22124-L31136 | B1               | 4           |
|          | SMN1/7            | 274 nt | 21488-L30891 | B1               | 2           |
|          | SMN2/7            | 281 nt | 21489-L30892 | B1               | 2           |
|          | SMN1/SMN2/3       | 288 nt | 21516-L30893 | B1               | 4           |
|          | SMN1/8            | 295 nt | 21490-L29983 | B1               | 2           |
|          | SMN2/8            | 301 nt | 21491-L29984 | B1               | 2           |
|          | Reference         | 310 nt | 20763-L28665 | B1               | 2           |
|          | SMN1/SMN2/2b      | 319 nt | 14132-L15557 | B1               | 4           |
|          | SMN1/SMN2/2b      | 328 nt | 21514-L30019 | B1               | 4           |
|          | Reference         | 337 nt | 19746-L26529 | B1               | 2           |
|          | SMN1/SMN2/3       | 346 nt | 21513-L30018 | B1               | 4           |
|          | Reference         | 355 nt | 19127-L25074 | B1               | 2           |
|          | SMN1/SMN2/8       | 364 nt | 01814-L00807 | B1               | 4           |
|          | Reference         | 373 nt | 18296-L25750 | B1               | 2           |
|          | SMN1/SMN2/1       | 382 nt | 22196-L31253 | B1               | 4           |
|          | SMN1/SMN2/intr. 7 | 391 nt | 22123-L31135 | B1               | 4           |
|          | SMN1/SMN2/8       | 400 nt | 22126-L31138 | B1               | 4           |
|          | SMN1/SMN2/4       | 409 nt | 01816-L30922 | B1               | 4           |
|          | SMN1/SMN2/6       | 418 nt | 22194-L31251 | B1               | 4           |
|          | SMN1/SMN2/intr. 7 | 427 nt | 22125-L31137 | B1               | 4           |
|          | Reference         | 436 nt | 19646-L26317 | B1               | 2           |
|          | Reference         | 445 nt | 20431-L27913 | B1               | 2           |

## Table 1. Probe targets in Reference Selection DNA SD082-S01

Note: Exon numbering used here may differ from literature! Please notify us of any mistakes: info@mlpa.com. For more information, like corresponding gene transcripts, please consult the respective probemix product description.

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

\*EUROPE:



**CE** \* comprising EU member states, EU member states, EU member state candidates and members of the European Free Trade Association (EFTA). The product is for RUO in all other countries within Europe

OUTSIDE EUROPE:

Implemented Changes – compared to the previous SD082 product description versions

Version S01-01 – 06 November 2018 (01)

- Not applicable, new document.