

## Product Description SALSA® Reference Selection DNA SD082-S01

### Version S01.

**Catalogue number: SD082:** SALSA® Reference Selection DNA, 6 reactions

To be used with the following SALSA MLPA probemixes: P060-B2 SMA Carrier and P021-B1 SMA, in combination with a SALSA® MLPA® 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](http://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](http://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](http://www.mlpa.com). **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 *SMN2Δ7-8* copy are expected in approximately 35% of Caucasian samples; 50% of Asian samples and 15-20% of African samples.

For SALSA MLPA Probemix P060-B2, the percentage of all samples that are suitable as reference sample is also population dependent. Based on analysis of cell lines from the 1000 genome collection, we estimate that samples with two copies *SMN1* and two copies *SMN2* are expected in approximately 46% of Caucasian samples; 54% of Asian samples and 23% of African samples.

**Experimental set up:** MLPA reactions for reference selection purposes should be performed with 5 µl of 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](http://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 *SMN2Δ7-8* 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 (for details, see table 1). The P060-B2 probemix contains four probes that are specific for either *SMN1* or *SMN2* and that detect 2 copies of that gene in SD082 DNA (for details, see table 2).

**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.

**Table 1. P021 Probe targets in Reference Selection DNA SD082-S01**

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

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

**Table 2. P060 Probe targets in Reference Selection DNA SD082-S01**

| Probemix | Gene/Exon   | Probe length | Probe ID     | Present in probemix version | Copy number |
|----------|-------------|--------------|--------------|-----------------------------|-------------|
| P060     | Reference   | 154 nt       | 02595-L17085 | B2                          | 2           |
|          | Reference   | 163 nt       | 02291-L17086 | B2                          | 2           |
|          | Reference   | 172 nt       | 02978-L17087 | B2                          | 2           |
|          | SMN1/exon 7 | 183 nt       | 14919-L17081 | B2                          | 2           |
|          | Reference   | 191 nt       | 00559-L17088 | B2                          | 2           |
|          | Reference   | 200 nt       | 00976-L17298 | B2                          | 2           |
|          | Reference   | 208 nt       | 12490-L17096 | B2                          | 2           |
|          | SMN1/exon 8 | 218 nt       | 14881-L17082 | B2                          | 2           |
|          | Reference   | 228 nt       | 14498-L17101 | B2                          | 2           |
|          | Reference   | 237 nt       | 02334-L17301 | B2                          | 2           |
|          | Reference   | 245 nt       | 14293-L17100 | B2                          | 2           |
|          | Reference   | 255 nt       | 13128-L17099 | B2                          | 2           |
|          | Reference   | 264 nt       | 07630-L17091 | B2                          | 2           |
|          | Reference   | 272 nt       | 14361-L17098 | B2                          | 2           |
|          | SMN2/exon 7 | 282 nt       | 14921-L17083 | B2                          | 2           |
|          | Reference   | 292 nt       | 00824-L17097 | B2                          | 2           |
|          | SMN2/exon 8 | 301 nt       | 14878-L17084 | B2                          | 2           |
|          | Reference   | 311 nt       | 06425-L17092 | B2                          | 2           |
|          | Reference   | 321 nt       | 01042-L17093 | B2                          | 2           |
|          | Reference   | 331 nt       | 01043-L17094 | B2                          | 2           |
|          | Reference   | 342 nt       | 13399-L17297 | B2                          | 2           |

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**More information:** [www.mlpa.com](http://www.mlpa.com); [www.mlpa.eu](http://www.mlpa.eu)

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|   |   |
|---|---|
|  | EUROPE*  |
|  | ALL OTHER COUNTRIES   |

\*comprising EU (candidate) member states and members of the European Free Trade Association (EFTA).  
The product is for RUO in all other European countries.

**Implemented Changes – compared to the previous SD082 product description versions**

*Version S01-02 – 29 April 2019 (02)*

- Product is now registered for IVD use with P060.
- Table 2 added.

*Version S01-01 – 06 November 2018 (01)*

- Not applicable, new document.