

Product Description

SALSA® Reference Selection & Binning DNA SD084-S02

Version S02

As compared to version S01, the content was modified by replacing synthetic DNA with plasmid DNA without changes in the target sequences.

Catalogue number

- **SD084:** SALSA Reference Selection & Binning DNA, 6 reactions

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 before use: www.mrcholland.com. Reference Selection & Binning DNA is not known to contain any harmful agents.

Safety data sheet

Based on the concentrations present, none of the ingredients are hazardous as defined by the Hazard Communication Standard. **A Safety Data Sheet (SDS) is not required for these products:** none of the preparations contain dangerous substances (as per Regulation (EC) No 1272/2008 [EU-GHS/CLP] and amendments) at concentrations requiring distribution of an SDS (as per Regulation (EC) No 1272/2008 [EU-GHS/CLP] and 1907/2006 [REACH] and amendments). If spills occur, clean with water and follow appropriate site procedures.

General information

The SALSA Reference Selection & Binning DNA SD084 is a research use only (RUO) reagent to be used in combination with SALSA MLPA probemix P460-A1 SMA (Silent) Carrier, a SALSA MLPA Reagent Kit, and Coffalyser.Net™ analysis software for the selection of suitable reference samples and the processes of linking all probe signals to their identity by use of the probe lengths. SD084 contains the targets of all probes included in the above-listed probemix, including two polymorphism-specific probe targets g.27134T>G and g.27706-27707delAT in *SMN1* intron 7 and exon 8, respectively.

We recommend the use of Reference Selection & Binning DNA SD084 only for initial experiments on DNA samples from healthy individuals with the intention of selecting suitable reference samples. Reference Selection & Binning DNA should never be used as a reference sample in the MLPA data analysis of patient samples. Neither should it be used in quantification of polymorphism signals.

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 samples is complicated. Suitable reference samples for use with the P460-A1 SMA (Silent) Carrier probemix should be diploid for *SMN1* and *SMN2*, but should not contain the g.27134T>G and g.27706-27707delAT polymorphisms.

Experimental set up

Reference Selection & Binning DNA SD084 is a hybrid product with dual functionality: (1) selection of suitable reference samples, and (2) linking of probe signals to their identity (i.e. binning).

Experiments for the selection of suitable reference samples should include three reactions with SALSA Reference Selection & Binning DNA SD084 as well as reactions on a number of independent DNA samples from healthy individuals. Patient samples should not be included in the experiment.

For binning purposes, inclusion of one reaction with SALSA Reference Selection & Binning DNA SD084 in the initial MLPA experiment is essential as it can aid in data binning of the peak pattern when using Coffalyser.Net software. Furthermore, one reaction with Reference Selection & Binning DNA should be included in the experiment whenever changes have been applied to the set-up of the capillary electrophoresis device (e.g. when a different polymer type is used).

Individual MLPA reactions for reference selection and binning purposes should be performed with 5 µl of Reference Selection & Binning DNA. Please note that the SD084 reaction for binning purposes in the initial

MLPA experiment can be one of the three SD084 reactions included in an experiment for the selection of suitable reference samples.

Data analysis

Coffalyser.Net software should be used for analysis of MLPA experiments. Coffalyser.Net software is freely available at www.mrcholland.com.

To select suitable reference samples, the three SD084 reactions should be set as reference samples in the data analysis. Suitable reference samples will be those samples from healthy individuals that have no signal for the polymorphism-specific probes and a final probe ratio between 0.80 and 1.20 for all other probes included in the probemix. Suitable reference samples selected as described can subsequently be used in experiments with patient samples.

For binning purposes, select SD084 in the *bin smpl* –column when performing the fragment analysis step in Coffalyser.Net. By selecting the SD084 sample as your binning sample, probes will be correctly identified in the peak pattern across all samples.

Reference Selection & Binning DNA content

SD084 consists of a mixture of human genomic DNA purified from a selected cell line and a titrated amount of plasmid DNA.

The cell line has two copies of the *SMN1* and *SMN2* genes and the P460-A1 probemix contains two probes specific for *SMN1* and one probe specific for *SMN2* (for details, see Table 1).

The plasmid DNA included in SD084 contains partial sequences of the *SMN1* gene. These partial sequences include two different polymorphisms that will be detected by the polymorphism-specific probes present in the above-listed probemix. See Table 1 and the corresponding probemix product description for more details on the polymorphism-specific probe targets present. The indicated polymorphism-specific probes will generate a signal on SD084.

Please note that the plasmid DNA also contains the target sequence of the 105 nt chromosome Y specific control fragment. As a result, the 100 and 105 nt control fragments indicate the presence of two copies chromosome X and one copy chromosome Y.

Table 1. P460 SMA (Silent) Carrier probe targets in Reference Selection & Binning DNA SD084-S02

| Probe length (nt) | Gene/Exon | Probe ID | Probemix version | Copy number | Polymorphism details |
|-------------------|------------------------|--------------|------------------|-------------|----------------------|
| 131 | Reference | 00797-L25925 | A1 | 2 | |
| 136 | Reference | 18457-L23634 | A1 | 2 | |
| 143 | SMN1 / intron 7 | S0938-L26163 | A1 | n.a. | g.27134T>G |
| 148 | SMN1 / exon 8 | S0961-L25586 | A1 | n.a. | g.27706-27707delAT |
| 154 | SMN1 / exon 8 | S0960-L25957 | A1 | 2 | |
| 163 | Reference | 02291-L17086 | A1 | 2 | |
| 172 | Reference | 02978-L17087 | A1 | 2 | |
| 183 | SMN1 / exon 7 | 14919-L17081 | A1 | 2 | |
| 191 | Reference | 00559-L17088 | A1 | 2 | |
| 200 | Reference | 00976-L17298 | A1 | 2 | |
| 208 | Reference | 12490-L17096 | A1 | 2 | |
| 228 | Reference | 14498-L17101 | A1 | 2 | |
| 237 | Reference | 02334-L17301 | A1 | 2 | |
| 245 | Reference | 14293-L17100 | A1 | 2 | |
| 255 | Reference | 13128-L17099 | A1 | 2 | |
| 264 | Reference | 07630-L17091 | A1 | 2 | |
| 272 | Reference | 14361-L17098 | A1 | 2 | |
| 282 | SMN2 / exon 7 | 14921-L17083 | A1 | 2 | |
| 292 | Reference | 18491-L23716 | A1 | 2 | |
| 301 | Reference | 12783-L13918 | A1 | 2 | |
| 311 | Reference | 06425-L17092 | A1 | 2 | |
| 321 | Reference | 01042-L17093 | A1 | 2 | |
| 331 | Reference | 01043-L17094 | A1 | 2 | |

Note: Mutation nomenclature used here may differ from literature! The exon numbering used in this SD084-S02 Reference Selection & Binning DNA product description is the traditional exon numbering (exons 1, 2a, 2b, and 3-8). This exon numbering is different from the NCBI reference sequences for **SMN1** and **SMN2**. Please consult the corresponding probemix product description for more information about exon numbering and gene transcripts used.

| More information: www.mrcholland.com ; www.mrcholland.eu | |
|---|---|
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| Implemented changes in the product description |
|---|
| <p><i>Version S02-01 - 23 October 2024 (01)</i></p> <ul style="list-style-type: none"> - Product description adapted to the new version of SD084. - Regulatory status has changed: SD084 is for research use only from version S02 onwards. <p><i>Version S01-03 - 02 June 2022 (03)</i></p> <ul style="list-style-type: none"> - Product description adapted to a new template. <p><i>Version S01-02 - 13 July 2021 (02)</i></p> <ul style="list-style-type: none"> - Name of P460 probemix changed from 'SMA' to 'SMA (Silent) Carrier'. <p><i>Version S01-01 - 25 February 2021 (02)</i></p> <ul style="list-style-type: none"> - Not applicable, new document. |