

Product Description SALSA® Reference Selection DNA SD072-S01

Version S01.

Catalogue number: SD072: SALSA® Reference Selection DNA, 20 reactions

To be used with the following SALSA MLPA probemixes: P008-C1 PMS2 and P236-B1 CFH Region, 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).

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 before use: www.mlpa.com. **Reference Selection DNA should never be used as a reference sample in the MLPA data analysis.**

Intended use: The SALSA Reference Selection DNA SD072 is an in vitro diagnostic (IVD)¹ or research use only (RUO) reagent to be used in combination with SALSA MLPA Probemix P008-C1 PMS2 or SALSA MLPA Probemix P236-B1 CFH Region, a SALSA MLPA Reagent Kit and Coffalyser.Net analysis software for the selection of suitable reference DNA samples. We recommend the use of this Reference Selection DNA SD072 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. When testing DNA samples from healthy Caucasian individuals, approximately 25% of all DNA samples tested are suitable as reference DNA for use with SALSA MLPA Probemix P008-C1 PMS2 and approximately 50% of all DNA samples tested are suitable as reference DNA for use with SALSA MLPA Probemix P236-B1 CFH Region.

¹Please note that this Reference Selection DNA is for in vitro diagnostic (IVD) use in the countries specified at the end of this product description. In all other countries, the product is for research use only (RUO).

Experimental set up: MLPA reactions for reference selection purposes should be performed with 5 µl of Reference Selection DNA. Initial experiments for the selection of suitable reference DNA samples should include three reactions with SALSA Reference Selection DNA SD072 as well as reactions on a number of DNA samples from healthy individuals. Patient samples should not be included in the experiment.

Data analysis: Coffalyser.Net software must be used for analysis of MLPA experiments. This software is available free of charge on www.mlpa.com. When the SD072 reactions are set as reference samples in the data analysis, suitable reference DNA samples will be those samples from healthy individuals that have a final probe ratio between 0.80 and 1.20 for all probes included in the probemix. Suitable reference DNA samples selected as described can subsequently be used as reference DNA samples in experiments with patient samples.

Reference Selection DNA content: SD072 Reference Selection DNA is human genomic DNA purified from a selected cell line. This cell line has two copies each of both alleles for five different SNPs that are present in the *PMS2* gene and the *PMS2CL* pseudogene. Probes for these 10 alleles are present in SALSA MLPA probemix P008-C1 PMS2 (for details, see Table 1). The cell line also has two copies of the *CFH*, *CFHR1*,

CFHR2, *CFHR3*, *CFHR4* and *CFHR5* genes. Probes for these six genes are included in SALSA MLPA probemix P236-B1 CFH Region (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. P008 Probe targets in Reference Selection DNA SD072-S01

Probemix	Gene/Exon	Probe length	Probe ID	Present in probemix version	Copy number	Remarks
P008	Reference	128 nt	00797-L00093	C1	2	
	PMS2 exon 11	133 nt	14452-L00900	C1	2	
	PMS2 exon 9	140 nt	14448-L16160	C1	2	
	PMS2 exon 1	146 nt	07935-L16148	C1	2	
	Reference	154 nt	02417-L04306	C1	2	
	Reference	160 nt	08583-L08584	C1	2	
	PMS2/PMS2CL exon 11	165 nt	14453-L16164	C1	2	SNP probe
	PMS2/PMS2CL exon 11	171 nt	14453-L16165	C1	2	SNP probe
	Reference	177 nt	04359-L03779	C1	2	
	PMS2 exon 2	184 nt	01176-L16620	C1	2	
	PMS2/PMS2CL exon 14	190 nt	15768-L18167	C1	4	Detects gene and pseudogene
	Reference	196 nt	07510-L07172	C1	2	
	PMS2/PMS2CL exon 14	202 nt	14458-L16176	C1	2	SNP probe
	PMS2/PMS2CL exon 14	208 nt	14458-L16177	C1	2	SNP probe
	PMS2/PMS2CL exon 13	214 nt	14456-L16511	C1	2	SNP probe
	PMS2/PMS2CL exon 13	220 nt	14456-L16512	C1	2	SNP probe
	Reference	226 nt	07083-L06712	C1	2	
	PMS2 exon 5	232 nt	14445-L16154	C1	2	
	PMS2/PMS2CL intron 12	238 nt	14455-L16168	C1	2	SNP probe
	PMS2/PMS2CL intron 12	244 nt	14455-L16169	C1	2	SNP probe
	PMS2/exon 6	250 nt	01180-L16157	C1	2	
	PMS2/PMS2CL exon 13	261 nt	15767-L17448	C1	4	Detects gene and pseudogene
	Reference	268 nt	19040-L09299	C1	2	
	PMS2 exon 7	276 nt	01181-L16158	C1	2	
	PMS2/PMS2CL exon 12	283 nt	15769-L17786	C1	4	Detects gene and pseudogene
	Reference	292 nt	11087-L11770	C1	2	
	PMS2 exon 8	299 nt	01182-L16159	C1	2	
	PMS2 exon 3	310 nt	19910-L26895	C1	2	
	PMS2 exon 10	319 nt	01184-L00745	C1	2	
	Reference	328 nt	08543-L08544	C1	2	
	PMS2 exon 1	338 nt	07934-L16147	C1	2	
	PMS2/PMS2CL exon 15	349 nt	14460-L04046	C1	2	SNP probe
	PMS2/PMS2CL exon 15	356 nt	14460-L16180	C1	2	SNP probe
	PMS2 exon 11	364 nt	14451-L16163	C1	2	
	Reference	373 nt	02528-L01959	C1	2	
	PMS2/PMS2CL exon 14	382 nt	15293-L17051	C1	4	Detects gene and pseudogene
	PMS2 exon 3	390 nt	19915-L26898	C1	2	
	PMS2 exon 2	400 nt	14441-L16150	C1	2	
	PMS2/PMS2CL exon 15	409 nt	01189-L00750	C1	4	Detects gene and pseudogene
	PMS2 exon 4	418 nt	19906-L26893	C1	2	
	Reference	427 nt	06029-L05485	C1	2	
	PMS2 exon 6	436 nt	14447-L16623	C1	2	
	PMS2 exon 9	445 nt	14449-L16622	C1	2	
	PMS2 exon 5	454 nt	14446-L16621	C1	2	
	PMS2 exon 10	463 nt	14450-L16162	C1	2	
	Reference	472 nt	15978-L18133	C1	2	
	Reference	483 nt	08480-L08491	C1	2	

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.

Table 2. P236 Probe targets in Reference Selection DNA SD072-S01

Probemix	Gene/Exon	Probe length	Probe ID	Present in probemix version	Copy number
P236	Reference	130 nt	00797-L00463	B1	2
	CFHR3 upstream	135 nt	22996-L32432	B1	2
	CFH downstream	139 nt	22043-L08618	B1	2
	CFH exon 2	142 nt	07821-L07575	B1	2
	CFHR4 exon 10	148 nt	22111-L31098	B1	2
	CFHR3 exon 4	154 nt	22069-L31040	B1	2
	Reference	157 nt	02731-L01824	B1	2
	CFHR3 exon 1	164 nt	07832-L07588	B1	2
	CFHR3 exon 6	168 nt	08218-L09921	B1	2
	CFH exon 15	172 nt	22071-L31042	B1	2
	CFH exon 3	179 nt	07822-L07576	B1	2
	CFHR2 exon 4	184 nt	07844-L07600	B1	2
	Reference	190 nt	03915-L03370	B1	2
	CFHR1 exon 4	196 nt	22072-L31043	B1	2
	CFH exon 1	202 nt	07820-L07574	B1	2
	CFH exon 18	208 nt	22073-L31044	B1	2
	CFH exon 14	214 nt	22074-L31045	B1	2
	Reference	220 nt	08879-L08935	B1	2
	CFHR2 exon 3	226 nt	21368-L31327	B1	2
	CFHR5 exon 3	232 nt	07847-L07603	B1	2
	CFHR3 upstream	238 nt	22997-L32433	B1	2
	CFHR1 exon 5	244 nt	22076-L31047	B1	2
	CFHR5 exon 8	253 nt	22077-L31048	B1	2
	Reference	258 nt	16472-L26940	B1	2
	CFHR2 exon 2	265 nt	07842-L07598	B1	2
	CFHR3 exon 2	274 nt	07833-L07589	B1	2
	CFHR1 intron 1	283 nt	22112-L31100	B1	2
	CFH intron 9	292 nt	22079-L31050	B1	2
	Reference	301 nt	02767-L02196	B1	2
	CFH exon 12	310 nt	07828-L07583	B1	2
	CFHR4 exon 5	317 nt	22994-L32539	B1	2
	CFH exon 22	324 nt	22044-L31698	B1	2
	CFHR5 exon 1	330 nt	07845-L30998	B1	2
	CFH exon 6	337 nt	07824-L07578	B1	2
	CFHR1 intron 3	346 nt	07839-L07595	B1	2
	Reference	355 nt	05991-L05416	B1	2
	CFHR3 intron 4	364 nt	07835-L07591	B1	2
	CFH intron 11	373 nt	07827-L07582	B1	2
	CFH exon 17	382 nt	07830-L07586	B1	2
	CFHR3 exon 3	392 nt	07834-L07590	B1	2
	CFHR4 exon 6	400 nt	22558-L31052	B1	2
	CFHR2 intron 1	406 nt	22113-L31101	B1	2
	Reference	414 nt	12787-L20671	B1	2
	CFH exon 4	419 nt	07823-L16758	B1	2
	CFHR5 exon 2	427 nt	07846-L16757	B1	2
	CFH exon 21	436 nt	22082-L31053	B1	2
	CFHR4 exon 1	445 nt	22084-L31055	B1	2
	CFHR1 exon 6	454 nt	22995-L32431	B1	2
	Reference	463 nt	12460-L13461	B1	2
	CFH exon 19	472 nt	22559-L31056	B1	2
	CFHR5 exon 10	481 nt	22086-L31057	B1	2
	CFHR1 exon 2	494 nt	22087-L31058	B1	2
	Reference	500 nt	09682-L22509	B1	2

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*  ISRAEL
	ALL OTHER COUNTRIES

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

Implemented Changes – compared to the previous SD072 product description versions
<p><i>Version S01-04 – 12 May 2021 (02)</i></p> <ul style="list-style-type: none"> - Intended use updated. - Product can now be used with P236-B1. - UK has been added to the list of countries in Europe that accept the CE mark. <p><i>Version S01-03 – 15 January 2021 (02)</i></p> <ul style="list-style-type: none"> - Intended use updated. - Product description rewritten and adapted to a new template. - More details on the selection of suitable reference DNA samples added to the experimental set-up and data analysis sections. - Minor textual changes in Table 1. <p><i>Version S01-02 – 11 May 2020 (01)</i></p> <ul style="list-style-type: none"> - Product is now registered for IVD use in Israel. <p><i>Version S01-01 – 03 October 2017 (01)</i></p> <ul style="list-style-type: none"> - Not applicable, new document.