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MC002 SMA Newborn Screen: study finds 100% sensitivity & specificity on DBS cards of SMA patients

MRC-Holland recently launched SALSA® MC002 SMA Newborn Screen, an affordable tool to perform neonatal population screening for Spinal Muscular Atrophy (SMA). The MC002 assay was developed in collaboration with Dutch neonatal screening lab Isala. A clinical performance study by Isala and two other labs using MC002 SMA Newborn Screen on anonymised dried blood spot (DBS) cards of 47 SMA patients and 375 controls found a 100% diagnostic sensitivity and specificity. MC002 was able to detect the absence of the *SMN1* exon 7 DNA sequence (which plays a pivotal role in SMA etiology), thereby reliably discriminating *SMN1* from its paralog gene *SMN2*. Secondly, the assay did not detect asymptomatic carriers - an added advantage in newborn screening. The MC002 test's concordance with the second-tier 'golden standard' P021 SMA MLPA test was 100%. The authors concluded "the MC002 test showed the feasibility and accuracy of SMA screening in a neonatal screening program".

Read the original article here, or read more about MC002 SMA Newborn Screen here. Interested in a free trial kit? Contact info@mlpa.com.

For more on SMA research, check out our posters at ESHG 2019 – see below.

ESHG 2019: meet us at the MRC-Holland booth – posters on SMA and more

MRC-Holland will be present at ESHG 2019 in Gothenburg. Come and visit us at stand #548! We will also present three posters based on our research collaborations:

1. ***Validation of a fast, robust, inexpensive, two-tiered neonatal screening test algorithm on dried blood spots for spinal muscular atrophy.*** A poster on the SMA newborn screening study described above. P10.22A.

2. ***Screening multiple populations reveals large differences in the prevalence of a truncated SMN gene lacking exon 7 and 8.*** In-house testing of a large set of samples with our SMA products revealed a relatively frequent version of the SMN genes: a truncated SMN gene lacking exons 7 and 8. This has some relevancy for Spinal Muscular Atrophy, as the presence of such a truncated copy of the gene may mask *SMN1* ex1-6 deletions. P10.29D.

3. *Copy number analysis of the OPN1LW and OPN1MW genes by MLPA.* A novel MLPA assay enables copy number quantification and identification of gene conversions in the *OPN1LW* and *OPN1MW* genes involved in eye cone disorders. Adding MLPA to routine sequencing of *OPN1LW* and *OPN1MW* improves detection of the cause of X-linked cone dysfunction disorders. Note: this product is currently in a test phase. P02.46D.

Avoid false results – use TE to dissolve your DNA samples

Are your DNA samples dissolved in water? Then you risk obtaining false MLPA results due to the effects of DNA depurination. In depurination, a purine base (adenine or guanine) is removed from the DNA deoxyribose backbone. If this happens to the target DNA sequence, this can affect MLPA probe binding and final results. The effects of depurination may differ per probe and per sample, and they may therefore appear intermittently. Dissolving samples in TE or adding Tris-HCl to your sample prevents this issue.

Depurination happens if a DNA sample has insufficient buffering capacity to maintain the pH at 8.0-8.5. Water has insufficient buffering capacity to prevent depurination during the heating of DNA in the first step of MLPA. Problems may be exacerbated by other sample treatments (formalin fixation/paraffin embedding, prolonged heating), and therefore may not affect all samples. Secondly, depurination does not affect all MLPA probes equally. Unavoidable differences in probe target sequence or design can make a probe more sensitive to depurination. If DNA depurination affects a probe in a test sample, this may present itself as a deletion. If depurination affects the reference samples, this may manifest itself as an apparent gain of the target probe in the test sample.

See the MLPA protocol section *2.2. Sample treatment and storage* for details, or learn more about depurination here.

Have you tried our HhaI enzyme yet?

In 2018, we introduced our own HhaI enzyme and many customers made the switch to SALSA HhaI. If you are also interested in trying it, please note that the enzyme needs to be explicitly included on your orders (product code: SMR51) because it is not a part of the standard MLPA reagent kits.

Czech customers now order directly from MRC-Holland

MRC-Holland has terminated the agreement with Czech distributor Biogen. All Czech orders can be placed with us at order@mlpa.com. In case of questions, contact info@mlpa.com.

Reminder: New MLPA buffer and Ligase-65

As communicated previously, we have improved two components of our reagent kit: SALSA MLPA Buffer and SALSA Ligase-65. The old buffer and enzyme will be supplied free of charge with orders of reagent kits until 31 May 2019. From 1 June to 31 August, the old reagents are only supplied on request (still free of charge); please mention “old version” on your order. After 31 August, only the new MLPA buffer and Ligase-65 will be available.

Come and meet us

▸ **June 15-18**
ESHG 2019
Gothenburg, Sweden
Booth #548

MLPA workshops 2019

▸ **Sweden**
Gothenburg (14 June)
prior to ESHG*
open for registration

▸ **Switzerland****
Bern (11 September)
open for registration

▸ **Norway**
Bergen (31 October)
open for registration

▸ **Israel*****
Tel Aviv (20 and 21 November)
open for registration

* Registration is independent from ESHG and participation at ESHG is not required.

** Workshop held in English, materials also available in French.

*** This workshop is organised with our local partner: Pronto Diagnostics (Israel).

Some workshops may be open to local customers only.

The general workshop programme is available on our website. For more information, please contact us.

CE marked

▸**EK20 MLPA reagent kit** (for 2000 reactions)

Recently improved

▸**ME001 Tumour suppressor mix1 probemix** (Tumour suppressor genes)
▸**ME034 Multi-locus Imprinting probemix** (Multi-locus imprinting defects, To distinguish maternal and paternal triploidies)
▸**P090 BRCA2 probemix** (Breast and ovarian cancer, hereditary)
▸**P188 22q13 probemix** (Phelan-Mcdermid syndrome)
▸**P239 BRCA1 region probemix** (Breast cancer)

To be discontinued

- **P096 Mental retardation-2** (Mental retardation syndromes) sold until July 2019
- **P208 Human Telomere-6** (Subtelomeric testing) sold until June 2019
- **P230 Human Telomere-7** (Subtelomeric testing) sold until June 2019
- **P286 Human Telomere-11** (Subtelomeric testing) sold until July 2019
- **P291 Human Telomere-12** (Subtelomeric testing) sold until July 2019

To receive automatic notifications about product improvements and the release of new related products, add products that interest you to your MyMLPA account!