



qPCR ASSAY FOR THE DETECTION OF SMN1 MUTATION FROM DRIED BLOOD SPOTS OF NEONATES



Targeted qPCR™ SMA kit

Spinal Muscular Atrophy (SMA) is an autosomal recessive disease caused by a deletion in the **7th exon of SMN1 gene**.

The Targeted qPCR™ SMA kit is designed to genotype the mutation site associated with SMA without differentiation between heterozygous and wild type and is provided in an easy-to-use format.



Identifies only homozygous SMA



Pre-dispensed plate with MasterMix



30min extraction time



Test description

The test is divided into **two steps**: an initial **extraction and lysis step** and a second **DNA amplification step** via qPCR.

The extraction and lysis of the sample on filter paper is carried out by adding two buffers and a 30-minute incubation step on microtitration plates (not included).

The extracted sample is then transferred to the Master Mix **pre-filled microtitration plate** provided in the kit.

The amplification step is completed using the Biorad CFX96 thermocycler and the qPCR programme lasts 1 hour. The FAM fluorochrome is used for detecting the **wild-type SMN1 allele** and the HEX fluorochrome for detecting the **RPP30** 'housekeeping gene'.

The sample from a healthy patient will generate a HEX and FAM signal and an SMA patient will generate a HEX signal only.

A synthetic DNA test, recognised by HEX and FAM, is provided with the kit to ensure proper function of the amplification step.



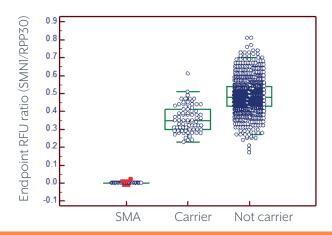
Benefits



- Specificity: detects homozygous only
- Saves time: plates pre-filled with the Master Mix
- **Speed:** result in approximately 90 minutes
- Quality control: positive control provided in the kit
- Practical: extraction and lysis buffers included and ready to use

Performances

No differenciation between heterozygous SMA (carrier) and healthy patient (not carrier)



General product information

- Storage at -20°C
- Ordering information:

Targeted qPCR™ SMA

Product code: K-LT-480

Size: 480 determinations





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