RefGen

RefKit Spinal Muscular Atrophy qPCR Screening Kit SMN1/SMN2

Spinal muscular atrophy (SMA) is a neurodegenerative disease that affects spinal and brainstem motor neurons and results in progressive, symmetrical muscle weakness. SMA is caused by mutations in the survival motor neuron (SMN) gene and represents itself as an autosomal recessive disorder. Located on chromosome 5q13, SMN gene is found in two forms as SMN1 and SMN2. While SMN1 produces functional protein, transcripts of SMN2 mostly generate unfunctional version. Therefore, most of the SMA cases are a result of the loss of SMN1 gene (caused by mutations on the gene). Increased copy number of SMN2, on the other hand, can result in a milder phenotype in SMA patients. Given that infant deaths caused by SMA have a high incidence worldwide and that the carrier frequencies are at a considerable rate, routine SMA screening becomes a high priority of the countries.

The RefKit Spinal Muscular Atrophy Screening Kit SMN1/SMN2 detects c.840C>T on exon 7 in the SMN1 and SMN2 genes of sample DNAs isolated from peripheral blood samples and from dry blood spots by using Quantitative Real Time PCR (qPCR) to reveal the copy number of SMN2 and copy number of wild type, carrier and sick individuals in terms of SMN1 gene. Based on probe-based RT-PCR technology, three different primer-probe sets are used, two of which detect SMN1 and SMN2 genes, while one of which targets CFTR gene that is used as an internal control. Kit also includes a wild type control as an external control to determine the copy number of extracted samples.

Ordering Information:

RP21-A48	RefKit Spinal Muscular Atrophy qPCR Screening Kit - SMN1/SMN2	48 rxns
RP21-A480	RefKit Spinal Muscular Atrophy qPCR Screening Kit - SMN1/SMN2	480 rxns

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