TRIMARIS

SMA REAL-TIME PCR ASSAY

- SMA is a severe autosomal neurodegenerative genetic disorder, that leads to disability and eventually death. SMA has an estimated incidence of 1:10000 live births and carrier frequencies ranging from 1:40-1:70 in various populations [1].
- Detection of SMA affected and SMA carrier status are possible through genetic testing, even at the pre-conception stage.

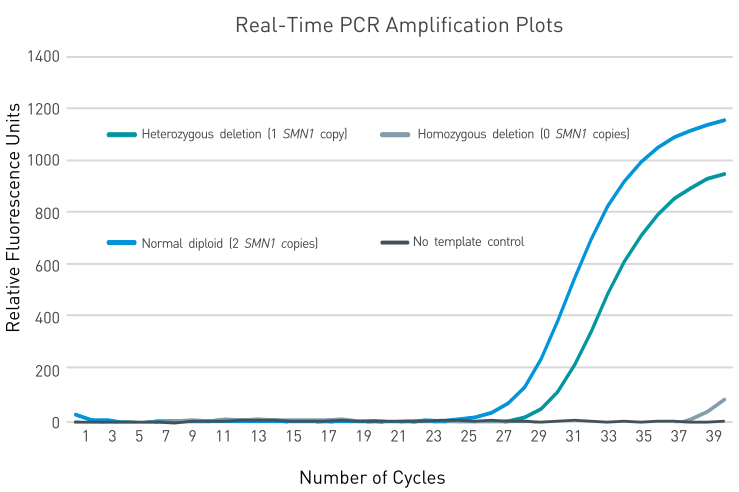
REAL-TIME PCR (5'exonuclease assay)

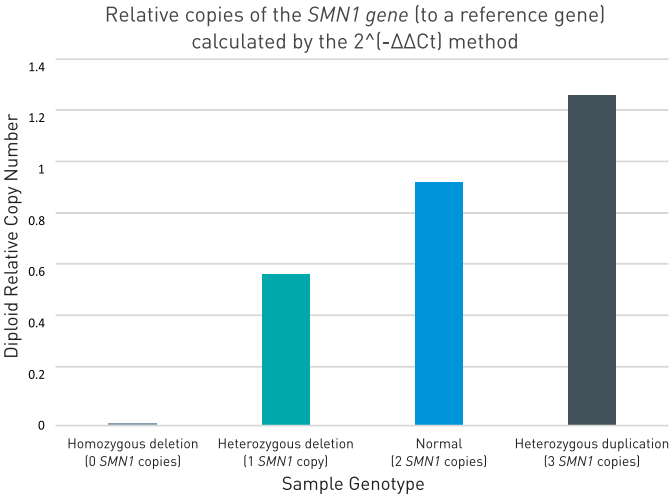
- SMA REAL-TIME PCR is a high performance molecular genetic test.
- The test is used to identify SMA affected individuals (*SMN1* homozygous deletions) using easily available laboratory instrumentation; it can also be used for the detection of SMA carriers (*SMN1* homozygous deletions).
- It can be used to test from small amounts of DNA e.g. from samples such as Dried Blood Spots (DBS).

Causes of SMA:

- Most cases (95 – 98%) of SMA are caused by homozygous deletions in exon 7 (often extending to exon 8) of the *SMN1* gene.
- The remaining 2-5% cases are caused by small sequence variants in the *SMN1* gene.
- All SMA carriers harbor only one functional copy of the *SMN1* gene i.e. heterozygous deletions of exon 7 (and exon 8) in 92-95% of cases [1,2].
- Silent carriers may have a "2+0" genotype where two copies of *SMN1* gene are present on the same chromosome and no copies on the other one [1, 2, 3, 4].

Accurate detection of *SMN1* homozygous deletions (affected individuals); can also be used to detect heterozygous deletions (carriers):





Limitations of the PCR Assay:

- PCR based methods like real-time/ quantitative PCR are effective for diagnosis of affected individuals, however, accurate determination of carrier status is challenging.
- This PCR assay cannot reveal absolute *SMN1* and *SMN2* copy numbers nor can it detect the "2+0" genotype. The SMA CODE-SEQ assay is recommended for the detection of these types of variations.
- SMA caused by small sequence variants in the *SMN1* gene cannot be detected.

Ordering details for SMA REAL-TIME PCR kits:

Product Code	Number of Tests
BR110020	24
BR110021	96

References:

1 Prior TW, Finanger E. Spinal Muscular Atrophy. 2000 Feb 24 [Updated 2016 Dec 22]. In: Adam MP, Ardinger HH, Pagon RA, et al., editors. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2019. Available from: https://www.ncbi.nlm.nih.gov/books/NBK1352/

2 Prior TW, Nagan N, Sugarman EA, Batish SD, Braastad C. Technical standards and guidelines for spinal muscular atrophy testing. Genet Med. 2011 Jul;13(7):686-94. doi: 10.1097/GIM.0b013e318220d523.

3 Phenotype-driven gene target definition in clinical genome-wide sequencing data interpretation. Genet Med. 2016 Jul;18(7):752. doi:10.1038/gim.2016.64. PubMed PMID: 27359096

4 Luo M, Liu L, Peter I, Zhu J, Scott SA, Zhao G, Eversley C, Kornreich R, Desnick RJ, Edelmann L. An Ashkenazi Jewish SMN1 haplotype specific to duplication alleles improves pan-ethnic carrier screening for spinal muscular atrophy. Genet Med. 2014 Feb;16(2):149-56. doi: 10.1038/gim.2013.84. Epub 2013. Jun 20.

5 Strom, C. , Anderson, B. , Peng, M. , Patel, U. , Braastad, C. and Sun, W. 1000 sample comparison of MLPA and RT-PCR for carrier detection and diagnostic testing for Spinal Muscular Atrophy Type 1. Open Journal of Genetics, 3, 111-114. doi: 10.4236/ojgen.2013.32014

This SMA REAL-TIME PCR assay (*Gene*Path Diagnostics Inc., Ann Arbor, MI, USA) is manufactured under license by Bome Trivitron in Turkey.

GENEPATH DX™

BOME TRIVITRON
speaking your language

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