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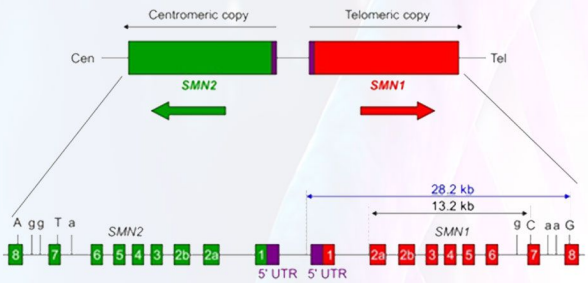
TECHNOLOGY

INOGene-SMA PCR Detection Kit

Spinal Muscular Atrophy (SMA) is a neurodegenerative, monogenic, and autosomal recessive disease that occurs by decreasing SMN protein levels in motor neurons depending on a single nucleotide mutation in the SMN1 gene. SMA can be seen by the ratio of 1/10000 and depending on its type it can result in death, life-long disability, and muscular atrophy because of the death of motor neurons in the body. Classificication of SMA is shown in the table below.

Type	Age of Onset	Maximal Motor Milestone	Motor Ability and Additional Features	Prognosis
SMA 0	Before Birth	None	Unable to sit or roll	Death within weeks
SMA I	2 weeks or 3 month	None	Unable to sit or roll	Death by to 2 years
SMA II	6 to 18 motnhs	Sitting	Unable to walk independently	Survival into adulthood
SMA III	<3 years, <12 years	Walking	May lose ability to walk	Normal life span
SMA IV	>30 years or 10 to 30	Normal	Mild motor impairment	Normal life span



SMA is caused by recessive mutations in the survival motor neuron 1 (SMN1) gene. About 95% of SMA cases are caused by homozygous deletions of exon 7 in SMN1, whereas the remaining cases exhibit a heterozygous mutation on one allele and other deleterious variants on the other. The human genome harbors a paralogous gene, SMN2, that differs from SMN1 by only a few nucleotides including a C to T transition in exon 7. This base change causes the skipping of exon 7 in most SMN2 transcripts. Approximately 90% of transcript isoforms encode a truncated unstable protein; full-length, functional SMN protein results from approximately 10% of SMN2 transcripts.

INOGene-SMA PCR Detection Kit

Nucleic acid extraction can be performed from whole blood samples with any commercial DNA isolation / extraction kits.

The Kit provides detection of the single nucleotide mutation occurs in the 840th base which causes skipping exon 7 and thus, spinal muscular atrophy. This single nucleotide change in the exon 7 is detected by using specially designed oligonucleotides and MGB tagged probes. The curves and Ct values of FAM, HEX, and CY5 fluorophores are analyzed at the end of real-time PCR to determine homozygous and heterozygous mutations in the individual.

The kit comes with reaction mix, primer probe mix, negative template control, and positive controls.

Product	Product Size	REF No.
INOGene-SMA PCR Detection Kit	25 rxns	INOSMA-0025
INOGene-SMA PCR Detection Kit	50 rxns	INOSMA-0050

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