

Bring **Technology** to **Life!**



# SPINAL MUSCULAR ATROPHY

Newborn screening  
Carrier screening

## Human Survival Motor Neuron 1 (SMN1) Gene Detection Kit

### (PCR-Melting Curve Method)

Spinal muscular atrophy (SMA) is an autosomal recessive genetic disease characterized by degeneration of motor neurons in the anterior horn of the spinal cord, which clinically manifests as progressive and symmetrical muscle weakness, atrophy, and paralysis in the proximal extremity and trunk. SMN1 is the main pathogenic gene of SMA. 95% of SMA patients show homozygous deletion of SMN1 in exon 7. The remaining 5% show heterozygous deletion of SMN1 in exon 7 and point mutation compound heterozygous deletion of SMN1.

Based on PCR melting curve method, Tianlong's Human Survival Motor Neuron 1 (SMN1) Gene Detection Kit is used for detecting the copy number of SMN1 in exon 7 and exon 8 of human genomic DNA. This kit is suitable for the auxiliary diagnosis of SMN1 patients and screening SMN1 gene carriers.

Technique	PCR-Melting Curve Method	
Used for	Neonatal Screening	✓
	Patient	✓
	Carrier	✓
Coverage	SMN1 exon 7	✓
	SMN1 exon 8	✓



### Features



#### Quality Performance

Ability to differentiate between 0, 1, and  $\geq 2$  copies for SMN1 in exon 7 and exon 8, detection of SMA carrier and SMA patients



#### Reduced Complexity

Tianlong Gentier96E/R automates the original data into multiple melting curve analysis software for data analysis



#### Optimized Workflow

DNA-to-data in 2-3 hours with only 60 minutes of hands-on-time



#### Low Sample Concentration

The minimum sample detection concentration of the kit is 10ng/ $\mu$ L



#### High Precision

The coefficient of variation (CV,%) of the R value is  $\leq 6\%$



#### More Accessible

CE and NMPA marked, accessible for more counties

## Data Interpretation

Figure 1: SMN1 exon 7

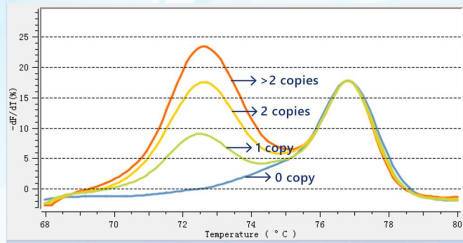
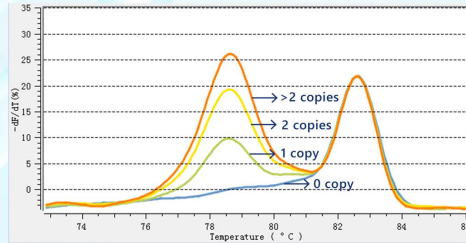


Figure 2: SMN1 exon 8



SMA patient: SMN1=0 copy, homozygous deletion  
SMA carrier: SMN1=1 copy, heterozygosity deletion

Note: Tianlong has a patent for multiple melting curve analysis software, which can achieve standardized data interpretation and avoid calculation error. The copy number can be provided and the results can be more accurate and reliable.

Figure 3: Tianlong's SMN 1 kit and kit using MLPA method (SMN1 exon 7)

	MLPA method			Total
	0 copy	1 copy	2 copies and above	
Tianlong SMN 1 kit	0 copy	261	0	261
	1 copy	0	31	31
	2 copies and above	0	0	1171
<b>Total</b>	<b>261</b>	<b>31</b>	<b>1171</b>	<b>1463</b>

Figure 4: Tianlong's SMN 1 kit and kit using MLPA method (SMN1 exon 8)

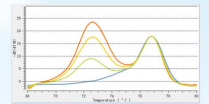
	MLPA method			Total
	0 copy	1 copy	2 copies and above	
Tianlong SMN 1 kit	0 copy	236	0	236
	1 copy	0	54	54
	2 copies and above	0	0	1173
<b>Total</b>	<b>236</b>	<b>54</b>	<b>1171</b>	<b>1463</b>

Note: The results show that the coincidence rate of the two products in the samples with different SMN1 copy number is 100%.

## Ordering Information

Product Name	Human Survival Motor Neuron 1 (SMN1) Gene Detection Kit (PCR-Melting Curve Method)
Cat.No	P124H
Specification	32T/Kit
Specimen	EDTA anticoagulant whole blood sample
Target Gene	SMN1 in exon 7, SMN1 in exon 8
Storage & Validity	-25°C~-15°C for 10 months
Applicable Equipment	Roche Light Cycler® 480, Tianlong Gentier 96E/96R real time PCR system
Certification	CE/NMPA

## Assay workflow



❖ Extract 2~5mL of venous blood with a disposable sterile syringe and inject into a glass tube containing EDTA anticoagulant

❖ Tianlong Whole Blood Nucleic Acid Extraction Kit  
❖ Tianlong Nucleic Acid Extractors

❖ Tianlong Human Survival Motor Neuron 1 (SMN1) Gene Detection Kit  
❖ Tianlong Gentier 96E/R Real-time PCR Systems

❖ Tianlong's SMN 1 detection kit can differentiate 0, 1, and ≥2copies for SMN1 in exon 7 and exon 8

version 2.0

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