

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	V	
	Carrier	V	
Coverage	SMN1 exon 7	V	
	SMN1 exon 8	V	



Features



Quality Performance

Ability to differentiate between 0, 1, and ≥2copies 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/μL



High Precision

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



More Accessible

CE and NMPA marked, accessible for more counties

Date Interpretation /

Figure 1: SMN1 exon 7

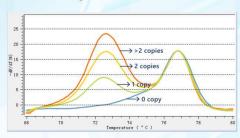
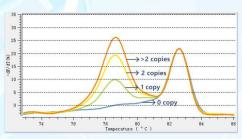


Figure 2: SMN1 exon 8



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				
		0 сору	1 сору	2 copies and above	Total
Tianlong SMN 1 kit	0 сору	261	0	0	261
	1 сору	0	31	0	31
	2 copies and above	0	0	1171	1171
	Total	261	31	1171	1463

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

	MLPA method				
		0 сору	1 сору	2 copies and above	Total
Tianlong SMN 1 kit	0 сору	236	0	0	236
	1 сору	0	54	0	54
	2 copies and above	0	0	1173	1173
	Total	236	54	1171	1463

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

Ordering Information

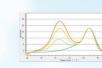
Product Name	Human Survival Motor Neuron 1 (SMN1) Gene Detection Kit (PCR-Melting Curve Method)		
Cat.No	P124H		
Specification	32T/Kit		
Specimen	EDTA anticoagulant whole blood sample		
Target Gene	SMN1 in exon 7, SMN1 in exon 8		
Storage & Validity	-25°C∼-15°C for 10 months		
Applicable Equipment	Roche 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
- * Tianlong Nucleic Acid Extractors
- Tianlong Human Survival Motor Neuron 1 (SMN1) Gene Detection Kit
- Tianlong Gentier 96E/R Real-time PCR
- Tianlong's SMN 1 detection kit can differentiate 0, 1, and ≥2copies for SMN1 in exon 7 and exon 8

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