

Table 1. Repeatability evaluation of qPCR method for different samples

Sample	tests	SMN1-7	SMN1-8
		R value	Rvalue
Single copy	1	0.53	0.57
	2	0.56	0.57
	3	0.56	0.56
	4	0.54	0.57
	5	0.55	0.55
	6	0.56	0.54
	7	0.55	0.55
	8	0.56	0.59
Average value		0.55	0.56
Standard deviation		0.01	0.02
CV		2.04%	2.81%
Two copies	1	0.98	1.01
	2	0.94	0.98
	3	0.97	1
	4	0.94	0.99
	5	0.95	0.99
	6	0.96	1.01
	7	0.92	1.03
	8	0.94	1.02
Average value		0.95	1.00
Standard deviation		0.02	0.02
CV		2.03%	1.68%

Table 2. SMN1 Exon 7 and Exon 8 Copy Number of 2563 Samples

SMN1 exon 7 copy	SMN1 exon 8 copy			Total	Rate,%	95%CI,%
	0 copy	1 copy	≥2 copies			
0 copy	236	19	6	261	10.18	9.07-11.41
1 copy	0	61	1	62	2.42	1.89-3.09
≥2 copies	0	11	2229	2240	87.40	86.06-88.63
Total	236	91	2236	2563	100	

Table 3. The Copy Number of SMN1 in different groups

copy number	SMA-positive		SMA-suspected		normal screening		
	number	frequency (%)	number	frequency (%)	number	frequency (%)	
exon 7	0	260	98.86	1	0.33	0	0
	1	3	1.14	12	4.00	43	2.15
	2	0	0	287	95.67	1957	97.85
exon 8	0	235	89.35	1	0.33	0	0
	1	22	8.37	11	3.67	54	2.7
	2	6	2.28	288	96.00	1946	97.30