Case: 2_to_2_of_SMN1_to_SMN2-normal Folder: P021-vs03-examples Date: 03-03-2009

P021-vs03 SMA Spinal Muscular Atrophy (lot 0807) Female & male ref.: P021-vs03_001 Ver.: 5.11

No.	Label	Size	Ref. size	Size diff.	MRC size	Peak Height	Peak Width	Peak Area	Peak Area	Ref. Mean	Ref. SD	Ref. Weigh	Position p-tel	Position band	Ratio	Dist. in SD	1.0 low high
	64 -		62.12		64					0.740	0.130			64 nt			
	70 -		67.96		70					0.733	0.341			70 nt			
	76 -		73.59		76					0.807	0.333			76 nt			
	82 -		79.77		82					1.720	0.170			82 nt			
Ctrl: Q-fragments					Mean							1.00		(CV:)			
1	6 a	85.55	85.57	-0.02	88	1260	15.9 *	19990	0.586	0.658	0.061	1.17		6p21.3 CpG is.	0.89	-1.2	
2	2 a	90.90	90.93	-0.03	92	1316	15.4 *	20294	0.594	0.658	0.053	1.34		2q14 synt.	0.90	-1.2	
3	1 a	96.84	96.83	0.01	96	1099	18.1 *	19902	0.583	0.632	0.140	0.49		1p36 CpG is.	0.92	-0.3	
Ctrl: D-fragments					Mean	1225	16.5	20062	0.588	0.649	0.085	1.00		(CV: **0.02**)	**0.90**		
4	12 A	137.34	137.38	-0.04	139	2548	15.8	40366	1.182	1.193	0.035	1.39	66.8	12q14	0.99	-0.3	
6	3 A	155.67	155.70	-0.03	157	2226	16.5	36759	1.077	1.094	0.075	0.59	99.8	3q12	0.98	-0.2	
7	17 A	164.29	164.36	-0.07	166	1395	16.6	23133	0.678	0.744	0.062	0.48	16.8	17p11.2	0.91	-1.1	
8	18 A	173.67	173.77	-0.10	175	1851	16.4	30354	0.889	0.887	0.038	0.95	55.7	18q21	1.00	0.1	
10	7 A	191.45	191.49	-0.04	193	2299	17.0	39063	1.144	1.124	0.034	1.33	86-9	7q21	1.02	0.6	
12	10 A	208.76	208.80	-0.04	211	2032	17.3	35144	1.030	0.959	0.060	0.64	1.45	10p15	1.07	1.2	
14	11 B	228.22	228.24	-0.02	229	2604	18.9	49198	1.006	0.959	0.032	1.21	65.2	11q13	1.05	1.5	
16	21 B	245.65	245.66	-0.01	247	2510	18.5	46371	0.948	0.996	0.054	0.75	14.67	21q11	0.95	-0.9	
17	18 B	253.21	253.20	0.01	256	2736	20.1	54973	1.124	1.136	0.043	1.08	75.32	18p11	0.99	-0.3	
20	3 B	283.75	283.78	-0.03	286	2988	20.2	60489	1.237	1.233	0.050	1.01	156.3	3q21	1.00	0.1	
23	13 B	309.63	309.72	-0.09	310	1562	21.5	33512	0.685	0.676	0.030	0.91	110.6	13q34	1.01	0.3	
24	8 C	319.62	319.65	-0.03	319	1240	21.6	26771	0.812	0.769	0.037	0.85	141.9	8q24	1.06	1.2	
26	21 C	336.85	336.90	-0.05	337	1304	22.4	29225	0.887	0.855	0.047	0.73	26.3	21q21	1.04	0.7	
28	2 C	352.81	352.79	0.02	355	1954	23.2	45382	1.377	1.406	0.036	1.56	71.5	2p24	0.98	-0.8	
30	8 C	371.79	371.79	0.00	373	1119	24.7	27627	0.838	0.864	0.043	0.82	38.4	8p11.2	0.97	-0.6	
32	3 C	389.37	389.31	0.06	391	1391	25.8	35827	1.087	1.107	0.059	0.76	10.2	3p26	0.98	-0.3	
34	2 D	407.24	407.19	0.05	409	1579	26.7	42218	1.219	1.235	0.032	1.57	55.2	2p14	0.99	-0.5	
36	8 D	423.95	423.83	0.12	427	1387	28.7 *	39792	1.149	1.097	0.042	1.05	117.8	8q24	1.05	1.2	
37	17 D	432.21	432.10	0.11	436	1501	27.7	41636	1.202	1.160	0.033	1.42	35.0	17q12	1.04	1.3	
38	13 D	442.22	442.08	0.14	445	887	28.2	25009	0.722	0.786	0.038	0.84	110.2	13q34	0.92	-1.7	
40	12 D	460.21	460.11	0.10	463	836	29.3	24525	0.708	0.722	0.028	1.05	120.9	12q24	0.98	-0.5	
Reference fragments					Mean	1807	21.8	37494	1.000	1.000	0.043	1.00		(CV: **0.04**)	**1.00**		
5	5 a	146.46	146.50	-0.04	148	1987	16.0	31697	0.929	0.950	0.041	1.00	31.3	CDH6-D01	0.98	-0.5	
5p15.1-14					Mean	1987	16.0	31697	0.929	0.950	0.041	1.00		(CV:)	**0.98**		
11	5 a	200.03	200.06	-0.03	202	1437	17.2	24677	0.723	0.738	0.034	1.00	68.7	RAD17-D02	0.98	-0.5	
Before SMN					Mean	1437	17.2	24677	0.723	0.738	0.034	1.00		(CV:)	**0.98**		
19	5 b	274.48	274.50	-0.02	276	1648	19.2	31565	0.645	0.660	0.046	0.84	69.4	SMN2 ex 7	0.98	-0.3	
22	5 b	298.44	298.46	-0.02	301	837	21.5	17957	0.367	0.375	0.019	1.16	69.4	SMN2 ex 8	0.98	-0.4	
SMN2	**>>>>>>>>>>>>**				Mean	1243	20.3	24761	0.506	0.518	0.033	1.00		(CV: **0.00**)	**0.98**		
15	5 b	236.01	236.02	-0.01	238	1569	18.1	28379	0.580	0.679	0.063	1.00	70.3	BIRC1 ex 5	0.85	-1.6	
Before SMN1					Mean	1569	18.1	28379	0.580	0.679	0.063	1.00		(CV:)	**0.85**		
18	5 b	268.22	268.23	-0.01	270	1163	19.9	23086	0.472	0.452	0.024	0.91	70.3	SMN1 ex 7	1.04	0.8	
21	5 b	292.22	292.25	-0.03	295	897	19.8	17779	0.364	0.385	0.017	1.09	70.3	SMN1 ex 8	0.95	-1.2	
SMN1	**>>>>>>>>>>>>**				Mean	1030	19.8	20433	0.418	0.418	0.021	1.00		(CV: **0.07**)	**0.99**		
31	5 c	379.74	379.68	0.06	382	1847	24.1	44589	1.353	1.448	0.132	0.61	70.3	SMN1/2 ex 1	0.93	-0.7	
33	5 d	397.82	397.78	0.04	400	1543	26.5	40906	1.181	1.160	0.061	1.07	70.3	SMN1/2 ex 4	1.02	0.4	
35	5 d	417.02	417.02	0.00	418	1377	27.1	37352	1.078	1.049	0.039	1.50	70.3	SMN1/2 ex 6	1.03	0.8	
29	5 c	361.29	361.21	0.08	364	1838	23.9	43986	1.334	1.390	0.095	0.82	70.3	SMN1/2 ex 8	0.96	-0.6	
SMN1/2	**>>>>>>>>>>>>**				Mean	1651	25.4	41708	1.237	1.262	0.082	1.00		(CV: **0.04**)	**1.00**		
25	5 c	328.27	328.38	-0.11	328	1374	21.7	29761	0.903	0.913	0.114	1.25	70.4	GTF2H2ex 4	0.99	-0.1	
13	5 b	216.06	216.10	-0.04	220	1284	18.0	23075	0.472	0.535	0.084	0.99	70.4	GTF2H2ex 7	0.88	-0.8	
9	5 a	183.79	183.90	-0.11	184	1558	16.5	25747	0.754	0.817	0.170	0.75	70.4	GTF2H2ex10	0.92	-0.4	
GTF2H2					Mean	1405	18.7	26194	0.710	0.755	0.123	1.00		(CV: **0.06**)	**0.94**		
39	5 d	451.88	451.84	0.04	454	2406	30.1 *	72347	2.089	2.133	0.075	1.27	69.3	N-Cadherin-like	0.98	-0.6	
27	5 c	344.79	344.84	-0.05	346	2640	22.9	60344	1.830	1.802	0.110	0.73	70.5	BIRC1 ex 13	1.02	0.3	
SMN1/2 like					Mean	2523	26.5	66346	1.960	1.967	0.092	1.00		(CV: **0.02**)	**0.99**		
Mean values				-0.01		1712	21.6	36233	**0.975**	0.986	0.055	**3**			0.99	Total of all except	
Standard deviations				0.06		(Coef. of variance:		0.342)	0.365	0.362					0.05	Ctrl and '?' peaks	

Quality assessment	**Quality limits**	**Quality**
Not evaluated as < 2 Q-fragments	>0.65 (1.50)	
Mean CpG-area / mean A-group area	>0.35 (0.55)	0.58
Mean height of first probes AB	> 450 (800)	2250
Mean height of last probes CD	> 280 (500)	1320
Ratio of mean heights AB/CD ('slope')	<3.00 (2.50)	1.70
CV of Control Probes	<0.20 (0.15)	0.04
0 uinidentified peak areas / 40 peak areas	< (0.02)	0.00

SMN1 and SMN2 peaks are assigned the nearest ploidy.
Probe ratios close (±0.025) to 0.75 or to 1.25 are difficult.
Difference to predicted SMN1/2 level should be inside ±0.125,
or at least SMN1/2ex8 should be predicted well.

SMN1 to SMN2 copies look like 2:2 (exon 7) and 2:2 (exon 8)

2:2 and 2:2 for exon 7 & 8 predicts SMN1/2 to 1.00 diff.=-0.003

An "*" marks: Size Diff.>0.5, Peak Height>7000, unexpected peak width, and "Dist. in SD">4.0.
Ratio group mean and coefficient of variance (CV) are weighted by the ref. weights
Labels A,B,... define normalization groups; a,b,... labeled probes do not contribute to normalization.
(No Rox peaks are avaliable in the raw data)

(Ctrl probes are used for quality evaluation only)