Case: 2_to_0-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	Norm. 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	85.56	85.57	-0.01	88	1396	16.3 *	22805	0.712	0.658	0.061	1.17		6p21.3 CpG is.	1.08	0.9	·|
2	2 a	90.93	90.93	0.00	92	1330	16.1 *	21397	0.668	0.658	0.053	1.34		2q14 synt.	1.02	0.2	·
3	1 a	96.82	96.83	-0.01	96	1411	17.6 *	24859	0.776	0.632	0.140	0.49		1p36 CpG is.	1.23	1.0	·||
Ctrl: D-fragments					Mean	1379	16.7	23020	0.718	0.649	0.085	1.00		(CV: **0.08**)	**1.08**		
4	12 A	137.32	137.38	-0.06	139	2394	16.6	39778	1.241	1.193	0.035	1.39	66.8	12q14	1.04	1.4	·
6	3 A	155.71	155.70	0.01	157	2065	16.6	34263	1.069	1.094	0.075	0.59	99.8	3q12	0.98	-0.3	·
7	17 A	164.33	164.36	-0.03	166	1464	16.8	24620	0.768	0.744	0.062	0.48	16.8	17p11.2	1.03	0.4	·
8	18 A	173.75	173.77	-0.02	175	1698	17.0	28875	0.901	0.887	0.038	0.95	55.7	18q21	1.02	0.4	·
10	7 A	191.43	191.49	-0.06	193	2045	17.2	35149	1.097	1.124	0.034	1.33	86-9	7q21	0.98	-0.8	·
12	10 A	208.69	208.80	-0.11	211	1724	17.2	29581	0.923	0.959	0.060	0.64	1.45	10p15	0.96	-0.6	·
14	11 B	228.16	228.24	-0.08	229	2307	18.0	41440	0.957	0.959	0.032	1.21	65.2	11q13	1.00	-0.1	·
16	21 B	245.57	245.66	-0.09	247	2560	18.0	46097	1.064	0.996	0.054	0.75	14.67	21q11	1.07	1.3	·|
17	18 B	253.10	253.20	-0.10	256	2697	18.5	50018	1.155	1.136	0.043	1.08	75.32	18p11	1.02	0.4	·
19	3 B	283.70	283.78	-0.08	286	2692	18.7	50404	1.164	1.233	0.050	1.01	156.3	3q21	0.94	-1.4	|·
21	13 B	309.71	309.72	-0.01	310	1427	20.1	28640	0.661	0.676	0.030	0.91	110.6	13q34	0.98	-0.5	·
22	8 C	319.72	319.65	0.07	319	1099	20.5	22543	0.724	0.769	0.037	0.85	141.9	8q24	0.94	-1.2	|·
24	21 C	336.99	336.90	0.09	337	1206	20.8	25069	0.806	0.855	0.047	0.73	26.3	21q21	0.94	-1.0	|·
26	2 C	352.76	352.79	-0.03	355	1906	22.0	41981	1.349	1.406	0.036	1.56	71.5	2p24	0.96	-1.6	·
28	8 C	371.83	371.79	0.04	373	1145	24.0	27514	0.884	0.864	0.043	0.82	38.4	8p11.2	1.02	0.5	·
30	3 C	389.31	389.31	0.00	391	1382	27.8 *	38471	1.236	1.107	0.059	0.76	10.2	3p26	1.12	2.2	·|
32	2 D	407.22	407.19	0.03	409	1550	28.7 *	44504	1.271	1.235	0.032	1.57	55.2	2p14	1.03	1.1	·
34	8 D	423.75	423.83	-0.08	427	1221	30.6 *	37419	1.069	1.097	0.042	1.05	117.8	8q24	0.97	-0.7	·
35	17 D	431.99	432.10	-0.11	436	1411	30.0 *	42321	1.209	1.160	0.033	1.42	35.0	17q12	1.04	1.5	·
36	13 D	441.97	442.08	-0.11	445	886	30.6 *	27133	0.775	0.786	0.038	0.84	110.2	13q34	0.99	-0.3	·
38	12 D	460.01	460.11	-0.10	463	755	31.4 *	23712	0.677	0.722	0.028	1.05	120.9	12q24	0.94	-1.6	|·
Reference fragments					Mean	1697	22.0	35216	1.000	1.000	0.043	1.00		(CV: **0.04**)	**1.00**		
5	5 a	146.47	146.50	-0.03	148	1874	16.4	30800	0.961	0.950	0.041	1.00	31.3	CDH6-D01	1.01	0.3	·
5p15.1-14					Mean	1874	16.4	30800	0.961	0.950	0.041	1.00		(CV:)	**1.01**		
11	5 a	200.00	200.06	-0.06	202	1258	17.2	21642	0.675	0.738	0.034	1.00	68.7	RAD17-D02	0.91	-1.8	|·
Before SMN					Mean	1258	17.2	21642	0.675	0.738	0.034	1.00		(CV:)	**0.91**		
39	5 b	0.00	274.50	------	276	0	0.0	0	0.000	0.660	0.046	0.84	69.4	SMN2 ex 7	0.00	-14.2 *	|||||||||·
40	5 b	0.00	298.46	------	301	0	0.0	0	0.000	0.375	0.019	1.16	69.4	SMN2 ex 8	0.00	-19.7 *	|||||||||·
SMN2		**>>>>>>>>>>>>**			Mean	0	0.0	0	0.000	0.518	0.033	1.00		(CV:)	**0.00**		
15	5 b	235.90	236.02	-0.12	238	1580	17.8	28094	0.649	0.679	0.063	1.00	70.3	BIRC1 ex 5	0.95	-0.5	·
Before SMN1					Mean	1580	17.8	28094	0.649	0.679	0.063	1.00		(CV:)	**0.95**		
18	5 b	268.14	268.23	-0.09	270	1098	19.0	20830	0.481	0.452	0.024	0.91	70.3	SMN1 ex 7	1.06	1.2	·|
20	5 b	292.21	292.25	-0.04	295	865	19.3	16671	0.385	0.385	0.017	1.09	70.3	SMN1 ex 8	1.00	0.0	·
SMN1		**>>>>>>>>>>>>**			Mean	982	19.1	18751	0.433	0.418	0.021	1.00		(CV: **0.04**)	**1.03**		
29	5 c	379.72	379.68	0.04	382	988	26.5 *	26152	0.840	1.448	0.132	0.61	70.3	SMN1/2 ex 1	0.58	-4.6 *	||||·
31	5 d	397.80	397.78	0.02	400	708	26.5	18780	0.536	1.160	0.061	1.07	70.3	SMN1/2 ex 4	0.46	-10.3 *	|||||·
33	5 d	417.01	417.02	-0.01	418	695	27.0	18757	0.536	1.049	0.039	1.50	70.3	SMN1/2 ex 6	0.51	-13.2 *	|||||·
27	5 c	361.18	361.21	-0.03	364	984	22.3	21960	0.706	1.390	0.095	0.82	70.3	SMN1/2 ex 8	0.51	-7.2 *	|||||·
SMN1/2		**>>>>>>>>>>>>**			Mean	844	25.6	21412	0.655	1.262	0.082	1.00		(CV: **0.08**)	**0.51**		
23	5 c	328.42	328.38	0.04	328	1232	20.5	25215	0.810	0.913	0.114	1.25	70.4	GTF2H2ex 4	0.89	-0.9	|·
13	5 b	216.03	216.10	-0.07	220	1416	17.5	24816	0.573	0.535	0.084	0.99	70.4	GTF2H2ex 7	1.07	0.4	·|
9	5 a	183.83	183.90	-0.07	184	1410	16.7	23573	0.736	0.817	0.170	0.75	70.4	GTF2H2ex10	0.90	-0.5	|·
GTF2H2					Mean	1353	18.2	24535	0.706	0.755	0.123	1.00		(CV: **0.11**)	**0.95**		
37	5 d	451.74	451.84	-0.10	454	1570	32.1 *	50367	1.438	2.133	0.075	1.27	69.3	N-Cadherin-like	0.67	-9.3 *	|||·
25	5 c	344.88	344.84	0.04	346	1723	21.3	36726	1.180	1.802	0.110	0.73	70.5	BIRC1 ex 13	0.65	-5.6 *	|||·
SMN1/2 like					Mean	1647	26.7	43547	1.309	1.967	0.092	1.00		(CV: **0.02**)	**0.67**		
Mean values				-0.04		1433	20.6	29836	**0.852**	0.986	0.055	**3**			0.87	Total of all except	
Standard deviations				0.06		(Coef. of variance:		0.401)	0.334	0.362					0.27	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)	0.74
Mean height of first probes AB	> 450 (800)	2098
Mean height of last probes CD	> 280 (500)	1256
Ratio of mean heights AB/CD ('slope')	<3.00 (2.50)	1.67
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 2:0 (exon 7) and 2:0 (exon 8)

2:0 and 2:0 for exon 7 & 8 predicts SMN1/2 to 0.50 diff.= 0.008

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)