Case: 1_to_2-of_SMN1-to-SMN2-carrier 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	85.53	85.57	-0.04	88	1201	15.6 *	18677	0.633	0.658	0.061	1.17		6p21.3 CpG is.	0.96	-0.4	·
2	2 a	90.93	90.93	0.00	92	1224	15.7 *	19209	0.651	0.658	0.053	1.34		2q14 synt.	0.99	-0.1	·
3	1 a	96.81	96.83	-0.02	96	1235	17.7 *	21822	0.740	0.632	0.140	0.49		1p36 CpG is.	1.17	0.8	·||
Ctrl: D-fragments					Mean	1220	16.3	19903	0.675	0.649	0.085	1.00		(CV: **0.09**)	**1.01**		
4	12 A	137.37	137.38	-0.01	139	2232	16.4	36653	1.242	1.193	0.035	1.39	66.8	12q14	1.04	1.4	·
6	3 A	155.70	155.70	0.00	157	2005	16.3	32695	1.108	1.094	0.075	0.59	99.8	3q12	1.01	0.2	·
7	17 A	164.36	164.36	0.00	166	1259	16.6	20857	0.707	0.744	0.062	0.48	16.8	17p11.2	0.95	-0.6	·
8	18 A	173.71	173.77	-0.06	175	1616	16.3	26272	0.891	0.887	0.038	0.95	55.7	18q21	1.00	0.1	·
10	7 A	191.47	191.49	-0.02	193	1993	16.8	33433	1.133	1.124	0.034	1.33	86-9	7q21	1.01	0.3	·
12	10 A	208.80	208.80	0.00	211	1577	17.2	27087	0.918	0.959	0.060	0.64	1.45	10p15	0.96	-0.7	·
14	11 B	228.23	228.24	-0.01	229	2213	18.1	39948	0.934	0.959	0.032	1.21	65.2	11q13	0.97	-0.8	·
16	21 B	245.66	245.66	0.00	247	2302	18.2	41871	0.979	0.996	0.054	0.75	14.67	21q11	0.98	-0.3	·
17	18 B	253.20	253.20	0.00	256	2557	19.5	49987	1.169	1.136	0.043	1.08	75.32	18p11	1.03	0.8	·
20	3 B	283.72	283.78	-0.06	286	2535	20.1	50983	1.193	1.233	0.050	1.01	156.3	3q21	0.97	-0.8	·
23	13 B	309.57	309.72	-0.15	310	1443	21.5	30954	0.724	0.676	0.030	0.91	110.6	13q34	1.07	1.6	·|
24	8 C	319.49	319.65	-0.16	319	1077	21.4	23093	0.809	0.769	0.037	0.85	141.9	8q24	1.05	1.1	·|
26	21 C	336.71	336.90	-0.19	337	1183	22.0	26083	0.914	0.855	0.047	0.73	26.3	21q21	1.07	1.3	·|
28	2 C	352.84	352.79	0.05	355	1700	22.9	39010	1.367	1.406	0.036	1.56	71.5	2p24	0.97	-1.1	·
30	8 C	371.82	371.79	0.03	373	1019	23.8	24215	0.849	0.864	0.043	0.82	38.4	8p11.2	0.98	-0.4	·
32	3 C	389.41	389.31	0.10	391	1216	24.9	30276	1.061	1.107	0.059	0.76	10.2	3p26	0.96	-0.8	·
34	2 D	407.26	407.19	0.07	409	1336	26.4	35331	1.219	1.235	0.032	1.57	55.2	2p14	0.99	-0.5	·
36	8 D	423.90	423.83	0.07	427	1154	28.0	32327	1.115	1.097	0.042	1.05	117.8	8q24	1.02	0.4	·
37	17 D	432.21	432.10	0.11	436	1218	27.4	33373	1.151	1.160	0.033	1.42	35.0	17q12	0.99	-0.3	·
38	13 D	442.18	442.08	0.10	445	839	27.6	23137	0.798	0.786	0.038	0.84	110.2	13q34	1.02	0.3	·
40	12 D	460.35	460.11	0.24	463	716	29.0	20775	0.717	0.722	0.028	1.05	120.9	12q24	0.99	-0.2	·
Reference fragments					Mean	1580	21.4	32303	1.000	1.000	0.043	1.00		(CV: **0.03**)	**1.00**		
5	5 a	146.51	146.50	0.01	148	1758	16.3	28614	0.970	0.950	0.041	1.00	31.3	CDH6-D01	1.02	0.5	·
5p15.1-14					Mean	1758	16.3	28614	0.970	0.950	0.041	1.00		(CV:)	**1.02**		
11	5 a	200.06	200.06	0.00	202	1237	17.2	21228	0.720	0.738	0.034	1.00	68.7	RAD17-D02	0.97	-0.5	·
Before SMN					Mean	1237	17.2	21228	0.720	0.738	0.034	1.00		(CV:)	**0.97**		
19	5 b	274.55	274.50	0.05	276	1380	19.2	26515	0.620	0.660	0.046	0.84	69.4	SMN2 ex 7	0.94	-0.9	|·
22	5 b	298.43	298.46	-0.03	301	770	20.1	15504	0.363	0.375	0.019	1.16	69.4	SMN2 ex 8	0.97	-0.6	·
SMN2 >>>>>>>>>>>>					Mean	1075	19.7	21010	0.491	0.518	0.033	1.00		(CV: **0.02**)	**0.96**		
15	5 b	236.00	236.02	-0.02	238	721	18.0	12975	0.304	0.679	0.063	1.00	70.3	BIRC1 ex 5	0.45	-5.9 *	||||||·
Before SMN1					Mean	721	18.0	12975	0.304	0.679	0.063	1.00		(CV:)	**0.45**		
18	5 b	268.29	268.23	0.06	270	533	19.9	10613	0.248	0.452	0.024	0.91	70.3	SMN1 ex 7	0.55	-8.4 *	|||||·
21	5 b	292.21	292.25	-0.04	295	377	20.3	7648	0.179	0.385	0.017	1.09	70.3	SMN1 ex 8	0.47	-11.8 *	|||||·
SMN1 >>>>>>>>>>>>					Mean	455	20.1	9131	0.214	0.418	0.021	1.00		(CV: **0.12**)	**0.50**		
31	5 c	379.72	379.68	0.04	382	1180	24.0	28274	0.991	1.448	0.132	0.61	70.3	SMN1/2 ex 1	0.68	-3.5	|||·
33	5 d	397.84	397.78	0.06	400	988	26.2	25873	0.893	1.160	0.061	1.07	70.3	SMN1/2 ex 4	0.77	-4.4 *	||·
35	5 d	417.04	417.02	0.02	418	931	25.2	23455	0.809	1.049	0.039	1.50	70.3	SMN1/2 ex 6	0.77	-6.2 *	||·
29	5 c	361.29	361.21	0.08	364	1188	24.1	28687	1.005	1.390	0.095	0.82	70.3	SMN1/2 ex 8	0.72	-4.0 *	|||·
SMN1/2 >>>>>>>>>>>>					Mean	1072	24.9	26572	0.924	1.262	0.082	1.00		(CV: **0.05**)	**0.75**		
25	5 c	328.18	328.38	-0.20	328	1234	21.6	26695	0.936	0.913	0.114	1.25	70.4	GTF2H2ex 4	1.02	0.2	·
13	5 b	216.08	216.10	-0.02	220	1124	17.6	19800	0.463	0.535	0.084	0.99	70.4	GTF2H2ex 7	0.87	-0.9	|·
9	5 a	183.85	183.90	-0.05	184	1419	16.7	23739	0.805	0.817	0.170	0.75	70.4	GTF2H2ex10	0.98	-0.1	·
GTF2H2					Mean	1259	18.7	23411	0.734	0.755	0.123	1.00		(CV: **0.09**)	**0.96**		
39	5 d	451.90	451.84	0.06	454	1817	29.8 *	54149	1.868	2.133	0.075	1.27	69.3	N-Cadherin-like	0.88	-3.5	|·
27	5 c	344.76	344.84	-0.08	346	2000	22.4	44772	1.569	1.802	0.110	0.73	70.5	BIRC1 ex 13	0.87	-2.1	|·
SMN1/2 like					Mean	1909	26.1	49461	1.718	1.967	0.092	1.00		(CV: **0.00**)	**0.87**		
Mean values			0.00			1401	21.3	29105	**0.912**	0.986	0.055	3			0.92	Total of all except	
Standard deviations			0.09		(Coef. of variance:			0.365)	0.345	0.362					0.16	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)	0.69
Mean height of first probes AB	> 450 (800)	1976
Mean height of last probes CD	> 280 (500)	1146
Ratio of mean heights AB/CD ('slope')	<3.00 (2.50)	1.72
CV of Control Probes	<0.20 (0.15)	0.03
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 1:2 (exon 7) and 1:2 (exon 8)

1:2 and 1:2 for exon 7 & 8 predicts SMN1/2 to 0.75 diff.=-0.002

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)