Case: P064-B2-Normal-sample-1 Folder: P064-B2-vs01-examples Date: 11-03-2010

P064-B2-vs01 MR1 MLPA Kit (lot 0208) Female & male ref.: P064-B2-vs01_t03 Ver.: 5.21

			Peak Data						Normalized Peak Area						
No.	Label	Size	Ref. size	Size diff.	MRC size	Height	Width	Area	Peak Area	Ref. Mean	Ref. SD	Ref. Weigh	Position p-tel band	Ratio	Dist. in SD	1.0 low high
15	64 -	60.90	60.68	0.22	64	465	11.2	5198	1.666	1.349	0.175	1.31	64 nt	1.23	1.8	·||
16	70 -	67.31	66.72	0.59 *	70	169	13.3	2254	0.722	0.874	0.137	1.09	70 nt	0.83	-1.1	||·
17	76 -	73.16	72.98	0.18	76	191	14.3	2723	0.873	0.886	0.129	1.17	76 nt	0.98	-0.1	·
18	82 -	79.36	79.33	0.03	82	196	11.8	2308	0.740	0.887	0.351	0.43	82 nt	0.83	-0.4	||·
Ctrl: Q-fragments					Mean	255	12.6	3121	1.000	0.999	0.198	1.00	(CV: **0.20**)	**1.01**		
19	6 a	85.96	85.75	0.21	88	1968	7.0 *****	13733	1.394	1.378	0.697	0.62	6p21.3 CpG isl.	1.01	0.0	·
21	2 a	91.31	91.11	0.20	94	1712	5.6	9633	0.978	0.907	0.174	1.63	MV 36 2q14 synt.	1.08	0.4	·|
22	1 a	97.39	97.18	0.21	96	1237	6.2	7726	0.784	0.948	0.392	0.76	mv36 1p36.3 CpG isl.	0.83	-0.4	||·
Ctrl: D-fragments					Mean	1639	6.3	10364	1.052	1.078	0.421	1.00	(CV: **0.13**)	**1.00**		
55	1 D	380.90	380.84	0.06	382	826	7.5	6215	0.845	0.848	0.051	1.09	1.1 1p36.3	1.00	-0.1	·
23	1 A	126.93	126.84	0.09	130	2028	5.9	11914	1.209	1.246	0.092	0.89	1.1 1p36.33	0.97	-0.4	·
24	1 A	132.33	132.22	0.11	136	1570	6.0	9397	0.954	0.892	0.112	0.53	1.2 1p36.33	1.07	0.6	·|
31	1 A	177.45	177.41	0.04	178	1537	6.2	9533	0.968	1.003	0.070	0.95	1.8 1p36.33	0.96	-0.5	·
61	1 D	434.90	434.69	0.21	436	1042	8.1	8465	1.151	1.108	0.071	1.03	2.2 1p36	1.04	0.6	·
40	1 B	246.16	246.14	0.02	247	1633	6.6	10745	1.152	1.151	0.046	1.66	2.4 1p36.33	1.00	0.0	·
50	1 C	336.22	336.29	-0.07	337	1098	7.3	8053	0.950	0.931	0.074	0.83	3.6 1p36.3	1.02	0.3	·
1p36 (1p-deletion)					Mean	1391	6.8	9189	1.033	1.026	0.074	1.00	(CV: **0.03**)	**1.00**		
25	5 A	140.08	139.97	0.11	142	2135	5.9	12592	1.278	1.261	0.059	1.53	176.6 NSD1 exon 4	1.01	0.3	·
62	5 D	444.70	444.48	0.22	445	434	8.2	3557	0.484	0.495	0.057	0.62	176.6 NSD1 exon 12	0.98	-0.2	·
36	5 B	210.23	210.17	0.06	211	1088	6.6	7143	0.766	0.769	0.064	0.85	176.6 NSD1 exon 17	1.00	0.0	·
5q35.3 (Sotos)					Mean	1219	6.9	7764	0.843	0.842	0.060	1.00	(CV: **0.02**)	**1.00**		
48	7 C	319.82	319.89	-0.07	319	1052	6.9	7310	0.862	0.832	0.057	0.89	19.1 7p21.2	1.04	0.5	·
35	7 B	202.22	202.22	0.00	202	1448	6.3	9055	0.971	0.975	0.053	1.11	19.7 7p21.2	1.00	-0.1	·
7p21.2 (Saethre-Chotzen)					Mean	1250	6.6	8183	0.917	0.904	0.055	1.00	(CV: **0.03**)	**1.01**		
41	7 B	255.96	255.81	0.15	256	1722	6.6	11284	1.210	1.140	0.074	0.94	72.5 7q11.23	1.06	0.9	·|
45	7 C	289.85	289.72	0.13	292	1611	6.8	10881	1.283	1.312	0.069	1.18	72.8 7q11.23	0.98	-0.4	·
47	7 C	310.38	310.39	-0.01	310	1717	6.9	11807	1.392	1.433	0.095	0.93	73.1 7q11.23	0.97	-0.4	·
56	7 D	389.92	389.89	0.03	391	923	7.8	7174	0.976	1.014	0.057	1.10	73.2 7q11.23	0.96	-0.7	·
38	7 B	229.80	229.70	0.10	229	1334	6.4	8504	0.912	0.924	0.055	1.04	73.4 7q11.23	0.99	-0.2	·
59	7 D	416.37	416.29	0.08	418	1089	8.0	8735	1.188	1.323	0.100	0.81	73.4 7q11.23	0.90	-1.3	|·
7q11.23 (Williams)					Mean	1399	7.1	9731	1.160	1.191	0.075	1.00	(CV: **0.05**)	**0.98**		
30	15 A	171.25	171.19	0.06	172	960	6.1	5820	0.591	0.627	0.051	0.92	21.4 15q11.2	0.94	-0.7	|·
37	15 B	219.01	218.87	0.14	220	1335	6.2	8339	0.894	0.894	0.067	1.00	21.5 15q11.2	1.00	0.0	·
28	15 A	158.96	158.89	0.07	160	1579	6.1	9601	0.974	0.946	0.048	1.46	23.2 15q12	1.03	0.6	·
57	15 D	399.11	399.01	0.10	400	1719	8.3	14252	1.939	1.768	0.156	0.85	23.2 15q12	1.10	1.1	·|
58	15 D	407.86	407.68	0.18	409	901	7.9	7078	0.963	1.002	0.096	0.78	24.5 15q12	0.96	-0.4	·
15q11q12 (Prader-Willi)					Mean	1299	6.9	9018	1.072	1.047	0.084	1.00	(CV: **0.06**)	**1.01**		
53	17 C	363.53	363.42	0.11	364	749	7.4	5506	0.649	0.658	0.060	0.73	1.9 17p13.3	0.99	-0.1	·
32	17 A	184.08	184.00	0.08	184	1726	6.1	10512	1.067	1.123	0.070	1.08	1.9 17p13.3	0.95	-0.8	·
60	17 D	425.29	425.19	0.10	427	783	8.1	6327	0.861	0.861	0.090	0.64	2.3 17p13.3	1.00	0.0	·
39	17 B	237.04	236.93	0.11	238	1339	6.6	8772	0.941	0.969	0.055	1.17	2.5 17p13.3	0.97	-0.5	·
44	17 C	282.15	282.10	0.05	283	1217	6.7	8105	0.956	1.031	0.088	0.79	2.5 17p13.3	0.93	-0.9	|·
42	17 B	264.09	263.98	0.11	265	1559	6.5	10177	1.091	1.117	0.068	1.10	3.3 17p13.3	0.98	-0.4	·
26	17 A	147.25	147.04	0.21	148	1865	6.1	11306	1.148	1.140	0.051	1.49	3.4 17p13.3	1.01	0.1	·
17p13.3 (Miller-Dieker)					Mean	1320	6.8	8672	0.959	0.986	0.069	1.00	(CV: **0.03**)	**0.98**		
29	17 A	165.58	165.51	0.07	166	1473	6.3	9283	0.942	0.901	0.055	1.03	16.8 17p11.2	1.05	0.8	·
43	17 B	272.56	272.45	0.11	274	1429	6.7	9558	1.025	0.946	0.084	0.72	17.8 17p11.2	1.08	0.9	·|
46	17 C	301.50	301.46	0.04	301	1413	6.8	9657	1.139	1.052	0.061	1.09	18.1 17p11.2	1.08	1.4	·|
49	17 C	329.12	329.21	-0.09	328	1145	7.1	8098	0.955	0.994	0.052	1.21	18.7 17p11.2	0.96	-0.7	·
52	17 C	354.86	354.84	0.02	355	979	7.4	7248	0.855	0.799	0.053	0.95	19.2 17p11.2	1.07	1.1	·|
17p11.2 (Smith-Magenis)					Mean	1288	6.9	8769	0.983	0.938	0.061	1.00	(CV: **0.05**)	**1.04**		
51	20 C	345.07	345.09	-0.02	346	1132	7.4	8391	0.989	0.959	0.059	1.23	10.6 20p12.2 JAG1	1.03	0.5	·
65	20 D	470.90	470.71	0.19	472	579	8.5	4901	0.667	0.609	0.060	0.77	10.6 20p12.2 JAG1	1.09	1.0	·|
20p12.2 (Alagille)					Mean	856	7.9	6646	0.828	0.784	0.060	1.00	(CV: **0.04**)	**1.06**		
33	22 B	191.45	191.36	0.09	190	1715	6.1	10431	1.119	1.125	0.081	1.06	17.6 22q11.21	0.99	-0.1	·
64	22 D	463.15	463.02	0.13	463	1186	8.5	10068	1.370	1.346	0.094	1.08	17.9 22q11.21	1.02	0.3	·
34	22 B	196.52	196.48	0.04	196	1538	6.0	9230	0.990	0.989	0.075	1.00	17.9 22q11.21	1.00	0.0	·
27	22 A	154.12	153.98	0.14	154	1527	6.0	9198	0.934	0.862	0.071	0.92	18.4 22q11.21	1.08	1.0	·|
63	22 D	452.86	452.66	0.20	454	557	8.3	4611	0.627	0.626	0.050	0.94	19.2 22q11.21	1.00	0.0	·
54	22 C	371.70	371.62	0.08	373	1074	8.2	8823	1.040	0.999	0.076	0.99	19.6 22q11.21	1.04	0.5	·
22q11.21 (DiGeorge)					Mean	1266	7.2	8727	1.013	0.991	0.075	1.00	(CV: **0.03**)	**1.02**		
Mean values			0.08			1297	7.0	8783	**1.006**	1.000	0.070	**3**		1.01	Total of all except	
Standard deviations			0.07			(Coef. of variance:		0.251)	0.247	0.239				0.04	Ctrl and '?' peaks	

Quality assessment	**Quality limits**	**Quality**
Mean A-group area / mean Q-frag. area	>0.65 (1.50)	3.16
Mean CpG-area / mean A-group area	>0.30 (0.65)	1.09
Mean height of first probes AB	> 450 (800)	1550
Mean height of last probes CD	> 280 (500)	1056
Ratio of mean heights AB/CD ('slope')	<3.00 (2.50)	1.47
Mean group CV of weighted ratio	<0.20 (0.15)	0.04
0 uinidentified peak areas / 46 peak areas	< (0.02)	0.00

Weighted mean ratios are tested for being outside ratio 1±0.13
One-tailed significance is high for p<=1%, and low for p<=5%.
Individual peaks having normalized area > 4.0 SD from the ref. mean and ratio <0.65 or >1.3 indicate 'abnormal' probe area.

Female & male ref.

Normal probes

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.
Mean Rox height is 215 (14 peaks). CV of ROX heights for peaks above 100 nt is: 0.06

(Ctrl probes are used for quality evaluation only)