

Peak Data										Normalized Peak Area							
No.	Label	Size	Ref. size	Size diff.	MRC size	Height	Width	Area	Area	Ref. Mean	Ref. SD	Ref. Weigh	Position p-tel band	Ratio	Dist. in SD	1.0 low high	
3	64 -	60.70	60.65	0.05	64	154	7.5	1159	0.495	1.190	0.371	0.75	64 nt	0.42	-1.9	.	
4	70 -	66.42	66.46	-0.04	70	132	19.2	2533	1.081	0.913	0.264	0.80	70 nt	1.18	0.6		
5	76 -	72.57	72.37	0.20	76	171	20.8	3564	1.521	1.138	0.170	1.55	76 nt	1.34	2.3		
6	82 -	78.89	78.56	0.33	82	117	18.1	2116	0.903	0.787	0.204	0.90	82 nt	1.15	0.6		
Ctrl: Q-fragments					Mean	144	16.4	2343	1.000	1.007	0.252	1.00	(CV: 0.35)	1.09			
7	6 a	85.21	84.95	0.26	88	1546	11.2*	17347	0.876	0.981	0.152	1.02	6p21.3 CpG isl.	0.89	-0.7	.	
9	2 a	90.40	90.17	0.23	92	1728	10.3	17728	0.895	0.967	0.135	1.13	2q14 synt.	0.93	-0.5	.	
10	1 a	96.79	96.50	0.29	96	2354	11.3*	26649	1.346	1.641	0.304	0.85	MV 36 1p36 CpG isl.	0.82	-1.0	.	
Ctrl: D-fragments					Mean	1876	10.9	20575	1.039	1.197	0.197	1.00	(CV: 0.06)	0.88			
14	Y a	114.65	114.50	0.15	118	1229	10.2	12522	0.632	0.811	0.088	1.03	13.54 Yq11	0.78	-2.0	.	
12	Y a	104.44	104.34	0.10	108	911	10.3	9365	0.473	0.566	0.065	0.97	14.10 Yq11	0.84	-1.4	.	
ctrl: Y-fragments (male ref.)					Mean	1070	10.2	10944	0.553	0.688	0.076	1.00	(CV: 0.05)	0.81			
15	1 A	126.24	126.04	0.20	130	2282	10.8	24623	1.243	1.200	0.054	1.30	1.14 1p36.33	1.04	0.8	.	
23	1 A	176.61	176.49	0.12	178	1804	10.8	19482	0.984	0.942	0.074	0.75	1.75 1p36.33	1.04	0.6	.	
21	1 A	164.60	164.53	0.07	166	2239	10.9	24467	1.235	1.181	0.073	0.95	1.95 1p36.33	1.05	0.7	.	
1p36 (1p-deletion)					Mean	2108	10.8	22857	1.154	1.108	0.067	1.00	(CV: 0.01)	1.04			
37	2 B	264.95	264.89	0.06	267	1165	12.5	14606	0.877	0.948	0.040	1.36	58.30 2p16.1	0.92	-1.8	.	
63	2 D	481.59	481.31	0.28	486	780	18.1	14123	1.027	1.121	0.101	0.64	61.00 2p16.1	0.92	-0.9	.	
"2p16.1 deletion syndrome"					Mean	973	15.3	14365	0.952	1.034	0.071	1.00	(CV: 0.01)	0.92			
56	3 D	416.61	416.46	0.15	418	1103	16.0	17663	1.284	1.268	0.073	0.96	198.51 3q29	1.01	0.2	.	
49	3 C	354.47	354.58	-0.11	359	1058	13.9	14747	0.873	1.039	0.056	1.04	198.28 3q29	0.84	-3.0	.	
"3q29 deletion syndrome"					Mean	1081	15.0	16205	1.078	1.154	0.064	1.00	(CV: 0.13)	0.92			
32	4 B	230.71	230.47	0.24	232	1692	11.7	19751	1.186	0.944	0.063	0.88	1.81 4p16.3	1.26	3.9		
59	4 D	443.25	443.03	0.22	445	706	16.5	11620	0.845	0.879	0.046	1.12	1.90 4p16.3	0.96	-0.8	.	
4p16.3 Wolf-Hirschhorn region					Mean	1199	14.1	15686	1.015	0.912	0.054	1.00	(CV: 0.19)	1.09			
58	5 D	435.74	435.58	0.16	436	585	16.1	9438	0.686	0.659	0.053	0.88	1.34 5p15.33	1.04	0.5	.	
39	5 C	281.42	281.39	0.03	283	1595	13.1	20942	1.239	1.115	0.071	1.12	1.40 5p15.33	1.11	1.8		
Cri du Chat syndrome					Mean	1090	14.6	15190	0.963	0.887	0.062	1.00	(CV: 0.05)	1.08			
19	5 A	151.57	151.42	0.15	154	2170	10.3	22328	1.127	1.127	0.052	1.03	176.62 5q35.3	1.00	0.0	.	
60	5 D	453.07	453.02	0.05	454	818	17.2	14039	1.020	1.123	0.055	0.97	176.65 5q35.3	0.91	-1.9	.	
Sotos syndrome					Mean	1494	13.7	18184	1.074	1.125	0.054	1.00	(CV: 0.07)	0.96			
43	7 C	310.29	310.40	-0.11	310	1421	13.4	19109	1.131	1.116	0.056	0.99	73.08 7q11.23	1.01	0.3	.	
50	7 C	363.44	363.46	-0.02	364	1205	15.0	18056	1.069	1.029	0.040	1.27	73.11 7q11.23	1.04	1.0	.	
53	7 D	390.06	389.95	0.11	391	730	15.7	11463	0.833	0.748	0.050	0.74	73.15 7q11.23	1.11	1.7		
Williams syndrome					Mean	1119	14.7	16209	1.011	0.964	0.049	1.00	(CV: 0.05)	1.05			
54	8 D	400.00	399.69	0.31	401	674	15.9	10700	0.778	0.776	0.036	1.06	116.75 8q24.12	1.00	0.0	.	
57	8 D	424.80	424.33	0.47	427	700	16.9	11864	0.862	0.895	0.047	0.94	117.73 8q24.11	0.96	-0.7	.	
Langer-Giedion syndrome					Mean	687	16.4	11282	0.820	0.835	0.042	1.00	(CV: 0.03)	0.98			
44	9 C	319.30	319.40	-0.10	319	1616	13.5	21820	1.291	1.292	0.072	1.05	100.95 9q22.33	1.00	0.0	.	
55	9 D	408.28	408.03	0.25	409	946	15.5	14686	1.067	1.119	0.069	0.95	100.95 9q22.33	0.95	-0.8	.	
"9q22.3 deletion syndrome"					Mean	1281	14.5	18253	1.179	1.206	0.070	1.00	(CV: 0.03)	0.98			
16	10 A	132.85	132.56	0.29	136	1900	10.8	20436	1.032	0.948	0.043	1.16	8.14 10p	1.09	2.0		
48	10 C	349.22	349.23	-0.01	349	1119	14.4	16068	0.951	0.979	0.060	0.84	10.59 10p15.1	0.97	-0.5	.	
DiGeorge region 2 (10p)					Mean	1510	12.6	18252	0.991	0.963	0.052	1.00	(CV: 0.08)	1.04			
30	11 B	218.77	218.68	0.09	220	1957	11.5	22553	1.354	1.246	0.061	1.00	31.78 11p13	1.09	1.8		
WAGR syndrome					Mean	1957	11.5	22553	1.354	1.246	0.061	1.00	(CV:)	1.09			
29	15 B	213.58	213.54	0.04	214	999	11.2	11151	0.669	1.226	0.047	1.34	21.48 15q11.2	0.55	-11.8*	.	
34	15 B	245.23	245.06	0.17	247	673	12.1	8132	0.488	0.962	0.058	0.86	22.65 15q12	0.51	-8.2*	.	
40	15 C	289.89	289.75	0.14	292	660	12.8	8434	0.499	1.018	0.061	0.86	22.76 15q12	0.49	-8.5*	.	
20	15 A	158.13	158.00	0.13	160	862	10.9	9430	0.476	0.930	0.051	0.94	23.17 15q12	0.51	-8.9*	.	
Prader-Willi / Angelman					Mean	799	11.7	9287	0.533	1.034	0.054	1.00	(CV: 0.05)	0.52			
25	15 A	189.82	189.75	0.07	190	2477	11.0	27154	1.371	1.299	0.130	0.78	72.50 15q24.1	1.06	0.6		
45	15 C	326.22	326.43	-0.21	325	1072	13.9	14878	0.881	0.771	0.049	1.22	72.80 15q24.1	1.14	2.2		
"15q24 deletion syndrome"					Mean	1775	12.4	21016	1.126	1.035	0.090	1.00	(CV: 0.05)	1.11			
22	16 A	170.94	170.91	0.03	172	1798	10.5	18873	0.953	1.018	0.063	1.00	3.87 16p13.3	0.94	-1.0	.	
Rubinstein-Taybi syndrome					Mean	1798	10.5	18873	0.953	1.018	0.063	1.00	(CV:)	0.94			
17	17 A	139.18	138.97	0.21	142	2378	11.1	26424	1.334	1.248	0.053	1.27	2.51 17p13.3	1.07	1.6		
33	17 B	236.25	236.22	0.03	238	1345	11.7	15712	0.943	0.900	0.067	0.73	2.52 17p13.3	1.05	0.6	.	
Miller-Dieker region					Mean	1862	11.4	21068	1.139	1.074	0.060	1.00	(CV: 0.01)	1.06			
61	17 D	463.34	463.07	0.27	465	825	17.1	14074	1.023	1.331	0.092	0.83	17.53 17p11.2-##	0.77	-3.4	.	
38	17 B	271.94	271.90	0.04	274	1424	12.6	17991	1.080	0.940	0.043	1.25	17.83 17p11.2	1.15	3.2		
42	17 C	302.93	302.90	0.03	303	1188	13.1	15564	0.921	0.839	0.053	0.91	18.08 17p11.2	1.10	1.6		
Smith-Magenis syndrome					Mean	1146	14.3	15876	1.008	1.037	0.063	1.00	(CV: 0.19)	1.03			
36	17 B	258.46	258.33	0.13	260	881	12.7	11160	0.670	0.621	0.038	0.88	26.56 17q11.2	1.08	1.3		
46	17 C	334.40	334.45	-0.05	335	1323	14.2	18838	1.115	1.118	0.053	1.12	26.58 17q11.2	1.00	-0.1	.	
NF1 microdeletion syndrome					Mean	1102	13.5	14999	0.892	0.870	0.046	1.00	(CV: 0.05)	1.03			
62	17 D	470.73	470.58	0.15	472	595	16.9	10030	0.729	0.719	0.047	0.70	41.26 17q21.31	1.01	0.2	.	
31	17 B	225.02	224.79	0.23	226	2086	11.7	24345	1.461	1.325	0.050	1.21	41.44 17q21.31	1.10	2.7		
47	17 C	341.66	341.78	-0.12	342	1074	14.5	15553	0.920	0.932	0.039	1.09	41.45 17q21.31	0.99	-0.3		
"17q21.31 microdeletion"					Mean	1252	14.3	16643	1.037	0.992	0.046	1.00	(CV: 0.06)	1.04			

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	1.0 high
26	22 A	195.37	195.29	0.08	196	1508	11.0	16653	0.841	0.890	0.065	0.83	17.89 22q11.21	0.95	-0.7	.	
28	22 B	208.16	207.99	0.17	208	1648	11.4	18802	1.129	1.109	0.061	1.11	18.09 22q11.21	1.02	0.3	.	
51	22 D	371.53	371.47	0.06	373	920	16.1	14836	1.078	0.920	0.053	1.06	19.57 22q11.21	1.17	3.0		
22q11.21 (DiGeorge)					Mean	1359	12.9	16764	1.016	0.973	0.060	1.00	(CV: 0.11)	1.05			
35	22 B	252.18	251.96	0.22	253	1675	12.3	20673	1.241	1.321	0.100	1.05	49.49 22q13.33-##	0.94	-0.8	.	
52	22 D	382.28	382.09	0.19	382	1228	15.7	19282	1.401	1.442	0.120	0.95	49.50 22q13.33-#	0.97	-0.3	.	
22q13 (Phelan-McDermid)					Mean	1452	14.0	19978	1.321	1.382	0.110	1.00	(CV: 0.02)	0.95			
41	X C	295.49	295.46	0.03	297	1017	12.8	13037	0.772	0.751	0.048	1.00	32.29 Xp21.2	1.03	0.4	.	
Chromosome X control probe					Mean	1017	12.8	13037	0.772	0.751	0.048	1.00	(CV:)	1.03			
27	X B	199.75	199.64	0.11	202	929	10.1	9412	0.565	0.457	0.047	0.99	153.02 Xq28	1.24	2.3		
18	X A	147.02	146.82	0.20	148	1001	9.1	9115	0.460	0.538	0.065	0.84	152.95 Xq28	0.85	-1.2	.	
24	X A	183.26	183.16	0.10	184	1102	10.9	11968	0.604	0.678	0.060	1.16	152.94 Xq28-#	0.89	-1.2	.	
Xq28 (RETT / MECP2)					Mean	1011	10.0	10165	0.543	0.558	0.057	1.00	(CV: 0.21)	0.99			
Mean values			0.11			1285	13.2	16247	0.972	1.000	0.060	4		0.98	Total of all except		
Standard deviations			0.13			(Coef. of variance: 0.310)			0.262	0.224				0.17	Ctrl and '?' peaks		

Quality assessment

	Quality limits	Quality
Mean A-group area / mean Q-frag. area	>1.00 (1.75)	8.45
Mean CpG-area / mean A-group area	>0.55 (0.85)	1.11
Mean height of first probes AB	> 450 (800)	1583
Mean height of last probes CD	> 280 (500)	998
Ratio of mean heights AB/CD ('slope')	<2.00 (1.55)	1.59 high
Mean group CV of weighted ratio	<0.20 (0.15)	0.07
3 unidentified peak areas / 54 peak areas	< (0.02)	0.01

Individual peaks having normalized area > 4.0 SD from the ref. mean and ratio <0.65 or >1.3 indicate 'abnormal' probe area.

Male Reference

Abn. peaks: 15q12 15q11.2 15q12 15q12

1 quality warning!

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 154 (14 peaks). CV of ROX heights for peaks above 100 nt is: 0.08

("#" marked probes are often low when CpG-D-fragments are low)