

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 -	60.79	60.65	0.14	64	156	10.7	1676	0.549	1.190	0.371	0.75	64 nt	0.46	-1.7		
4	70 -	66.81	66.46	0.35	70	171	19.4	3310	1.084	0.913	0.264	0.80	70 nt	1.19	0.6		
5	76 -	72.79	72.37	0.42	76	194	22.0	4262	1.396	1.138	0.170	1.55	76 nt	1.23	1.5		
6	82 -	78.65	78.56	0.09	82	161	18.4	2962	0.970	0.787	0.204	0.90	82 nt	1.23	0.9		
Ctrl: Q-fragments					Mean	171	17.6	3053	1.000	1.007	0.252	1.00	(CV: 0.32)	1.08			
7	6 a	85.17	84.95	0.22	88	1556	11.0*	17144	0.783	0.981	0.152	1.02	6p21.3 CpG isl.	0.80	-1.3		
9	2 a	90.42	90.17	0.25	92	1835	10.0	18406	0.841	0.967	0.135	1.13	2q14 synt.	0.87	-0.9		
10	1 a	96.80	96.50	0.30	96	3206	11.2*	36047	1.647	1.641	0.304	0.85	MV 36 1p36 CpG isl.	1.00	0.0	.	
Ctrl: D-fragments					Mean	2199	10.8	23866	1.090	1.197	0.197	1.00	(CV: 0.11)	0.88			
15	Y a	114.69	114.50	0.19	118	1406	10.3	14529	0.664	0.811	0.088	1.03	13.54 Yq11	0.82	-1.7		
12	Y a	104.43	104.34	0.09	108	947	10.5	9914	0.453	0.566	0.065	0.97	14.10 Yq11	0.80	-1.7		
Ctrl: Y-fragments (male ref.)					Mean	1177	10.4	12222	0.558	0.688	0.076	1.00	(CV: 0.02)	0.81			
16	1 A	126.31	126.04	0.27	130	2582	10.7	27669	1.264	1.200	0.054	1.30	1.14 1p36.33	1.05	1.2		
24	1 A	176.64	176.49	0.15	178	1962	10.7	21000	0.959	0.942	0.074	0.75	1.75 1p36.33	1.02	0.2	.	
22	1 A	164.58	164.53	0.05	166	2267	11.1	25229	1.153	1.181	0.073	0.95	1.95 1p36.33	0.98	-0.4	.	
1p36 (1p-deletion)					Mean	2270	10.8	24633	1.125	1.108	0.067	1.00	(CV: 0.04)	1.02			
38	2 B	265.06	264.89	0.17	267	1764	13.3	23438	1.013	0.948	0.040	1.36	58.30 2p16.1	1.07	1.6		
64	2 D	481.39	481.31	0.08	486	1090	19.8	21573	0.955	1.121	0.101	0.64	61.00 2p16.1	0.85	-1.6		
"2p16.1 deletion syndrome"					Mean	1427	16.5	22506	0.984	1.034	0.071	1.00	(CV: 0.14)	1.00			
57	3 D	416.58	416.46	0.12	418	1717	17.7	30404	1.346	1.268	0.073	0.96	198.51 3q29	1.06	1.1		
50	3 C	354.51	354.58	-0.07	359	1519	15.3	23257	0.943	1.039	0.056	1.04	198.28 3q29	0.91	-1.7		
"3q29 deletion syndrome"					Mean	1618	16.5	26831	1.144	1.154	0.064	1.00	(CV: 0.11)	0.98			
33	4 B	230.78	230.47	0.31	232	1884	12.2	22909	0.990	0.944	0.063	0.88	1.81 4p16.3	1.05	0.7	.	
60	4 D	443.21	443.03	0.18	445	1099	18.4	20223	0.895	0.879	0.046	1.12	1.90 4p16.3	1.02	0.4	.	
4p16.3 Wolf-Hirschhorn region					Mean	1492	15.3	21566	0.943	0.912	0.054	1.00	(CV: 0.02)	1.03			
59	5 D	435.75	435.58	0.17	436	786	17.9	14047	0.622	0.659	0.053	0.88	1.34 5p15.33	0.94	-0.7		
40	5 C	281.58	281.39	0.19	283	1821	14.1	25746	1.044	1.115	0.071	1.12	1.40 5p15.33	0.94	-1.0		
Cri du Chat syndrome					Mean	1304	16.0	19897	0.833	0.887	0.062	1.00	(CV: 0.01)	0.94			
20	5 A	151.56	151.42	0.14	154	2470	10.4	25707	1.174	1.127	0.052	1.03	176.62 5q35.3	1.04	0.9	.	
61	5 D	453.13	453.02	0.11	454	1355	18.7	25374	1.123	1.123	0.055	0.97	176.65 5q35.3	1.00	0.0	.	
Sotos syndrome					Mean	1913	14.6	25541	1.149	1.125	0.054	1.00	(CV: 0.03)	1.02			
44	7 C	310.41	310.40	0.01	310	2009	14.3	28805	1.168	1.116	0.056	0.99	73.08 7q11.23	1.05	0.9	.	
51	7 C	363.47	363.46	0.01	364	1655	16.0	26498	1.074	1.029	0.040	1.27	73.11 7q11.23	1.04	1.1	.	
54	7 D	389.99	389.95	0.04	391	1096	17.3	18934	0.838	0.748	0.050	0.74	73.15 7q11.23	1.12	1.8		
Williams syndrome					Mean	1587	15.9	24746	1.027	0.964	0.049	1.00	(CV: 0.04)	1.06			
55	8 D	399.81	399.69	0.12	401	923	17.0	15699	0.695	0.776	0.036	1.06	116.75 8q24.12	0.90	-2.2		
58	8 D	424.51	424.33	0.18	427	1095	17.9	19643	0.870	0.895	0.047	0.94	117.73 8q24.11	0.97	-0.5	.	
Langer-Giedion syndrome					Mean	1009	17.5	17671	0.782	0.835	0.042	1.00	(CV: 0.06)	0.93			
45	9 C	319.36	319.40	-0.04	319	2182	14.3	31275	1.268	1.292	0.072	1.05	100.95 9q22.33	0.98	-0.3	.	
56	9 D	408.22	408.03	0.19	409	1417	17.4	24601	1.089	1.119	0.069	0.95	100.95 9q22.33	0.97	-0.4	.	
"9q22.3 deletion syndrome"					Mean	1800	15.8	27938	1.178	1.206	0.070	1.00	(CV: 0.01)	0.98			
17	10 A	132.88	132.56	0.32	136	2099	10.9	22935	1.048	0.948	0.043	1.16	8.14 10p	1.11	2.3		
49	10 C	349.23	349.23	0.00	349	1659	15.4	25490	1.033	0.979	0.060	0.84	10.59 10p15.1	1.06	0.9		
DiGeorge region 2 (10p)					Mean	1879	13.1	24213	1.041	0.963	0.052	1.00	(CV: 0.03)	1.08			
31	11 B	218.84	218.68	0.16	220	2412	11.7	28331	1.224	1.246	0.061	1.00	31.78 11p13	0.98	-0.4	.	
WAGR syndrome					Mean	2412	11.7	28331	1.224	1.246	0.061	1.00	(CV:)	0.98			
30	15 B	213.67	213.54	0.13	214	2362	11.9	28047	1.212	1.226	0.047	1.34	21.48 15q11.2	0.99	-0.3	.	
35	15 B	245.39	245.06	0.33	247	1564	12.9	20117	0.869	0.962	0.058	0.86	22.65 15q12	0.90	-1.6		
41	15 C	289.89	289.75	0.14	292	1669	13.9	23242	0.942	1.018	0.061	0.86	22.76 15q12	0.93	-1.2		
21	15 A	158.14	158.00	0.14	160	1930	11.1	21380	0.977	0.930	0.051	0.94	23.17 15q12	1.05	0.9	.	
Prader-Willi / Angelman					Mean	1881	12.4	23197	1.000	1.034	0.054	1.00	(CV: 0.06)	0.97			
26	15 A	189.91	189.75	0.16	190	2802	11.3	31538	1.441	1.299	0.130	0.78	72.50 15q24.1	1.11	1.1		
46	15 C	326.33	326.43	-0.10	325	1291	14.8	19142	0.776	0.771	0.049	1.22	72.80 15q24.1	1.01	0.1	.	
"15q24 deletion syndrome"					Mean	2047	13.0	25340	1.108	1.035	0.090	1.00	(CV: 0.07)	1.05			
23	16 A	170.91	170.91	0.00	172	2109	10.5	22229	1.016	1.018	0.063	1.00	3.87 16p13.3	1.00	0.0	.	
Rubinstein-Taybi syndrome					Mean	2109	10.5	22229	1.016	1.018	0.063	1.00	(CV:)	1.00			
18	17 A	139.18	138.97	0.21	142	2638	11.2	29423	1.344	1.248	0.053	1.27	2.51 17p13.3	1.08	1.8		
34	17 B	236.38	236.22	0.16	238	1600	12.5	19922	0.861	0.900	0.067	0.73	2.52 17p13.3	0.96	-0.6	.	
Miller-Dieker region					Mean	2119	11.8	24673	1.103	1.074	0.060	1.00	(CV: 0.08)	1.03			
62	17 D	463.27	463.07	0.20	465	2258	19.1	43067	1.907	1.331	0.092	0.83	17.53 17p11.2-##	1.43	6.3*		
39	17 B	272.08	271.90	0.18	274	2442	13.5	32953	1.424	0.940	0.043	1.25	17.83 17p11.2	1.52	11.2*		
43	17 C	303.00	302.90	0.10	303	2058	14.2	29217	1.184	0.839	0.053	0.91	18.08 17p11.2	1.41	6.6*		
Smith-Magenis syndrome					Mean	2253	15.6	35079	1.505	1.037	0.063	1.00	(CV: 0.04)	1.46			
37	17 B	258.52	258.33	0.19	260	1029	13.3	13688	0.592	0.621	0.038	0.88	26.56 17q11.2	0.95	-0.8	.	
47	17 C	334.54	334.45	0.09	335	1844	15.2	27941	1.133	1.118	0.053	1.12	26.58 17q11.2	1.01	0.3	.	
NF1 microdeletion syndrome					Mean	1437	14.2	20815	0.862	0.870	0.046	1.00	(CV: 0.04)	0.99			
63	17 D	470.77	470.58	0.19	472	890	19.1	17035	0.754	0.719	0.047	0.70	41.26 17q21.31	1.05	0.8	.	
32	17 B	225.05	224.79	0.26	226	2527	12.1	30659	1.325	1.325	0.050	1.21	41.44 17q21.31	1.00	0.0	.	
48	17 C	341.73	341.78	-0.05	342	1551	15.6	24180	0.980	0.932	0.039	1.09	41.45 17q21.31	1.05	1.2		
"17q21.31 microdeletion"					Mean	1656	15.6	23958	1.020	0.992	0.046</						

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		
27	22 A	195.37	195.29	0.08	196	1838	11.1	20447	0.934	0.890	0.065	0.83	17.89 22q11.21	1.05	0.7	·		
29	22 B	208.20	207.99	0.21	208	2088	11.8	24726	1.069	1.109	0.061	1.11	18.09 22q11.21	0.96	-0.7	·		
52	22 D	371.47	371.47	0.00	373	1233	17.2	21195	0.938	0.920	0.053	1.06	19.57 22q11.21	1.02	0.3	·		
22q11.21 (DiGeorge)					Mean	1720	13.4	22123	0.980	0.973	0.060	1.00	(CV: 0.04)	1.01				
36	22 B	252.20	251.96	0.24	253	2247	12.7	28585	1.235	1.321	0.100	1.05	49.49 22q13.33-##	0.94	-0.9	·		
53	22 D	382.19	382.09	0.10	382	1703	17.1	29188	1.292	1.442	0.120	0.95	49.50 22q13.33-#	0.90	-1.3	·		
22q13 (Phelan-McDermid)					Mean	1975	14.9	28887	1.264	1.382	0.110	1.00	(CV: 0.03)	0.92				
42	X C	295.52	295.46	0.06	297	1396	13.3	18621	0.755	0.751	0.048	1.00	32.29 Xp21.2	1.01	0.1	·		
Chromosome X control probe					Mean	1396	13.3	18621	0.755	0.751	0.048	1.00	(CV:)	1.01				
28	X B	199.83	199.64	0.19	202	1067	10.5	11217	0.485	0.457	0.047	0.99	153.02 Xq28	1.06	0.6	·		
19	X A	147.05	146.82	0.23	148	1050	9.3	9800	0.448	0.538	0.065	0.84	152.95 Xq28	0.83	-1.4	·		
25	X A	183.20	183.16	0.04	184	1067	11.1	11869	0.542	0.678	0.060	1.16	152.94 Xq28-#	0.80	-2.3	·		
Xq28 (RETT / MECP2)					Mean	1061	10.3	10962	0.492	0.558	0.057	1.00	(CV: 0.16)	0.89				
Mean values			0.13			1737	14.1	23637	1.025	1.000	0.060	4		1.02	Total of all except			
Standard deviations			0.10			(Coef. of variance: 0.264)			0.271	0.224				0.13	Ctrl and '?' peaks			

Quality assessment	Quality limits	Quality
Mean A-group area / mean Q-frag. area	>1.00 (1.75)	7.17
Mean CpG-area / mean A-group area	>0.55 (0.85)	1.22
Mean height of first probes AB	> 450 (800)	1992
Mean height of last probes CD	> 280 (500)	1493
Ratio of mean heights AB/CD ('slope')	<2.00 (1.55)	1.33
Mean group CV of weighted ratio	<0.20 (0.15)	0.06
4 unidentified peak areas / 54 peak areas	< (0.02)	0.00

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: 17p11.2 17p11.2 17p11.2-##

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 168 (14 peaks). CV of ROX heights for peaks above 100 nt is: 0.09

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