

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	Dist. Ratio	1.0 in SD	low high
3	64 -	60.68	60.65	0.03	64	125	5.9	741	0.235	1.438	0.213	0.89	64 nt	0.16	-5.7 *	
4	70 -	66.31	66.46	-0.15	70	204	18.9	3865	1.228	0.773	0.104	0.98	70 nt	1.59	4.4 *	
5	76 -	72.63	72.37	0.26	76	231	19.7	4555	1.447	1.055	0.125	1.12	76 nt	1.37	3.1	
6	82 -	78.97	78.56	0.41	82	204	16.8	3433	1.090	0.734	0.097	1.00	82 nt	1.49	3.7	
Ctrl: Q-fragments					Mean	191	15.4	3149	1.000	1.000	0.134	1.00	(CV: 0.54)	1.18		
7	6 a	85.13	84.95	0.18	88	1616	11.6 *	18702	0.751	0.834	0.103	0.95	6p21.3 CpG isl.	0.90	-0.8	
9	2 a	90.32	90.17	0.15	92	2040	10.2 *	20903	0.840	0.824	0.081	1.20	2q14 synt.	1.02	0.2	.
10	1 a	96.72	96.50	0.22	96	3421	11.3 *	38814	1.559	1.685	0.232	0.85	MV 36 1p36 CpG isl.	0.93	-0.5	
Ctrl: D-fragments					Mean	2359	11.1	26140	1.050	1.115	0.138	1.00	(CV: 0.07)	0.95		
	Y a	114.50			118					0.811			13.54 Yq11			
	Y a	104.34			108					0.566			14.10 Yq11			
ctrl: Y-fragments (male ref.)					Mean							1.00	(CV:)			
11	1 A	126.23	126.04	0.19	130	2415	10.9	26248	1.054	1.133	0.040	1.25	1.14 1p36.33	0.93	-1.9	
19	1 A	176.55	176.49	0.06	178	2002	11.3	22533	0.905	0.839	0.046	0.81	1.75 1p36.33	1.08	1.4	
17	1 A	164.54	164.53	0.01	166	2550	10.8	27511	1.105	1.090	0.052	0.94	1.95 1p36.33	1.01	0.3	.
1p36 (1p-deletion)					Mean	2322	11.0	25431	1.022	1.021	0.046	1.00	(CV: 0.08)	1.00		
33	2 B	264.98	264.89	0.09	267	1975	12.6	24856	0.984	0.910	0.038	1.17	58.30 2p16.1	1.08	2.0	
59	2 D	481.36	481.31	0.05	486	1326	18.6	24643	1.126	1.072	0.063	0.83	61.00 2p16.1	1.05	0.9	.
"2p16.1 deletion syndrome"					Mean	1651	15.6	24750	1.055	0.991	0.050	1.00	(CV: 0.02)	1.07		
52	3 D	416.66	416.46	0.20	418	1767	16.8	29626	1.353	1.230	0.057	1.15	198.51 3q29	1.10	2.2	
45	3 C	354.72	354.58	0.14	359	1842	14.6	26863	1.023	0.959	0.061	0.85	198.28 3q29	1.07	1.1	
"3q29 deletion syndrome"					Mean	1805	15.7	28245	1.188	1.094	0.059	1.00	(CV: 0.02)	1.09		
28	4 B	230.73	230.47	0.26	232	1875	11.7	21996	0.871	0.896	0.058	0.88	1.81 4p16.3	0.97	-0.4	.
55	4 D	443.30	443.03	0.27	445	1123	17.4	19580	0.895	0.880	0.045	1.12	1.90 4p16.3	1.02	0.3	.
4p16.3 Wolf-Hirschhorn region					Mean	1499	14.6	20788	0.883	0.888	0.052	1.00	(CV: 0.03)	1.00		
54	5 D	435.74	435.58	0.16	436	835	16.6	13850	0.633	0.655	0.037	1.02	1.34 5p15.33	0.97	-0.6	.
35	5 C	281.43	281.39	0.04	283	1940	13.3	25818	0.984	1.053	0.061	0.98	1.40 5p15.33	0.93	-1.1	
Cri du Chat syndrome					Mean	1388	14.9	19834	0.808	0.854	0.049	1.00	(CV: 0.02)	0.95		
15	5 A	151.48	151.42	0.06	154	2713	9.9	26848	1.079	0.972	0.047	0.98	176.62 5q35.3	1.11	2.3	
56	5 D	453.26	453.02	0.24	454	1501	17.7	26512	1.211	1.134	0.052	1.02	176.65 5q35.3	1.07	1.5	
Sotos syndrome					Mean	2107	13.8	26680	1.145	1.053	0.049	1.00	(CV: 0.03)	1.09		
39	7 C	310.15	310.40	-0.25	310	1958	13.5	26525	1.011	1.047	0.040	1.13	73.08 7q11.23	0.97	-0.9	.
46	7 C	363.70	363.46	0.24	364	1668	15.3	25482	0.971	0.995	0.048	0.88	73.11 7q11.23	0.98	-0.5	.
49	7 D	390.12	389.95	0.17	391	1047	16.0	16798	0.767	0.774	0.034	0.99	73.15 7q11.23	0.99	-0.2	.
Williams syndrome					Mean	1558	15.0	22935	0.916	0.938	0.041	1.00	(CV: 0.01)	0.98		
50	8 D	400.00	399.69	0.31	401	1213	16.2	19652	0.898	0.753	0.051	0.88	116.75 8q24.12	1.19	2.9	
53	8 D	424.72	424.33	0.39	427	1253	17.1	21439	0.979	0.897	0.048	1.12	117.73 8q24.11	1.09	1.7	
Langer-Giedion syndrome					Mean	1233	16.7	20546	0.939	0.825	0.049	1.00	(CV: 0.06)	1.14		
40	9 C	318.98	319.40	-0.42	319	2274	13.9	31624	1.205	1.194	0.050	0.92	100.95 9q22.33	1.01	0.2	.
51	9 D	408.25	408.03	0.22	409	1718	16.5	28372	1.296	1.115	0.040	1.08	100.95 9q22.33	1.16	4.5 *	
"9q22.3 deletion syndrome"					Mean	1996	15.2	29998	1.251	1.155	0.045	1.00	(CV: 0.10)	1.09		
12	10 A	132.88	132.56	0.32	136	2002	10.7	21519	0.864	0.895	0.048	0.90	8.14 10p	0.97	-0.6	.
44	10 C	349.37	349.23	0.14	349	1692	14.5	24571	0.936	0.938	0.041	1.10	10.59 10p15.1	1.00	-0.1	.
DiGeorge region 2 (10p)					Mean	1847	12.6	23045	0.900	0.917	0.044	1.00	(CV: 0.02)	0.98		
26	11 B	218.81	218.68	0.13	220	2605	11.3	29552	1.170	1.184	0.062	1.00	31.78 11p13	0.99	-0.2	.
WAGR syndrome					Mean	2605	11.3	29552	1.170	1.184	0.062	1.00	(CV:)	0.99		
25	15 B	213.67	213.54	0.13	214	3540	11.6	41023	1.624	1.162	0.060	0.90	21.48 15q11.2	1.40	7.7 *	
30	15 B	245.33	245.06	0.27	247	2790	12.6	35042	1.388	0.909	0.047	0.90	22.65 15q12	1.53	10.1 *	
36	15 C	289.85	289.75	0.10	292	2906	13.3	38677	1.474	0.932	0.035	1.25	22.76 15q12	1.58	15.5 *	
16	15 A	158.04	158.00	0.04	160	2977	11.2	33212	1.334	0.838	0.041	0.95	23.17 15q12	1.59	12.0 *	
Prader-Willi / Angelman					Mean	3053	12.2	36989	1.455	0.960	0.046	1.00	(CV: 0.06)	1.53		
21	15 A	189.79	189.75	0.04	190	2793	11.0	30685	1.233	1.228	0.057	1.07	72.50 15q24.1	1.00	0.1	.
41	15 C	326.05	326.43	-0.38	325	1307	13.9	18131	0.691	0.753	0.040	0.93	72.80 15q24.1	0.92	-1.5	
"15q24 deletion syndrome"					Mean	2050	12.4	24408	0.962	0.990	0.049	1.00	(CV: 0.06)	0.96		
18	16 A	170.84	170.91	-0.07	172	2061	10.7	22135	0.889	0.921	0.053	1.00	3.87 16p13.3	0.97	-0.6	.
Rubinstein-Taybi syndrome					Mean	2061	10.7	22135	0.889	0.921	0.053	1.00	(CV:)	0.97		
13	17 A	139.18	138.97	0.21	142	2840	11.1	31401	1.261	1.138	0.031	1.42	2.51 17p13.3	1.11	4.0	
29	17 B	236.30	236.22	0.08	238	1877	11.9	22360	0.885	0.859	0.058	0.58	2.52 17p13.3	1.03	0.5	.
Miller-Dieker region					Mean	2359	11.5	26881	1.073	0.999	0.044	1.00	(CV: 0.05)	1.09		
57	17 D	463.31	463.07	0.24	465	1531	18.2	27906	1.275	1.385	0.097	0.75	17.53 17p11.2-##	0.92	-1.1	
34	17 B	271.91	271.90	0.01	274	1712	12.8	21868	0.866	0.934	0.038	1.30	17.83 17p11.2	0.93	-1.8	
38	17 C	302.81	302.90	-0.09	303	1530	13.7	20905	0.796	0.796	0.044	0.95	18.08 17p11.2	1.00	0.0	.
Smith-Magenis syndrome					Mean	1591	14.9	23560	0.979	1.038	0.060	1.00	(CV: 0.05)	0.95		
32	17 B	258.36	258.33	0.03	260	1134	12.8	14489	0.574	0.578	0.041	0.94	26.56 17q11.2	0.99	-0.1	.
42	17 C	334.23	334.45	-0.22	335	1913	14.3	27395	1.044	1.044	0.065	1.06	26.58 17q11.2	1.00	0.0	.
NF1 microdeletion syndrome					Mean	1524	13.5	20942	0.809	0.811	0.053	1.00	(CV: 0.00)	1.00		
58	17 D	470.71	470.58	0.13	472	886	18.1	16014	0.732	0.728	0.046	0.78	41.26 17q21.31	1.01	0.1	.
27	17 B	225.10	224.79	0.31	226	2550	11.7	29951	1.186	1.273	0.049	1.26	41.44 17q21.31	0.93	-1.8	
43	17 C	341.54	341.78	-0.24	342	1643	14.6	23947	0.912	0.896	0.046	0.95	41.45 17q21.31	1.02	0.3	.
"17q21.31 microdeletion"					Mean	1693	14.8	23304	0.943	0.966	0.047	1.00	(CV: 0.05)	0.98		

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	
22	22 A	195.32	195.29	0.03	196	1804	10.3	18638	0.749	0.767	0.034	1.08	17.89 22q11.21	0.98	-0.5	.	
24	22 B	208.21	207.99	0.22	208	2066	11.2	23083	0.914	1.070	0.053	0.96	18.09 22q11.21	0.85	-2.9	.	
47	22 D	371.76	371.47	0.29	373	1407	16.6	23315	1.065	0.942	0.047	0.96	19.57 22q11.21	1.13	2.6	-.	
22q11.21 (DiGeorge)					Mean	1759	12.7	21679	0.909	0.926	0.044	1.00	(CV: 0.14)	0.99			
31	22 B	252.22	251.96	0.26	253	2208	12.5	27700	1.097	1.330	0.090	0.75	49.49 22q13.33-##	0.82	-2.6	.	
48	22 D	382.44	382.09	0.35	382	1733	16.4	28391	1.297	1.436	0.059	1.25	49.50 22q13.33-#	0.90	-2.4	.	
22q13 (Phelan-McDermid)					Mean	1971	14.5	28046	1.197	1.383	0.075	1.00	(CV: 0.06)	0.87			
37	X C	295.37	295.46	-0.09	297	2884	13.1	37817	1.441	1.392	0.054	1.00	32.29 Xp21.2	1.03	0.9	.	
Chromosome X control probe					Mean	2884	13.1	37817	1.441	1.392	0.054	1.00	(CV:)	1.03			
23	X B	199.83	199.64	0.19	202	2214	10.6	23435	0.928	0.895	0.045	1.00	153.02 Xq28	1.04	0.7	.	
14	X A	147.02	146.82	0.20	148	2106	9.5	20074	0.806	0.964	0.056	0.86	152.95 Xq28	0.84	-2.8	.	
20	X A	183.18	183.16	0.02	184	2675	11.2	30029	1.206	1.214	0.053	1.14	152.94 Xq28-#	0.99	-0.2	.	
Xq28 (RETT / MECP2)					Mean	2332	10.4	24513	0.980	1.025	0.051	1.00	(CV: 0.10)	0.96			
Mean values			0.10			1967	13.5	25511	1.041	1.000	0.050	4		1.05	Total of all except		
Standard deviations			0.18			(Coef. of variance: 0.232)			0.230	0.196				0.17	Ctrl and '?' peaks		

Quality assessment	Quality limits	Quality
Mean A-group area / mean Q-frag. area	>1.00 (1.75)	7.91
Mean CpG-area / mean A-group area	>0.55 (0.85)	1.16
Mean height of first probes AB	> 450 (800)	2312
Mean height of last probes CD	> 280 (500)	1636
Ratio of mean heights AB/CD ('slope')	<2.00 (1.55)	1.41
Mean group CV of weighted ratio	<0.20 (0.15)	0.05
1 unidentified peak area / 52 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.

Female Reference

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

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 244 (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)