

Peak Data										Normalized Peak Area						
No.	Label	Size	Ref. size	Size diff.	MRC	Height	Width	Area	Peak Area	Ref. Mean	Ref. SD	Ref. Weigh	Position p-tel band	Dist. Ratio	1.0 in SD	low high
3	64 -	60.75	60.65	0.10	64	69	5.7	390	0.350	1.438	0.213	0.89	64 nt	0.24	-5.1*	.
4	70 -	66.73	66.46	0.27	70	84	18.3	1535	1.378	0.773	0.104	0.98	70 nt	1.78	5.8*	
5	76 -	72.54	72.37	0.17	76	77	18.8	1444	1.297	1.055	0.125	1.12	76 nt	1.23	1.9	
6	82 -	78.54	78.56	-0.02	82	75	14.5	1086	0.975	0.734	0.097	1.00	82 nt	1.33	2.5	
Ctrl: Q-fragments					Mean	76	14.3	1114	1.000	1.000	0.134	1.00	(CV: 0.53)	1.17		
7	6 a	85.07	84.95	0.12	88	1164	12.0*	13933	0.796	0.834	0.103	0.95	6p21.3 CpG isl.	0.95	-0.4	.
9	2 a	90.27	90.17	0.10	92	1417	10.5	14927	0.853	0.824	0.081	1.20	2q14 synt.	1.03	0.4	.
10	1 a	96.67	96.50	0.17	96	2506	11.9*	29736	1.700	1.685	0.232	0.85	MV 36 1p36 CpG isl.	1.01	0.1	.
Ctrl: D-fragments					Mean	1696	11.5	19532	1.117	1.115	0.138	1.00	(CV: 0.04)	1.00		
	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	126.13	126.04	0.09	130	1711	11.3	19365	1.107	1.133	0.040	1.25	1.14 1p36.33	0.98	-0.6	.
19	1 A	176.53	176.49	0.04	178	1226	11.8	14445	0.826	0.839	0.046	0.81	1.75 1p36.33	0.98	-0.3	.
17	1 A	164.53	164.53	0.00	166	1650	11.6	19087	1.091	1.090	0.052	0.94	1.95 1p36.33	1.00	0.0	.
1p36 (1p-deletion)					Mean	1529	11.6	17632	1.008	1.021	0.046	1.00	(CV: 0.01)	0.99		
33	2 B	264.93	264.89	0.04	267	1116	13.2	14770	0.932	0.910	0.038	1.17	58.30 2p16.1	1.02	0.6	.
59	2 D	481.37	481.31	0.06	486	790	19.0	14989	1.012	1.072	0.063	0.83	61.00 2p16.1	0.94	-1.0	.
"2p16.1 deletion syndrome"					Mean	953	16.1	14880	0.972	0.991	0.050	1.00	(CV: 0.06)	0.99		
52	3 D	416.41	416.46	-0.05	418	1047	17.4	18193	1.228	1.230	0.057	1.15	198.51 3q29	1.00	0.0	.
45	3 C	354.63	354.58	0.05	359	1034	15.3	15843	0.944	0.959	0.061	0.85	198.28 3q29	0.98	-0.3	.
"3q29 deletion syndrome"					Mean	1041	16.3	17018	1.086	1.094	0.059	1.00	(CV: 0.01)	0.99		
28	4 B	230.59	230.47	0.12	232	1211	12.7	15382	0.971	0.896	0.058	0.88	1.81 4p16.3	1.08	1.3	.
55	4 D	442.91	443.03	-0.12	445	747	17.9	13366	0.902	0.880	0.045	1.12	1.90 4p16.3	1.03	0.5	.
4p16.3 Wolf-Hirschhorn region					Mean	979	15.3	14374	0.937	0.888	0.052	1.00	(CV: 0.04)	1.05		
54	5 D	435.48	435.58	-0.10	436	578	17.2	9935	0.671	0.655	0.037	1.02	1.34 5p15.33	1.02	0.4	.
35	5 C	281.47	281.39	0.08	283	1401	13.7	19222	1.145	1.053	0.061	0.98	1.40 5p15.33	1.09	1.5	.
Cri du Chat syndrome					Mean	990	15.5	14579	0.908	0.854	0.049	1.00	(CV: 0.04)	1.05		
15	5 A	151.54	151.42	0.12	154	1592	10.3	16430	0.939	0.972	0.047	0.98	176.62 5q35.3	0.97	-0.7	.
56	5 D	452.91	453.02	-0.11	454	918	18.3	16757	1.131	1.134	0.052	1.02	176.65 5q35.3	1.00	0.0	.
Sotos syndrome					Mean	1255	14.3	16594	1.035	1.053	0.049	1.00	(CV: 0.02)	0.98		
39	7 C	310.37	310.40	-0.03	310	1425	14.5	20602	1.227	1.047	0.040	1.13	73.08 7q11.23	1.17	4.5*	
46	7 C	363.55	363.46	0.09	364	1093	16.0	17514	1.043	0.995	0.048	0.88	73.11 7q11.23	1.05	1.0	.
49	7 D	390.10	389.95	0.15	391	684	16.6	11350	0.766	0.774	0.034	0.99	73.15 7q11.23	0.99	-0.2	.
Williams syndrome					Mean	1067	15.7	16489	1.012	0.938	0.041	1.00	(CV: 0.09)	1.08		
50	8 D	399.71	399.69	0.02	401	713	16.7	11908	0.804	0.753	0.051	0.88	116.75 8q24.12	1.07	1.0	.
53	8 D	424.31	424.33	-0.02	427	758	17.7	13431	0.907	0.897	0.048	1.12	117.73 8q24.11	1.01	0.2	.
Langer-Giedion syndrome					Mean	736	17.2	12670	0.855	0.825	0.049	1.00	(CV: 0.04)	1.04		
40	9 C	319.45	319.40	0.05	319	1371	14.3	19651	1.170	1.194	0.050	0.92	100.95 9q22.33	0.98	-0.5	.
51	9 D	408.18	408.03	0.15	409	1005	16.9	16972	1.146	1.115	0.040	1.08	100.95 9q22.33	1.03	0.8	.
"9q22.3 deletion syndrome"					Mean	1188	15.6	18312	1.158	1.155	0.045	1.00	(CV: 0.03)	1.01		
12	10 A	132.71	132.56	0.15	136	1341	11.3	15112	0.864	0.895	0.048	0.90	8.14 10p	0.97	-0.7	.
44	10 C	349.24	349.23	0.01	349	1050	14.9	15680	0.934	0.938	0.041	1.10	10.59 10p15.1	1.00	-0.1	.
DiGeorge region 2 (10p)					Mean	1196	13.1	15396	0.899	0.917	0.044	1.00	(CV: 0.02)	0.98		
26	11 B	218.78	218.68	0.10	220	1599	12.0	19186	1.211	1.184	0.062	1.00	31.78 11p13	1.02	0.4	.
WAGR syndrome					Mean	1599	12.0	19186	1.211	1.184	0.062	1.00	(CV:)	1.02		
25	15 B	213.59	213.54	0.05	214	1627	12.1	19700	1.244	1.162	0.060	0.90	21.48 15q11.2	1.07	1.3	.
30	15 B	245.20	245.06	0.14	247	1140	13.0	14856	0.938	0.909	0.047	0.90	22.65 15q12	1.03	0.6	.
36	15 C	289.79	289.75	0.04	292	1168	13.4	15618	0.930	0.932	0.035	1.25	22.76 15q12	1.00	-0.1	.
16	15 A	157.99	158.00	-0.01	160	1291	11.5	14793	0.846	0.838	0.041	0.95	23.17 15q12	1.01	0.2	.
Prader-Willi / Angelman					Mean	1307	12.5	16242	0.989	0.960	0.046	1.00	(CV: 0.03)	1.02		
21	15 A	189.80	189.75	0.05	190	1855	11.5	21342	1.220	1.228	0.057	1.07	72.50 15q24.1	0.99	-0.1	.
41	15 C	326.46	326.43	0.03	325	865	14.5	12564	0.748	0.753	0.040	0.93	72.80 15q24.1	0.99	-0.1	.
"15q24 deletion syndrome"					Mean	1360	13.0	16953	0.984	0.990	0.049	1.00	(CV: 0.00)	0.99		
18	16 A	170.87	170.91	-0.04	172	1423	11.3	16031	0.916	0.921	0.053	1.00	3.87 16p13.3	1.00	-0.1	.
Rubinstein-Taybi syndrome					Mean	1423	11.3	16031	0.916	0.921	0.053	1.00	(CV:)	1.00		
13	17 A	139.09	138.97	0.12	142	1642	11.5	18956	1.084	1.138	0.031	1.42	2.51 17p13.3	0.95	-1.8	.
29	17 B	236.29	236.22	0.07	238	1021	12.4	12631	0.797	0.859	0.058	0.58	2.52 17p13.3	0.93	-1.1	.
Miller-Dieker region					Mean	1332	12.0	15794	0.940	0.999	0.044	1.00	(CV: 0.02)	0.95		
57	17 D	463.03	463.07	-0.04	465	1107	18.4	20346	1.374	1.385	0.097	0.75	17.53 17p11.2-##	0.99	-0.1	.
34	17 B	271.89	271.90	-0.01	274	1106	13.6	15077	0.952	0.934	0.038	1.30	17.83 17p11.2	1.02	0.5	.
38	17 C	302.99	302.90	0.09	303	1005	13.7	13796	0.822	0.796	0.044	0.95	18.08 17p11.2	1.03	0.6	.
Smith-Magenis syndrome					Mean	1073	15.2	16406	1.049	1.038	0.060	1.00	(CV: 0.02)	1.02		
32	17 B	258.37	258.33	0.04	260	720	13.3	9553	0.603	0.578	0.041	0.94	26.56 17q11.2	1.04	0.6	.
42	17 C	334.59	334.45	0.14	335	1188	15.0	17845	1.063	1.044	0.065	1.06	26.58 17q11.2	1.02	0.3	.
NF1 microdeletion syndrome					Mean	954	14.1	13699	0.833	0.811	0.053	1.00	(CV: 0.02)	1.03		
58	17 D	470.56	470.58	-0.02	472	311	17.6	5488	0.371	0.728	0.046	0.78	41.26 17q21.31	0.51	-7.8*	.
27	17 B	224.92	224.79	0.13	226	869	12.1	10529	0.665	1.273	0.049	1.26	41.44 17q21.31	0.52	-12.3*	.
43	17 C	341.87	341.78	0.09	342	577	15.4	8894	0.530	0.896	0.046	0.95	41.45 17q21.31	0.59	-8.0*	.
"17q21.31 microdeletion"					Mean	586	15.1	8304	0.522	0.966	0.047	1.00	(CV: 0.08)	0.54		

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
22	22 A	195.35	195.29	0.06	196	1215	11.0	13396	0.766	0.767	0.034	1.08	17.89 22q11.21	1.00	0.0	.	.
24	22 B	208.10	207.99	0.11	208	1525	11.9	18180	1.148	1.070	0.053	0.96	18.09 22q11.21	1.07	1.5	↓	.
47	22 D	371.54	371.47	0.07	373	878	16.9	14798	0.999	0.942	0.047	0.96	19.57 22q11.21	1.06	1.2	↓	.
22q11.21 (DiGeorge)					Mean	1206	13.3	15458	0.971	0.926	0.044	1.00	(CV: 0.04)	1.04			
31	22 B	252.03	251.96	0.07	253	1732	13.1	22611	1.427	1.330	0.090	0.75	49.49 22q13.33-##	1.07	1.1	↓	.
48	22 D	382.31	382.09	0.22	382	1397	17.1	23945	1.617	1.436	0.059	1.25	49.50 22q13.33-#	1.13	3.1	↓	.
22q13 (Phelan-McDermid)					Mean	1565	15.1	23278	1.522	1.383	0.075	1.00	(CV: 0.03)	1.11			
37	X C	295.47	295.46	0.01	297	1678	13.8	23133	1.378	1.392	0.054	1.00	32.29 Xp21.2	0.99	-0.3	.	.
Chromosome X control probe					Mean	1678	13.8	23133	1.378	1.392	0.054	1.00	(CV:)	0.99			
23	X B	199.76	199.64	0.12	202	1487	11.1	16572	1.046	0.895	0.045	1.00	153.02 Xq28	1.17	3.4	↓	↓
14	X A	146.98	146.82	0.16	148	1704	10.3	17601	1.006	0.964	0.056	0.86	152.95 Xq28	1.04	0.8	.	.
20	X A	183.17	183.16	0.01	184	1911	11.6	22202	1.269	1.214	0.053	1.14	152.94 Xq28-#	1.05	1.0	.	.
Xq28 (RETT / MECP2)					Mean	1701	11.0	18792	1.107	1.025	0.051	1.00	(CV: 0.07)	1.09			
Mean values			0.05			1194	14.0	16115	0.994	1.000	0.050	4		0.99	Total of all except		
Standard deviations			0.07			(Coef. of variance: 0.242)			0.238	0.196				0.13	Ctrl and '?' peaks		

Quality assessment	Quality limits	Quality
Mean A-group area / mean Q-frag. area	>1.00 (1.75)	15.71
Mean CpG-area / mean A-group area	>0.55 (0.85)	1.25
Mean height of first probes AB	> 450 (800)	1405
Mean height of last probes CD	> 280 (500)	992
Ratio of mean heights AB/CD ('slope')	<2.00 (1.55)	1.42
Mean group CV of weighted ratio	<0.20 (0.15)	0.04
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: 17q21.31 17q21.31 17q21.31

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: 0.07

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