

----- 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
4	64 -	60.89	60.65	0.24	64	69	3.5	241	0.116	1.438	0.213	0.89	64 nt	0.08	-6.2*	.
5	70 -	66.64	66.46	0.18	70	185	18.4	3403	1.636	0.773	0.104	0.98	70 nt	2.12	8.3*	.
6	76 -	72.61	72.37	0.24	76	152	14.1	2145	1.031	1.055	0.125	1.12	76 nt	0.98	-0.2	.
8	82 -	78.54	78.56	-0.02	82	157	16.1	2529	1.216	0.734	0.097	1.00	82 nt	1.66	5.0*	.
<b>Ctrl: Q-fragments</b>					Mean	141	13.0	2080	1.000	1.000	0.134	1.00	(CV: 0.70)	1.23		
9	6 a	85.15	84.95	0.20	88	1219	11.3*	13806	0.635	0.834	0.103	0.95	6p21.3 CpG isl.	0.76	-1.9	.
11	2 a	90.35	90.17	0.18	92	1660	10.5*	17499	0.805	0.824	0.081	1.20	2q14 synt.	0.98	-0.2	.
12	1 a	96.75	96.50	0.25	96	2872	11.6*	33410	1.537	1.685	0.232	0.85	MV 36 1p36 CpG isl.	0.91	-0.6	.
<b>Ctrl: D-fragments</b>					Mean	1917	11.2	21572	0.992	1.115	0.138	1.00	(CV: 0.13)	0.89		
	Y a	114.50			118					0.811			13.54 Yq11			
	Y a	104.34			108					0.566			14.10 Yq11			
<b>ctrl: Y-fragments (male ref.)</b>					Mean							1.00	(CV: )			
13	1 A	126.21	126.04	0.17	130	2170	10.8	23472	1.080	1.133	0.040	1.25	1.14 1p36.33	0.95	-1.3	.
21	1 A	176.59	176.49	0.10	178	1599	11.5	18428	0.848	0.839	0.046	0.81	1.75 1p36.33	1.01	0.2	.
19	1 A	164.61	164.53	0.08	166	2209	11.3	24954	1.148	1.090	0.052	0.94	1.95 1p36.33	1.05	1.1	.
<b>1p36 (1p-deletion)</b>					Mean	1993	11.2	22285	1.025	1.021	0.046	1.00	(CV: 0.05)	1.00		
35	2 B	264.92	264.89	0.03	267	1457	13.5	19685	0.913	0.910	0.038	1.17	58.30 2p16.1	1.00	0.1	.
61	2 D	481.49	481.31	0.18	486	915	20.6	18825	1.026	1.072	0.063	0.83	61.00 2p16.1	0.96	-0.7	.
<b>"2p16.1 deletion syndrome"</b>					Mean	1186	17.0	19255	0.969	0.991	0.050	1.00	(CV: 0.03)	0.98		
54	3 D	416.56	416.46	0.10	418	1404	18.1	25413	1.385	1.230	0.057	1.15	198.51 3q29	1.13	2.7	.
47	3 C	354.54	354.58	-0.04	359	1331	15.8	21088	0.952	0.959	0.061	0.85	198.28 3q29	0.99	-0.1	.
<b>"3q29 deletion syndrome"</b>					Mean	1368	17.0	23251	1.168	1.094	0.059	1.00	(CV: 0.09)	1.07		
30	4 B	230.61	230.47	0.14	232	1572	12.8	20044	0.930	0.896	0.058	0.88	1.81 4p16.3	1.04	0.6	.
57	4 D	443.24	443.03	0.21	445	786	18.9	14887	0.811	0.880	0.045	1.12	1.90 4p16.3	0.92	-1.5	.
<b>4p16.3 Wolf-Hirschhorn region</b>					Mean	1179	15.8	17466	0.870	0.888	0.052	1.00	(CV: 0.08)	0.97		
56	5 D	435.74	435.58	0.16	436	310	18.0	5593	0.305	0.655	0.037	1.02	1.34 5p15.33	0.47	-9.6*	.
37	5 C	281.38	281.39	-0.01	283	893	14.1	12635	0.570	1.053	0.061	0.98	1.40 5p15.33	0.54	-7.9*	.
<b>Cr du Chat syndrome</b>					Mean	602	16.1	9114	0.438	0.854	0.049	1.00	(CV: 0.11)	0.50		
17	5 A	151.54	151.42	0.12	154	2131	10.2	21770	1.001	0.972	0.047	0.98	176.62 5q35.3	1.03	0.6	.
58	5 D	453.09	453.02	0.07	454	1051	19.2	20184	1.100	1.134	0.052	1.02	176.65 5q35.3	0.97	-0.6	.
<b>Sotos syndrome</b>					Mean	1591	14.7	20977	1.051	1.053	0.049	1.00	(CV: 0.04)	1.00		
41	7 C	310.16	310.40	-0.24	310	1541	15.0	23122	1.044	1.047	0.040	1.13	73.08 7q11.23	1.00	-0.1	.
48	7 C	363.55	363.46	0.09	364	1310	16.5	21604	0.975	0.995	0.048	0.88	73.11 7q11.23	0.98	-0.4	.
51	7 D	389.99	389.95	0.04	391	854	17.1	14629	0.797	0.774	0.034	0.99	73.15 7q11.23	1.03	0.7	.
<b>Williams syndrome</b>					Mean	1235	16.2	19785	0.939	0.938	0.041	1.00	(CV: 0.02)	1.00		
52	8 D	399.81	399.69	0.12	401	854	17.6	15011	0.818	0.753	0.051	0.88	116.75 8q24.12	1.09	1.3	.
55	8 D	424.53	424.33	0.20	427	869	19.1	16617	0.906	0.897	0.048	1.12	117.73 8q24.11	1.01	0.2	.
<b>Langer-Giedion syndrome</b>					Mean	862	18.3	15814	0.862	0.825	0.049	1.00	(CV: 0.05)	1.04		
42	9 C	319.26	319.40	-0.14	319	1891	15.1	28545	1.289	1.194	0.050	0.92	100.95 9q22.33	1.08	1.9	.
53	9 D	408.25	408.03	0.22	409	1240	18.3	22658	1.235	1.115	0.040	1.08	100.95 9q22.33	1.11	3.0	.
<b>"9q22.3 deletion syndrome"</b>					Mean	1566	16.7	25602	1.262	1.155	0.045	1.00	(CV: 0.02)	1.09		
14	10 A	132.79	132.56	0.23	136	1652	11.1	18376	0.845	0.895	0.048	0.90	8.14 10p	0.94	-1.0	.
46	10 C	349.32	349.23	0.09	349	1514	15.7	23695	1.070	0.938	0.041	1.10	10.59 10p15.1	1.14	3.2	.
<b>DiGeorge region 2 (10p)</b>					Mean	1583	13.4	21036	0.958	0.917	0.044	1.00	(CV: 0.13)	1.05		
28	11 B	218.74	218.68	0.06	220	2174	12.2	26523	1.230	1.184	0.062	1.00	31.78 11p13	1.04	0.7	.
<b>WAGR syndrome</b>					Mean	2174	12.2	26523	1.230	1.184	0.062	1.00	(CV: )	1.04		
27	15 B	213.57	213.54	0.03	214	2145	12.0	25685	1.191	1.162	0.060	0.90	21.48 15q11.2	1.02	0.5	.
32	15 B	245.17	245.06	0.11	247	1526	13.1	19927	0.924	0.909	0.047	0.90	22.65 15q12	1.02	0.3	.
38	15 C	289.67	289.75	-0.08	292	1506	14.1	21207	0.957	0.932	0.035	1.25	22.76 15q12	1.03	0.7	.
18	15 A	158.07	158.00	0.07	160	1607	11.2	18017	0.829	0.838	0.041	0.95	23.17 15q12	0.99	-0.2	.
<b>Prader-Willi / Angelman</b>					Mean	1696	12.6	21209	0.975	0.960	0.046	1.00	(CV: 0.02)	1.02		
23	15 A	189.83	189.75	0.08	190	2487	11.5	28676	1.319	1.228	0.057	1.07	72.50 15q24.1	1.07	1.6	.
43	15 C	326.13	326.43	-0.30	325	1070	15.1	16205	0.732	0.753	0.040	0.93	72.80 15q24.1	0.97	-0.5	.
<b>"15q24 deletion syndrome"</b>					Mean	1779	13.3	22441	1.025	0.990	0.049	1.00	(CV: 0.07)	1.03		
20	16 A	170.93	170.91	0.02	172	1751	11.2	19665	0.905	0.921	0.053	1.00	3.87 16p13.3	0.98	-0.3	.
<b>Rubinstein-Taybi syndrome</b>					Mean	1751	11.2	19665	0.905	0.921	0.053	1.00	(CV: )	0.98		
15	17 A	139.18	138.97	0.21	142	2273	11.4	25887	1.191	1.138	0.031	1.42	2.51 17p13.3	1.05	1.7	.
31	17 B	236.20	236.22	-0.02	238	1478	12.8	18958	0.879	0.859	0.058	0.58	2.52 17p13.3	1.02	0.4	.
<b>Miller-Dieker region</b>					Mean	1876	12.1	22423	1.035	0.999	0.044	1.00	(CV: 0.01)	1.04		
59	17 D	463.25	463.07	0.18	465	1200	20.2	24217	1.320	1.385	0.097	0.75	17.53 17p11.2-##	0.95	-0.7	.
36	17 B	271.84	271.90	-0.06	274	1511	14.0	21194	0.983	0.934	0.038	1.30	17.83 17p11.2	1.05	1.3	.
40	17 C	302.86	302.90	-0.04	303	1198	14.5	17419	0.786	0.796	0.044	0.95	18.08 17p11.2	0.99	-0.2	.
<b>Smith-Magenis syndrome</b>					Mean	1303	16.2	20943	1.030	1.038	0.060	1.00	(CV: 0.05)	1.01		
34	17 B	258.29	258.33	-0.04	260	912	14.1	12842	0.596	0.578	0.041	0.94	26.56 17q11.2	1.03	0.4	.
44	17 C	334.33	334.45	-0.12	335	1678	15.6	26170	1.182	1.044	0.065	1.06	26.58 17q11.2	1.13	2.1	.
<b>NF1 microdeletion syndrome</b>					Mean	1295	14.8	19506	0.889	0.811	0.053	1.00	(CV: 0.07)	1.08		
60	17 D	470.80	470.58	0.22	472	777	19.6	15224	0.830	0.728	0.046	0.78	41.26 17q21.31	1.14	2.2	.
29	17 B	224.95	224.79	0.16	226	2069	12.5	25958	1.204	1.273	0.049	1.26	41.44 17q21.31	0.95	-1.4	.
45	17 C	341.63	341.78	-0.15	342	1201	15.8	18922	0.854	0.896	0.046	0.95	41.45 17q21.31	0.95	-0.9	.
<b>"17q21.31 microdeletion"</b>					Mean	1349	16.0	20035	0.963	0.966	0.047	1.00	(CV: 0.10)	1.00		

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	
24	22 A	195.36	195.29	0.07	196	1641	10.9	17830	0.820	0.767	0.034	1.08	17.89 22q11.21	1.07	1.6	↓	
26	22 B	208.09	207.99	0.10	208	1852	12.1	22323	1.035	1.070	0.053	0.96	18.09 22q11.21	0.97	-0.7	↓	
49	22 D	371.70	371.47	0.23	373	1020	18.0	18345	1.000	0.942	0.047	0.96	19.57 22q11.21	1.06	1.2	↓	
<b>22q11.21 (DiGeorge)</b>					Mean	1504	13.6	19499	0.952	0.926	0.044	1.00	(CV: <b>0.05</b> )	<b>1.03</b>			
33	22 B	252.06	251.96	0.10	253	1814	13.4	24262	1.125	1.330	0.090	0.75	49.49 22q13.33-##	0.85	-2.3		
50	22 D	382.16	382.09	0.07	382	1406	17.8	25002	1.362	1.436	0.059	1.25	49.50 22q13.33-#	0.95	-1.2		
<b>22q13 (Phelan-McDermid)</b>					Mean	1610	15.6	24632	1.244	1.383	0.075	1.00	(CV: <b>0.08</b> )	<b>0.91</b>			
39	X C	295.41	295.46	-0.05	297	2281	14.5	33006	1.490	1.392	0.054	1.00	32.29 Xp21.2	1.07	1.8	↓	
<b>Chromosome X control probe</b>					Mean	2281	14.5	33006	1.490	1.392	0.054	1.00	(CV: )	<b>1.07</b>			
25	X B	199.76	199.64	0.12	202	1718	11.2	19207	0.891	0.895	0.045	1.00	153.02 Xq28	0.99	-0.1	↓	
16	X A	146.98	146.82	0.16	148	1952	9.9	19305	0.888	0.964	0.056	0.86	152.95 Xq28	0.92	-1.4		
22	X A	183.21	183.16	0.05	184	1963	11.4	22352	1.028	1.214	0.053	1.14	152.94 Xq28-#	0.85	-3.5		
<b>Xq28 (RETT / MECP2)</b>					Mean	1878	10.8	20288	0.936	1.025	0.051	1.00	(CV: <b>0.08</b> )	<b>0.92</b>			
<b>Mean values</b>			0.06			1505	14.5	20726	<b>0.992</b>	1.000	0.050	4		0.99	Total of all except		
<b>Standard deviations</b>			0.12			(Coef. of variance: 0.233)			0.225	0.196				0.12	Ctrl and '?' peaks		

Quality assessment	Quality limits	Quality
Mean A-group area / mean Q-frag. area	>1.00 (1.75)	10.45
Mean CpG-area / mean A-group area	>0.55 (0.85)	1.09
Mean height of first probes AB	> 450 ( 800)	1819
Mean height of last probes CD	> 280 ( 500)	1204
Ratio of mean heights AB/CD ('slope')	<2.00 (1.55)	1.51
Mean group CV of weighted ratio	<0.20 (0.15)	0.06
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: 5p15.33 5p15.33**

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 225 (14 peaks). CV of ROX heights for peaks above 100 nt is: 0.10

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