

----- 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
2	64 -	60.84	60.65	0.19	64	71	5.4	385	0.208	1.190	0.371	0.75	64 nt	0.17	-2.6	
3	70 -	66.67	66.46	0.21	70	133	18.4	2451	1.323	0.913	0.264	0.80	70 nt	1.45	1.5	
4	76 -	72.44	72.37	0.07	76	131	20.2	2643	1.426	1.138	0.170	1.55	76 nt	1.25	1.7	
5	82 -	78.57	78.56	0.01	82	124	15.6	1934	1.044	0.787	0.204	0.90	82 nt	1.33	1.3	
<b>Ctrl: Q-fragments</b>					Mean	115	14.9	1853	1.000	1.007	0.252	1.00	(CV: 0.47)	1.11		
6	6 a	85.14	84.95	0.19	88	1728	11.6*	20112	0.822	0.981	0.152	1.02	6p21.3 CpG isl.	0.84	-1.0	
8	2 a	90.38	90.17	0.21	92	2082	10.8	22424	0.917	0.967	0.135	1.13	2q14 synt.	0.95	-0.4	
9	1 a	96.73	96.50	0.23	96	3084	11.6*	35751	1.462	1.641	0.304	0.85	MV 36 1p36 CpG isl.	0.89	-0.6	
<b>Ctrl: D-fragments</b>					Mean	2298	11.3	26096	1.067	1.197	0.197	1.00	(CV: 0.06)	0.89		
13	Y a	114.63	114.50	0.13	118	1511	10.8	16388	0.670	0.811	0.088	1.03	13.54 Yq11	0.83	-1.6	
11	Y a	104.36	104.34	0.02	108	911	11.1	10109	0.413	0.566	0.065	0.97	14.10 Yq11	0.73	-2.4	
<b>ctrl: Y-fragments (male ref.)</b>					Mean	1211	11.0	13249	0.542	0.688	0.076	1.00	(CV: 0.09)	0.78		
14	1 A	126.25	126.04	0.21	130	2947	11.2	33084	1.353	1.200	0.054	1.30	1.14 1p36.33	1.13	2.8	
22	1 A	176.56	176.49	0.07	178	1997	11.6	23091	0.944	0.942	0.074	0.75	1.75 1p36.33	1.00	0.0	.
20	1 A	164.56	164.53	0.03	166	2852	11.3	32302	1.321	1.181	0.073	0.95	1.95 1p36.33	1.12	1.9	
<b>1p36 (1p-deletion)</b>					Mean	2599	11.4	29492	1.206	1.108	0.067	1.00	(CV: 0.06)	1.09		
36	2 B	264.95	264.89	0.06	267	1606	13.4	21509	0.931	0.948	0.040	1.36	58.30 2p16.1	0.98	-0.4	.
62	2 D	481.59	481.31	0.28	486	964	19.8	19057	0.930	1.121	0.101	0.64	61.00 2p16.1	0.83	-1.9	
<b>"2p16.1 deletion syndrome"</b>					Mean	1285	16.6	20283	0.931	1.034	0.071	1.00	(CV: 0.11)	0.93		
55	3 D	416.72	416.46	0.26	418	1496	17.8	26563	1.296	1.268	0.073	0.96	198.51 3q29	1.02	0.4	.
48	3 C	354.60	354.58	0.02	359	1429	15.2	21771	0.875	1.039	0.056	1.04	198.28 3q29	0.84	-3.0	
<b>"3q29 deletion syndrome"</b>					Mean	1463	16.5	24167	1.086	1.154	0.064	1.00	(CV: 0.14)	0.93		
31	4 B	230.66	230.47	0.19	232	1937	12.7	24551	1.063	0.944	0.063	0.88	1.81 4p16.3	1.13	1.9	
58	4 D	443.28	443.03	0.25	445	1007	18.9	19021	0.928	0.879	0.046	1.12	1.90 4p16.3	1.06	1.1	
<b>4p16.3 Wolf-Hirschhorn region</b>					Mean	1472	15.8	21786	0.996	0.912	0.054	1.00	(CV: 0.05)	1.09		
57	5 D	435.80	435.58	0.22	436	812	18.2	14776	0.721	0.659	0.053	0.88	1.34 5p15.33	1.09	1.2	
38	5 C	281.48	281.39	0.09	283	2032	14.2	28838	1.159	1.115	0.071	1.12	1.40 5p15.33	1.04	0.6	.
<b>Cri du Chat syndrome</b>					Mean	1422	16.2	21807	0.940	0.887	0.062	1.00	(CV: 0.04)	1.06		
18	5 A	151.47	151.42	0.05	154	2558	10.8	27511	1.125	1.127	0.052	1.03	176.62 5q35.3	1.00	0.0	.
59	5 D	453.22	453.02	0.20	454	1162	18.9	22009	1.074	1.123	0.055	0.97	176.65 5q35.3	0.96	-0.9	.
<b>Sotos syndrome</b>					Mean	1860	14.8	24760	1.099	1.125	0.054	1.00	(CV: 0.03)	0.98		
42	7 C	310.32	310.40	-0.08	310	3004	14.8	44422	1.785	1.116	0.056	0.99	73.08 7q11.23	1.60	12.0*	
49	7 C	363.63	363.46	0.17	364	2377	16.4	39038	1.568	1.029	0.040	1.27	73.11 7q11.23	1.52	13.5*	
52	7 D	390.14	389.95	0.19	391	1573	17.1	26903	1.313	0.748	0.050	0.74	73.15 7q11.23	1.76	11.2*	
<b>Williams syndrome</b>					Mean	2318	16.1	36788	1.555	0.964	0.049	1.00	(CV: 0.07)	1.61		
53	8 D	399.90	399.69	0.21	401	836	17.5	14593	0.712	0.776	0.036	1.06	116.75 8q24.12	0.92	-1.8	
56	8 D	424.77	424.33	0.44	427	922	17.9	16473	0.804	0.895	0.047	0.94	117.73 8q24.11	0.90	-1.9	
<b>Langer-Giedion syndrome</b>					Mean	879	17.7	15533	0.758	0.835	0.042	1.00	(CV: 0.02)	0.91		
43	9 C	319.29	319.40	-0.11	319	1957	14.9	29179	1.172	1.292	0.072	1.05	100.95 9q22.33	0.91	-1.7	
54	9 D	408.33	408.03	0.30	409	1279	17.6	22573	1.102	1.119	0.069	0.95	100.95 9q22.33	0.98	-0.3	.
<b>"9q22.3 deletion syndrome"</b>					Mean	1618	16.3	25876	1.137	1.206	0.070	1.00	(CV: 0.06)	0.94		
15	10 A	132.81	132.56	0.25	136	2088	11.3	23616	0.966	0.948	0.043	1.16	8.14 10p	1.02	0.4	.
47	10 C	349.31	349.23	0.08	349	1403	15.1	21150	0.850	0.979	0.060	0.84	10.59 10p15.1	0.87	-2.1	
<b>DiGeorge region 2 (10p)</b>					Mean	1746	13.2	22383	0.908	0.963	0.052	1.00	(CV: 0.11)	0.95		
29	11 B	218.76	218.68	0.08	220	2363	12.3	28953	1.253	1.246	0.061	1.00	31.78 11p13	1.01	0.1	.
<b>WAGR syndrome</b>					Mean	2363	12.3	28953	1.253	1.246	0.061	1.00	(CV: )	1.01		
28	15 B	213.62	213.54	0.08	214	2324	12.0	27910	1.208	1.226	0.047	1.34	21.48 15q11.2	0.99	-0.4	.
33	15 B	245.28	245.06	0.22	247	1648	13.2	21762	0.942	0.962	0.058	0.86	22.65 15q12	0.98	-0.3	.
39	15 C	289.84	289.75	0.09	292	1627	14.1	22960	0.922	1.018	0.061	0.86	22.76 15q12	0.91	-1.6	
19	15 A	158.05	158.00	0.05	160	1674	11.7	19509	0.798	0.930	0.051	0.94	23.17 15q12	0.86	-2.6	
<b>Prader-Willi / Angelman</b>					Mean	1818	12.7	23035	0.968	1.034	0.054	1.00	(CV: 0.07)	0.94		
24	15 A	189.82	189.75	0.07	190	3256	11.6	37856	1.548	1.299	0.130	0.78	72.50 15q24.1	1.19	1.9	
44	15 C	326.31	326.43	-0.12	325	1220	15.1	18362	0.738	0.771	0.049	1.22	72.80 15q24.1	0.96	-0.7	.
<b>"15q24 deletion syndrome"</b>					Mean	2238	13.3	28109	1.143	1.035	0.090	1.00	(CV: 0.15)	1.05		
21	16 A	170.94	170.91	0.03	172	2261	11.3	25624	1.048	1.018	0.063	1.00	3.87 16p13.3	1.03	0.5	.
<b>Rubinstein-Taybi syndrome</b>					Mean	2261	11.3	25624	1.048	1.018	0.063	1.00	(CV: )	1.03		
16	17 A	139.18	138.97	0.21	142	2757	11.6	32110	1.313	1.248	0.053	1.27	2.51 17p13.3	1.05	1.2	
32	17 B	236.22	236.22	0.00	238	1541	12.6	19490	0.844	0.900	0.067	0.73	2.52 17p13.3	0.94	-0.9	
<b>Miller-Dieker region</b>					Mean	2149	12.1	25800	1.078	1.074	0.060	1.00	(CV: 0.08)	1.01		
60	17 D	463.41	463.07	0.34	465	1427	19.2	27409	1.338	1.331	0.092	0.83	17.53 17p11.2-##	1.00	0.1	.
37	17 B	271.94	271.90	0.04	274	1775	13.6	24133	1.045	0.940	0.043	1.25	17.83 17p11.2	1.11	2.4	
41	17 C	302.88	302.90	-0.02	303	1524	14.5	22158	0.890	0.839	0.053	0.91	18.08 17p11.2	1.06	1.0	
<b>Smith-Magenis syndrome</b>					Mean	1575	15.8	24567	1.091	1.037	0.063	1.00	(CV: 0.05)	1.07		
35	17 B	258.44	258.33	0.11	260	1032	13.5	13966	0.605	0.621	0.038	0.88	26.56 17q11.2	0.97	-0.4	.
45	17 C	334.38	334.45	-0.07	335	1592	15.4	24508	0.985	1.118	0.053	1.12	26.58 17q11.2	0.88	-2.5	
<b>NF1 microdeletion syndrome</b>					Mean	1312	14.5	19237	0.795	0.870	0.046	1.00	(CV: 0.07)	0.92		
61	17 D	470.87	470.58	0.29	472	870	19.2	16672	0.814	0.719	0.047	0.70	41.26 17q21.31	1.13	2.0	
30	17 B	225.03	224.79	0.24	226	2610	12.7	33167	1.436	1.325	0.050	1.21	41.44 17q21.31	1.08	2.2	
46	17 C	341.73	341.78	-0.05	342	1483	15.6	23145	0.930	0.932	0.039	1.09	41.45 17q21.31	1.00	-0.1	.
<b>"17q21.31 microdeletion"</b>					Mean	1654	15.8	24328	1.060	0.992	0.046	1.00	(CV: 0.06)	1.06		

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	
25	22 A	195.24	195.29	-0.05	196	1936	11.5	22327	0.913	0.890	0.065	0.83	17.89 22q11.21	1.03	0.4	.	
27	22 B	208.17	207.99	0.18	208	2300	12.2	28018	1.213	1.109	0.061	1.11	18.09 22q11.21	1.09	1.7	↓	
50	22 D	371.61	371.47	0.14	373	1170	17.4	20322	0.992	0.920	0.053	1.06	19.57 22q11.21	1.08	1.4	↓	
<b>22q11.21 (DiGeorge)</b>					Mean	1802	13.7	23556	1.039	0.973	0.060	1.00	(CV: 0.03)	<b>1.07</b>			
34	22 B	252.15	251.96	0.19	253	2462	13.5	33263	1.440	1.321	0.100	1.05	49.49 22q13.33-##	1.09	1.2	↓	
51	22 D	382.38	382.09	0.29	382	1781	17.2	30654	1.496	1.442	0.120	0.95	49.50 22q13.33-#	1.04	0.4	.	
<b>22q13 (Phelan-McDermid)</b>					Mean	2122	15.4	31959	1.468	1.382	0.110	1.00	(CV: 0.03)	<b>1.06</b>			
40	X C	295.45	295.46	-0.01	297	1117	13.5	15092	0.606	0.751	0.048	1.00	32.29 Xp21.2	0.81	-3.0	.	
<b>Chromosome X control probe</b>					Mean	1117	13.5	15092	0.606	0.751	0.048	1.00	(CV: )	<b>0.81</b>			
26	X B	199.75	199.64	0.11	202	1080	10.7	11578	0.501	0.457	0.047	0.99	153.02 Xq28	1.10	0.9	↓	
17	X A	146.98	146.82	0.16	148	1273	10.0	12683	0.519	0.538	0.065	0.84	152.95 Xq28	0.96	-0.3	.	
23	X A	183.16	183.16	0.00	184	1352	11.5	15534	0.635	0.678	0.060	1.16	152.94 Xq28-#	0.94	-0.7	↓	
<b>Xq28 (RETT / MECP2)</b>					Mean	1235	10.7	13265	0.552	0.558	0.057	1.00	(CV: 0.09)	<b>1.00</b>			
<b>Mean values</b>			0.12			1749	14.4	24106	<b>1.040</b>	1.000	0.060	4		1.04	Total of all except		
<b>Standard deviations</b>			0.13			(Coef. of variance: 0.294)			0.290	0.224				0.17	Ctrl and '?' peaks		

Quality assessment	Quality limits	Quality
Mean A-group area / mean Q-frag. area	>1.00 (1.75)	13.20
Mean CpG-area / mean A-group area	>0.55 (0.85)	1.14
Mean height of first probes AB	> 450 ( 800)	2068
Mean height of last probes CD	> 280 ( 500)	1443
Ratio of mean heights AB/CD ('slope')	<2.00 (1.55)	1.43
Mean group CV of weighted ratio	<0.20 (0.15)	0.07
3 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: 7q11.23 7q11.23 7q11.23**

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 199 (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)