

----- 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	in SD	1.0 low high		
3	64 -	60.76	60.65	0.11	64	390	10.8	4212	0.605	1.438	0.213	0.89	64 nt	0.42	-3.9	.		
4	70 -	66.47	66.46	0.01	70	282	17.8	5023	0.722	0.773	0.104	0.98	70 nt	0.93	-0.5	.		
5	76 -	72.34	72.37	-0.03	76	456	20.9	9540	1.371	1.055	0.125	1.12	76 nt	1.30	2.5			
6	82 -	78.61	78.56	0.05	82	445	20.3	9055	1.301	0.734	0.097	1.00	82 nt	1.77	5.9*			
<b>Ctrl: Q-fragments</b>					Mean	393	17.5	6958	1.000	1.000	0.134	1.00	(CV: 0.49)		1.13			
7	6 a	85.12	84.95	0.17	88	1379	11.2*	15468	0.738	0.834	0.103	0.95	6p21.3 CpG isl.	0.89	-0.9	.		
9	2 a	90.38	90.17	0.21	92	1761	10.7*	18758	0.895	0.824	0.081	1.20	2q14 synt.	1.09	0.9	.		
10	1 a	96.76	96.50	0.26	96	3146	11.6*	36476	1.741	1.685	0.232	0.85	MV 36 1p36 CpG isl.	1.03	0.2	.		
<b>Ctrl: D-fragments</b>					Mean	2095	11.2	23567	1.125	1.115	0.138	1.00	(CV: 0.10)		1.01			
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: )					
11	1 A	126.30	126.04	0.26	130	2079	11.2*	23234	1.109	1.133	0.040	1.25	1.14 1p36.33	0.98	-0.6	.		
19	1 A	176.56	176.49	0.07	178	1554	11.9	18446	0.881	0.839	0.046	0.81	1.75 1p36.33	1.05	0.9	.		
17	1 A	164.58	164.53	0.05	166	1883	12.0	22597	1.079	1.090	0.052	0.94	1.95 1p36.33	0.99	-0.2	.		
<b>1p36 (1p-deletion)</b>					Mean	1839	11.7	21426	1.023	1.021	0.046	1.00	(CV: 0.04)		1.00			
33	2 B	265.09	264.89	0.20	267	1410	14.1	19823	0.915	0.910	0.038	1.17	58.30 2p16.1	1.00	0.1	.		
59	2 D	481.66	481.31	0.35	486	846	21.9	18552	1.003	1.072	0.063	0.83	61.00 2p16.1	0.94	-1.1	.		
<b>"2p16.1 deletion syndrome"</b>					Mean	1128	18.0	19188	0.959	0.991	0.050	1.00	(CV: 0.05)		0.98			
52	3 D	416.61	416.46	0.15	418	1309	19.4	25346	1.371	1.230	0.057	1.15	198.51 3q29	1.11	2.5	.		
45	3 C	354.59	354.58	0.01	359	1315	16.5	21731	0.954	0.959	0.061	0.85	198.28 3q29	1.00	-0.1	.		
<b>"3q29 deletion syndrome"</b>					Mean	1312	17.9	23539	1.163	1.094	0.059	1.00	(CV: 0.08)		1.06			
28	4 B	230.67	230.47	0.20	232	1420	12.4	17671	0.816	0.896	0.058	0.88	1.81 4p16.3	0.91	-1.4	.		
55	4 D	443.29	443.03	0.26	445	810	20.4	16564	0.896	0.880	0.045	1.12	1.90 4p16.3	1.02	0.3	.		
<b>4p16.3 Wolf-Hirschhorn region</b>					Mean	1115	16.4	17118	0.856	0.888	0.052	1.00	(CV: 0.08)		0.97			
54	5 D	435.65	435.58	0.07	436	552	19.2	10584	0.572	0.655	0.037	1.02	1.34 5p15.33	0.87	-2.3	.		
35	5 C	281.53	281.39	0.14	283	1421	15.0	21310	0.936	1.053	0.061	0.98	1.40 5p15.33	0.89	-1.9	.		
<b>Cri du Chat syndrome</b>					Mean	987	17.1	15947	0.754	0.854	0.049	1.00	(CV: 0.01)		0.88			
15	5 A	151.53	151.42	0.11	154	998	10.0	10003	0.478	0.972	0.047	0.98	176.62 5q35.3	0.49	-10.6*	.		
56	5 D	453.10	453.02	0.08	454	584	20.5	11959	0.647	1.134	0.052	1.02	176.65 5q35.3	0.57	-9.3*	.		
<b>Sotos syndrome</b>					Mean	791	15.3	10981	0.562	1.053	0.049	1.00	(CV: 0.11)		0.53			
39	7 C	310.37	310.40	-0.03	310	1658	15.8	26144	1.148	1.047	0.040	1.13	73.08 7q11.23	1.10	2.5	.		
46	7 C	363.62	363.46	0.16	364	1293	17.6	22815	1.002	0.995	0.048	0.88	73.11 7q11.23	1.01	0.1	.		
49	7 D	390.01	389.95	0.06	391	727	18.6	13491	0.730	0.774	0.034	0.99	73.15 7q11.23	0.94	-1.3	.		
<b>Williams syndrome</b>					Mean	1226	17.3	20817	0.960	0.938	0.041	1.00	(CV: 0.08)		1.02			
50	8 D	399.91	399.69	0.22	401	761	18.9	14376	0.777	0.753	0.051	0.88	116.75 8q24.12	1.03	0.5	.		
53	8 D	424.61	424.33	0.28	427	852	19.9	16966	0.918	0.897	0.048	1.12	117.73 8q24.11	1.02	0.4	.		
<b>Langer-Giedion syndrome</b>					Mean	807	19.4	15671	0.848	0.825	0.049	1.00	(CV: 0.01)		1.03			
40	9 C	319.39	319.40	-0.01	319	1666	15.4	25630	1.126	1.194	0.050	0.92	100.95 9q22.33	0.94	-1.4	.		
51	9 D	408.28	408.03	0.25	409	1179	19.0	22408	1.212	1.115	0.040	1.08	100.95 9q22.33	1.09	2.4	.		
<b>"9q22.3 deletion syndrome"</b>					Mean	1423	17.2	24019	1.169	1.155	0.045	1.00	(CV: 0.10)		1.02			
12	10 A	132.75	132.56	0.19	136	1954	11.2*	21931	1.047	0.895	0.048	0.90	8.14 10p	1.17	3.2			
44	10 C	349.33	349.23	0.10	349	1309	16.5	21627	0.950	0.938	0.041	1.10	10.59 10p15.1	1.01	0.3	.		
<b>DiGeorge region 2 (10p)</b>					Mean	1632	13.9	21779	0.998	0.917	0.044	1.00	(CV: 0.10)		1.08			
26	11 B	218.80	218.68	0.12	220	2203	12.6	27653	1.276	1.184	0.062	1.00	31.78 11p13	1.08	1.5	.		
<b>WAGR syndrome</b>					Mean	2203	12.6	27653	1.276	1.184	0.062	1.00	(CV: )		1.08			
25	15 B	213.65	213.54	0.11	214	1929	12.4	23938	1.105	1.162	0.060	0.90	21.48 15q11.2	0.95	-1.0	.		
30	15 B	245.31	245.06	0.25	247	1385	13.9	19198	0.886	0.909	0.047	0.90	22.65 15q12	0.97	-0.5	.		
36	15 C	289.93	289.75	0.18	292	1506	14.4	21613	0.949	0.932	0.035	1.25	22.76 15q12	1.02	0.5	.		
16	15 A	158.09	158.00	0.09	160	1639	11.4	18734	0.894	0.838	0.041	0.95	23.17 15q12	1.07	1.4	.		
<b>Prader-Willi / Angelman</b>					Mean	1615	13.0	20871	0.959	0.960	0.046	1.00	(CV: 0.05)		1.00			
21	15 A	189.88	189.75	0.13	190	2213	11.7	25975	1.240	1.228	0.057	1.07	72.50 15q24.1	1.01	0.2	.		
41	15 C	326.47	326.43	0.04	325	903	15.8	14304	0.628	0.753	0.040	0.93	72.80 15q24.1	0.83	-3.1	.		
<b>"15q24 deletion syndrome"</b>					Mean	1558	13.8	20140	0.934	0.990	0.049	1.00	(CV: 0.13)		0.93			
18	16 A	170.94	170.91	0.03	172	1900	11.5	21894	1.045	0.921	0.053	1.00	3.87 16p13.3	1.14	2.3	.		
<b>Rubinstein-Taybi syndrome</b>					Mean	1900	11.5	21894	1.045	0.921	0.053	1.00	(CV: )		1.14			
13	17 A	139.18	138.97	0.21	142	2015	11.3	22795	1.088	1.138	0.031	1.42	2.51 17p13.3	0.96	-1.6	.		
29	17 B	236.40	236.22	0.18	238	1426	13.0	18583	0.858	0.859	0.058	0.58	2.52 17p13.3	1.00	0.0	.		
<b>Miller-Dieker region</b>					Mean	1721	12.2	20689	0.973	0.999	0.044	1.00	(CV: 0.03)		0.97			
57	17 D	463.27	463.07	0.20	465	1300	21.0	27243	1.473	1.385	0.097	0.75	17.53 17p11.2-##	1.06	0.9	.		
34	17 B	272.06	271.90	0.16	274	1468	14.5	21285	0.982	0.934	0.038	1.30	17.83 17p11.2	1.05	1.3	.		
38	17 C	303.03	302.90	0.13	303	1131	15.1	17129	0.752	0.796	0.044	0.95	18.08 17p11.2	0.95	-1.0	.		
<b>Smith-Magenis syndrome</b>					Mean	1300	16.9	21886	1.069	1.038	0.060	1.00	(CV: 0.06)		1.02			
32	17 B	258.43	258.33	0.10	260	921	13.9	12836	0.592	0.578	0.041	0.94	26.56 17q11.2	1.02	0.4	.		
42	17 C	334.49	334.45	0.04	335	1459	16.5	24041	1.056	1.044	0.065	1.06	26.58 17q11.2	1.01	0.2	.		
<b>NF1 microdeletion syndrome</b>					Mean	1190	15.2	18439	0.824	0.811	0.053	1.00	(CV: 0.01)		1.02			
58	17 D	470.92	470.58	0.34	472	714	21.1	15031	0.813	0.728	0.046	0.78	41.26 17q21.31	1.12	1.9	.		
27	17 B	224.97	224.79	0.18	226	2275	13.4	30392	1.403	1.273	0.049	1.26	41.44 17q21.31	1.10	2.6	.		
43	17 C	341.77	341.78	-0.01	342	1229	16.6	20437	0.898	0.896	0.046	0.95	41.45 17q21.31	1.00	0.0	.		
<b>"17q21.31 microdeletion"</b>					Mean	1406	17.0	21953	1.038	0.966	0.047	1.00	(CV: 0.06)		1.07			

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	195.31	195.29	0.02	196	1368	11.2	15343	0.732	0.767	0.034	1.08	17.89 22q11.21	0.96	-1.0	.	
24	22 B	208.20	207.99	0.21	208	1803	12.4	22295	1.029	1.070	0.053	0.96	18.09 22q11.21	0.96	-0.8	.	
47	22 D	371.62	371.47	0.15	373	981	18.6	18286	0.989	0.942	0.047	0.96	19.57 22q11.21	1.05	1.0	.	
<b>22q11.21 (DiGeorge)</b>					Mean	1384	14.1	18641	0.917	0.926	0.044	1.00	(CV: <b>0.05</b> )	<b>0.99</b>			
31	22 B	252.15	251.96	0.19	253	1903	13.9	26516	1.224	1.330	0.090	0.75	49.49 22q13.33-##	0.92	-1.2	.	
48	22 D	382.20	382.09	0.11	382	1378	18.6	25606	1.385	1.436	0.059	1.25	49.50 22q13.33-#	0.96	-0.9	.	
<b>22q13 (Phelan-McDermid)</b>					Mean	1641	16.3	26061	1.304	1.383	0.075	1.00	(CV: <b>0.03</b> )	<b>0.95</b>			
37	X C	295.53	295.46	0.07	297	2161	14.8	31935	1.403	1.392	0.054	1.00	32.29 Xp21.2	1.01	0.2	.	
<b>Chromosome X control probe</b>					Mean	2161	14.8	31935	1.403	1.392	0.054	1.00	(CV: )	<b>1.01</b>			
23	X B	199.84	199.64	0.20	202	1400	11.1	15542	0.717	0.895	0.045	1.00	153.02 Xq28	0.80	-4.0	.	
14	X A	147.00	146.82	0.18	148	1987	10.8	21521	1.027	0.964	0.056	0.86	152.95 Xq28	1.07	1.1	.	
20	X A	183.23	183.16	0.07	184	2015	12.3	24759	1.182	1.214	0.053	1.14	152.94 Xq28-#	0.97	-0.6	.	
<b>Xq28 (RETT / MECP2)</b>					Mean	1801	11.4	20607	0.976	1.025	0.051	1.00	(CV: <b>0.14</b> )	<b>0.94</b>			
<b>Mean values</b>			0.14			1432	15.1	20573	<b>0.984</b>	1.000	0.050	4		0.98	Total of all except		
<b>Standard deviations</b>			0.09			(Coef. of variance: 0.243)			0.231	0.196				0.12	Ctrl and '?' peaks		

Quality assessment	Quality limits	Quality
Mean A-group area / mean Q-frag. area	>1.00 (1.75)	3.01
Mean CpG-area / mean A-group area	>0.55 (0.85)	1.24
Mean height of first probes AB	> 450 ( 800)	1715
Mean height of last probes CD	> 280 ( 500)	1162
Ratio of mean heights AB/CD ('slope')	<2.00 (1.55)	1.48
Mean group CV of weighted ratio	<0.20 (0.15)	0.07
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: 5q35.3 5q35.3**

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 113 (14 peaks). CV of ROX heights for peaks above 100 nt is: 0.12

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