

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	Dist. Ratio	1.0 in SD	low high	
14	64 -	61.07	60.69	0.38	64	320	10.7	3428	1.217	1.269	0.229	1.20	64 nt	0.96	-0.2	.	
15	70 -	67.10	66.75	0.35	70	174	12.2	2122	0.753	0.817	0.150	1.18	70 nt	0.92	-0.4	.	
16	76 -	73.31	72.95	0.36	76	237	11.6	2749	0.976	0.997	0.203	1.06	76 nt	0.98	-0.1	.	
17	82 -	79.56	79.25	0.31	82	247	12.0	2971	1.054	0.918	0.357	0.56	82 nt	1.15	0.4	.	
Ctrl: Q-fragments					Mean	245	11.6	2818	1.000	1.000	0.235	1.00	(CV: 0.08)	0.98			
18	6 a	85.96	85.72	0.24	88	2398	5.8	13992	1.128	1.405	1.008	0.34	6p21.3 CpG isl.	0.80	-0.3	.	
20	2 a	91.38	91.11	0.27	92	1869	5.3	9964	0.804	0.782	0.193	1.00	2q14 synt.	1.03	0.1	.	
21	1 a	97.41	97.14	0.27	96	3525	6.0	20997	1.693	1.615	0.241	1.65	MV 36 1p36 CpG isl.	1.05	0.3	.	
Ctrl: D-fragments					Mean	2597	5.7	14984	1.209	1.267	0.481	1.00	(CV: 0.09)	1.01			
22	X a	101.13	100.95	0.18	100	2069	5.8	12034	0.971	0.912	0.183	1.00	Xq23	1.06	0.3	.	
	Y a		105.47		105					0.796			Yq11				
	Y a		115.28		118					0.637			Yq11				
Ctrl: X- & Y-fragments					Mean	2069	5.8	12034	0.971	0.912	0.183	1.00	(CV:)	1.06			
23	1 A	126.99	126.81	0.18	130	1672	5.7	9605	0.775	0.744	0.092	0.57	1.14 1p36.33	1.04	0.3	.	
31	1 A	177.56	177.39	0.17	178	1937	5.9	11335	0.914	0.983	0.094	0.74	1.75 1p36.33	0.93	-0.7	.	
29	1 A	165.39	165.19	0.20	166	2420	5.9	14386	1.160	1.117	0.046	1.69	1.95 1p36.33	1.04	0.9	.	
1p36 (1p-deletion)					Mean	2010	5.8	11775	0.950	0.948	0.077	1.00	(CV: 0.06)	1.01			
45	2 B	266.16	266.00	0.16	267	1276	6.2	7904	0.839	0.833	0.066	1.18	58.30 2p16.1	1.01	0.1	.	
71	2 D	481.99	481.94	0.05	486	572	7.9	4534	0.852	0.878	0.099	0.82	61.00 2p16.1	0.97	-0.3	.	
"2p16.1 deletion syndrome"					Mean	924	7.1	6219	0.845	0.856	0.083	1.00	(CV: 0.03)	0.99			
64	3 D	417.03	416.93	0.10	418	1070	7.2	7713	1.449	1.278	0.111	0.77	198.51 3q29	1.13	1.5	.	
57	3 C	355.21	355.13	0.08	359	1021	6.8	6972	0.855	1.036	0.057	1.23	198.28 3q29	0.82	-3.2	.	
"3q29 deletion syndrome"					Mean	1046	7.0	7343	1.152	1.157	0.084	1.00	(CV: 0.23)	0.94			
40	4 B	231.95	231.76	0.19	232	1554	5.9	9239	0.981	0.892	0.068	0.88	1.81 4p16.3	1.10	1.3	.	
67	4 D	444.14	443.99	0.15	445	623	7.5	4684	0.880	0.869	0.052	1.12	1.90 4p16.3	1.01	0.2	.	
4p16.3 Wolf-Hirschhorn region					Mean	1089	6.7	6962	0.930	0.881	0.060	1.00	(CV: 0.06)	1.05			
66	5 D	436.85	436.65	0.20	436	533	7.6	4026	0.756	0.643	0.044	1.05	1.34 5p15.33	1.18	2.6	.	
47	5 C	282.08	282.02	0.06	283	1422	6.5	9174	1.125	0.971	0.074	0.95	1.40 5p15.33	1.16	2.1	.	
Cri du Chat syndrome					Mean	978	7.0	6600	0.941	0.807	0.059	1.00	(CV: 0.01)	1.17			
27	5 A	152.77	152.53	0.24	154	2481	5.8	14469	1.167	1.143	0.048	1.08	176.62 5q35.3	1.02	0.5	.	
68	5 D	453.47	453.34	0.13	454	711	7.6	5389	1.012	1.108	0.055	0.92	176.65 5q35.3	0.91	-1.7	.	
Sotos syndrome					Mean	1596	6.7	9929	1.090	1.126	0.052	1.00	(CV: 0.08)	0.97			
51	7 C	310.49	310.43	0.06	310	1400	6.6	9242	1.133	1.142	0.075	1.12	73.08 7q11.23	0.99	-0.1	.	
58	7 C	363.65	363.45	0.20	364	1033	7.1	7302	0.895	1.042	0.062	1.23	73.11 7q11.23	0.86	-2.4	.	
61	7 D	389.97	389.86	0.11	391	1090	7.3	7929	1.489	1.455	0.163	0.66	73.15 7q11.23	1.02	0.2	.	
Williams syndrome					Mean	1174	7.0	8158	1.173	1.213	0.100	1.00	(CV: 0.09)	0.94			
62	8 D	400.35	400.27	0.08	401	553	7.2	3988	0.749	0.703	0.052	1.06	116.75 8q24.12	1.06	0.9	.	
65	8 D	425.88	425.76	0.12	427	565	7.8	4389	0.824	0.865	0.073	0.94	117.73 8q24.11	0.95	-0.6	.	
Langer-Giedion syndrome					Mean	559	7.5	4189	0.787	0.784	0.062	1.00	(CV: 0.08)	1.01			
52	9 C	319.58	319.60	-0.02	319	1521	6.5	9897	1.214	1.152	0.066	1.26	100.95 9q22.33	1.05	0.9	.	
63	9 D	408.37	408.22	0.15	409	848	7.4	6242	1.172	1.067	0.104	0.74	100.95 9q22.33	1.10	1.0	.	
"9q22.3 deletion syndrome"					Mean	1185	6.9	8070	1.193	1.110	0.085	1.00	(CV: 0.03)	1.07			
24	10 A	133.50	133.29	0.21	136	2269	5.9	13335	1.076	0.951	0.046	0.97	8.14 10p	1.13	2.7	.	
56	10 C	349.78	349.68	0.10	349	1054	6.8	7178	0.880	0.982	0.045	1.03	10.59 10p15.1	0.90	-2.3	.	
DiGeorge region 2 (10p)					Mean	1662	6.3	10257	0.978	0.967	0.046	1.00	(CV: 0.16)	1.01			
38	11 B	219.58	219.49	0.09	220	1800	6.1	10931	1.161	1.145	0.057	1.00	31.78 11p13	1.01	0.3	.	
WAGR syndrome					Mean	1800	6.1	10931	1.161	1.145	0.057	1.00	(CV:)	1.01			
37	15 B	214.55	214.40	0.15	214	1774	5.9	10493	1.114	1.185	0.064	1.19	21.48 15q11.2	0.94	-1.1	.	
42	15 B	246.42	246.25	0.17	247	1368	6.3	8644	0.918	0.876	0.058	0.97	22.65 15q12	1.05	0.7	.	
48	15 C	290.10	290.07	0.03	292	1401	6.4	9024	1.107	0.920	0.069	0.86	22.76 15q12	1.20	2.7	.	
28	15 A	159.08	158.88	0.20	160	1794	5.9	10527	0.849	0.894	0.058	0.98	23.17 15q12	0.95	-0.8	.	
Prader-Willi / Angelman					Mean	1584	6.1	9672	0.997	0.969	0.062	1.00	(CV: 0.12)	1.02			
33	15 A	190.80	190.64	0.16	190	2448	5.8	14150	1.141	1.183	0.086	0.87	72.50 15q24.1*	0.97	-0.5	.	
53	15 C	325.60	325.70	-0.10	325	771	6.6	5056	0.620	0.678	0.038	1.13	72.80 15q24.1	0.91	-1.5	.	
"15q24 deletion syndrome"					Mean	1610	6.2	9603	0.881	0.930	0.062	1.00	(CV: 0.04)	0.94			
30	16 A	171.67	171.56	0.11	172	2065	6.0	12358	0.997	0.952	0.045	1.00	3.87 16p13.3	1.05	1.0	.	
Rubinstein-Taybi syndrome					Mean	2065	6.0	12358	0.997	0.952	0.045	1.00	(CV:)	1.05			
25	17 A	140.33	140.07	0.26	142	2742	5.9	16137	1.301	1.196	0.072	1.05	2.51 17p13.3	1.09	1.4	.	
41	17 B	236.99	236.92	0.07	238	1396	6.2	8597	0.913	0.858	0.057	0.95	2.52 17p13.3	1.06	1.0	.	
Miller-Dieker region					Mean	2069	6.0	12367	1.107	1.027	0.065	1.00	(CV: 0.02)	1.08			
69	17 D	463.64	463.52	0.12	465	809	7.6	6179	1.161	1.276	0.165	0.48	17.53 17p11.2-##	0.91	-0.7	.	
46	17 B	272.52	272.44	0.08	274	1282	6.2	7924	0.841	0.860	0.045	1.20	17.83 17p11.2	0.98	-0.4	.	
50	17 C	303.65	303.54	0.11	303	1044	6.5	6821	0.836	0.779	0.036	1.33	18.08 17p11.2	1.07	1.6	.	
Smith-Magenis syndrome					Mean	1045	6.8	6975	0.946	0.972	0.082	1.00	(CV: 0.07)	1.01			
44	17 B	259.14	258.97	0.17	260	1584	6.3	9970	1.059	1.082	0.046	0.88	26.56 17q11.2	0.98	-0.5	.	
54	17 C	334.79	334.83	-0.04	335	1066	6.7	7160	0.878	1.001	0.034	1.12	26.58 17q11.2	0.88	-3.6	.	
NF1 microdeletion syndrome					Mean	1325	6.5	8565	0.968	1.041	0.040	1.00	(CV: 0.08)	0.92			
70	17 D	471.78	471.67	0.11	472	424	7.7	3246	0.610	0.755	0.046	0.89	41.26 17q21.31	0.81	-3.1	.	
39	17 B	226.59	226.43	0.16	226	1967	6.0	11830	1.256	1.279	0.076	0.91	41.44 17q21.31	0.98	-0.3	.	
55	17 C	341.67	341.67	0.00	342	981	6.8	6623	0.812	0.891	0.040	1.20	41.45 17q21.31	0.91	-1.9	.	
"17q21.31 microdeletion"					Mean	1124	6.8	7233	0.893	0.975	0.054	1.00	(CV: 0.09)	0.90			

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	Dist. in SD	1.0 low	1.0 high
34	22 A	196.57	196.48	0.09	196	844	5.7	4844	0.391	0.834	0.046	1.15	17.89 22q11.21	0.47	-9.7*	.
36	22 B	209.41	209.20	0.21	208	814	6.0	4862	0.516	0.940	0.056	1.05	18.09 22q11.21	0.55	-7.5*	.
59	22 D	371.80	371.60	0.20	373	364	8.0*	2912	0.547	0.913	0.071	0.81	19.57 22q11.21	0.60	-5.2*	.
22q11.21 (DiGeorge)					Mean	674	6.6	4206	0.485	0.896	0.058	1.00	(CV: 0.12)	0.53		
43	22 B	253.21	253.12	0.09	253	1603	6.3	10164	1.079	1.130	0.129	0.91	49.49 22q13.33-##	0.96	-0.4	.
60	22 D	382.70	382.54	0.16	382	897	7.4	6602	1.240	1.189	0.114	1.09	49.50 22q13.33-#	1.04	0.4	.
22q13 (Phelan-McDermid)					Mean	1250	6.9	8383	1.160	1.160	0.122	1.00	(CV: 0.06)	1.00		
49	X C	295.71	295.60	0.11	297	1798	6.4	11466	1.406	1.406	0.082	1.00	32.29 Xp21.2	1.00	0.0	.
Chromosome X control probe					Mean	1798	6.4	11466	1.406	1.406	0.082	1.00	(CV:)	1.00		
35	X B	201.31	201.09	0.22	202	1686	6.0	10194	1.083	0.920	0.062	1.02	153.02 Xq28	1.18	2.6	..
26	X A	147.93	147.68	0.25	148	2139	5.8	12433	1.003	0.963	0.052	1.27	152.95 Xq28	1.04	0.8	.
32	X A	184.65	184.46	0.19	184	2084	5.9	12246	0.988	1.039	0.101	0.71	152.94 Xq28-#	0.95	-0.5	.
Xq28 (RETT / MECP2)					Mean	1970	5.9	11624	1.024	0.974	0.071	1.00	(CV: 0.10)	1.07		
Mean values				0.13		1359	6.6	8578	0.980	1.000	0.069	4		0.98	Total of all except	
Standard deviations				0.08		(Coef. of variance:	0.379)		0.235	0.183				0.15	Ctrl and '?' peaks	

Quality assessment		Quality limits	Quality
Mean A-group area / mean Q-frag. area		>0.65 (1.50)	4.40
Mean CpG-area / mean A-group area		>0.30 (0.85)	1.41
Mean height of first probes AB		> 450 (800)	1792
Mean height of last probes CD		> 280 (500)	943
Ratio of mean heights AB/CD ('slope')		<2.00 (1.55)	1.90 high
Mean group CV of weighted ratio		<0.20 (0.15)	0.08
0 unidentified peak areas / 53 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: 22q11.21 22q11.21 22q11.21

1 quality warning!

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 218 (14 peaks). CV of ROX heights for peaks above 100 nt is: 0.05

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