

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	
4	64 -	60.67	60.62	0.05	64	149	18.1	2697	0.590	0.653	0.326	0.43	64 nt	0.90	-0.2	.	
5	70 -	66.41	66.60	-0.19	70	224	19.3	4324	0.946	0.883	0.166	1.15	70 nt	1.07	0.4	.	
7	76 -	72.60	72.49	0.11	76	273	18.2	4964	1.086	1.154	0.214	1.17	76 nt	0.94	-0.3	.	
9	82 -	78.46	78.66	-0.20	82	306	20.6	6296	1.378	1.145	0.201	1.24	82 nt	1.20	1.2	.	
<b>Ctrl: Q-fragments</b>					Mean	238	19.0	4570	1.000	0.959	0.227	1.00	(CV: 0.13)	<b>1.06</b>			
12	2 A	90.16	90.38	-0.22	94	3018	11.0	33336	0.927	0.959	0.074	1.00	MV 36 2q14 synt.	0.97	-0.4	.	
<b>Synthetic control probe</b>					Mean	3018	11.0	33336	0.927	0.959	0.074	1.00	(CV: )	<b>0.97</b>			
13	1 A	126.00	126.22	-0.22	130	3917	11.4	44491	1.238	1.115	0.071	0.88	1.1 1p36.33	1.11	1.7	.	
45	1 D	380.59	380.60	-0.01	382	1334	15.7	20971	1.094	0.914	0.062	0.82	1.1 1p36.3	1.20	2.9	.	
14	1 A	131.32	131.51	-0.19	136	3443	11.1	38239	1.064	0.964	0.085	0.63	1.2 1p36.33	1.10	1.2	.	
21	1 A	176.51	176.62	-0.11	178	4011	11.8	47244	1.314	1.378	0.055	1.39	1.8 1p36.33	0.95	-1.2	.	
51	1 D	433.13	433.48	-0.35	436	1540	17.1	26258	1.370	1.277	0.074	0.96	2.2 1p36	1.07	1.3	.	
30	1 B	245.35	245.37	-0.02	247	2796	12.9	35976	1.155	1.189	0.046	1.43	2.4 1p36.33	0.97	-0.7	.	
40	1 C	336.94	336.88	0.06	337	1994	15.0	29881	1.018	0.993	0.061	0.89	3.6 1p36.3	1.03	0.4	.	
<b>1p36 (1p-deletion)</b>					Mean	2719	13.6	34723	1.179	1.119	0.065	1.00	(CV: 0.08)	<b>1.04</b>			
52	5 D	443.19	443.40	-0.21	445	688	17.5	12027	0.627	0.576	0.044	0.91	176.6 NSD1 exon 16	1.09	1.2	.	
15	5 A	140.04	140.22	-0.18	142	3715	11.2	41459	1.153	1.155	0.069	1.16	176.6 NSD1 exon 11	1.00	0.0	.	
26	5 B	209.44	209.52	-0.08	211	1786	13.4	23848	0.765	0.801	0.059	0.93	176.6 NSD1 exon 8	0.96	-0.6	.	
<b>5q35.3 (Sotos)</b>					Mean	2063	14.0	25778	0.849	0.844	0.057	1.00	(CV: 0.06)	<b>1.01</b>			
38	7 C	320.89	320.79	0.10	319	1708	14.2	24260	0.827	0.897	0.050	1.02	19.1 7p21.2	0.92	-1.4	.	
25	7 B	201.05	201.14	-0.09	202	2650	11.8	31319	1.005	0.979	0.056	0.98	19.7 7p21.2	1.03	0.5	.	
<b>7p21.2 (Saethre-Chotzen)</b>					Mean	2179	13.0	27790	0.916	0.938	0.053	1.00	(CV: 0.08)	<b>0.97</b>			
31	7 B	254.46	254.55	-0.09	256	2977	13.1	39020	1.252	1.238	0.072	0.83	72.5 7q11.23	1.01	0.2	.	
35	7 C	289.57	289.57	0.00	292	2686	13.8	37087	1.264	1.299	0.061	1.03	72.8 7q11.23	0.97	-0.6	.	
37	7 C	310.63	310.44	0.19	310	2653	14.2	37705	1.285	1.379	0.054	1.22	73.1 7q11.23	0.93	-1.7	.	
46	7 D	390.02	390.01	0.01	391	1383	16.0	22131	1.154	1.120	0.059	0.92	73.2 7q11.23	1.03	0.6	.	
28	7 B	228.86	228.90	-0.04	229	2249	12.7	28531	0.916	0.943	0.043	1.06	73.4 7q11.23	0.97	-0.6	.	
49	7 D	416.02	416.13	-0.11	418	1680	16.7	28057	1.464	1.440	0.075	0.93	73.4 7q11.23	1.02	0.3	.	
<b>7q11.23 (Williams)</b>					Mean	2271	14.4	32089	1.222	1.236	0.061	1.00	(CV: 0.04)	<b>0.99</b>			
20	15 A	170.45	170.56	-0.11	172	1101	11.1	12199	0.339	0.578	0.040	1.01	21.4 15q11.2	0.59	-6.0 *	.	
27	15 B	217.99	218.10	-0.11	220	1204	12.3	14777	0.474	0.931	0.057	1.13	21.5 15q11.2	0.51	-8.1 *	.	
18	15 A	158.00	158.11	-0.11	160	1458	11.2	16311	0.454	0.941	0.066	0.98	23.2 15q12	0.48	-7.4 *	.	
47	15 D	399.34	399.38	-0.04	400	649	16.6	10757	0.561	0.824	0.094	0.60	23.2 15q12**	0.68	-2.8	.	
48	15 D	407.42	407.57	-0.15	409	692	16.4	11370	0.593	1.078	0.058	1.27	24.5 15q12	0.55	-8.3 *	.	
<b>15q11q12 (Prader-Willi)</b>					Mean	1021	13.5	13083	0.484	0.870	0.063	1.00	(CV: 0.12)	<b>0.55</b>	<b>P= 0.02%</b>		
43	17 C	363.22	363.32	-0.10	364	1178	15.4	18084	0.616	0.713	0.054	0.79	1.9 17p13.3	0.86	-1.8	.	
22	17 A	183.11	183.15	-0.04	184	3274	11.5	37777	1.051	1.038	0.068	0.92	1.9 17p13.3	1.01	0.2	.	
50	17 D	424.40	424.61	-0.21	427	1031	17.6	18145	0.947	0.847	0.070	0.72	2.3 17p13.3	1.12	1.4	.	
29	17 B	236.31	236.37	-0.06	238	1690	12.5	21118	0.678	0.792	0.050	0.95	2.5 17p13.3	0.86	-2.3	.	
34	17 C	281.54	281.46	0.08	283	1999	13.3	26581	0.906	0.901	0.057	0.95	2.5 17p13.3	1.01	0.1	.	
32	17 B	264.01	263.99	0.02	265	2491	13.2	32883	1.055	0.959	0.047	1.23	3.3 17p13.3	1.10	2.0	.	
16	17 A	145.80	145.97	-0.17	148	3482	11.0	38163	1.062	1.040	0.043	1.44	3.4 17p13.3	1.02	0.5	.	
<b>17p13.3 (Miller-Dieker)</b>					Mean	2164	13.5	27536	0.902	0.898	0.056	1.00	(CV: 0.10)	<b>1.00</b>			
19	17 A	164.46	164.62	-0.16	166	2511	11.9	29797	0.829	0.781	0.049	0.93	16.8 17p11.2	1.06	1.0	.	
33	17 B	272.00	271.98	0.02	274	2548	13.3	33829	1.086	0.995	0.055	1.05	17.8 17p11.2	1.09	1.6	.	
36	17 C	300.96	300.85	0.11	301	2172	13.9	30293	1.032	1.031	0.045	1.34	18.1 17p11.2	1.00	0.0	.	
39	17 C	328.88	328.70	0.18	328	1844	14.6	26984	0.920	0.955	0.064	0.88	18.7 17p11.2	0.96	-0.6	.	
42	17 C	354.02	354.04	-0.02	355	1521	15.2	23120	0.788	0.793	0.058	0.80	19.2 17p11.2	0.99	-0.1	.	
<b>17p11.2 (Smith-Magenis)</b>					Mean	2119	13.8	28805	0.931	0.911	0.054	1.00	(CV: 0.05)	<b>1.02</b>			
41	20 C	345.38	345.23	0.15	346	1901	15.0	28589	0.974	0.969	0.050	1.23	10.6 20p12.2 JAG1	1.01	0.1	.	
55	20 D	470.10	470.28	-0.18	472	793	18.1	14350	0.749	0.681	0.057	0.77	10.6 20p12.2 JAG1	1.10	1.2	.	
<b>20p12.2 (Alagille)</b>					Mean	1347	16.6	21470	0.861	0.825	0.053	1.00	(CV: 0.06)	<b>1.04</b>			
23	22 B	190.41	190.45	-0.04	190	3190	11.5	36678	1.177	1.215	0.065	1.14	17.6 22q11.21	0.97	-0.6	.	
54	22 D	462.30	462.52	-0.22	463	1522	17.7	26896	1.403	1.510	0.085	1.08	17.9 22q11.21	0.93	-1.3	.	
24	22 B	195.37	195.38	-0.01	196	2722	11.6	31519	1.012	0.959	0.061	0.96	17.9 22q11.21	1.06	0.9	.	
17	22 A	152.46	152.65	-0.19	154	3633	11.3	41162	1.145	1.051	0.078	0.82	18.4 22q11.21	1.09	1.2	.	
53	22 D	451.67	451.94	-0.27	454	654	18.0	11752	0.613	0.734	0.053	0.85	19.2 22q11.21	0.83	-2.3	.	
44	22 C	371.52	371.53	-0.01	373	1662	16.7	27710	0.944	1.069	0.056	1.17	19.6 22q11.21	0.88	-2.2	.	
<b>22q11.21 (DiGeorge)</b>					Mean	2231	14.5	29286	1.049	1.090	0.066	1.00	(CV: 0.10)	<b>0.96</b>			
<b>Mean values</b>				-0.07		2117	13.9	27788	<b>0.961</b>	1.000	0.060	2		0.95	Total of all except		
<b>Standard deviations</b>				0.12		(Coef. of variance: 0.351)			0.270	0.216				0.16	Ctrl and '?' peaks		

Quality assessment	Quality limits	Quality
Mean A-group area / mean Q-frag. area	>0.65 (1.50)	7.87
Mean height of first probes AB	> 450 ( 800)	2721
Mean height of last probes CD	> 280 ( 500)	1513
Ratio of mean heights AB/CD ('slope')	<3.00 (2.50)	1.80
Mean group CV of weighted ratio	<0.20 (0.15)	0.08
0 unidentified peak areas / 44 peak areas	< (0.02)	0.00

Weighted mean ratios are tested for being outside ratio 1±0.13  
One-tailed significance is high for p<=1%, and low for p<=5%.  
Individual peaks having normalized area > 4.0 SD from the ref.  
mean and ratio <0.65 or >1.3 indicate 'abnormal' probe area.

**High significance P= 0.022%** **Female & male ref.**  
**Loss of 15q11q12**

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 186 (14 peaks). 100\*CV of ROX heights for peaks above 100 nt is: 3.77