

| 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 |
| 64 | - | | 60.65 | | 64 | | | | | 1.190 | 0.371 | | 64 nt | | | |
| 70 | - | | 66.46 | | 70 | | | | | 0.913 | 0.264 | | 70 nt | | | |
| 76 | - | | 72.37 | | 76 | | | | | 1.138 | 0.170 | | 76 nt | | | |
| 82 | - | | 78.56 | | 82 | | | | | 0.787 | 0.204 | | 82 nt | | | |
| Ctrl: Q-fragments | | | | | Mean | | | | | | | 1.00 | (CV:) | | | |
| 6 | 6 a | 85.21 | 84.95 | 0.26 | 88 | 455 | 11.8* | 5353 | 0.370 | 0.981 | 0.152 | 1.02 | 6p21.3 CpG isl. | 0.38 | -4.0* | |
| 8 | 2 a | 90.45 | 90.17 | 0.28 | 92 | 597 | 10.7 | 6386 | 0.442 | 0.967 | 0.135 | 1.13 | 2q14 synt. | 0.46 | -3.9 | |
| 10 | 1 a | 96.86 | 96.50 | 0.36 | 96 | 532 | 10.9 | 5780 | 0.400 | 1.641 | 0.304 | 0.85 | MV 36 1p36 CpG isl. | 0.24 | -4.1* | |
| Ctrl: D-fragments | | | | | Mean | 528 | 11.1 | 5840 | 0.404 | 1.197 | 0.197 | 1.00 | (CV: 0.29) | 0.37 | | |
| 14 | Y a | 114.83 | 114.50 | 0.33 | 118 | 429 | 9.8 | 4193 | 0.290 | 0.811 | 0.088 | 1.03 | 13.54 Yq11 | 0.36 | -5.9* | |
| 12 | Y a | 104.56 | 104.34 | 0.22 | 108 | 335 | 10.0 | 3335 | 0.231 | 0.566 | 0.065 | 0.97 | 14.10 Yq11 | 0.41 | -5.2* | |
| ctrl: Y-fragments (male ref.) | | | | | Mean | 382 | 9.9 | 3764 | 0.260 | 0.688 | 0.076 | 1.00 | (CV: 0.09) | 0.38 | | |
| 15 | 1 A | 126.41 | 126.04 | 0.37 | 130 | 1668 | 11.0 | 18341 | 1.268 | 1.200 | 0.054 | 1.30 | 1.14 1p36.33 | 1.06 | 1.3 | - |
| 26 | 1 A | 176.70 | 176.49 | 0.21 | 178 | 1349 | 11.1 | 14934 | 1.033 | 0.942 | 0.074 | 0.75 | 1.75 1p36.33 | 1.10 | 1.2 | - |
| 24 | 1 A | 164.69 | 164.53 | 0.16 | 166 | 1507 | 10.7 | 16200 | 1.120 | 1.181 | 0.073 | 0.95 | 1.95 1p36.33 | 0.95 | -0.8 | - |
| 1p36 (1p-deletion) | | | | | Mean | 1508 | 10.9 | 16492 | 1.140 | 1.108 | 0.067 | 1.00 | (CV: 0.07) | 1.03 | | |
| 40 | 2 B | 265.07 | 264.89 | 0.18 | 267 | 1326 | 12.2 | 16153 | 1.175 | 0.948 | 0.040 | 1.36 | 58.30 2p16.1 | 1.24 | 5.6* | |
| 66 | 2 D | 481.52 | 481.31 | 0.21 | 486 | 617 | 15.2 | 9409 | 0.770 | 1.121 | 0.101 | 0.64 | 61.00 2p16.1 | 0.69 | -3.5 | |
| "2p16.1 deletion syndrome" | | | | | Mean | 972 | 13.7 | 12781 | 0.973 | 1.034 | 0.071 | 1.00 | (CV: 0.34) | 1.06 | | |
| 59 | 3 D | 416.62 | 416.46 | 0.16 | 418 | 1328 | 14.3 | 19056 | 1.560 | 1.268 | 0.073 | 0.96 | 198.51 3q29 | 1.23 | 4.0 | |
| 52 | 3 C | 354.66 | 354.58 | 0.08 | 359 | 1241 | 13.7 | 17035 | 1.142 | 1.039 | 0.056 | 1.04 | 198.28 3q29 | 1.10 | 1.9 | - |
| "3q29 deletion syndrome" | | | | | Mean | 1285 | 14.0 | 18046 | 1.351 | 1.154 | 0.064 | 1.00 | (CV: 0.08) | 1.16 | | |
| 35 | 4 B | 230.78 | 230.47 | 0.31 | 232 | 1340 | 11.8 | 15854 | 1.154 | 0.944 | 0.063 | 0.88 | 1.81 4p16.3 | 1.22 | 3.3 | |
| 62 | 4 D | 443.41 | 443.03 | 0.38 | 445 | 786 | 14.7 | 11526 | 0.943 | 0.879 | 0.046 | 1.12 | 1.90 4p16.3 | 1.07 | 1.4 | - |
| 4p16.3 Wolf-Hirschhorn region | | | | | Mean | 1063 | 13.2 | 13690 | 1.048 | 0.912 | 0.054 | 1.00 | (CV: 0.09) | 1.14 | | |
| 61 | 5 D | 435.72 | 435.58 | 0.14 | 436 | 639 | 14.7 | 9400 | 0.769 | 0.659 | 0.053 | 0.88 | 1.34 5p15.33 | 1.17 | 2.1 | |
| 42 | 5 C | 281.53 | 281.39 | 0.14 | 283 | 1034 | 12.6 | 12995 | 0.871 | 1.115 | 0.071 | 1.12 | 1.40 5p15.33 | 0.78 | -3.5 | |
| Cri du Chat syndrome | | | | | Mean | 837 | 13.6 | 11198 | 0.820 | 0.887 | 0.062 | 1.00 | (CV: 0.29) | 0.95 | | |
| 21 | 5 A | 151.60 | 151.42 | 0.18 | 154 | 1664 | 10.8 | 17944 | 1.241 | 1.127 | 0.052 | 1.03 | 176.62 5q35.3 | 1.10 | 2.2 | - |
| 63 | 5 D | 453.27 | 453.02 | 0.25 | 454 | 965 | 15.2 | 14677 | 1.201 | 1.123 | 0.055 | 0.97 | 176.65 5q35.3 | 1.07 | 1.4 | - |
| Sotos syndrome | | | | | Mean | 1315 | 13.0 | 16311 | 1.221 | 1.125 | 0.054 | 1.00 | (CV: 0.02) | 1.09 | | |
| 46 | 7 C | 310.49 | 310.40 | 0.09 | 310 | 1255 | 12.8 | 16067 | 1.077 | 1.116 | 0.056 | 0.99 | 73.08 7q11.23 | 0.97 | -0.7 | · |
| 53 | 7 C | 363.59 | 363.46 | 0.13 | 364 | 1209 | 13.7 | 16566 | 1.111 | 1.029 | 0.040 | 1.27 | 73.11 7q11.23 | 1.08 | 2.0 | - |
| 56 | 7 D | 390.05 | 389.95 | 0.10 | 391 | 907 | 14.3 | 12933 | 1.058 | 0.748 | 0.050 | 0.74 | 73.15 7q11.23 | 1.42 | 6.2* | |
| Williams syndrome | | | | | Mean | 1124 | 13.6 | 15189 | 1.082 | 0.964 | 0.049 | 1.00 | (CV: 0.19) | 1.12 | | |
| 57 | 8 D | 399.91 | 399.69 | 0.22 | 401 | 752 | 14.2 | 10677 | 0.874 | 0.776 | 0.036 | 1.06 | 116.75 8q24.12 | 1.13 | 2.7 | - |
| 60 | 8 D | 424.68 | 424.33 | 0.35 | 427 | 781 | 14.5 | 11319 | 0.926 | 0.895 | 0.047 | 0.94 | 117.73 8q24.11 | 1.04 | 0.7 | · |
| Langer-Giedion syndrome | | | | | Mean | 767 | 14.3 | 10998 | 0.900 | 0.835 | 0.042 | 1.00 | (CV: 0.06) | 1.08 | | |
| 47 | 9 C | 319.46 | 319.40 | 0.06 | 319 | 1681 | 12.7 | 21287 | 1.427 | 1.292 | 0.072 | 1.05 | 100.95 9q22.33 | 1.10 | 1.9 | - |
| 58 | 9 D | 408.24 | 408.03 | 0.21 | 409 | 1051 | 14.2 | 14915 | 1.221 | 1.119 | 0.069 | 0.95 | 100.95 9q22.33 | 1.09 | 1.5 | - |
| "9q22.3 deletion syndrome" | | | | | Mean | 1366 | 13.4 | 18101 | 1.324 | 1.206 | 0.070 | 1.00 | (CV: 0.01) | 1.10 | | |
| 17 | 10 A | 132.97 | 132.56 | 0.41 | 136 | 1257 | 10.9 | 13645 | 0.944 | 0.948 | 0.043 | 1.16 | 8.14 10p | 1.00 | -0.1 | · |
| 51 | 10 C | 349.26 | 349.23 | 0.03 | 349 | 1091 | 13.1 | 14320 | 0.960 | 0.979 | 0.060 | 0.84 | 10.59 10p15.1 | 0.98 | -0.3 | · |
| DiGeorge region 2 (10p) | | | | | Mean | 1174 | 12.0 | 13983 | 0.952 | 0.963 | 0.052 | 1.00 | (CV: 0.01) | 0.99 | | |
| 33 | 11 B | 218.89 | 218.68 | 0.21 | 220 | 1463 | 11.5 | 16796 | 1.222 | 1.246 | 0.061 | 1.00 | 31.78 11p13 | 0.98 | -0.4 | · |
| WAGR syndrome | | | | | Mean | 1463 | 11.5 | 16796 | 1.222 | 1.246 | 0.061 | 1.00 | (CV:) | 0.98 | | |
| 32 | 15 B | 213.65 | 213.54 | 0.11 | 214 | 1293 | 11.4 | 14754 | 1.074 | 1.226 | 0.047 | 1.34 | 21.48 15q11.2 | 0.88 | -3.2 | - |
| 37 | 15 B | 245.40 | 245.06 | 0.34 | 247 | 1031 | 11.9 | 12304 | 0.895 | 0.962 | 0.058 | 0.86 | 22.65 15q12 | 0.93 | -1.2 | - |
| 43 | 15 C | 289.99 | 289.75 | 0.24 | 292 | 1301 | 12.6 | 16425 | 1.101 | 1.018 | 0.061 | 0.86 | 22.76 15q12 | 1.08 | 1.4 | - |
| 22 | 15 A | 158.19 | 158.00 | 0.19 | 160 | 1262 | 11.0 | 13909 | 0.962 | 0.930 | 0.051 | 0.94 | 23.17 15q12 | 1.03 | 0.6 | · |
| Prader-Willi / Angelman | | | | | Mean | 1222 | 11.7 | 14348 | 1.008 | 1.034 | 0.054 | 1.00 | (CV: 0.10) | 0.97 | | |
| 28 | 15 A | 189.94 | 189.75 | 0.19 | 190 | 1951 | 11.1 | 21739 | 1.503 | 1.299 | 0.130 | 0.78 | 72.50 15q24.1 | 1.16 | 1.6 | |
| 48 | 15 C | 326.52 | 326.43 | 0.09 | 325 | 825 | 12.8 | 10575 | 0.709 | 0.771 | 0.049 | 1.22 | 72.80 15q24.1 | 0.92 | -1.2 | - |
| "15q24 deletion syndrome" | | | | | Mean | 1388 | 12.0 | 16157 | 1.106 | 1.035 | 0.090 | 1.00 | (CV: 0.16) | 1.01 | | |
| 25 | 16 A | 171.07 | 170.91 | 0.16 | 172 | 1084 | 10.9 | 11853 | 0.820 | 1.018 | 0.063 | 1.00 | 3.87 16p13.3 | 0.81 | -3.1 | |
| Rubinstein-Taybi syndrome | | | | | Mean | 1084 | 10.9 | 11853 | 0.820 | 1.018 | 0.063 | 1.00 | (CV:) | 0.81 | | |
| 18 | 17 A | 139.35 | 138.97 | 0.38 | 142 | 1577 | 10.1 | 15952 | 1.103 | 1.248 | 0.053 | 1.27 | 2.51 17p13.3 | 0.88 | -2.8 | - |
| 36 | 17 B | 236.35 | 236.22 | 0.13 | 238 | 1016 | 11.7 | 11894 | 0.865 | 0.900 | 0.067 | 0.73 | 2.52 17p13.3 | 0.96 | -0.5 | · |
| Miller-Dieker region | | | | | Mean | 1297 | 10.9 | 13923 | 0.984 | 1.074 | 0.060 | 1.00 | (CV: 0.06) | 0.91 | | |
| 64 | 17 D | 463.31 | 463.07 | 0.24 | 465 | 454 | 14.8 | 6705 | 0.549 | 1.331 | 0.092 | 0.83 | 17.53 17p11.2-## | 0.41 | -8.5* | |
| 41 | 17 B | 272.03 | 271.90 | 0.13 | 274 | 1199 | 12.3 | 14709 | 1.070 | 0.940 | 0.043 | 1.25 | 17.83 17p11.2 | 1.14 | 3.0 | - |
| 45 | 17 C | 303.08 | 302.90 | 0.18 | 303 | 1068 | 12.9 | 13770 | 0.923 | 0.839 | 0.053 | 0.91 | 18.08 17p11.2 | 1.10 | 1.6 | - |
| Smith-Magenis syndrome | | | | | Mean | 907 | 13.3 | 11728 | 0.847 | 1.037 | 0.063 | 1.00 | (CV: 0.42) | 0.93 | | |
| 39 | 17 B | 258.59 | 258.33 | 0.26 | 260 | 760 | 12.6 | 9574 | 0.697 | 0.621 | 0.038 | 0.88 | 26.56 17q11.2 | 1.12 | 2.0 | - |
| 49 | 17 C | 334.58 | 334.45 | 0.13 | 335 | 1087 | 13.3 | 14478 | 0.971 | 1.118 | 0.053 | 1.12 | 26.58 17q11.2 | 0.87 | -2.8 | - |
| NF1 microdeletion syndrome | | | | | Mean | 924 | 13.0 | 12026 | 0.834 | 0.870 | 0.046 | 1.00 | (CV: 0.18) | 0.98 | | |
| 65 | 17 D | 470.75 | 470.58 | 0.17 | 472 | 757 | 14.9 | 11310 | 0.926 | 0.719 | 0.047 | 0.70 | 41.26 17q21.31 | 1.29 | 4.4* | |
| 34 | 17 B | 225.06 | 224.79 | 0.27 | 226 | 1762 | 11.9 | 21040 | 1.531 | 1.325 | 0.050 | 1.21 | 41.44 17q21.31 | 1.16 | 4.1* | |
| 50 | 17 C | 341.82 | 341.78 | 0.04 | 342 | 899 | 13.4 | 12082 | 0.810 | 0.932 | 0.039 | 1.09 | 41.45 17q21.31 | 0.87 | -3.1 | - |
| "17q21.31 microdeletion" | | | | | Mean | 1139 | 13.4 | 14811 | 1.089 | 0.992 | 0.046 | 1.00 | (CV: 0.19) | 1.08 | | |

| 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 | | |
| 29 | 22 A | 195.37 | 195.29 | 0.08 | 196 | 1411 | 11.1 | 15602 | 1.079 | 0.890 | 0.065 | 0.83 | 17.89 22q11.21 | 1.21 | 2.9 | .. | | |
| 31 | 22 B | 208.18 | 207.99 | 0.19 | 208 | 1205 | 11.6 | 13922 | 1.013 | 1.109 | 0.061 | 1.11 | 18.09 22q11.21 | 0.91 | -1.6 | .. | | |
| 54 | 22 D | 371.60 | 371.47 | 0.13 | 373 | 877 | 14.7 | 12930 | 1.058 | 0.920 | 0.053 | 1.06 | 19.57 22q11.21 | 1.15 | 2.6 | .. | | |
| 22q11.21 (DiGeorge) | | | | | Mean | 1164 | 12.5 | 14151 | 1.050 | 0.973 | 0.060 | 1.00 | (CV: 0.15) | 1.08 | | | | |
| 38 | 22 B | 252.23 | 251.96 | 0.27 | 253 | 842 | 11.9 | 10051 | 0.731 | 1.321 | 0.100 | 1.05 | 49.49 22q13.33-## | 0.55 | -5.9 * | | | |
| 55 | 22 D | 382.23 | 382.09 | 0.14 | 382 | 842 | 14.4 | 12110 | 0.991 | 1.442 | 0.120 | 0.95 | 49.50 22q13.33-# | 0.69 | -3.8 | | | |
| 22q13 (Phelan-McDermid) | | | | | Mean | 842 | 13.2 | 11081 | 0.861 | 1.382 | 0.110 | 1.00 | (CV: 0.15) | 0.62 | | | | |
| 44 | X C | 295.69 | 295.46 | 0.23 | 297 | 902 | 12.5 | 11252 | 0.754 | 0.751 | 0.048 | 1.00 | 32.29 Xp21.2 | 1.00 | 0.1 | .. | | |
| Chromosome X control probe | | | | | Mean | 902 | 12.5 | 11252 | 0.754 | 0.751 | 0.048 | 1.00 | (CV:) | 1.00 | | | | |
| 30 | X B | 199.92 | 199.64 | 0.28 | 202 | 602 | 9.8 | 5926 | 0.431 | 0.457 | 0.047 | 0.99 | 153.02 Xq28 | 0.94 | -0.6 | .. | | |
| 20 | X A | 147.14 | 146.82 | 0.32 | 148 | 633 | 9.1 | 5787 | 0.400 | 0.538 | 0.065 | 0.84 | 152.95 Xq28 | 0.74 | -2.1 | | | |
| 27 | X A | 183.29 | 183.16 | 0.13 | 184 | 529 | 10.6 | 5594 | 0.387 | 0.678 | 0.060 | 1.16 | 152.94 Xq28-# | 0.57 | -4.9 * | | | |
| Xq28 (RETT / MECP2) | | | | | Mean | 588 | 9.9 | 5769 | 0.406 | 0.558 | 0.057 | 1.00 | (CV: 0.26) | 0.74 | | | | |
| Mean values | | | 0.20 | | | 1104 | 12.6 | 13639 | 0.988 | 1.000 | 0.060 | | The small 2q14 area indicates poor hybridization! | 1.00 | Total of all except | | | |
| Standard deviations | | | 0.09 | | | (Coef. of variance: 0.276) | | | 0.261 | 0.224 | | | | 0.19 | Ctrl and '?' peaks | | | |

Quality assessment

| | Quality limits | Quality |
|---|----------------|----------------------|
| Not evaluated as < 2 Q-fragments | >1.00 (1.75) | |
| Mean CpG-area / mean A-group area | >0.55 (0.85) | 0.38 Too low! |
| Mean height of first probes AB | > 450 (800) | 1239 |
| Mean height of last probes CD | > 280 (500) | 974 |
| Ratio of mean heights AB/CD ('slope') | <2.00 (1.55) | 1.27 |
| Mean group CV of weighted ratio | <0.20 (0.15) | 0.16 high |
| 7 unidentified peak areas / 54 peak areas | < (0.02) | 0.01 |

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: Xq28-# 22q13.33-## 7q11.23 17p11.2-##

Poor Quality!

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 201 (14 peaks). CV of ROX heights for peaks above 100 nt is: 0.09

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