

| Sample | Reference sample | QF-PCR/<br>karyotype | [pmol/l] | Sequencing results |       |            |                     |            |                    |                            |                    |
|--------|------------------|----------------------|----------|--------------------|-------|------------|---------------------|------------|--------------------|----------------------------|--------------------|
|        |                  |                      |          | Reads              | %chip | NRA TMAP   | % reads mapped TMAP | NRA HPG    | % reads mapped HPG | TR+FGR TMAP                | TR+FGR HPG         |
| 10_5   | CVS              | 47,XX+18             | 193.8    | 7,021,412          | 9.16  | 6,672,572  | 95.03               | 5,937,822  | 84.57              | Incorrectly identified T21 | Trisomy 18         |
| 15_3   | AF               | 46,XY                | 27.1     | 5,538,611          | 6.50  | 5,286,437  | 95.45               | 4,670,419  | 84.32              | False Positive T18         | Euploid            |
| 18_33  | AF               | 46,XY                | 7.97     | 2,774,019          | 9.52  | 2,595,204  | 92.47               | 2,084,577  | 75.15              | Euploid                    | False Positive T21 |
| 19_4   | AF               | 46,XX                | 32.4     | 5,596,764          | 7.49  | 5,417,211  | 96.79               | 4,778,337  | 85.38              | False Positive T18         | Euploid            |
| 20_22  | AF               | 46,XY                | 50.8     | 11,323,073         | 14.51 | 11,020,461 | 97.33               | 9,531,601  | 84.18              | False Positive T18         | False Positive T21 |
| 21_31  | CVS              | 46,XX                | 10.87    | 15,562,358         | 20.94 | 15,231,058 | 97.87               | 13,212,672 | 84.90              | False Positive T18         | Euploid            |
| 22_20  | AF               | 46,XY                | 5.59     | 9,509,391          | 11.95 | 9,186,265  | 96.60               | 8,192,557  | 86.15              | Euploid                    | False Positive T18 |
| 25_25  | AF               | 46,XY                | 8.47     | 11,788,291         | 14.85 | 11,436,216 | 97.01               | 10,151,560 | 86.12              | Euploid                    | False Positive T13 |
| 26_30  | AF               | 46,XY                | 28.8     | 1,089,537          | 1.43  | 1,050,484  | 96.42               | 923,187    | 84.73              | False Positive T18         | Euploid            |
| 3_4    | PH               | Euploid              | 651.9    | 6,457,932          | 7.63  | 6,077,578  | 94.11               | 5,119,264  | 79.27              | False Positive T18         | Euploid            |
| 3_6    | AF               | 46, XY               | 6061.1   | 4,478,886          | 5.27  | 4,202,795  | 93.84               | 3,579,404  | 79.92              | False Positive T18         | False Positive T18 |
| 3_7    | AF               | 46, XY               | 208.7    | 8,242,646          | 9.67  | 7,709,927  | 93.54               | 6,570,885  | 79.72              | False Positive T18         | Euploid            |
| 3_9    | CVS              | 47,XY+21             | 7477.7   | 3,307,346          | 4.60  | 3,032,347  | 91.69               | 2,491,380  | 75.33              | Incorrectly identified T18 | Trisomy 21         |
| 4_2    | CVS              | 46,XY                | 297.2    | 4,809,940          | 8.43  | 4,595,986  | 95.55               | 3,899,508  | 81.07              | False Positive T18         | Euploid            |
| 6_7    | CVS              | 46,XY                | 594.6    | 2,131,689          | 2.80  | 2,066,603  | 96.95               | 1,821,440  | 85.45              | False Positive T18         | Euploid            |
| 7_5    | AF               | 46,XY                | 1209     | 7,322,010          | 9.44  | 6,982,254  | 95.36               | 6,243,977  | 85.28              | False Positive T18         | Euploid            |
| 9_11   | CVS              | 47,XY+18             | 432.1    | 8,557,813          | 9.31  | 8,125,563  | 94.95               | 7,244,362  | 84.65              | False Negative T18         | Trisomy 18         |

CVS: Chorionic villus sampling

AF: amniotic fluid

PH: phenotypic features in new born

NRA: number of reads aligned

[pmol/l]: library concentration

| Sample | Resequencing results |       |            |                     |            |                    |                            |                    |
|--------|----------------------|-------|------------|---------------------|------------|--------------------|----------------------------|--------------------|
|        | Reads                | %chip | NRA TMAP   | % reads mapped TMAP | NRA HPG    | % reads mapped HPG | TR+FGR TMAP                | TR+FGR HPG         |
| 10_5   | 20,423,254           | 26.79 | 19,897,179 | 97.42               | 17,333,555 | 84.87              | Trisomy 18                 | Trisomy 18         |
| 15_3   | 13,487,649           | 17.66 | 13,118,185 | 97.26               | 11,478,468 | 85.10              | False Positive T21         | Euploid            |
| 18_33  | 8,318,533            | 10.85 | 8,111,860  | 97.52               | 7,015,960  | 84.34              | Euploid                    | False Positive T21 |
| 19_4   | 14,479,851           | 18.99 | 14,055,006 | 97.07               | 12,243,590 | 84.56              | False Positive T18         | Euploid            |
| 20_22  | 18,788,320           | 24.75 | 18,225,235 | 97.00               | 15,803,561 | 84.11              | False Positive T21         | False Positive T21 |
| 21_31  | 15,983,445           | 21.10 | 15,598,679 | 97.59               | 13,569,724 | 84.90              | Euploid                    | Euploid            |
| 22_20  | -                    | -     | -          | -                   | -          | -                  | -                          | -                  |
| 25_25  | 15,246,382           | 19.88 | 14,845,806 | 97.37               | 13,092,597 | 85.87              | Euploid                    | Euploid            |
| 26_30  | -                    | -     | -          | -                   | -          | -                  | -                          | -                  |
| 3_4    | 12,302,992           | 14.70 | 11,714,013 | 95.21               | 10,189,946 | 82.82              | Euploid                    | Euploid            |
| 3_6    | 17,201,588           | 28.39 | 16,284,267 | 94.67               | 14,483,653 | 84.20              | False Positive T18         | Euploid            |
| 3_7    | 12,352,404           | 15.58 | 11,757,551 | 95.18               | 10,463,484 | 84.71              | Euploid                    | Euploid            |
| 3_9    | 4,304,845            | 5.09  | 4,056,035  | 94.22               | 3,432,628  | 79.74              | Incorrectly identified T18 | Trisomy 21         |
| 4_2    | 15,376,969           | 19.37 | 14,611,825 | 95.02               | 13,142,061 | 85.47              | False Positive T21         | Euploid            |
| 6_7    | 6,557,563            | 8.74  | 6,355,032  | 96.91               | 5,599,505  | 85.39              | False Positive T18         | Euploid            |
| 7_5    | 19,365,845           | 13.18 | 18,478,540 | 95.42               | 15,908,609 | 82.15              | False Positive T18         | Euploid            |
| 9_11   | 15,573,621           | 25.70 | 14,737,517 | 94.63               | 13,078,644 | 83.98              | Trisomy 18                 | Trisomy 18         |

| Sample | Main indication             | Risk of T21 | Risk of T18 | Maternal age | MBI (kg/m2) | Weigh (Kg) | Current smoker | Maternal diabetes mellitus | Racial origin | Gestational age (weeks) |
|--------|-----------------------------|-------------|-------------|--------------|-------------|------------|----------------|----------------------------|---------------|-------------------------|
| 10_5   | Abnormal ultrasound scan    | 1:50        | 1:50        | 34           | 19.6        | 52         | No             | No                         | Caucasian     | 11                      |
| 15_3   | Positive combined screening | 1:23        | <1:10000    | 34           | 21.6        | 52         | No             | No                         | Caucasian     | 12                      |
| 18_33  | Advanced maternal age       | -           | -           | 40           | 34.4        | 88         | No             | Sí                         | Caucasian     | 15                      |
| 19_4   | Positive combined screening | 1:161       | <1:10000    | 36           | 30.0        | 72         | No             | No                         | Caucasian     | 17                      |
| 20_22  | Other                       | 1:317       | <1:10000    | 40           | 24.6        | 54         | No             | No                         | Caucasian     | 18                      |
| 21_31  | Positive combined screening | 1:16        | <1:10000    | 35           | 17.8        | 55         | No             | No                         | Caucasian     | 13                      |
| 22_20  | Other: maternal anxiety     | 1:3499      | <1:10000    | 38           | 20.5        | 48         | No             | No                         | Caucasian     | 15                      |
| 25_25  | Positive combined screening | 1:150       | <1:10000    | 38           | 24.8        | 65         | No             | No                         | Caucasian     | 15                      |
| 26_30  | Positive family history     | 1:868       | <1:10000    | 34           | 24.0        | 60         | No             | No                         | South America | 14                      |
| 3_4    | Anti-D isoimmunization      | <1:10000    | <1:10000    | 29           | 26.3        | 64         | No             | No                         | South America | 19                      |
| 3_6    | Advanced maternal age       | 1:1582      | <1:10000    | 41           | 22.5        | 57         | No             | No                         | Caucasian     | 15                      |
| 3_7    | Positive combined screening | 1:618       | 1:62        | 36           | 26.1        | 66         | No             | No                         | Caucasian     | 16                      |
| 3_9    | Positive combined screening | 1:45        | 1:41        | 39           | 32.0        | 73         | No             | No                         | Caucasian     | 12                      |
| 4_2    | Positive combined screening | 1:116       | <1:10000    | 26           | 27.1        | 66         | No             | No                         | Caucasian     | 12                      |
| 6_7    | Positive combined screening | 1:5         | <1:10000    | 42           | 26.1        | 60         | No             | No                         | Caucasian     | 13                      |
| 7_5    | Positive family history     | -           | -           | 30           | 23.4        | 60         | No             | No                         | -             | 16                      |
| 9_11   | Abnormal ultrasound scan    | 1:6         | 1:3         | 40           | 28.1        | 72         | No             | No                         | Caucasian     | 12                      |