ESHRE PGD consortium data collection X: cycles from January to December 2007 with pregnancy follow-up to October 2008†

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ABSTRACT: The 10th report of the European Society of Human Reproduction and Embryology (ESHRE) PGD Consortium is presented, documenting cycles collected for the calendar year 2007 and follow-up of the pregnancies and babies born until October 2008 which resulted from these cycles. Since the beginning of the data collections there has been a steady increase in the number of cycles, pregnancies and babies reported annually. For data collection X, 57 centres participated, reporting on 5887 cycles to oocyte retrieval (OR), along with details of the follow-up on 1516 pregnancies and 1206 babies born. A total of 729 OR were reported for chromosomal abnormalities, 110 OR for sexing for X-linked diseases, 1203 OR for monogenic diseases, 3753 OR for preimplantation genetic screening and 92 OR for social sexing. Data X is compared with the cumulative data for data collections I–IX.

Key words: PGD / preimplantation genetic screening / fluorescence in situ hybridization / PCR / ESHRE PGD Consortium

Introduction

The European Society of Human Reproduction and Embryology (ESHRE) PGD Consortium was established in 1997. Since 1999, nine data collections of PGD for autosomal and sex-linked monogenic diseases and chromosome abnormalities, preimplantation genetic screening (PGS) and social sex selection have been published (ESHRE PGD Consortium Steering Committee, 1999, 2000, 2002; Sermon et al., 2005; Harper et al., 2006; 2008a,b; Sermon et al., 2007; Goossens et al., 2008, 2009). This report summarizes data X collected for the calendar year 2007 and the subsequent pregnancies. As it was reported for the first time in data VIII, data X also includes the delivery rate for each indication.

Materials and Methods

Data were collected using a FileMaker Pro 5, 6 or 8 database, consisting of files for cycle, pregnancy and baby records. The submitted data were thoroughly analyzed to identify omissions and any ambivalent data. Corrections were requested from the participating centres. Records with insufficient data, e.g. with no cycle or patient identification, no clear indication or from the wrong time period were excluded from the calculations. In-depth corrections and tables were made by expert co-authors. Clinical pregnancies were defined as the presence of one or more fetal hearts at ~6 weeks gestation. Implantation rate was defined as the number of fetal hearts per 100 embryos transferred. Delivery rate was defined as the percentage of pregnancies with delivery per oocyte retrieval (OR) and per embryo transfer procedure.

Results

The number of centres that become members of the PGD Consortium increases annually. Data from 57 centres were included in this report. The results are represented in tables according to an established lay-out. The accompanying text is deliberately concise and seven tables are available in an electronic version only: Supplementary
### Table Ia Overall cycle data collection I–IX.

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<td>21/29</td>
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OR, oocyte retrieval; AT, acid Tyrode’s; COC, cumulus-oocyte complexes; SS, social sexing; PGS, preimplantation genetic screening; FISH, fluorescence in situ hybridization; ET, embryo transfer; ART, assisted reproduction technology; PB, polar body.

The PGD column includes PGD for chromosome abnormalities, sexing for X-linked disease and PGD for monogenic disorders.

1Includes two cycles with PGD on frozen embryos only. These cycles were not counted in the cycles with OR.

2Twelve cycles had PB biopsy and cleavage stage biopsy.
### Table Ib  Overall cycle data collection X.

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FHB, fetal heartbeats; FU, follow-up.

PGD column includes PGD for chromosome abnormalities, sexing for X-linked disease and PGD for monogenic disorders.

*% per number of clinical pregnancies minus the number of pregnancies that were lost to FU.
### Table IIa PGD for chromosomal abnormalities, data collection I–IX.

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<td>182</td>
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<td>47</td>
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Table SIIc lists the abnormal karyotypes carried by the patients undergoing PGD, Supplementary Table SIIIc lists the X-linked diseases for which sexing was carried out, Supplementary Table SIVc lists the monogenic diseases for which PGD was carried out, Supplementary Tables SVIIa (data I–IX) and SVIIIb (data X) list the complications of pregnancy and Supplementary Tables SXIIa (data I–IX) and SXIIb (data X) list the congenital malformations and the neonatal complications.

An overview of all cycles collected previously in data collections I–IX can be found in Table Ia, while an overview of the current data collection can be found in Table Ib.

For all indications for PGD/PGS (data I–IX and X), ICSI was the most often used method of fertilization and cleavage stage aspiration was the most commonly used method of biopsy. Overall zona pellucida drilling was more commonly performed using a laser.

### PGD cycles for chromosomal abnormalities

Tables IIa and IIb summarize the 3524 and 729 cycles to OR collected for data collection I–IX and X, respectively. As for previous years, data X showed that PGD for reciprocal translocations was performed more often than for Robertsonian translocations or other types of chromosome abnormalities. For data X 9045 oocytes were collected, 69% (5325/7727) fertilized, 74% (3947/5325) embryos were biopsied and 99% (3902/3947) embryos were successfully biopsied. Of the embryos successfully biopsied 94% (3652/3902) gave a diagnostic result, of which only 26% (938/3652) were transferable. From 729 OR procedures only 62% (450/729) resulted in an embryo transfer procedure. This is in agreement with previous data showing that a high level of chromosomally abnormal embryos is found in these patients. A positive hCG was obtained in 184 cycles, with a positive heart beat in 152 cycles [21% per OR (152/729) and 34% per embryo transfer (152/450)]. This gave an implantation rate of 26% (176/681). Finally, the delivery rate was 16% per OR (18/110) and 27% per embryo transfer (18/66). There were 18/138 miscarriages (13% per clinical pregnancy) and 9% (2/22) pregnancies were lost to follow-up.

### PGD cycles for sexing for X-linked diseases

Tables IIIa and IIIb summarize the 1057 and 110 cycles to OR collected for data collection I–IX and X, respectively. This year, again, fluorescence in situ hybridization (FISH) was the only method used for sexing cycles. For data X, 1485 oocytes were collected, 70% (866/1238) fertilized, 79% (685/866) embryos were biopsied and 99% (681/685) were successfully biopsied. Of the embryos successfully biopsied 94% (638/681) gave a diagnostic result, of which only 37% (236/638) were transferable (female). From 110 OR procedures only 78% (86/110) resulted in an embryo transfer procedure. A positive hCG was obtained in 25 cycles, with a positive heart beat in 22 cycles [20% per OR (22/110) and 26% per embryo transfer (22/86)]. This gave an implantation rate of 19% (27/141). Finally, the delivery rate was 16% per OR (18/110) and 21% per embryo transfer (18/86). There were 2/20 miscarriages (10% per clinical pregnancy) and 9% (2/22) pregnancies were lost to follow-up.
Table IIb  PGD for chromosomal abnormalities, data collection X.

<table>
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<tr>
<th>Indication</th>
<th>Robertsonian translocation, male carrier</th>
<th>Robertsonian translocation, female carrier</th>
<th>Reciprocal translocation, male carrier</th>
<th>Reciprocal translocation, female carrier</th>
<th>Sex chromosome aneuploidy</th>
<th>Others</th>
<th>Total</th>
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<td>211</td>
<td>229</td>
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<td>729</td>
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<td>113</td>
<td>32</td>
<td>24</td>
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<td>148</td>
<td>150</td>
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<td>534</td>
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<td>9</td>
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<td>3</td>
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<td>22/33</td>
<td>16/28</td>
<td>19/34</td>
<td>29/45</td>
<td>18/23</td>
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<td>19/29</td>
<td>13/22</td>
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<td>22/34</td>
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<td>(% per clinical pregnancy – pregnancy lost to FU)</td>
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PGD for monogenic diseases

Tables IVa and IVb summarize the 3530 and 1203 cycles to OR collected for data collection I–IX and X, respectively. The most common indications for PGD for autosomal recessive diseases were β-thalassemia and/or sickle cell syndromes (135 cycles), plus 115 cycles for β-thalassemia/sickle cell with HLA typing, cystic fibrosis (CF) (107 cycles, including 4 cycles for CF and a second indication) and spinal muscular atrophy (SMA) (51 cycles, of which 1 was for SMA and a second indication). Amongst the autosomal dominant...
Table IVa  Cycles performed for single gene disorders, data collection I–IX.

<table>
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<tr>
<th>Indication</th>
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<th>Others(^8)</th>
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<td>SMA and SMA + Retinitis pigmentosa(^3)</td>
<td>HLA compatibility HLA + specific disease</td>
<td>DM1(^4)</td>
<td>HD and HD exclusion</td>
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<td>47</td>
<td>491</td>
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<td>481</td>
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<td>Cancelled after IVF/ICSI</td>
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<td>13(^10)</td>
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<td>195</td>
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<td>3</td>
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<td>25/31</td>
<td>54/27</td>
<td>4/9</td>
<td>17/23</td>
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- Frozen: cystic fibrosis (various mutations); b-thal, b-thalassaemia; SMA, spinal muscular atrophy; SC, sickle cell anaemia; DM1, myotonic dystrophy type 1; HD, Huntington’s disease; FRAXA, fragile X syndrome; DMD, Duchenne muscular dystrophy (specific); BMD, Becker muscular dystrophy; Haem, haemophilia.
- CF, cystic fibrosis; b-thal, b-thalassaemia; SMA, spinal muscular atrophy; SC, sickle cell anaemia; DM1, myotonic dystrophy type 1; HD, Huntington’s disease; FRAXA, fragile X syndrome; DMD, Duchenne muscular dystrophy (specific); BMD, Becker muscular dystrophy; Haem, haemophilia.

1Five cycles for two indications: CF and FRAXA; CF and SS, CF + PGS for diabetes insipidus, CF + diabetes insipidus (sexing) and CF + PGS.
2Includes two cycles performed also with FISH for a Robertsonian translocation.
3Includes three cycles for SMA and PGS, and 5 cycles performed also for retinitis pigmentosa.
4Includes one cycle also for DMD.
5Includes one cycle for BMD and PGS.
6Includes three cycles for FRAXA testing and PGS.
7Includes one cycle for Haem A and PGS.
8Includes one cycle for Tuberous Sclerosis and PGS + 3 cycles using FISH for a microdeletion.
9Two cycles were on frozen-thawed embryos only so they were not counted as cycles with an OR, but were counted as cycles going to PGD.
10Eleven cycles had both PB biopsy and cleavage stage biopsy.

### Table IVb Cycles performed for single gene disorders, data collection X.

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<th>Indication</th>
<th>Beta-Thal and/or SC (+ HLA)</th>
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<th>Specific sex-linked</th>
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<td>292</td>
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<td>25</td>
<td>17</td>
<td>8 (8)</td>
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<td>Clinical pregnancy rate (%)</td>
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<td>23/26</td>
<td>33/37</td>
<td>22/38 (28/44)</td>
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<tr>
<td>Implantation rate (% fetal hearts/embryos transferred)</td>
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<td>17</td>
<td>23</td>
<td>27 (32)</td>
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<td>Deliveries</td>
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<td>Delivery rate (% per OR/% per ET)</td>
<td>26/31 (19/27)</td>
<td>19/21</td>
<td>32/35</td>
<td>22/38 (17/28)</td>
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</table>
diseases, the most PGD cycles were performed for Huntington disease (HD) (106 cycles, including 5 cycles for HD and a second indication) and myotonic dystrophy type I (DM1) (95 cycles), neurofibromatosis (25 cycles) and Charcot Marie-Tooth (17 cycles). For a specific diagnosis of X-linked diseases the most common indications were for fragile X syndrome (FRAXA) (77 cycles, of which 1 was for FRAXA and a second indication), Duchenne and Becker muscular dystrophy (DMD/BMD) (24 cycles and 2 cycles, respectively) and haemophilia A and B (15 cycles). PGD cycles for an additional 115 monogenic diseases were initiated in 369 cycles (included under ‘others’ in Table IVb) and they are listed in Supplementary Table SIVc. Besides the 115 cycles for β-thalassemia and/or sickle cell syndromes with HLA typing, there were 36 cycles for HLA compatibility typing plus a further 29 cycles for HLA typing along with a specific disorder. The most common indications here were Fanconi anaemia, Gaucher disease, adrenoleukodystrophy and osteopetrosis.

For data X, 16 005 oocytes were collected and 76% (9943/13 056) fertilized. ICSI was used in 1185 cycles (of which 15 were subsequently frozen) and IVF in 3 cycles. A total of 76% (7568/9943) of the embryos were biopsied and 99% (7495/7568) were successfully biopsied. Of the embryos successfully biopsied, 90% (6725/7495) gave a diagnostic result, of which 42% (2799/6725) were transferable.

From 1203 OR procedures 79% (952/1203) resulted in an embryo transfer procedure. A positive hCG was obtained in 374 cycles, with a positive heart beat in 298 cycles [25% per OR (298/1203) and 31% per embryo transfer (298/952)] and 366 fetal hearts, giving an overall implantation rate of 22% (366/1660). These pregnancy rates were notably higher than in the previous data collections. Finally, the delivery rate was 21% per OR (253/1203) and 27% per embryo transfer (253/952). There were 37/290 miscarriages (13% per clinical pregnancy) and 3% (8/298) clinical pregnancies were lost to follow-up.

Overall, the number of PGD cycles performed for monogenic disorders between January and December 2007 further increased compared with data collection IX. This increase is primarily a result of a marked increase in the cycles for β-thalassemia and/or sickle cell syndromes (with HLA typing) (245 cycles in data X versus 110 cycles in data IX) and an increase in cycles for less frequent monogenic disorders (column ‘others’ in Table IVb). Overall, there were no marked changes with respect to the progress and outcome of cycles, including the embryology, rates of diagnosis and clinical outcome, such as clinical pregnancy and embryo implantation rates (Goossens et al., 2009).

Preimplantation genetic screening

Tables Va and Vb summarize the 13 053 and 3753 cycles to OR reported for data collection I–IX and X, respectively. For data X, 40 656 oocytes were collected, 72% (23 713/33 129) fertilized, 80% (18 964/23 713) embryos were biopsied and 99% (18 750/18 964) were successfully biopsied. Of the embryos successfully biopsied, 93% (17 415/18 750) gave a diagnostic result, of which only 34% (5898/17 415) were transferable. From 3753 OR procedures only 70% (2638/3753) resulted in an embryo transfer procedure. A positive hCG was obtained in 940 cycles, with a positive heart beat in 781.
### Table Va Cycles performed for PGS, data collection I–IX.

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<th>AMA + miscarriage(^1)</th>
<th>AMA + RIF (^1)</th>
<th>Recurrent miscarriage</th>
<th>Recurrent IVF failure</th>
<th>SMF(^2)</th>
<th>Oocyte donation(^1)</th>
<th>Prev abn preg(^3)</th>
<th>No indication</th>
<th>Others(^4)</th>
<th>Total</th>
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<td>1063</td>
<td>1696</td>
<td>3380</td>
<td>1164</td>
<td>67</td>
<td>25</td>
<td>357</td>
<td>752</td>
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<td>47</td>
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<td>3063</td>
<td>7561</td>
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<td>36 878</td>
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Biopsied 18 910 1984 5776 9884 20 818 7141 436 136 1950 4405 71 440
Successfully biopsied 18 640 1981 5725 9760 20 575 7105 435 133 1906 4363 70 623
Diagnosed 17 042 1851 5371 89357 19 2597 6637 433 129 1637 7 38877 65 1817
Transferable 5050 550 1870 32497 72677 2675 204 51 8117 16537 23 3807
Transferred 4394 408 1397 23407 48737 1824 106 26 4907 11177 16 9757
Frozen 553 63 171 468 1026 342 73 11 100 358 3165
Clinical outcome
Cycles to ET 2577 248 814 1282 2614 953 61 16 277 591 9433
hCG positive 749 56 172 495 887 401 37 5 109 234 3145
Positive heartbeat 557 38 140 384 685 328 30 5 93 173 2433
Clinical pregnancy rate (% per OR/% per ET)

AMA, advanced maternal age; RIF, repeated implantation failure; SMF, severe male factor.

1These data were not extracted from I to IV.
2These data were not extracted from I to III.
3These data were not extracted from data I–VIII.
4‘Others’ contains also cycles with multiple indications and previous abnormal (prev abn) pregnancies (data I–VIII).
5Several cycles had incomplete results.
6One cycle had cleavage stage biopsy and PB biopsy.
7Several cycles from one centre had no information on the number of embryos diagnosed as transferable, but patients did have embryos transferred. In these cases, undiagnosed/failed or abnormal embryos were transferred.

### Table Vb Cycles performed for PGS, data collection X.

<table>
<thead>
<tr>
<th>Indication</th>
<th>AMA</th>
<th>RIF</th>
<th>Recurrent miscarriage</th>
<th>AMA + RIF</th>
<th>AMA + miscarriage</th>
<th>SMF</th>
<th>AMA + SMF</th>
<th>RIF + SMF</th>
<th>Miscarriage + SMF</th>
<th>Oocyte donation</th>
<th>Prev abn preg</th>
<th>No indication</th>
<th>Others</th>
<th>Total</th>
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<td>410</td>
<td>147</td>
<td>334</td>
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<td>23</td>
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<td>381</td>
<td>70</td>
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<td>56</td>
<td>9</td>
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<td>2726</td>
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<td>Female age (years)</td>
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<td>36</td>
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<td>41</td>
<td>34</td>
<td>40</td>
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<td>41</td>
<td>37</td>
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<td>112</td>
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<td>75</td>
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<td>3261</td>
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<td>IVF + ICSI</td>
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<td>13</td>
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<td>9</td>
<td>0</td>
<td>1</td>
<td>1</td>
<td>0</td>
<td>1</td>
<td>0</td>
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<td>16</td>
<td>99</td>
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<td>IVF + Frozen embryos</td>
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<td>0</td>
<td>0</td>
<td>1</td>
<td>0</td>
<td>0</td>
<td>0</td>
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<td>0</td>
<td>0</td>
<td>0</td>
<td>0</td>
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<tr>
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<td>4</td>
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<td>2</td>
<td>3</td>
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<td>409</td>
<td>146</td>
<td>330</td>
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<td>5</td>
<td>88</td>
<td>23</td>
<td>105</td>
<td>195</td>
<td>3733</td>
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<td>Zona breaching</td>
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<td>77</td>
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<td>16</td>
<td>76</td>
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Continued
Table Vb  Continued

<table>
<thead>
<tr>
<th>Indication</th>
<th>AMA</th>
<th>RIF</th>
<th>Recurrent miscarriage</th>
<th>AMA + RIF</th>
<th>AMA + miscarriage</th>
<th>SMF</th>
<th>AMA + SMF</th>
<th>RIF + SMF</th>
<th>Miscarriage + SMF</th>
<th>Oocyte donation</th>
<th>Prev abn preg</th>
<th>No indication</th>
<th>Others</th>
<th>Total</th>
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<td>169</td>
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<td>265</td>
<td>37</td>
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<td>102</td>
<td>308</td>
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<td>1</td>
<td>0</td>
<td>2</td>
<td>129</td>
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</tbody>
</table>

**Embryology**

| COCs                        | 11516| 8676| 4893                   | 3587      | 1446              | 4719| 393       | 514       | 59                | 1115           | 248          | 1224         | 2266   | 40656 |
| Inseminated                 | 9576 | 6963| 4019                   | 2766      | 1164              | 3739| 322       | 445       | 47                | 941            | 192          | 1025         | 1930   | 33129 |
| Fertilized                  | 6725 | 5082| 2957                   | 1975      | 833               | 2637| 204       | 312       | 37                | 713            | 139          | 714          | 1385   | 23713 |
| Biopsed                     | 5072 | 4245| 2223                   | 1998      | 745               | 1923| 173       | 270       | 29                | 536            | 110          | 556          | 1084   | 18964 |
| Successfully biopsed        | 5036 | 4150| 2206                   | 1975      | 738               | 1911| 173       | 267       | 29                | 532            | 108          | 554          | 1071   | 18750 |
| Diagnosed                   | 4742 | 3832| 2073                   | 1734      | 705               | 1807| 152       | 238       | 29                | 519            | 101          | 508          | 975    | 17415 |
| Transferable                | 1267 | 1393| 711                    | 655       | 192               | 673 | 47        | 98        | 9                 | 231            | 35           | 201          | 386    | 5898  |
| Transferred\(^1\)           | 1068 | 1047| 507                    | 594       | 162               | 461 | 46        | 76        | 8                 | 138            | 28           | 140          | 293    | 4568  |
| Frozen                      | 122  | 170 | 108                    | 68        | 17                | 69  | 1         | 18        | 1                 | 66             | 9            | 35           | 35     | 719   |

**Clinical outcome**

| Cycles to ET                | 683  | 572 | 297                    | 317       | 91                | 271 | 29        | 37        | 5                 | 76             | 17           | 84           | 159    | 2638  |
| hCG positive                | 199  | 224 | 132                    | 71        | 20                | 110 | 6         | 21        | 3                 | 46             | 6            | 33           | 69     | 940   |
| Positive heartbeat          | 160  | 175 | 110                    | 65        | 15                | 98  | 6         | 21        | 4                 | 36             | 6            | 27           | 58     | 781   |
| Clinical pregnancy rate (% per OR/\% per ET) | 13/23 | 24/31 | 27/37                    | 16/20     | 10/16              | 29/36| 15/21     | 52/57     | 80/80              | 41/47          | 26/35        | 60/36         | 30/36  | 21/30  |
| Number of fetal hearts      | 190  | 213 | 142                    | 79        | 19                | 129 | 6         | 30        | 3                 | 49             | 10           | 31           | 70     | 971   |
| Implantation rate (fetal hearts/100 embryos transferred) | 18   | 20  | 28                     | 13        | 12                | 28  | 13        | 40        | 37                | 35             | 36           | 22           | 24     | 21    |
| Deliveries                 | 118  | 134 | 90                     | 37        | 7                 | 80  | 4         | 13        | 2                 | 31             | 6            | 17           | 44     | 586   |
| Delivery rate (% per OR/\% per ET) | 9/17 | 19/23 | 22/30                    | 9/12      | 7/11               | 24/30| 10/14     | 32/35     | 40/40              | 35/41          | 26/35        | 16/20         | 23/28  | 16/22  |
| Miscarriages               | 29   | 10  | 8                      | 9         | 4                 | 13  | 2         | 4         | 2                 | 2              | 0            | 4            | 6      | 93    |
| Miscarriage rate (% per clinical pregnancy – pregnancy lost to FU) | 20   | 7   | 8                      | 20        | 29                | 14  | 33        | 23        | 50                | 6              | 0            | 19           | 12     | 14    |
| Clinical pregnancies lost to FU | 13   | 31  | 12                     | 19        | 1                 | 5   | 0         | 4         | 0                 | 3              | 0            | 6            | 8      | 102   |

\(^1\)Others’ contains also cycles with multiple indications.

\(^2\)Failed embryos were also transferred.
Table VIa PGD for social sexing, data collection I–IX.

<table>
<thead>
<tr>
<th>Method for sexing</th>
<th>FISH (SS only)</th>
<th>FISH (SS + AS)</th>
<th>PCR</th>
<th>Unknown</th>
<th>Total</th>
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<td>89</td>
<td>189</td>
<td>5²</td>
<td>579²</td>
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<td>5</td>
<td>16</td>
<td>1</td>
<td>47</td>
</tr>
<tr>
<td>Female age (years)</td>
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<td>39</td>
<td>37</td>
<td>35</td>
<td>36</td>
</tr>
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<td>ART method</td>
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</tr>
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<td>IVF</td>
<td>123</td>
<td>10</td>
<td>10</td>
<td>3</td>
<td>146</td>
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<td>ICSI</td>
<td>168</td>
<td>78</td>
<td>168</td>
<td>2</td>
<td>416</td>
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<td>Frozen</td>
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<td>2</td>
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<td>5</td>
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<td>Frozen + IVF + ICSI + unknown</td>
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<td>563</td>
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<td>Zona breaching</td>
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<td></td>
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</tr>
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<td>AT drilling</td>
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<td>10</td>
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<td>19</td>
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<td>Laser drilling</td>
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<td>4573</td>
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<td>34</td>
<td>86</td>
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<td>Clinical outcome</td>
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<td>60</td>
<td>138</td>
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<td>58</td>
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<td>161</td>
</tr>
<tr>
<td>Positive heartbeat</td>
<td>68</td>
<td>13</td>
<td>39</td>
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<td>120</td>
</tr>
<tr>
<td>Clinical pregnancy rate (% per OR/% per ET)</td>
<td>23/31</td>
<td>15/22</td>
<td>21/28</td>
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<td>21/29</td>
</tr>
</tbody>
</table>

AS, aneuploidy screening.
¹These data were not extracted from I to VII.
²One natural cycle included.
³Eleven cycles with embryos frozen without biopsy or failed diagnosis included.
⁴Three embryos frozen without biopsy were not included.
cycles [21% per OR (781/3753) and 30% per embryo transfer (781/2638)]. This gave an implantation rate of 21% (971/4568). These pregnancy rates were similar to the previous data collections. Finally, the delivery rate was 16% per OR (586/3753) and 22% per embryo transfer (586/2638). There were 93/679 miscarriages (14% per clinical pregnancy) and 13% (102/781) clinical pregnancies lost to follow-up.

The main indications were advanced maternal age (AMA) (1250 OR) and repeated implantation failure (RIF) (713 OR). There were still a number of cycles reported where no indication was given (105 OR). All indications involving AMA showed a somewhat lower pregnancy rate (between 10 and 16% per OR) in comparison with the other indications, although the pregnancy rates were higher than in previous data collections. Patients with severe male factor (SMF) showed a relatively high pregnancy rate [29% per OR (98/334)] as did patients where oocyte donation was performed [41% per OR (36/88)]. Patients with no indication had a pregnancy rate of 26% per OR (27/105).

From 3753 cycles, 387 involved the biopsy of only one embryo and 555 involved the biopsy of two embryos. As stated in data VII (Harper et al., 2008a,b), in the majority of cases these embryos should be replaced without biopsy.

There was only one cycle where PCR and FISH was used, in all other 3752 cycles to OR FISH was used.

In Table Vb, the column ‘others’ contains various indications such as mosaic embryos and single or double embryo transfer, as well as cycles with multiple indications.

The PGD Consortium recently published a position statement on the use of PGS (Harper et al., 2010a). All RCTs for PGS using FISH and mainly cleavage-stage biopsy show no improvements in success rates. The Consortium recommendation was that the use of arrays on either polar bodies or trophectoderm biopsies should be validated and appropriate RCTs performed. The ESHRE PGS Task Force has conducted a pilot into the feasibility of using arrays for polar body biopsy (Geraedts et al., 2010) and is in the process of setting up an RCT.

### PGD cycles for social sexing

Tables VIa and VIb summarize the 579 and 92 cycles to OR collected for data collection I–IX and X, respectively. For data X, 1377 oocytes were collected, 74% (866/1175) fertilized, 81% (703/866) embryos were biopsied and 98% (692/703) were successfully biopsied. Of the embryos successfully biopsied 82% (568/692) gave a diagnostic result, of which only 38% (213/568) were transferable (of the desired sex). From 92 OR procedures only 79% (73/92) resulted in an embryo transfer procedure. A positive hCG was obtained in 36 cycles, with a positive heart beat in 23 cycles [25% per OR (23/92) and 32% per embryo transfer (23/73)]. This gave an implantation rate of 23% (31/133). These pregnancy rates were similar to the previous data collections. Finally, the delivery rate was 20% per OR (18/92) and 25% per embryo transfer (18/73). There were 4/22 miscarriages (18% per clinical pregnancy) and 4% (1/23) was lost to follow-up.

### Pregnancies and babies

Tables VIIa, VIIb, IXa–Xlb, and Supplementary Tables SVIIIa, SVIIIb, SXIIa, SXIIb summarize the pregnancy and baby data. Data X was comparable to previous data collections. Data X included 1516 clinical pregnancies which resulted in 1609 fetal sacs (Table VIIb). There were 977 deliveries of 1206 babies. Of the 1291 cycles ending in a
### Table VIIa Evolution of pregnancy, data I–IX.

<table>
<thead>
<tr>
<th>Category</th>
<th>n pregnancies</th>
<th>n fetal sacs</th>
</tr>
</thead>
<tbody>
<tr>
<td>Pregnanies</td>
<td>4595</td>
<td>4874</td>
</tr>
<tr>
<td>FISH cycles</td>
<td>3688</td>
<td></td>
</tr>
<tr>
<td>PCR cycles</td>
<td>902</td>
<td></td>
</tr>
<tr>
<td>FISH + PCR</td>
<td>5</td>
<td></td>
</tr>
<tr>
<td>Subclinical pregnancies¹</td>
<td>699</td>
<td></td>
</tr>
<tr>
<td>Clinical pregnancies</td>
<td>3896</td>
<td>4874</td>
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<tr>
<td>Singletons</td>
<td>2822</td>
<td>2822</td>
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<tr>
<td>Twins</td>
<td>877</td>
<td>1754</td>
</tr>
<tr>
<td>Triplets</td>
<td>91</td>
<td>273</td>
</tr>
<tr>
<td>Quadruplet</td>
<td>6</td>
<td>24</td>
</tr>
<tr>
<td>Unknown</td>
<td>100</td>
<td>1³</td>
</tr>
<tr>
<td>Lost to FU during first trimester</td>
<td>42</td>
<td>43</td>
</tr>
<tr>
<td>First trimester loss</td>
<td>528</td>
<td>653</td>
</tr>
<tr>
<td>Miscarriage</td>
<td>417³</td>
<td>461</td>
</tr>
<tr>
<td>TOP</td>
<td>8⁴</td>
<td>9</td>
</tr>
<tr>
<td>Extra-uterine pregnancy</td>
<td>45⁵</td>
<td>35</td>
</tr>
<tr>
<td>Vanishing twins/triplets or miscarriage multiple</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Reduction of multiple pregnancies</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Quadruplet to twin</td>
<td>8</td>
<td></td>
</tr>
<tr>
<td>Triplet to twin</td>
<td>12</td>
<td></td>
</tr>
<tr>
<td>Triplet to singleton</td>
<td>8</td>
<td></td>
</tr>
<tr>
<td>Twin to singleton</td>
<td>7⁸</td>
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<tr>
<td>Unknown</td>
<td>58</td>
<td>3</td>
</tr>
<tr>
<td>Ongoing pregnancies &gt; 12 weeks</td>
<td>3326</td>
<td>4178</td>
</tr>
<tr>
<td>Second trimester loss</td>
<td>78</td>
<td>141</td>
</tr>
<tr>
<td>Miscarriage</td>
<td>57⁶</td>
<td>83</td>
</tr>
<tr>
<td>Miscarriage twin to singleton</td>
<td></td>
<td>3</td>
</tr>
<tr>
<td>TOP</td>
<td>20⁷</td>
<td>20</td>
</tr>
<tr>
<td>Twin to twin transfusion</td>
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<td>2</td>
</tr>
<tr>
<td>Reduction of multiple pregnancies</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Quadruplet to twin</td>
<td>4</td>
<td></td>
</tr>
<tr>
<td>Triplet to twin</td>
<td>11</td>
<td></td>
</tr>
<tr>
<td>Triplet to singleton</td>
<td>14</td>
<td></td>
</tr>
<tr>
<td>Twin to singleton</td>
<td>4</td>
<td></td>
</tr>
<tr>
<td>Lost to FU during second trimester</td>
<td>85⁹</td>
<td>108</td>
</tr>
<tr>
<td>Deliveries</td>
<td>3163</td>
<td>3929</td>
</tr>
<tr>
<td>Singletons</td>
<td>2424</td>
<td>2424</td>
</tr>
<tr>
<td>Twins</td>
<td>712</td>
<td>1424</td>
</tr>
<tr>
<td>Triplets</td>
<td>27</td>
<td>81</td>
</tr>
</tbody>
</table>

¹Subclinical pregnancy defined as pregnancy without any other clinical signs, but positive serum hCG.
²Number of FHBs not known for data I–VIII. Counted further as one fetal heart.
³One miscarriage after amniocentesis.
⁴TOP, termination of pregnancy. Two TOPs for aneuploidy, one TOP for social reasons, one TOP of twin with misdiagnosis for CMT disease 1a, one TOP for 47,XY + 13, one TOP for encephalocele and one TOP for 47,XY + 21.
⁵One heterotrophic gestation continued as singleton after reduction of extra-uterine gestation at 6 weeks.
⁶One triplet: fetal reduction, followed by amniocentesis and loss of remaining twin at 16 weeks (1 fetal sac counted in reduction, 2 in miscarriage, 1 s trimester pregnancy loss after miscarriage counted).
⁷TOP after misdiagnosis: one misdiagnosis for sexing, FISH, female fetus, indication SS; one misdiagnosis for 8-Thu, PCR; one misdiagnosis for MD, PCR; one misdiagnosis after PGS, karyotype 45,X; one misdiagnosis for a reciprocal translocation 46,XY,der(15)(13;15)(q25.1; q26.3); TOP after ultrasound (four): enlarged lateral ventricle, two singletons with cardiopathy, one singleton with tetralogy of Fallot. TOP after amniocentesis, not related to the PGD: trisomy 18, indication for PGD parent carrier of reciprocal translocation not involving chromosome 18; one polymalformation; one cystic hygroma, failed karyotype; one Turner mosaic, one spina bifida, one trisomy 21, one mosaic 46,XY/47,XY + 18 (misdiagnosis), one Hemivertebrae, hypoplastic cerebellum, hydrocephaly (46,XX), one abnormal chromosome 15, one polycystic kidney.
⁸One misdiagnosis for sexing, PCR, indication Duchenne, twin pregnancy, selective termination of male fetus. Cycle done in 1996, Y-specific amplification only.
⁹One misdiagnosis (47,XXX after PGS for RIF) lost to FU.
pregnancy with a positive heartbeat, follow-up data on 1271 pregnancies were reported. Of the 977 pregnancies reported to have ended with a delivery (total number of babies: 1206), neonatal data on 1206 babies were submitted. The delivery rates per indication are reported in Tables IIb, IIIb, IVb, Vb and VIb. Fifty per cent of the deliveries were by Caesarean section (489/977) (Table IXb). In 132 cases the method of delivery was not known.

Table VIIb  Evolution of pregnancy, data X.

<table>
<thead>
<tr>
<th>n pregnancies</th>
<th>n fetal sacs</th>
</tr>
</thead>
<tbody>
<tr>
<td>Preganancies</td>
<td>1516</td>
</tr>
<tr>
<td>FISH only cycles</td>
<td>1151</td>
</tr>
<tr>
<td>PCR only cycles</td>
<td>362</td>
</tr>
<tr>
<td>FISH + PCR</td>
<td>3</td>
</tr>
<tr>
<td>Subclinical pregnancies</td>
<td>225</td>
</tr>
<tr>
<td>Clinical pregnancies</td>
<td>1291</td>
</tr>
<tr>
<td>Singletons</td>
<td>973</td>
</tr>
<tr>
<td>Twins</td>
<td>270</td>
</tr>
<tr>
<td>Triplets</td>
<td>21</td>
</tr>
<tr>
<td>Quadruplet</td>
<td>2</td>
</tr>
<tr>
<td>Unknown</td>
<td>25</td>
</tr>
<tr>
<td>Lost to FU during first trimester</td>
<td>20</td>
</tr>
<tr>
<td>First trimester loss</td>
<td>211</td>
</tr>
<tr>
<td>Miscarriage</td>
<td>198</td>
</tr>
<tr>
<td>TOP</td>
<td>5</td>
</tr>
<tr>
<td>Extra-uterine pregnancy</td>
<td>8</td>
</tr>
<tr>
<td>Vanishing/miscarriage multiples</td>
<td></td>
</tr>
<tr>
<td>Twin to singleton</td>
<td>33</td>
</tr>
<tr>
<td>Triplet to twin or singleton</td>
<td>3</td>
</tr>
<tr>
<td>Quadruplet to twin</td>
<td>1</td>
</tr>
<tr>
<td>Reduction of multiple pregnancies</td>
<td>9</td>
</tr>
<tr>
<td>Quadruplet to twin</td>
<td>1</td>
</tr>
<tr>
<td>Triplet to twin</td>
<td>3</td>
</tr>
<tr>
<td>Triplet to singleton</td>
<td>2</td>
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<tr>
<td>Twin to singleton</td>
<td>3</td>
</tr>
<tr>
<td>Ongoing pregnancies (&gt;12 weeks)</td>
<td>1060</td>
</tr>
<tr>
<td>Second trimester loss</td>
<td>37</td>
</tr>
<tr>
<td>Miscarriage</td>
<td>29</td>
</tr>
<tr>
<td>TOP</td>
<td>8</td>
</tr>
<tr>
<td>Miscarriage twin to singleton</td>
<td>1</td>
</tr>
<tr>
<td>Lost to FU during second trimester</td>
<td>46</td>
</tr>
<tr>
<td>Deliveries</td>
<td>977</td>
</tr>
<tr>
<td>Singletons</td>
<td>758</td>
</tr>
<tr>
<td>Twins</td>
<td>209</td>
</tr>
<tr>
<td>Triplets</td>
<td>10</td>
</tr>
</tbody>
</table>

Table IXa  Method of delivery and gestational age, data collection I–IX.

<table>
<thead>
<tr>
<th></th>
<th>Total</th>
<th>Singletons</th>
<th>Twins</th>
<th>Triplets</th>
</tr>
</thead>
<tbody>
<tr>
<td>No deliveries</td>
<td>3163</td>
<td>2424</td>
<td>712</td>
<td>11</td>
</tr>
<tr>
<td>Method of delivery</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Vaginal</td>
<td>1315</td>
<td>1169</td>
<td>145</td>
<td>1</td>
</tr>
<tr>
<td>Caesarean</td>
<td>1490</td>
<td>988</td>
<td>482</td>
<td>20</td>
</tr>
<tr>
<td>Vaginal and Caesarean</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Unknown</td>
<td>351</td>
<td>265</td>
<td>80</td>
<td>6</td>
</tr>
<tr>
<td>Term at delivery</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Preterm</td>
<td>898</td>
<td>402</td>
<td>476</td>
<td>20</td>
</tr>
<tr>
<td>Term</td>
<td>1985</td>
<td>1807</td>
<td>176</td>
<td>2</td>
</tr>
<tr>
<td>Unknown</td>
<td>279</td>
<td>215</td>
<td>59</td>
<td>5</td>
</tr>
</tbody>
</table>

1For one twin there was only partial information: pregnancy was reported as a twin, the birth and baby as a singleton.

Table IXb  Method of delivery and gestational age, data X.

<table>
<thead>
<tr>
<th></th>
<th>Total</th>
<th>Singleton</th>
<th>Twin</th>
<th>Triplet</th>
</tr>
</thead>
<tbody>
<tr>
<td>No deliveries</td>
<td>977</td>
<td>758</td>
<td>209</td>
<td>10</td>
</tr>
<tr>
<td>Method of delivery</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Vaginal</td>
<td>356</td>
<td>328</td>
<td>27</td>
<td>1</td>
</tr>
<tr>
<td>Caesarean</td>
<td>489</td>
<td>327</td>
<td>153</td>
<td>9</td>
</tr>
<tr>
<td>Unknown</td>
<td>132</td>
<td>103</td>
<td>29</td>
<td>0</td>
</tr>
<tr>
<td>Term at delivery</td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Preterm</td>
<td>206</td>
<td>89</td>
<td>112</td>
<td>5</td>
</tr>
<tr>
<td>Term</td>
<td>705</td>
<td>628</td>
<td>75</td>
<td>3</td>
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<tr>
<td>Unknown</td>
<td>66</td>
<td>41</td>
<td>22</td>
<td>2</td>
</tr>
</tbody>
</table>

Confirmation of the diagnosis was performed prenatally (441/1609) and/or postnatally (401/1609) (Table Xb). Supplementary Table SXIIb shows the abnormalities found during or after the pregnancy. Several abnormalities were found that were not related to the PGD.

This report again confirms that pregnancies and babies born after PGD are very similar to the pregnancies obtained and babies born after ICSI treatment (Bonduelle et al., 2002). In our series, the number of multiple pregnancies remains high (293/1291, 23%). This means 37% (448/1206) of the babies born are part of a multiplet at birth.

Misdiagnoses

Table Xilla summarizes the misdiagnoses reported for data I–IX. For data X, no misdiagnoses have been reported. The Consortium has published a paper on the possible causes of misdiagnosis in PGD (Wilton et al., 2009).
<table>
<thead>
<tr>
<th>Method</th>
<th>Result</th>
<th>n</th>
<th>Normal</th>
<th>Abnormal</th>
<th>Failed</th>
</tr>
</thead>
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<td></td>
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<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td><strong>Prenatal diagnosis</strong></td>
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</tr>
<tr>
<td>FISH</td>
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<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>CVS</td>
<td>106</td>
<td>105</td>
<td>1</td>
<td>0</td>
<td></td>
</tr>
<tr>
<td>Amniocentesis</td>
<td>593</td>
<td>578</td>
<td>12</td>
<td>3</td>
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</tr>
<tr>
<td>Ultrasound</td>
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<td>961</td>
<td>11</td>
<td>4</td>
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</tr>
<tr>
<td>Unknown</td>
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<td>0</td>
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</tr>
<tr>
<td>Total</td>
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</tr>
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<td>PCR</td>
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<td></td>
</tr>
<tr>
<td>CVS</td>
<td>145</td>
<td>141</td>
<td>4</td>
<td>0</td>
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</tr>
<tr>
<td>Amniocentesis</td>
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<td>159</td>
<td>10</td>
<td>1</td>
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<tr>
<td>Ultrasound</td>
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<td>31</td>
<td>3</td>
<td>0</td>
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<td>2</td>
<td>0</td>
<td>0</td>
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<tr>
<td>Total</td>
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<td>16</td>
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<tr>
<td><strong>Post-natal diagnosis</strong></td>
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</tr>
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<td>FISH</td>
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<td>Karyotype miscarriage</td>
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<tr>
<td>Karyotype post-natal</td>
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</tr>
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<td>FISH microdeletion</td>
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</tr>
<tr>
<td>Physical examination</td>
<td>1142</td>
<td>1137</td>
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<td>0</td>
<td></td>
</tr>
<tr>
<td>Karyo post-natal + physical examination</td>
<td>15</td>
<td>15</td>
<td>0</td>
<td>0</td>
<td></td>
</tr>
<tr>
<td>Unknown</td>
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<td>2</td>
<td>0</td>
<td>0</td>
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</tr>
<tr>
<td>Total</td>
<td>1430</td>
<td>1379</td>
<td>53</td>
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</tr>
<tr>
<td>PCR</td>
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<td></td>
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<td></td>
<td></td>
</tr>
<tr>
<td>Karyotype miscarriage</td>
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<td>4</td>
<td>2</td>
<td>0</td>
<td></td>
</tr>
<tr>
<td>DNA test miscarriage</td>
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<td>0</td>
<td></td>
</tr>
<tr>
<td>DNA test post-natal</td>
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<td>Sweat test</td>
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<td>0</td>
<td>0</td>
<td></td>
</tr>
<tr>
<td>Physical examination</td>
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<td>82</td>
<td>1</td>
<td>0</td>
<td></td>
</tr>
<tr>
<td>Karyotype</td>
<td>15</td>
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<td>0</td>
<td></td>
</tr>
<tr>
<td>Karyotype + DNA</td>
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<td>3</td>
<td>0</td>
<td>0</td>
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</tr>
<tr>
<td>Karyotype + phys exam</td>
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<td>1</td>
<td>0</td>
<td>0</td>
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<tr>
<td>Algo test</td>
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<td>0</td>
<td></td>
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<tr>
<td>Unknown</td>
<td>11</td>
<td>11</td>
<td>0</td>
<td>0</td>
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</tr>
<tr>
<td>Total</td>
<td>218</td>
<td>214</td>
<td>4</td>
<td>0</td>
<td></td>
</tr>
</tbody>
</table>

1. Total 3 miscarriages after normal outcome amniocentesis (1 FISH, 2 PCR), one miscarriage after normal outcome (CVS) (FISH).
2. XY, + 21 \rightarrow TOP (AS maternal age, repeated IVF failure).
3. Three fetal sacs with abnormalities on ultrasound (enlarged lateral ventricle, cardiopathy, hydrocephalus) with normal result on amniocentesis.
4. 9% mosaic XY/XXY (FISH AS), abnormal chromosome 15 and skeletal displasia \rightarrow TOP (AS maternal age); Mosaic: 46,XY/47,XY+18 \rightarrow TOP (AS repeated IVF failures); 21 trisomy \rightarrow TOP (AS maternal age, repeated IVF failures).
5. Encephalocele \rightarrow TOP (AS repeated miscarriage); hemivertebrae, hypoplastic cerebellum, hydrocephaly \rightarrow TOP; cystic hygroma 1 twin miscarriage \rightarrow ongoing singleton (rec. translocation FISH).
6. Three fetal sacs had PCR and FISH at PGD.
7. 47,XY, + 13 \rightarrow TOP (PCR, not affected of Zellweger).
8. Mosaic 4n/2n (AS oocyte donation recurrent miscarriage); trisomy 20 (AS maternal age recurrent miscarriage); 92,XXXX (AS maternal age repeated IVF failures); 47,XX, + 10 (AS recurrent miscarriages maternal age); 46,XY/45,X0 (AS oocyte donation); 45,X,t(2;4)(q11.2;q13) (FISH reciprocal translocation); 47,XY,t(11;22)(q13;q13.2),+i(1)(t11)(t11p;11q) (FISH reciprocal translocation).
10. Two children had unknown check and karyotype.
Success of individual centres

Figure 1 shows the pregnancy rate for each centre for data X. The average pregnancy rate is 21.73%. Success rate tends to be higher and more homogenous in the most active centres (performing more than 100 OR per year). Centres carrying out lower numbers of cycles may have lower rates owing to less experience. The findings, however, indicate that some of the most active centres fall below the average 22% pregnancy rate and even have pregnancy rates lower than some of the centres performing few cycles. A more detailed statistical analysis comparing success rates according to various factors (indication, women age at OR, etc.) should be performed to confirm these differences.

Discussion

This 10th report of the ESHRE PGD Consortium demonstrates, as in previous years, the continuing increase in the number of PGD cycles, with subsequent pregnancies and babies. The number of centres participating in the data collection of 2007 was equal to 2006. There are still two levels of membership of the

### Table Xb Confirmation of diagnosis per fetal sac, data collection X.

<table>
<thead>
<tr>
<th>Method</th>
<th>Result</th>
<th></th>
<th></th>
<th></th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td></td>
<td>n</td>
<td>Normal</td>
<td>Abnormal</td>
</tr>
<tr>
<td>Prenatal diagnosis</td>
<td>FISH</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>CVS</td>
<td>12</td>
<td>10</td>
<td>2(^1)</td>
<td>0</td>
</tr>
<tr>
<td>Amniocentesis</td>
<td>80</td>
<td>75</td>
<td>5(^2)</td>
<td>0</td>
</tr>
<tr>
<td>Ultrasound</td>
<td>250</td>
<td>247</td>
<td>3(^3)</td>
<td>0</td>
</tr>
<tr>
<td>Total</td>
<td>342</td>
<td>332</td>
<td>10</td>
<td>0</td>
</tr>
<tr>
<td>PCR</td>
<td>CVS</td>
<td>26</td>
<td>26</td>
<td>0</td>
</tr>
<tr>
<td>Amniocentesis</td>
<td>70</td>
<td>68</td>
<td>2(^4)</td>
<td>0</td>
</tr>
<tr>
<td>Ultrasound</td>
<td>3</td>
<td>3</td>
<td>0</td>
<td>0</td>
</tr>
<tr>
<td>Total</td>
<td>99</td>
<td>97</td>
<td>2</td>
<td>0</td>
</tr>
<tr>
<td>Post-natal diagnosis</td>
<td>FISH</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Karyotype miscarriage</td>
<td>18</td>
<td>8</td>
<td>10(^5)</td>
<td>0</td>
</tr>
<tr>
<td>Karyotype post-natal</td>
<td>55</td>
<td>55</td>
<td>0</td>
<td>0</td>
</tr>
<tr>
<td>Physical examination</td>
<td>221</td>
<td>221</td>
<td>0</td>
<td>0</td>
</tr>
<tr>
<td>Karyo post-natal + physical examination</td>
<td>3</td>
<td>3</td>
<td>0</td>
<td>0</td>
</tr>
<tr>
<td>Total</td>
<td>297</td>
<td>287</td>
<td>10</td>
<td>0</td>
</tr>
<tr>
<td>PCR</td>
<td>Karyotype miscarriage</td>
<td>2</td>
<td>2</td>
<td>0</td>
</tr>
<tr>
<td>Physical examination</td>
<td>21</td>
<td>21</td>
<td>0</td>
<td>0</td>
</tr>
<tr>
<td>DNA test post-natal</td>
<td>38</td>
<td>38</td>
<td>0</td>
<td>0</td>
</tr>
<tr>
<td>Karyotype post-natal</td>
<td>1</td>
<td>0</td>
<td>1(^6)</td>
<td>0</td>
</tr>
<tr>
<td>Karyo post-natal + physical examination</td>
<td>30</td>
<td>30</td>
<td>0</td>
<td>0</td>
</tr>
<tr>
<td>DNA test + karyotype</td>
<td>2</td>
<td>1</td>
<td>1(^7)</td>
<td>0</td>
</tr>
<tr>
<td>Unknown</td>
<td>10</td>
<td>10</td>
<td>0</td>
<td>0</td>
</tr>
<tr>
<td>Total</td>
<td>104</td>
<td>102</td>
<td>2</td>
<td>0</td>
</tr>
</tbody>
</table>

### Table Xla Data on live born children, data collection I–IX.

<table>
<thead>
<tr>
<th>Total children born</th>
<th>2841(^1)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Sex</td>
<td></td>
</tr>
<tr>
<td>Male</td>
<td>1723</td>
</tr>
<tr>
<td>Female</td>
<td>1964</td>
</tr>
<tr>
<td>Unknown</td>
<td>154</td>
</tr>
<tr>
<td>Mean birthweight (g)</td>
<td></td>
</tr>
<tr>
<td>Singletons</td>
<td>3217</td>
</tr>
<tr>
<td>Twins</td>
<td>2389</td>
</tr>
<tr>
<td>Triplets</td>
<td>1883</td>
</tr>
<tr>
<td>Mean birth length (cm)</td>
<td></td>
</tr>
<tr>
<td>Singletons</td>
<td>50</td>
</tr>
<tr>
<td>Twins</td>
<td>46</td>
</tr>
<tr>
<td>Triplets</td>
<td>44</td>
</tr>
</tbody>
</table>

\(^1\)Numbers in the right column indicate the number of newborns for whom information is available.
Table XIb Data on children born, data collection X.

<table>
<thead>
<tr>
<th>Total children born</th>
<th>1206</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Sex</strong></td>
<td></td>
</tr>
<tr>
<td>Male</td>
<td>551</td>
</tr>
<tr>
<td>Female</td>
<td>555</td>
</tr>
<tr>
<td>Unknown</td>
<td>100</td>
</tr>
<tr>
<td><strong>Mean birthweight (g)</strong></td>
<td></td>
</tr>
<tr>
<td>Singletons</td>
<td>3224</td>
</tr>
<tr>
<td>Twins</td>
<td>2374</td>
</tr>
<tr>
<td>Triplets</td>
<td>1922</td>
</tr>
<tr>
<td><strong>Mean birth length (cm)</strong></td>
<td></td>
</tr>
<tr>
<td>Singletons</td>
<td>49.9</td>
</tr>
<tr>
<td>Twins</td>
<td>45.8</td>
</tr>
<tr>
<td>Triplets</td>
<td>46.3</td>
</tr>
<tr>
<td><strong>Mean head circumference (cm)</strong></td>
<td></td>
</tr>
<tr>
<td>Singletons</td>
<td>34.3</td>
</tr>
<tr>
<td>Twins</td>
<td>32.8</td>
</tr>
<tr>
<td>Triplets</td>
<td>33.9</td>
</tr>
<tr>
<td><strong>Apgar scores after 1 min</strong></td>
<td></td>
</tr>
<tr>
<td>Good²</td>
<td>215</td>
</tr>
<tr>
<td>Poor²</td>
<td>7</td>
</tr>
<tr>
<td><strong>Apgar scores after 5 min</strong></td>
<td></td>
</tr>
<tr>
<td>Good²</td>
<td>215</td>
</tr>
<tr>
<td>Poor²</td>
<td>4</td>
</tr>
<tr>
<td><strong>Apgar scores after 10 min</strong></td>
<td></td>
</tr>
<tr>
<td>Good²</td>
<td>119</td>
</tr>
<tr>
<td>Poor²</td>
<td>2</td>
</tr>
</tbody>
</table>

¹Indicates the number of newborns for whom information is available out of the total number of newborns.
²Good is defined ≥7; poor is defined <7.

Consortium; full membership for centres who submit annual data and associate membership for centres who cannot submit data (including new clinics, IVF units who work with a diagnostic laboratory that is a member of the Consortium). Associate centres performing PGD must send in summary data. For data X, only six associate centres sent in summary data and so these data were not included in this report. Most associate centres are satellite PGD centres that work with many IVF centres and they have reported that they cannot obtain information about the IVF cycles. Therefore, we have amended the information we will collect from associate centres to just include data on the diagnosis.

As always, the centres who submit data have access to the raw data while the associate centres are allowed to participate in the annual Consortium meetings and they are sent the quarterly Consortium newsletter.

Besides data collection, the Consortium is involved with a number of activities through the working groups. Currently there are five working groups: accreditation, misdiagnosis auditing and monitoring, guidelines, database and molecular methods. The accreditation working group has organized two quality management meetings and has two more scheduled: 2011 (Athens) and 2012 (Istanbul) in collaboration with EuroGentest. The group has also written a paper on the accreditation process specifically relating to PGD and ISO15189 (Harper et al., 2010b). This group annually collects data on the number of centres accredited. In many countries it is now becoming mandatory for all diagnostic laboratories to be accredited and the Consortium supports this as accreditation ensures quality of treatment. The FISH and PCR external quality assessment schemes (EQA) continue to operate annually and it is hoped that an array EQA will be set up in the near future. The misdiagnosis auditing and monitoring group are conducting two studies to examine the follow-up of embryos after PGD; one for PCR and one for FISH diagnoses. It is key that all centres utilize their untransferred embryos to validate and audit the methods that they are using and to calculate the efficacy of their techniques. The misdiagnosis working group wrote a paper on the causes of misdiagnosis (Wilton et al., 2009).

The guidelines working group has almost finished its task. Four new specific guidelines have been written: organization of a PGD/PGS Centre, Amplification-based PGD, FISH-based PGD and Embryo Biopsy and Embryology. Three of the four documents are available on the ESHRE website for discussion and suggestion, whereas the Embryo Biopsy Guideline is in the final stages of preparation and will be available online soon. All four guideline documents should be published before the end of 2010. The database working group has been refining the data collection and developing a database for the frozen embryo data. This will become more important as centres move to arrays and vitrification for their diagnoses (Harper and Harton, 2010). The molecular methods working group has set up a database of primers which is only open to full Consortium members. It is essential that all primer sets are validated in individual laboratories before clinical use.

From the ten data collections, the Consortium now has detailed data on 27 630 cycles and 4047 babies born after PGD/PGS.

The large amount of detailed information the Consortium has collected is unique and studies are underway to analyse many aspects of the data in more depth.

Supplementary data
Supplementary data are available at http://humrep.oxfordjournals.org/.

Acknowledgements
Many thanks also to all of the centres who participated in data collection X. Argentina: Fecunditas; Australia: Melbourne IVF; Belgium: Department of Embryology and Genetics of the VUB and Centre for Medical Genetics of the Universitair Ziekenhuis Brussels; Hospital Erasme, ULB, Laboratoire FIV; Leuven Institute for Fertility and Embryology; GIFT, ZOL Ziekenhuis; Leuven University Fertility Centre; Brazil: Fertility - Assisted Reproductive Centre, Sao Paolo; Czech Republic: Sanatorium Repromeda; Institute Pronatal, Genetics; Denmark: Centre for Preimplantation Genetic Diagnosis, Aarhus University Hospital, Fertility Clinic; Fertility Clinic, University Hospital Copenhagen; Fertility Clinic, University of Odense; Finland: Helsinki University Central Hospital, Department of Obstetrics & Gynecology/IVF Unit; AVA-Clinic; France: SIHCUS-CMCO, Unité de
### Table XIIIa  Summary of misdiagnosis from data I–IX.

<table>
<thead>
<tr>
<th>Indication</th>
<th>Method used</th>
<th>PND-post-natal</th>
<th>Outcome</th>
<th>Reported in</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Monogenics</strong></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>DM 1</td>
<td>PCR</td>
<td>PND</td>
<td>TOP</td>
<td>I</td>
</tr>
<tr>
<td>β-Thal</td>
<td>PCR</td>
<td>PND</td>
<td>TOP</td>
<td>II</td>
</tr>
<tr>
<td>β-Thal</td>
<td>PCR</td>
<td>PND</td>
<td>TOP</td>
<td>VIII</td>
</tr>
<tr>
<td>Familial amyloid polyneuropathy</td>
<td>PCR</td>
<td>PND</td>
<td>Born</td>
<td>IV</td>
</tr>
<tr>
<td>CF</td>
<td>PCR</td>
<td>PND</td>
<td>Born</td>
<td>II</td>
</tr>
<tr>
<td>CF (one of twins)</td>
<td>PCR</td>
<td>Post</td>
<td>Born</td>
<td>IV</td>
</tr>
<tr>
<td>CMT1A</td>
<td>PCR</td>
<td>PND</td>
<td>born</td>
<td>Cycle reported in V but misdiagnosis in VII</td>
</tr>
<tr>
<td>SMA</td>
<td>PCR</td>
<td>Post</td>
<td>Born</td>
<td>Cycle reported IV but misdiagnosis in VII</td>
</tr>
<tr>
<td>CMT1A (twins)</td>
<td>PCR</td>
<td>PND</td>
<td>TOP of both twins</td>
<td>VII</td>
</tr>
<tr>
<td>FRAXA</td>
<td>PCR</td>
<td>PND</td>
<td>Born</td>
<td>VIII</td>
</tr>
<tr>
<td><strong>Sexing for X-linked disease</strong></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>46,XY in retinitis pigmentosa</td>
<td>PCR</td>
<td>PND</td>
<td>Born</td>
<td>IV</td>
</tr>
<tr>
<td>46,XY in DMD twin</td>
<td>PCR</td>
<td>PND</td>
<td>TOP of one twin</td>
<td>III</td>
</tr>
<tr>
<td>45,X, Haem A</td>
<td>FISH</td>
<td>PND</td>
<td>TOP</td>
<td>IV</td>
</tr>
<tr>
<td>46,XY, Haem A</td>
<td>FISH</td>
<td>Post</td>
<td>Born</td>
<td>VIII</td>
</tr>
<tr>
<td><strong>Translocations</strong></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Trisomy 13 after 45,XY,der(13;14)(q10;q10)</td>
<td>FISH</td>
<td>PND</td>
<td>Miscarried</td>
<td>Miscarried</td>
</tr>
<tr>
<td>47,XX, +der(22)(11;22)(q23.3;q11.2)mat</td>
<td>FISH</td>
<td>PND</td>
<td>TOP</td>
<td>III</td>
</tr>
<tr>
<td>46,XY,der(15)(13;15) (q25.1;q26.3)pat</td>
<td>FISH</td>
<td>PND</td>
<td>TOP</td>
<td>VII</td>
</tr>
<tr>
<td><strong>PGS</strong></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>47,XXX</td>
<td>FISH</td>
<td>PND</td>
<td>Lost to FU</td>
<td>VII</td>
</tr>
<tr>
<td>45,X</td>
<td>FISH</td>
<td>PND</td>
<td>Miscarried</td>
<td>Miscarried</td>
</tr>
<tr>
<td>Transomy 16 after 1st PB biopsy only</td>
<td>FISH</td>
<td>Miscarried</td>
<td>Miscarried</td>
<td>VI</td>
</tr>
<tr>
<td>Transomy 16 after 1st PB biopsy only</td>
<td>FISH</td>
<td>Miscarried</td>
<td>Miscarried</td>
<td>V</td>
</tr>
<tr>
<td>Transomy 16</td>
<td>FISH</td>
<td>Miscarried</td>
<td>Miscarried</td>
<td>VI</td>
</tr>
<tr>
<td>Transomy 16</td>
<td>FISH</td>
<td>Post</td>
<td>Born</td>
<td>III</td>
</tr>
<tr>
<td>Transomy 21</td>
<td>FISH</td>
<td>PND</td>
<td>TOP</td>
<td>IX</td>
</tr>
<tr>
<td>Transomy 21</td>
<td>FISH</td>
<td>PND</td>
<td>TOP</td>
<td>IX</td>
</tr>
<tr>
<td>46,XY/47,XY+18</td>
<td>FISH</td>
<td>PND</td>
<td>TOP</td>
<td>IX</td>
</tr>
<tr>
<td><strong>SS</strong></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Requested male but female fetus</td>
<td>FISH</td>
<td>PND</td>
<td>TOP</td>
<td>III</td>
</tr>
</tbody>
</table>

PND, prenatal diagnosis.

The numbers in the last column indicate the PGD Consortium report number.
University Medical Centre Utrecht; Turkey: Istanbul Memorial Hospital, Reproductive Endocrinology & ART Centre; Acibadem Genetic Diagnosis and Cell Therapy Centre, Acibadem Genel Mudurluk; UK: UCL Centre for PGD, Department of Cytogenetics and Centre for Preimplantation Genetic Diagnosis; Centre for PGD, Assisted Conception Unit, Guy’s Hospital; Institute of Ob/Gyn-RPMS, Hammer smith Hospital; Ukraine: Clinic of Reproductive Medicine ‘Nadiya’; USA: Jones Inst. for Reproductive Med; Genetics and IVF Institute; Reproductive Biology Associates Atlanta.

References


