**Supplemental Table 1.** Two excluded cases with unique and atypical results involving the sex chromosomes

|  |  |  |
| --- | --- | --- |
|  | **Case 1** | **Case 2** |
| Gestational age at screening - wks | 10 | 12 |
| Fetal fraction - % | 12.1 | 10.4 |
| Prenatal diagnosis of major ultrasound abnormality | No | No |
| cfDNA Result | High Risk MX(Fetal sex not requested) | Low Risk(Fetal sex not requested) |
| Clinical Genetic Test Source and Result(s) | Newborn peripheral bloodCytogenetics-32/50 cells 47,XXX-17/50 cells 45,X-1/50 cells 46,XXFISH-SRY neg 200/200-114/143 XXX-17/143 X-12/143 XX | Not Performed |
| Research Array Source and Result | Newborn Blood SpotNormal Female | Newborn Blood SpotXp22.33 dup 2.6MbXq21.31a21.32 dup 3.9MbYp11.2 dup 3.3 MbYq11.21p11.223 del 10.5Mb |
| Clinical Fetal Sex Report at Birth | Female | Male |
| Mother/Child concordance testinga | Match | Match |
| Additional Notes | No genetic testing of placenta |  |
| Reason for Exclusion | Double Aneuploidy and Conflicting Confirmatory Genetic Test Results | Copy number variations of uncertain clinical significance, outside scope of cfDNA screening |

aSNP-cfDNA compared with SNP-research array only

**Supplemental Table 2.** Characteristics and outcomes of pregnancies with cfDNA results for autosomal trisomies but no cfDNA result for MX (n=59); stratified by likelihood of redraw success

|  |  |  |
| --- | --- | --- |
|  | **No Result MX****High Likelihood of Successful Redraw****n=28** | **No Result MX****Low Likelihood of Successful Redraw****n=31** |
| Mean maternal age (SD) - year | 34.5 (4.4) | 33.6 (5.8) |
| Mean gestational age at screening (SD) - weeks | 14.5 (3.5) | 13.1 (2.4) |
| Median fetal fraction (IQR) - % | 5.7 (5.0 – 6.2) | 9.4 (7.0 – 13.3) |
| Ultrasound abnormality before cfDNA | 0 | 0 |
| Confirmatory Genetic Test Result | Normal XX, n=28Normal XY, n=0  | Normal XX, n=24Normal XY, n=6Abnormal XXY, n=1 |
| cfDNA fetal sex reporting | Fetal sex requested n=24cfDNA reported ‘female’ n=24 | Fetal sex requested n=29cfDNA ‘female’ n=22cfDNA no result fetal sex n=7  |

NA=not applicable

**Supplemental Table 3.** Demographics and pregnancy characteristics for total cohort and SCTs

|  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- |
| **Variable** | **Total Cohort n=17,538** | **Any SCT n=52** | **XXX n=8** | **XXY n=26** | **XYY n=18** | p valuea  |
| Mean maternal age (SD)– year | 33.5 (5.4) | 35.6 (5.9) | 37.5 (6.2) | 36.3 (5.6) | 33.7 (6.1) | 0.014 |
| Nulliparity, n (%) | 7743 (44.3) | 22 (42.3) | 5 (62.5) | 9 (34.6) | 8 (44.4) | 0.778 |
| Mean gestational age at screening (SD) - weeks | 13.3 (3.2) | 13.0 (2.6) | 15.4 (3.8) | 12.9 (2.0) | 12.0 (2.2) | 0.662 |
| Mean body mass index (SD) - kg/m2  | 26.3 (5.8) | 27.4 (6.0) | 30.9 (7.0) | 26.2 (4.4) | 28.0 (7.2) | 0.112 |
| Median fetal fraction (IQR) - % | 9.3 (7.0 – 12.2) | 9.1 (6.4 – 11.2) | 9.5 (7.7 – 13.4) | 8.4 (5.8 – 10.5) | 9.7 (6.5 – 11.5) | 0.257 |
| Race, n (%)\* |  |  |  |  |  | 0.973 |
|  Asian | 1516 (8.6) | 4 (7.7) | 1 (12.5) | 3 (11.5) | 0 |  |
|  Black | 1505 (8.6) | 4 (7.7) | 1 (12.5) | 2 (7.7) | 1 (5.6) |  |
|  Caucasian | 10,653 (60.7) | 31 (59.6) | 3 (37.5) | 15 (57.7) | 13 (72.2) |  |
|  Latina | 3,267 (18.6) | 11 (21.1) | 2 (25.0) | 6 (23.1) | 3 (16.7) |  |
|  Other/unknown | 599 (3.4) | 2 (3.8) | 1 (12.5) | 0 | 1 (5.6) |  |
| Ultrasound abnormality before cfDNA, n (%) | 98 (0.6) | 0 | 0 | 0 | 0 | 1.00 |
| Ultrasound abnormality at any time, n (%) | 667 (4.0) | 5 (10.2) | 1 (12.5) | 2 (7.7) | 2 (13.3) | 0.043 |
| Pregnancy outcome, n (%) Livebirth Intrauterine fetal death/stillbirth Spontaneous abortion Elective termination Unknown | 17341 (98.88)24 (0.14)26 (0.15)134 (0.76)13 (0.07) | 39 (75.0)0012 (23.1)1 (1.9) | 7 (87.5)001 (12.5)0 | 18 (69.2)007 (26.9)1 (3.9) | 14 (77.8) 004 (22.2) | <0.001 |

a comparing any SCT to No/ missing SCT

**Supplemental Table 4.** Demographics and pregnancy characteristics for study participants who declined or requested fetal sex reporting

|  |  |  |
| --- | --- | --- |
| **Variable** | **Declined fetal sex reporting****(n=2,052)** | **Requested fetal sex reporting****(n=10,745)** |
| Mean maternal age (SD)– year | 35.7 (4.5) | 33.4 (5.5) |
| Nulliparitya | 926 (45.5) | 4693 (43.8) |
| Mean gestational age at screening (SD) - weeks | 13.2 (3.9) | 13.1 (3.2) |
| Mean body mass index (SD) - kg/m2  | 25.4 (4.8) | 26.5 (5.9) |
| Median fetal fraction (IQR) - % | 9.7 (7.4 – 12.9) | 9.3 (6.9 – 12.3) |
| Race, n (%) b |  |  |
|  Asian | 94 (4.6) | 1136 (10.6) |
|  Black | 76 (3.7) | 1229 (11.4) |
|  White | 1670 (81.4) | 5223 (48.6) |
|  Latina | 192 (9.4) | 2737 (25.5) |
|  Other/unknown | 20 (1.0) | 420 (3.9) |
| Ultrasound abnormality before or concurrent with cfDNA test, n (%) \* | 15 (0.7) | 54 (0.5) |
| Ultrasound abnormality at anytime, n (%) | 101 (5.5) | 382 (3.7) |
| Pregnancy outcome, n (%) Livebirth Intrauterine fetal death/stillbirth Spontaneous abortion Pregnancy Interruption | 2011 (98.1)2 (0.1)8 (0.4)30 (1.5) | 10,625 (99.0)19 (0.2)16 (0.2)75 (0.7) |

aAll differences are significant at p<0.05, except for nulliparity and ultrasound abnormality before or concurrent with cfDNA test.

bRace and ethnicity as reported by participants. If the participant did not report the information, the information from the medical record was used.