

Supplementary Table 3. Reasons cited by ARCs choosing not to undergo prenatal diagnosis.

|  | All pregnancies,<br>n (%; CI) | Pregnancy during<br>screening,<br>n (%; CI) | Subsequent<br>pregnancy,<br>n (%; CI) |
|--|-------------------------------|---|---------------------------------------|
| Did not undergo prenatal diagnosis                       | 213 (100)                     | 95 (100)                                    | 118 (100)                             |
| Reason <sup>a</sup>                                      |                               |   |                                       |
| Risk of miscarriage                                      | 50 (23; 18-29)                | 33 (35; 26-45) <sup>b</sup>                 | 17 (14; 9-22) <sup>b</sup>            |
| Perception of low risk of pregnancy being affected       | 45 (21; 16-27)                | 25 (26; 18-36)                              | 20 (17; 11-24)                        |
| Would not have terminated the pregnancy                  | 38 (18; 13-23)                | 26 (27; 19-37) <sup>c</sup>                 | 12 (10; 6-17) <sup>c</sup>            |
| Not necessary because IVF with PGT-M was performed       | 31 (15; 11-21)                | 0 (0; 0-3)                                  | 31 (28; 20-37)                        |
| Pregnancy miscarried before testing could be performed   | 16 (8; 5-12)                  | 0 (0; 0-3)                                  | 16 (14; 8-21)                         |
| Condition phenotype not severe enough to warrant testing | 12 (6; 3-10)                  | 6 (6.3; 3-13)                               | 6 (5.1; 2-10)                         |
| Planning post-natal testing instead                      | 8 (4; 2-7)                    | 5 (5.3; 2-11)                               | 3 (2.5; 1-7)                          |
| No prenatal treatment options available                  | 4 (1.9; 1-4)                  | 3 (3.2; 1-8)                                | 1 (0.8; 0-4)                          |
| Too late in the pregnancy to terminate                   | 4 (1.9; 1-4)                  | 3 (3.2; 1-8)                                | 1 (0.8; 0-4)                          |
| Provider did not recommend testing                       | 4 (1.9; 1-4)                  | 2 (2.1; 0-7)                                | 2 (1.7; 0-5)                          |
| Cost was too high  | 3 (1.4; 0-4)                  | 1 (1.1; 0-5)                                | 2 (1.7; 0-5)                          |
| Pregnancy was terminated before pursuing testing         | 2 (0.9; 0-3)                  | 0 (0; 0-3)                                  | 2 (1.7; 0-5)                          |
| Too early in pregnancy to undergo testing                | 2 (0.9; 0-3)                  | 0 (0; 0-3)                                  | 2 (1.7; 0-5)                          |
| Other/Did not specify                                    | 24 (11; 8-26)                 | 7 (7.4; 3-14)                               | 17 (14; 9-22)                         |

ARC: At-risk couple. CI: confidence interval, 95%. IVF: In vitro fertilization. PGT-M: Preimplantation genetic testing for monogenic conditions.

- a. Percents will sum to greater than 100% because respondents could cite more than one reason.
- b. Significantly different at the 95% confidence level ( $p=0.0005$ ).
- c. Significantly different at the 95% confidence level ( $p=0.001$ ).