What test did I have? Patient uncertainty about prenatal genetic screening

OBJECTIVE: Pregnant patients often lack understanding about the genetic testing they undergo. We asked patients who received prenatal genetic screening to identify the tests they had undergone. We hypothesized that patient awareness about having undergone a specific genetic test would be associated with sociodemographic factors.

STUDY DESIGN: This was a planned secondary analysis of a randomized controlled trial in which the effect of video education about genetic privacy laws on patients who underwent cell-free DNA (cfDNA) screening was analyzed (WIH_IRB_#1500909). Inclusion criteria for the original study included English-speaking patients with a singleton gestation presenting between 17+0 and 23+6 weeks’ gestation for a detailed anatomy scan at a prenatal diagnosis unit of a tertiary academic medical center and who previously underwent cfDNA aneuploidy screening. All participants viewed an infographic about cfDNA screening and a description of carrier testing. Patients in the intervention arm also viewed a video about genetic privacy laws. All participants took an electronic survey about their knowledge of and attitudes toward genetic screening. For inclusion in this secondary analysis, participants must have completed the survey questions related to cfDNA and carrier screening. Only participants with carrier screening results in their medical record were included in the analyses regarding carrier screening. The outcome of interest was participant awareness of having undergone cfDNA or carrier screening.

<table>
<thead>
<tr>
<th>Characteristics</th>
<th>cfDNA screening (%)</th>
<th>Carrier screening (%)</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>Unaware (n=160)</td>
<td>Aware (n=128)</td>
</tr>
<tr>
<td>Age (y), median (IQR)</td>
<td>32 (28–36)</td>
<td>35 (32–38)</td>
</tr>
<tr>
<td>Insurance typec</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Private or commercial</td>
<td>20 (17.1)</td>
<td>97 (82.9)</td>
</tr>
<tr>
<td>Public or Medicaid</td>
<td>12 (27.9)</td>
<td>31 (72.1)</td>
</tr>
<tr>
<td>Self-reported race</td>
<td></td>
<td></td>
</tr>
<tr>
<td>White</td>
<td>22 (18.3)</td>
<td>98 (81.7)</td>
</tr>
<tr>
<td>Black</td>
<td>6 (42.9)</td>
<td>8 (57.1)</td>
</tr>
<tr>
<td>All others</td>
<td>4 (15.4)</td>
<td>22 (84.6)</td>
</tr>
<tr>
<td>Self-reported Hispanic</td>
<td>7 (21.9)</td>
<td>25 (78.1)</td>
</tr>
<tr>
<td>College education or higher</td>
<td>16 (17.4)</td>
<td>76 (82.6)</td>
</tr>
<tr>
<td>Nulliparous</td>
<td>9 (15.5)</td>
<td>49 (84.5)</td>
</tr>
<tr>
<td>High risk for fetal aneuploidyd</td>
<td>16 (15.0)</td>
<td>91 (85.0)</td>
</tr>
<tr>
<td>Genetic counseling visitf</td>
<td>13 (24.1)</td>
<td>41 (75.9)</td>
</tr>
<tr>
<td>Prenatal care provider</td>
<td></td>
<td></td>
</tr>
<tr>
<td>General OB/GYN</td>
<td>26 (21.3)</td>
<td>96 (78.7)</td>
</tr>
<tr>
<td>Maternal-fetal medicine</td>
<td>0 (0.0)</td>
<td>7 (100)</td>
</tr>
<tr>
<td>All others</td>
<td>6 (19.4)</td>
<td>25 (80.6)</td>
</tr>
</tbody>
</table>

Categorical data are presented as number (percentage). cfDNA, cell-free DNA; IQR, interquartile range; OB/GYN, obstetrician-gynecologist.

a Includes participants who incorrectly stated that they had not taken a test, as well as participants who were unsure; b Wilcoxon rank-sum tests were used to determine significance; c Fisher’s exact tests were used to determine significance; d Risk factors include advanced maternal age at time of delivery, ultrasound findings or serum screen with increased risk for aneuploidy, previous pregnancy affected by aneuploidy, family history of aneuploidy, and parental balanced Robertsonian translocation with increased risk of T13 or T21; e Genetic counseling visit must have occurred during the current pregnancy; f Other providers included nurse midwives, nurse practitioners, family practice providers, and residents.

Awareness was defined as correctly reporting that they received a specific genetic screen. Unawareness was defined as either reporting incorrectly that they did not undergo a specific genetic screen or being unsure. No a priori power calculation was performed for this secondary analysis.

RESULTS: Of the 213 patients approached, 161 (76%) were randomized and included in the original study, and 160 (75%) met the inclusion criteria for this secondary analysis. All participants had undergone cfDNA screening and 54 (33.7%) had undergone carrier screening. Of the patients included in the cohort, 30% (48 of 160) and 16.6% (9 of 54) did not remember discussing cfDNA or carrier screening, respectively, with their prenatal providers (data not shown).

Of those who underwent cfDNA screening, 20% (32 of 160) were unaware of doing so. The following sociodemographic differences were identified between the 2 groups: the frequency of being unaware of undergoing cfDNA screening was higher among younger women ($P = .02$) and women at low risk for fetal aneuploidy ($P = .04$) (Table). Among the 54 women who underwent carrier screening, 50% (27 of 54) were unaware of doing so. Although the difference was not statistically significant, a higher proportion of women who identified as racial minorities, had public insurance, or had lower educational attainment was unaware of undergoing carrier screening (Table). The outcomes did not differ by study arm (data not shown).

CONCLUSION: Overall, 1 in 5 pregnant patients presenting for level 2 ultrasound did not know that they underwent cfDNA testing and half were unaware that they had completed carrier screening. Rates of awareness were significantly lower among women who were younger and at lower risk for fetal aneuploidy. The lack of awareness highlights serious concerns about informed consent for prenatal genetic screening, particularly as this screening expands to younger and lower-risk populations. Providers should develop improved consenting strategies—perhaps with educational interventions—to improve patients’ awareness of, and consent for, prenatal genetic screening.

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This study was registered on ClinicalTrials.gov with the identifier NCT04420858.

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REFERENCES
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