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Sociodemographic and attitudinal predictors of simultaneous and redundant multiple marker and cell-free DNA screening among women aged 35 years and older

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Abstract

Objective—To identify characteristics associated with undergoing cell-free DNA (cfDNA) and multiple marker screening (MMS) simultaneously or redundantly (after receiving negative results from the first screening test) among women aged 35 and older.

Study Design—Participants presenting for prenatal testing completed a questionnaire which included measures of pregnancy worry and attitudes toward potential testing outcomes; data on prenatal test use was obtained via medical record review. We used multivariable logistic regression to identify factors associated with redundant or simultaneous screening.

Result—Among 164 participants, 69 (42.1%) had cfDNA redundantly (n=51) to, or simultaneously (n=18) with, MMS. Compared to the 46 MMS-negative women who did not undergo further testing, those who underwent redundant or simultaneous cfDNA/MMS screening were more likely to have annual family incomes >$150,000, to feel having a miscarriage would be worse than having an intellectually disabled child, to desire comprehensive testing for intellectual disability, and to have more pregnancy worry.

Conclusion—Providers who counsel patients on prenatal aneuploidy screening tests should explain the appropriate utilization of these screening tests to avoid unnecessary or minimally informative use of multiple tests.

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Conflicts of interest: From April 1, 2012 through March 31, 2014, Dr. Kuppermann was the UCSF site PI for a clinical study of cfDNA testing among average risk women (the Noninvasive Examination of Trisomy NEXT Study) funded by Ariosa Diagnostics, and her institution received unrestricted research funding for her research program from Natera in 2013.

Introduction

Current prenatal testing for fetal chromosomal abnormalities includes non-invasive screening and invasive diagnostic testing options.[1] Until recently, the only non-invasive screening option was multiple marker screening (MMS), incorporating maternal serum analyte screening with or without a first-trimester nuchal translucency ultrasound. In 2011, another non-invasive method, cell-free DNA (cfDNA) screening, which analyses cell-free fetal DNA in maternal circulation, became clinically available. Although tradeoffs between detection rates and false positive rates have always been central to prenatal testing decisions, the availability of cfDNA has increased not just the number of non-invasive options available but the complexity of prenatal testing decision-making.[1] Compared to MMS, cfDNA is a more accurate test for trisomies 13, 18, and 21 and sex chromosome anomalies; however, unlike MMS, cfDNA does not screen for other less common chromosomal conditions (or fetal anatomic anomalies, if second-trimester serum analytes are drawn).[2–8] Aggressive marketing of cfDNA as a simple blood test that detects Down syndrome[9]—and fetal sex[10]—much earlier in pregnancy has further contributed to uncertainty regarding the optimal use of these tests.

Although the American College of Medical Genetics and Genomics has issued a position statement permitting cfDNA screening for all women, including those at low-risk for aneuploidy,[11] the Society for Maternal-Fetal Medicine (SMFM),[12,13] the American College of Obstetricians and Gynecologists (ACOG),[1] the National Society of Genetic Counselors (NSGC),[14] and the International Society for Prenatal Diagnosis (ISPD)[15] prenatal aneuploidy screening algorithms support cfDNA as an option for prenatal screening only for women at increased risk of fetal aneuploidy. Increased risk has been defined as maternal age 35 or older at delivery, screen positive results from MMS, ultrasound findings associated with increased risk of aneuploidy, a history of pregnancy affected by aneuploidy, or having a balanced Robertsonian translocation.[1,12–15] Of note, ACOG and SMFM recommend MMS as first-line screening, with cfDNA or invasive testing recommended as follow-up tests among screen-positive women and no additional testing among screen-negative women.[1,12,13] Furthermore, ACOG's most recent guidelines explicitly state not only that women who have a negative screening test result should not be offered additional screening tests (referred to in this paper as "redundant screening") due to an increased risk for false-positive test results, but that simultaneous testing with multiple screening methodologies for aneuploidy is not cost-effective and should not be performed.[1]

Little is known about the utilization of cfDNA and MMS in combination in clinical practice. While prior studies have described cfDNA uptake rates among women with high-risk pregnancies worldwide,[16,17] to our knowledge, there are no published data describing cfDNA uptake rates among women of advanced maternal age who have already had or are having MMS. In addition, racial, ethnic, and socioeconomic differences in prenatal testing uptake have been reported, some of which is attributed to differences in attitudes[18] and some of which is not,[19–21] but these data are from the pre-cfDNA era. This study aims to determine the prevalence of redundant or simultaneous cfDNA screening and to identify sociodemographic and attitudinal predictors of cfDNA use in this context among women who would be aged 35 or older at the time of delivery.

Materials and Methods

This prospective cohort study was conducted at the University of California, San Francisco (UCSF), a tertiary care, academic medical center. During the study period (June 2013 to December 2014), the UCSF's Prenatal Diagnostic Center (PDC)'s protocol for prenatal aneuploidy screening required genetic counselors to discuss detection rates for trisomy 21 for cfDNA and MMS, to review the limitations of cfDNA and MMS, and to compare the limitations of screening in comparison to the rates of detection and miscarriage for invasive diagnostic testing. Genetic counselors at the study site were instructed to follow ACOG and SMFM recommendations, counseling women who screen negative with MMS against undergoing additional aneuploidy testing, while offering those who screen positive with MMS additional testing with either cfDNA or invasive testing if desired. Women were also counseled against use of simultaneous cfDNA and MMS screening. Despite these recommendations, some women at UCSF requested redundant or simultaneous cfDNA screening, and the policy was to honor these preferences. After their in-person consultation, genetic counselors ordered whichever aneuploidy testing that the patient requested.

Participants for this study included English- or Spanish-speaking women who would be aged 35 and older at the time of delivery, were at less than 20 weeks gestation with a singleton pregnancy, had an in-person consultation with a genetic counselor, had undergone a first-trimester ultrasound for viability, and had either not yet undergoing genetic screening, or, if they had undergone first- or second-trimester screening, had not yet received results.

Research coordinators approached potentially eligible women in the PDC at their scheduled appointments for nuchal translucency or anatomy ultrasound, prior to undergoing the ultrasound. Among the 595 women who were found to be eligible during the study period, 172 signed the consent form and completed and returned the questionnaire; of these, 8 did not undergo any prenatal testing or screening and therefore were excluded from the analysis, leaving a study population of 164 women. Participants were remunerated with a $5 gift card for a local coffee shop.

The questionnaire included standard sociodemographic questions regarding participants' age, race or ethnicity, family income, education level, number of prior pregnancies and outcomes of each pregnancy, and personal or family history of aneuploidy. Additionally, the questionnaire included items related to knowledge about prenatal testing, attitudes toward potential testing outcomes, and pregnancy worry, all of which our group has used in previous studies.[18,21] The knowledge scale was modified to include cfDNA as a screening option (see Appendix). Medical record review was conducted after delivery to obtain information on which prenatal tests the participant underwent, as well as the timing of these tests. We maximized our sample size based on available resources and remuneration of participants. The UCSF Committee for Human Subjects Research approved this study.

Negative MMS results were defined as negative first- and/or second-trimester serum analytes. Women were categorized as having redundant or simultaneous screening based on the timing of their cfDNA blood draw and the receipt of their MMS results: those who had cfDNA drawn prior to receiving results from MMS were included in the simultaneous

screening group while those who had a cfDNA blood draw after receiving negative results for first- or second-trimester MMS were included in the redundant screening group. The timing of study enrollment or survey completion did not impact this categorization.

The California Prenatal Screening program defines screen-positive MMS as having a calculated risk ratio for aneuploidy higher than 1:100 for first trimester MMS (serum analytes and nuchal translucency ultrasound, if done) and higher than 1:200 for integrated MMS. However, given that women may have received MMS results that were negative by the California Prenatal Screening program standards but considered clinically concerning for aneuploidy, additional screening may have been clinically justifiable. Therefore, beyond assessing whether results were screen positive or screen negative based on the California Prenatal Screening program standards, we also performed chart review to ascertain whether women received first-trimester risk ratios higher than the risk of miscarriage from invasive testing or their predicted age, had abnormal ultrasound findings concerning for aneuploidy, or received an integrated risk ratio after first and second trimester samples higher than their reported first trimester risk ratio. Bivariable predictors of redundant or simultaneous MMS and cfDNA screening were identified using chi-squared analysis and bivariate logistic regression. Multivariable logistic regression was then performed using backwards elimination, with the final model including all variables that had adjusted odds ratios (aOR) with p values less than 0.20. Candidate variables evaluated in the multivariable logistic regression model included maternal characteristics (age at due date, annual household income, and history of prior live birth, all self-reported), preferences regarding specific pregnancy outcomes (miscarriage versus intellectual disability and false-positive test versus undetected intellectual disability), and scales for knowledge and pregnancy worry.

Results

Figure 1 summarizes the prenatal testing strategies employed by our study participants. Among the 164 women who enrolled in this study, 142 women had MMS as a first-line screening strategy. Of these, 103 women received screen negative MMS results, 21 did not have MMS results when they underwent additional testing (i.e., they were MMS screen unknown), and 18 received positive MMS results. Among the 103 women who received negative MMS results, more than half (56%) underwent redundant screening with cfDNA (n=51) or amniocentesis (n=6) rather than foregoing additional testing. As such, our final study population was composed of 115 women: the 69 women who either pursued redundant (n=51) or simultaneous (n=18) screening with cfDNA were compared to 46 women who received negative MMS results and did not undergo additional testing.

Among the 51 women who pursued redundant screening with cfDNA and MMS, six received first-trimester MMS risk ratios that were higher than UCSF's quoted risk of miscarriage from invasive testing (1:350), three had combined MMS risk ratios higher for aneuploidy than their first trimester MMS risk, and three had first trimester risk ratios higher than their age-predicted risk ratio. None of the 69 women who underwent redundant or simultaneous cfDNA did so in the setting of abnormal ultrasound findings. As such, among women who underwent redundant cfDNA, 76% did so having received MMS results that

were screen negative and with risk ratios lower than age, lower than risk of miscarriage from invasive diagnostic testing, and lower for combined screening than first-trimester screening.

Because our primary interest was gaining an understanding of the characteristics of women who had redundant or simultaneous MMS/cfDNA screening, we focused our analysis on comparison of the 69 women who underwent redundant (n=51) or simultaneous (n=18) MMS and cfDNA screening in comparison to the 46 women who had negative MMS results and did not pursue further screening (Figure).

Bivariable analysis suggested that women were more likely to pursue redundant or simultaneous cfDNA screening if they scored higher on the knowledge scale or had annual incomes that were either less than $50,000 or more than $150,000 (Table 1). They also were more likely to pursue redundant or simultaneous cfDNA screening if they scored higher on the worry scale, rated the importance of knowing whether the baby would be born with any chromosomal cause of intellectual disability more highly, or thought that an undetected intellectual disability would be worse than a false-positive test result.

The final multivariable logistic regression model controlled for maternal race/ethnicity, education, and attitude towards having a false-positive test result. In these analyses, we observed that women whose annual household income was greater than $150,000 or less than $50,000 were more likely to undergo redundant or simultaneous screening than those whose annual household incomes were between $50,001 and $149,999 (adjusted odds ratio [aOR] 5.58 [95% Confidence Interval (CI): 1.51–20.59], p=0.01 and aOR 6.27 [95% CI: 1.06–37.06]; p=0.04 for each of these comparisons respectively; Table 1). We also observed non-significant trends towards undergoing redundant or simultaneous cfDNA screening among women who were older at their delivery date or who were nulliparous (aOR 1.29 [95% CI 1.00–1.65], p=0.05; aOR 2.92 [95% CI 0.99–8.58], p=0.05; respectively).

A number of attitudinal predictors of redundant or simultaneous cfDNA and MMS screening were also identified in multivariable logistic regression. Specifically, women who scored higher on the pregnancy worry scale or who indicated that having a miscarriage would be worse for them than having a child with an undiagnosed intellectual disability were at higher odds of undergoing redundant or simultaneous cfDNA screening (aOR 3.16 [95% CI 1.17–8.51] for every 1 point increase on a 4-point scale, p=0.02 and aOR 4.59 [95% CI 1.11–19.10] p=0.04, respectively; Table 1). Women who scored higher on the scale measuring the importance of knowing whether or not their baby would be born with any chromosomal cause of intellectual disability also were at increased odds of undergoing redundant or simultaneous cfDNA/MMS screening (aOR 6.06 [95% CI 1.81–19.97] for every 1-point increase on a 4-point scale, p=0.003; Table 1). There was also a trend towards higher prenatal knowledge scores among women who underwent redundant or simultaneous cfDNA and MMS (aOR 1.43 [95% CI 0.99–2.05], p= 0.06).

Discussion

In this cohort of women aged 35 and older seeking care at a single academic center in California, we found that a significant proportion of women underwent redundant screening

with cfDNA after receiving negative MMS results, or opted for simultaneous screening with both cfDNA and MMS. Women who underwent these non-recommended screening strategies scored higher on a pregnancy worry scale and were more likely to indicate that having a miscarriage would be worse for them than having a child with an undiagnosed intellectual disability. In addition, older women and nulliparous women also demonstrated a trend toward pursuing redundant or simultaneous cfDNA/MMS screening. While these strategies may provide additional reassurance that a pregnancy is low-risk for having common causes of intellectual disability, despite the fact that the second screening test does not contribute new information, our findings suggest that more targeted counseling regarding the advantages and disadvantages of both screening and diagnostic tests may be useful to help better align testing decisions with stated values, priorities, and concerns, as well as ACOG and SMFM recommendations for aneuploidy screening.

In addition to supporting prior research demonstrating that sociodemographic and attitudinal factors are associated with prenatal testing choices,[18–21] our study contributes to the limited published data on the prenatal screening choices being made in the era of cfDNA screening by women who will be 35 or older at the time of delivery. To be eligible for our study, participants had to have discussed prenatal aneuploidy screening options with a certified genetic counselor, consistent with SMFM, ACOG and NSGC's recommendations that prenatal screening, particularly cfDNA, should be offered only in the context of informed consent, education, and counseling by a qualified provider.[1,12–15] While the majority of the women in our study did undergo MMS screening, more than half of those who had MMS also opted to undergo cfDNA either after receiving negative MMS results or before they had even received their MMS results. We had hypothesized that women may have pursued redundant screening to receive additional reassurance from cfDNA because they had received first-trimester risk ratios higher than either the risk of miscarriage from invasive testing or their predicted age, had received abnormal ultrasound findings concerning for aneuploidy, or had received a combined risk ratio higher than their first trimester risk ratio. However, the majority of women who pursued redundant cfDNA screening did so without fitting into any of these scenarios.

It is important to note that women who underwent redundant or simultaneous cfDNA screening were more likely to strongly value the antenatal diagnosis of intellectual disabilities. Ultimately, these findings suggest that women may undergo further screening in the hope that additional screening will offer them comprehensive information regarding intellectual disability without their having to incur the risk of miscarriage. More focused counseling may be needed to clarify this misconception. Further qualitative research may allow a more nuanced understanding of the factors that motivate women to pursue dual screening.

Several limitations of our study deserve comment. First, our study population was composed of women presenting for care at a single academic prenatal diagnostic center in California, a state that has a comprehensive, formal program for prenatal screening utilizing MMS as well as universal health-care for pregnant women. Women who undergo prenatal genetic screening at UCSF either have private insurance (including integrated health plans) or are covered by Medi-Cal, California's Medicaid plan, and both cover cfDNA testing for women

aged 35 and older.[22,23] This is the likely explanation for the finding that women with an income more than $150,000 (who are likely to have private insurance, or to find the cost of the test, if it is not covered, less burdensome) or less than $50,000 (because they are more likely to have Medi-Cal coverage) were more likely to pursue simultaneous or redundant cfDNA testing.

The California Prenatal Program is unique in that it offers a unified approach to prenatal testing regardless of location, practice or insurance status. This unified approach and a desire by providers to support it may have encouraged patients and providers to view cell free DNA as an appropriate addition to the standard approach, rather than as an alternative of follow up test for screen positive patients as the guidelines recommend. As such, the high prevalence of combined cfDNA and MMS screening in our cohort may not be generalizable to women who live in states without such a program, particularly in settings where patients have to pay out-of-pocket for cfDNA or other prenatal tests.

Finally, the study participants may not be representative of the population of women in the US desiring prenatal testing: more than half of participants had a graduate or master's degree or had an annual household income of more than $150,000. More research is needed to determine the prenatal testing patterns among diverse groups of women.

Despite these limitations, to our knowledge, this study provides the first insight into both the prevalence of redundant or simultaneous cfDNA screening and the demographic and attitudinal predictors of these non-recommended screening algorithms. Though ACOG and SMFM have challenged obstetricians and perinatologists to avoid secondary screening with cfDNA or MMS after negative first-line screening,[24] our study suggests that dual aneuploidy screening may be a common clinical practice in some populations. Our findings not only have significant implications for the cost-effectiveness of prenatal screening, but underscore the need for more effective counselling on the part of obstetric providers and genetic counselors regarding the appropriate sequencing of these tests to avoid unnecessary testing.

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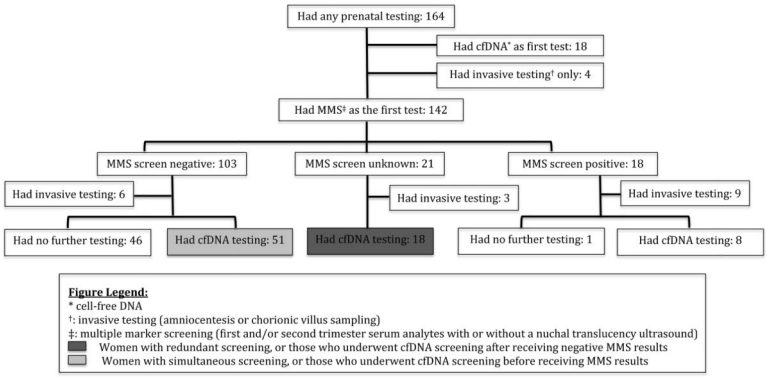

Figure 1.

Prenatal testing strategies of study participants

* cell-free DNA

†: invasive testing (amniocentesis or chorionic villus sampling)

‡: multiple marker screening (first and/or second trimester serum analytes with or without a nuchal translucency ultrasound)

Women with redundant screening, or those who underwent cfDNA screening after receiving negative MMS results

Women with simultaneous screening, or those who underwent cfDNA screening before receiving MMS results

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Table 1

Sociodemographic and attitudinal characteristics of women who underwent redundant or simultaneous cfDNA/MMS screening versus women who did not

	Total	Had redundant or simultaneous cfDNA screening	No redundant or simultaneous cfDNA screening	*p* value	Odds Ratio (95% CI)	p value	Adjusted Odds Ratio* (95% CI)	p value
Maternal age at due date (mean years (±SD))	37.7 (±2.2)	38.0 (±2.4)	37.3 (±2.0)	0.1	1.16 (0.97–1.38)	0.1	1.29 (1.00–1.65)	0.05
Nulliparous (n (%))	66 (56.9%)	45 (68.2%)	21 (31.8%)	0.05	2.14 (1.00–4.58)	0.05	2.92 (0.99–8.58)	0.05
Racial/Ethnic Group (n (%))				0.19				
African American/Black	6 (5.1%)	3 (50.0%)	3 (50.0%)		0.49 (0.09–2.61)	0.4	--	--
Asian	29 (24.8%)	16 (55.2%)	13 (44.8%)		0.60 (0.25–1.45)	0.26	--	--
Hispanic	9 (7.7%)	3 (33.3%)	6 (66.7%)		0.24 (0.06–1.06)	0.06	--	--
White	73 (62.4%)	49 (67.1%)	24 (32.9%)		Reference	--	--	--
Education (n (%))				0.09				
Less than college degree	14 (12.0%)	9 (64.3%)	5 (35.7%)		2.14 (0.59–7.68)	0.24	--	--
College degree	35 (29.9%)	16 (45.7%)	19 (54.3%)		Reference	--	--	--
Professional/Graduate degree	68 (58.1%)	46 (67.6%)	22 (32.4%)		2.48 (1.08–5.73)	0.03	--	--
Annual Household Income (n (%))				0.03				
<$50,000	12 (10.8%)	7 (58.3%)	5 (41.7%)		1.75 (0.47–6.57)	0.41	6.23 (1.06–36.78)	0.04
$50,001–149,999	36 (32.4%)	16 (44.4%)	20 (55.6%)		Reference	--		
>$150,000	63 (56.8%)	45 (71.4%)	18 (28.6%)		3.12 (1.33–7.35)	0.009	5.50 (1.49–20.36)	0.01
Knowledge Score[a] (mean scale (±SD))	79.8 (±16.5)	83.2 (±14.2)	74.6 (±18.4)	0.005	1.40 (1.09–1.81)	0.009	1.43 (0.99–2.05)	0.06
Worry Scale[b] (mean scale (±SD))	2.0 (±0.7)	2.1 (±0.7)	1.8 (±0.6)	0.02	2.00 (1.09–3.70)	0.03	3.16 (1.17–8.49)	0.02
Importance of knowing whether baby will be born with any chromosomal cause of intellectual disability (mean scale (±SD))	3.6 (±0.6)	3.8 (±0.4)	3.4 (±0.7)	0.001	3.63 (1.73–7.63)	<0.001	6.00 (1.82–19.77)	0.003
Importance of having test with low possibility of false positive result[d] (mean scale (±SD))	3.7 (±0.5)	3.8 (±0.5)	3.6 (±0.6)	0.05	2.04 (0.97–4.27)	0.06	--	--
Miscarriage worse than intellectual disability	43 (37.1%)	25 (58.1%)	18 (41.9%)	0.6	0.82 (0.38–1.76)	0.6	4.50 (1.08–18.85)	0.04
Undetected intellectual disability worse than false positive result	83 (72.8%)	57 (68.7%)	26 (31.3%)	0.02	2.66 (1.14–6.20)	0.02	2.47 (0.73–8.39)	0.15

*Adjusted for maternal race/ethnicity, education, and attitude towards having a false positive test result

[a] Knowledge: Total correct score of 20 items as 0–100 scale, with 100 as perfect score.

[b] Worry Scale: Average worry score of 6 items as 1–4 scale, with 4 as extremely worried.

[c] Importance of knowing whether fetus has Down Syndrome, classified on 1–4 scale, with higher scores denoting more importance.

[d] Importance of having a test with a low false positive rate, classified on 1–4 scale, with higher scores denoting more importance. Odds ratio and adjusted odds ratio calculated for every 1-point increment on a 4-point scale.

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