

OBJECTIVE AND METHODS

To describe the past medical history, sociodemographic and pregnancy characteristics of women at high risk for aneuploidy and to determine which factors are related to her choice of cell-free DNA (cfDNA) testing instead of invasive diagnostic testing.

We conducted a prospective descriptive study including pregnant women from the Western-Barcelona public health area at high risk for fetal aneuploidy, defined as a trisomy 21 or 18 risk between 1/10 and 1/250 at the combined first trimester or at the second trimester biochemical screening. During one year (December 2018 to November 2019) these women were asked to fill in a confidential questionnaire about her past medical history, demographic and pregnancy characteristics, and her opinion about termination of the pregnancy after a counseling consultation with a maternal-fetal medicine (MFM) specialist in which advantages and disadvantages of both testing methods, cell-free DNA (cfDNA) or diagnostic testing, were discussed. Logistic regression analysis was used to determine which factors were related with cfDNA uptake.

RESULTS

During the study period, 82 pregnant women filled the questionnaire. The median maternal age was 39.6 years (interquartile range (IQR) 37.3 - 40.9 years), and 73 (89%) of them were 35 or older. Forty-three (52%) women opted for cfDNA testing, while 39 (48%) chose invasive diagnosis. In a logistic regression analysis, the use of assisted reproductive techniques (OR 13.03; 95%CI: 1.47 to 115.56; $p = 0.021$) and Latin American origin (OR 6.66; 95%CI 1.73 to 25.66; $p = 0.006$) were shown to be related to a higher cfDNA uptake. In contrast, non religious women (OR 0.21; 95%CI: 0.06 to 0.72, $p = 0.013$) and a favorable opinion about termination of pregnancy (OR 0.23; 95%CI: 0.06 to 0.92, $p = 0.037$) were related with a lower uptake.

	Median (IQR); n (%)	
Maternal age (years)	39.6 (3.7)	
Origin	White European	56 (69%)
	Latin American	18 (22%)
	Non-White European, non-Latin American	8 (9.8%)
Educational level	Elementary	6 (7.3%)
	High school	10 (12%)
	Technical school	12 (15%)
	University	54 (66%)
Religion	Catholic	29 (36%)
	Muslim	2 (2.5%)
	No religious	19 (24%)
	Others	30 (38%)
Multiparous	56 (69%)	
Past history of miscarriages	32 (39%)	
Intended pregnancy	67 (85%)	
Use of assisted reproduction techniques	16 (20%)	
Favorable opinion about termination of pregnancy	67 (82%)	
Risk over 1 in 100 for T21/T18	23 (28%)	

Table 1. Baseline demographics of pregnant women at high risk for fetal aneuploidy included in the study. Continuous variables are shown as mean (standard deviation, sd) and categorical variables as number (n) and frequency (%).

	DNA uptake % (n)	Odds Ratio (95% CI)	p-value
Maternal age	-	0.94 (0.72 - 1.21)	0.613
Origin	White European	reference	-
	Latin American	6.66 (1.73 - 25.66)	0.006*
	Non-White European, non-Latin American	1 (0.20 - 4.89)	0.999
Educational level	Elementary	reference	-
	High school	0.73 (0.005 - 104.16)	0.91
	Technique school	0.093 (0.001 - 7.99)	0.296
	University	0.32 (0.05 - 18.81)	0.585
Religion	Catholic	reference	-
	No religious	0.21 (0.06-0.72)	0.013*
	Others	0.45 (0.16 - 1.30)	0.141
Multiparous	51 % (28)	2.14 (0.34 - 13.76)	0.420
Past history of miscarriages	65% (20)	2.31 (0.92 - 5.83)	0.075
Intended pregnancy	53% (35)	1.28 (0.15 - 11.2)	0.822
Use of assisted reproduction techniques	75% (12)	13.03 (1.47 - 115.56)	0.021*
Favorable opinion about termination of pregnancy	46% (31)	0.23 (0.06 - 0.92)	0.037*
Risk over 1/100 for T21/T18	35 % (8)	0.41 (0.08 - 2.03)	0.274
Total	52 % (43)		

Table 2. Determinants for cell-free DNA uptake. Data are shown as Odds ratio, 95% confidence interval and p-value. OR=Odds Ratio; ART = Assisted reproductive techniques.

CONCLUSION

Half of the pregnant women at high risk for fetal aneuploidy opted for cfDNA testing. The main reason to choose cfDNA was avoiding the risk of pregnancy loss. Women using assisted reproductive techniques and those of Latin-American origin preferred cfDNA testing, while non-religious women and those with a favorable opinion on termination pregnancy preferred invasive testing.

CONTACT

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