Impact of ethnicity and maternal age in the uptake of invasive prenatal testing for high-risk first trimester combined aneuploidy screening

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Objective
To investigate the impact of maternal ethnicity and age in the uptake of invasive prenatal testing for high-risk first trimester combined aneuploidy screening.

Methods
This is a population-based cohort study including all pregnant women undergoing first trimester combined aneuploidy screening in the Hospital General de l’Hospitalet’s reference area, between January 2018 and December 2021. Women at high-risk in the combined aneuploidy screening (<1/250) for trisomy 21, 13, and 18 were selected. Maternal age and ethnicity and data concerning the choice of prenatal testing (invasive, non-invasive prenatal testing or no further testing) were reviewed. In our setting, patients with screening results <1/10 (group 1) are offered invasive prenatal testing (chorion villous sampling (CVS)), patients with screening risks of 1/10 to 1/250 (group 2) are offered a choice between invasive genetic diagnosis or non-invasive prenatal testing according to their personal preferences.

Results
During the study period, 7026 pregnancies underwent first-trimester combined aneuploidy screening, of which 228 (3.2%) were at high-risk for trisomy 21, 13, or 18 (49 (0.7%) in group 1 and 179 (2.5%) in group 2. Mean maternal age was 36.3 (standard deviation 5.2) and ethnicity was distributed as follows: Caucasian/Hispanic 185 (77.4%), Maghrebis 28 (11.7%), Black 7 (2.9%), and Asian 6 (2.5%). There were no significant differences in maternal age or ethnicity distribution in groups 1 and 2 (p>0.05). Forty-one patients (83.7%) in group 1 underwent CVS while 8 (16.3%) refused the procedure. In group 2, 52 patients (29.1%) underwent CVS while the remaining 127 (70.9%) did not. Maternal age was not significantly different in patients undergoing invasive prenatal testing in both groups (group 1: CVS 34.3 (SD 5.9) vs no CVS 35.7 years (SD 6.0), p=0.535); group 2: CVS 35.8 (SD 5.2) vs. no CVS 37.2 years (SD 4.7), p=0.071). In group 1, but not in group 2, Maghrebi women were less likely to undergo invasive prenatal testing (OR 0.02 (confidence interval 0.003-0.195).

Conclusion
In our study, maternal age was not associated with the uptake of invasive diagnostic testing in patients at high risk of aneuploidy in the first-trimester combined screening. Maghrebi women at very high risk of aneuploidy (<1/10) were less likely to undergo an invasive procedure than women from other ethnicities.