

1- Introduction/objectives

The first trimester combined test allows the identification of cases with high or intermediate risk for trisomies 13, 18 and 21. In the presence of an intermediate risk, subsequent tests (namely cell-free DNA or an invasive test) are usually offered to these couples so that the result can be clarified.

The main goal of this paper was to analyse cases with an intermediate risk for trisomies on the first trimester combined test, namely the management and the outcomes of those pregnancies.

2- Methods

A retrospective analysis of all cases with an intermediate risk for trisomies on the first trimester combined test detected at our prenatal diagnostic centre between January and December 2022 (using a cut-off of $\geq 1:200$ and $< 1:1000$).

3- Results

During 2022, we obtain a total of 50 cases with an intermediate risk for trisomies, which corresponded to 4.3% of the first trimester combined tests performed at our centre during this year.

The mean maternal age was 34.5 years (SD = 5.49) and the mean gestational age at screening date was 13.1 weeks (SD = 0.65). All pregnant women evaluated were caucasian and only 14% of them were smokers. Just 2% of the evaluated pregnancies originated from medically assisted procreation techniques.

The average value of beta-hCG obtained was 1.37 MoM and average PAPP-A value was 0.71 MoM. The mean value of nuchal translucency was 1.70 mm and the mean fetal heart rate was 160 bpm – Table 1.

In 12% of cases nasal bones were absent and 4% of cases had an abnormal ductus venosus flow velocity, but all cases had an a-wave present.

In 86% of cases analysed a subsequent cell-free DNA test was done, of which only one case showed a high risk of aneuploidy, later confirmed with karyotype obtained by invasive testing. In the remaining 14%, an invasive test was initially done by maternal choice, confirming a normal karyotype in all these cases.

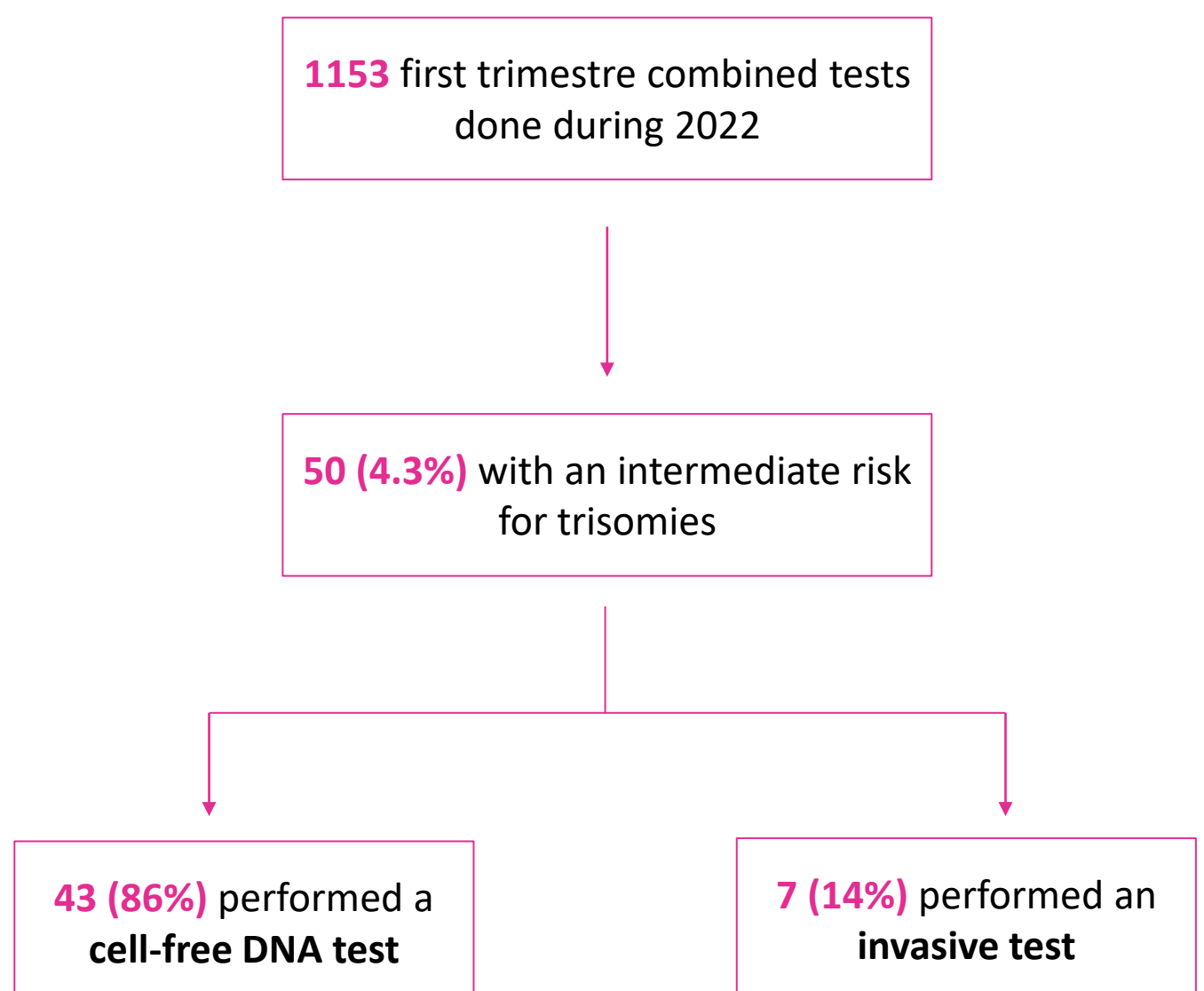


Figure 1. First trimester combined tests with an intermediate risk for trisomies performed during 2022.

	Mean (SD)
Maternal age (years)	34.54 (5.49)
Mean gestational age at screening date (weeks)	13.1 (0.65)
Beta-HCG (MoM)	1.37 (1.04)
PAPP-A (MoM)	0.71 (0.41)
Nuchal translucency (mm)	1.7 (0.47)
Fetal heart rate (bpm)	160 (8.02)

Table 1. Mean and standard deviation (SD) of some variables analysed.

4- Conclusions

In most cases in which the first trimester combined test reveals an intermediate risk for trisomies, there is no aneuploidy. With the current growth in cell-free DNA testing, it is possible to reduce the number of invasive tests performed, as well as the risks associated with them.