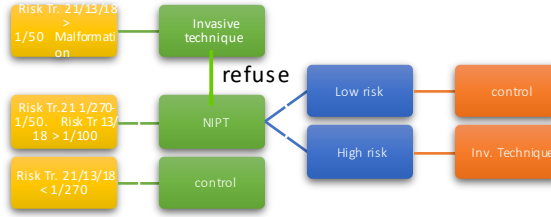
Efficacy of non-invasive prenatal test (NIPT) as a screening method in our hospital

Martínez-Hernández AI, Leal-Benavent A, Higueras-García G, Gil-Raga F.
Manises Hospital. Valencia. Spain

Objective: To assess the indications for NIPT, patient characteristics and results.

Method: In our hospital, NIPT was implemented in January 2021 as a test to be performed in patients whose combined risk in 1st trimester screening was between 1/50 and 1/270 for Trisomy 21 and >1/100 for Trisomy 13 and 18, without any ultrasound malformation. Patients with a risk > 1/50 of Trisomy 21 were offered an invasive technique as the first option and, those who refused it, NIPT was offered as a second option.



Pregnant women who had not undergone the 1st trimester screening at the time were offered the 2nd trimester screening, with the same indications for the NIPT according to the results obtained. The epidemiological characteristics of the patients to whom NIPT was indicated, from January 2021 to April 2023, the test indication and the result were analyzed.

Results: A total of 124 patients were analyzed. 17% of the patients who underwent NIPT were ≥ 40 years, with a mean age of 37.7 years. 7 patients obtained a combined risk of Trisomy 21 > 1/50 and refused an invasive test, therefore, NIPT was performed. Only one of them obtained high risk of Trisomy 21 as a result. 8 patients obtained a high risk of Trisomy 21 in screening of the 2nd trimester, calculating it based on AFP and total HCG. In 35% of the patients, the PAPP-A was less than 0.4 MoM and the BHCG was > 2.0 MoM in only 2.6%. 6 patients underwent NIPT due to high risk of Trisomy 18 and 13 and two more, due to high risk of the three aneuploidies studied. All of them obtained a low risk result in the NIPT. Of the 124 NIPTs analyzed, only 2 resulted in a high risk of Trisomy 21, which a later confirmed by amniocentesis. One of the patients, 45 years old, obtained a combined risk of Trisomy 21 of 1/78 and the other, 42 years old, > 1/50. Both, with a normal NT in ultrasound of 12 weeks and a normal BHCG and PAPP-A.

Conclusion: Thanks to the NIPT, an invasive technique in 122 women was avoided, with the consequent risk of abortion, therefore, we can affirm that the NIPT is an effective test to avoid invasive techniques in pregnant women who, not observing any ultrasound malformation in the screening at 1st trimester, obtain a risk of aneuploidy high enough to carry out additional studies.