

EFFICACY OF OUR CONTINGENT ANEUPLOIDY TEST IN RELATION TO THE UNIVERSAL NIPT TEST

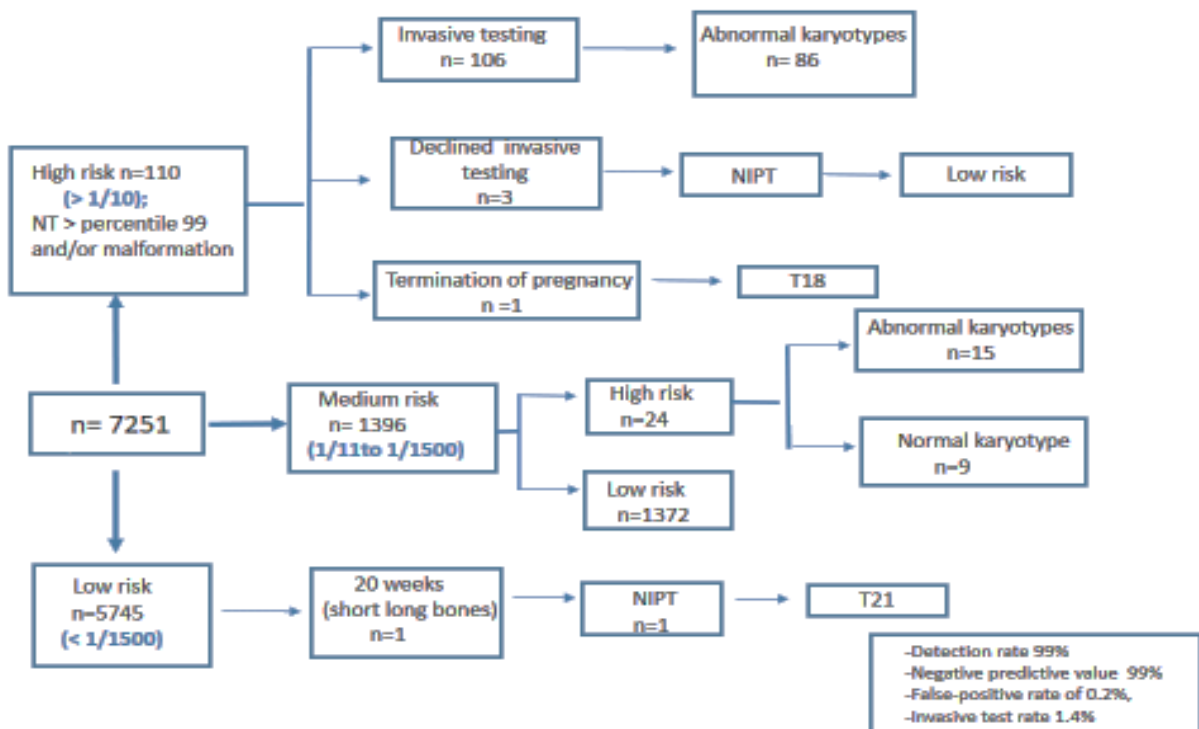
Loscalzo G, Izquierdo-Puchol M.T, Martinez-Hernandez A.I

Hospital Vithas Valencia 9 de Octubre, Valencia, Spain

Objective: To evaluate the efficacy of our NIPT contingent screening at our centre and compare it with the efficacy of universal NIPT screening.

Methods: This is a retrospective observational study in which the results of 7.251 screening tests performed at the Hospital Vithas Valencia 9 de Octubre (Spain) were evaluated and followed up postnatally. For risk calculation we combined in all cases maternal factors, ultrasound markers (NT, NB, DVPIV, FHR, Tricuspid flow) and biochemical factors (PAPP-A and free beta HCG). We grouped the patients into three categories: **High risk (> 1/10), NT >percentile 99 and/or malformation), Medium risk (1/11 to 1/1500) and Low risk (< 1/1500).** For high-risk patients we indicated invasive technique (chorionic biopsy), for medium-risk patients we indicated NIPT in maternal blood, and for those at low risk, ultrasound in week 20.

Results: Of the 7251 patients: 110 were high risk (0.01%), we performed chorionic biopsy on 106, obtaining 86 pathological cases, 3 rejected invasive test and opted for NIPT with a normal result and 1 was a malformation who opted for termination of pregnancy (Trisomy 18). 1396 patients were medium risk (19,2%) and underwent NIPT, of which 1372 were low risk and 24 were high risk and invasive test was indicated of which 15 were abnormal karyotype and 9 were normal karyotype. Of the remaining 5745 patients considered low risk (79,2%), only 1 was assessed at week 20 as suspicious for pathology (short long bones) with indication of invasive test and result of trisomy 21. Finally the detection rate (DR), invasive technique rate (IR) and false positive rate (FPR) (DR T21:99% IR:1.4% FPR:0.2%) were similar to that described in the literature with the implementation of universal NIPT (DR T21: 99.2% IR 1 % and FPR 0.3%).



Conclusion: Our contingent test for first trimester aneuploidy screening proves to be cost-effective compared to universal screening with NIPT.