NON-INVASIVE PRENATAL TESTING IN THE SCREENING OF THE MOST COMMON ANEUPLOIDIES: OUR EXPERIENCE

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INTRODUCTION

Prenatal diagnosis of chromosomopathies is an indispensable part of prenatal care and is performed in the first and second trimesters of pregnancy. It includes a range of invasive and non-invasive methods. Non-invasive screening methods are used to determine the risk of aneuploidies in the fetus, while invasive methods can be used to diagnostically confirm chromosomal abnormalities. A noninvasive method of prenatal screening that can reliably determine the risk of the most common chromosomal aneuploidies is noninvasive prenatal testing of cell-free circulating fetal DNA from maternal blood (NIPT).

The aim of this study was to present the results of NIPT analyses performed at the Department of Medical Biology and Genetics, Faculty of Medicine, University of Rijeka, in the period from January 2019 to August 2022, and to compare the obtained results with karyotype analyses.

RESULTS

During thie analysed period, NIPT analyses of 80 pregnant women were performed, with a significant annual increase in the number of subjects.

No statistically significant association was found between the proportion of fetal fraction and gestational week, body mass index, and maternal age.

NIPT was an independent indication for amniotic fluid cytogenetic analysis in 49% of cases, followed by a combination of NIPT and ultrasound and a combination of NIPT and maternal age. The combination of NIPT and ultrasound had the highest accuracy in assessing the risk of fetal aneuploidy.

NIPT analysis was the most reliable in assessing the risk for trisomy 21 while in assessing sex chromosome aneuploidies, it was less reliable. Altogether, invasive methods continue to be the basis for the diagnosis of chromosomal abnormalities in the prenatal period.

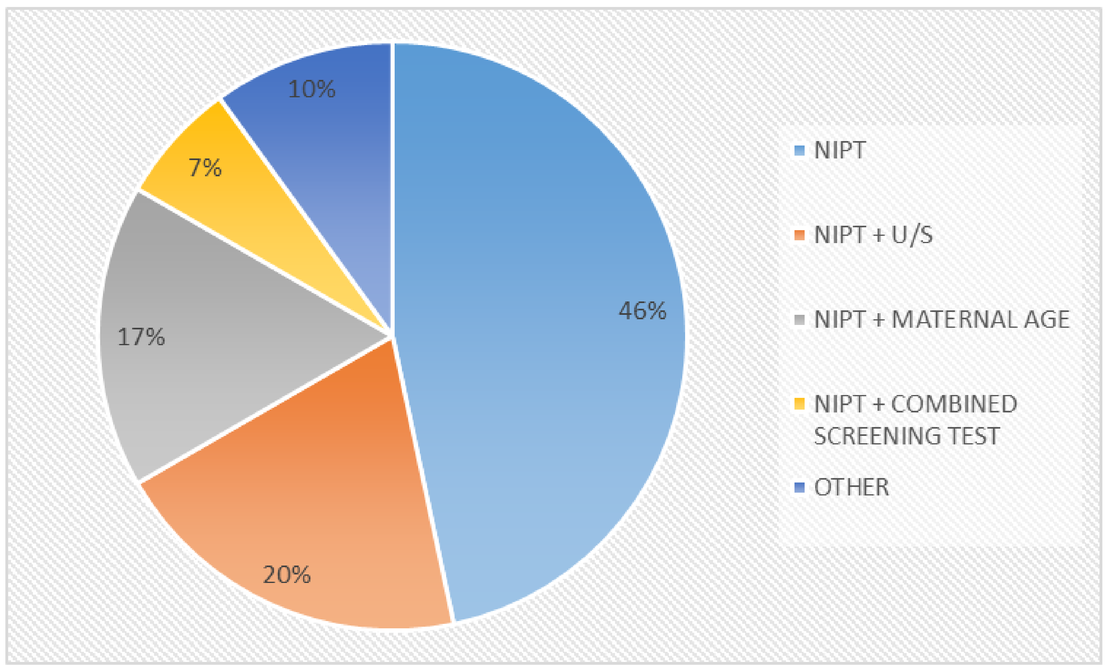

Figure 1. indications for amniotic fluid cytogenetic analysis.

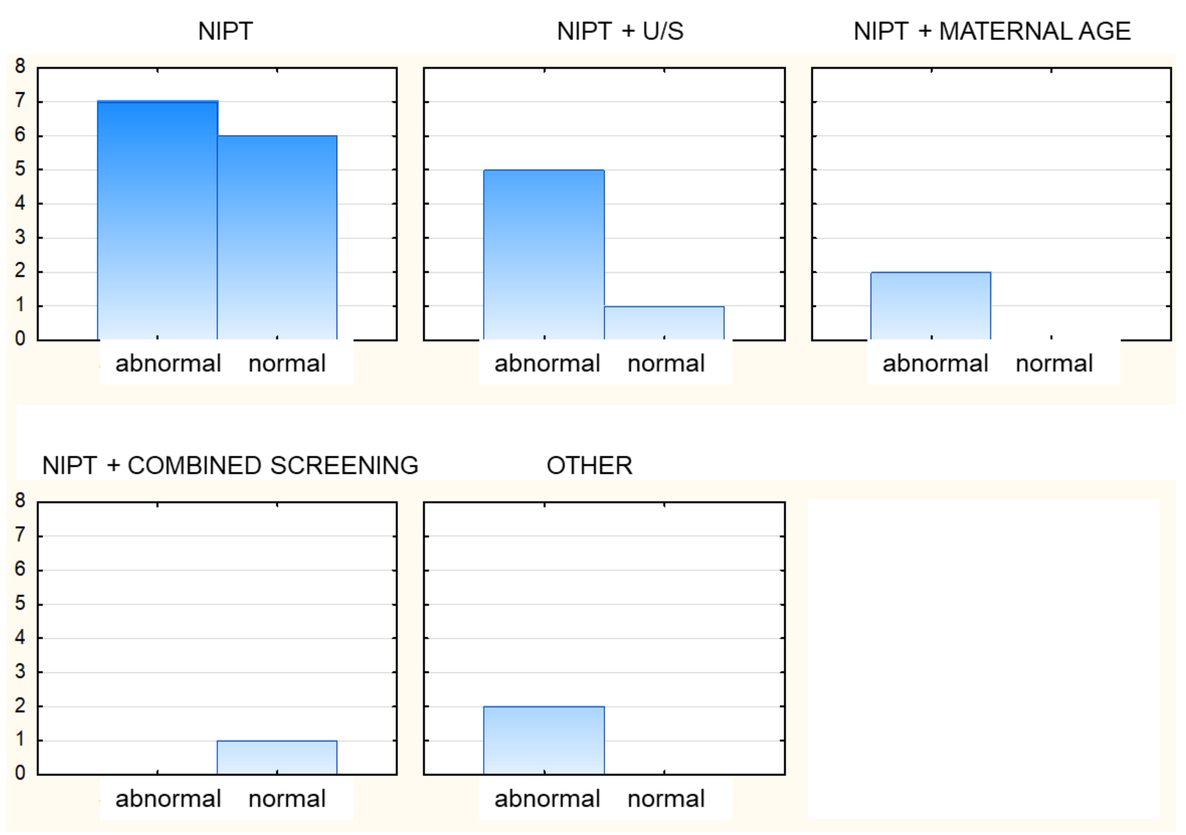

Figure 2. Accuracy in assessing the risk of fetal aneuploidy.

CONCLUSION

NIPT ANALYSES WERE MOST RELIABLE IN ASSESSING RISK FOR TRISOMY 21, ESPECIALLY IN A COMBINATION WITH ULTRASOUND, WHEREAS IT WAS LESS RELIABLE IN ASSESSING SEX CHROMOSOME ANEUPLOIDIES.

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