INTRODUCTION

The development of the genetic non-invasive prenatal tests (NIPT) has been an issue during the last years all over the world, in order to reduce the number of invasive investigations for fetal aneuploidies.

OBJECTIVES

The purpose of this study was to review the efficiency and limits of the genetic non-invasive prenatal tests, analyzing the ratio of the prenatal detection of fetal aneuploidies using different types of NIPTs.

MATERIAL AND METHOD

We performed a systematic research of relevant databases, using the MeSH words: noninvasive prenatal tests, DANSR, NIPT, cfDNA. Out of the 120 found papers, 23 presented data on the prenatal detection rate of the fetal aneuploidies.

RESULTS AND DISCUSSIONS

By combining the data of the 23 studies included in the study, the testing of 52486 patients have been provided, of which 833 with Down syndrome, 255 with Edwards syndrome, 54 with Patau syndrome and 59 with aneuploidies of the sexual chromosomes. When compared, the NIPT tests presented several advantages due to the direct analysis of the DNA fragments specific for certain chromosomes. The results of this study also indicate higher efficiency of NIPT in detecting the Down syndrome compared to other aneuploidies, specifically:

- trisomy 21 - sensitivity 98.56%, specificity 99.97%;
- trisomy 18 - sensitivity 96.86%, specificity 99.96%,
- trisomy 13 - sensitivity 87.04%, specificity 99.98%

CONCLUSIONS

The use of the non-invasive prenatal tests promises an encouraging accuracy and precision, the sensitivity and specificity in prenatal detection of the fetal aneuploidies being superior to the known screening tests. The limits of NIPT are that, even if these tests are accepted as very good screening methods, the invasive interventions are required to be performed in all cases with high risk and abnormal results.