17th World Congress in Fetal Medicine

NIPT is it a key of prenatal diagnosis or is a complement?

Granja MP
Hospital da Luz, Porto, Portugal

Objective

The changing landscape of prenatal screening. There are always challenges when integrating new and established screening and testing options into prenatal practice.

Methods

NIPT currently detects almost 75% of chromosomal abnormalities identified from invasive prenatal diagnostic testing, with a residual risk of abnormal karyotype of 1, 9% after a negative NIPT result. This residual risk would have been higher if prenatal samples have been analyzed using array technology. Limited data are available regarding NIPT detection rate of microdeletions, less common trisomies and mosaicism.

Results

The use of prenatal diagnosis tests is a choice closely related to the perception of the two types of risk: that of the invasive procedure - related miscarriage and that of carrying a Down Syndrome fetus.

Conclusion

Acceptability rather than the numerical relevance affects the interpretation of a given risk. A key role of caregiver during prenatal counselling is to facilitate a clear understanding of the different risks facing the patient and to incorporate their perception into the decision - making process.