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CONCORDANCE OF NIPT AND AMNIOTIC FLUID CELL KARYOTYPING RESULTS IN THE PRENATAL SCREENING OF FETAL CHROMOSOMAL ABNORMALITIES

Background. NIPT is a highly sensitive screening method for fetal chromosomal aneuploidies. According to ACOG and ISPD guidelines, invasive diagnostic testing, such as amniocentesis followed by amniotic fluid cell karyotyping, remains the "gold standard" for verifying fetal chromosomal abnormalities. This study compares amniotic fluid karyotyping with prior NIPT results to evaluate their concordance. **Methods.** This retrospective study included 124 clinical cases collected over a 7-year period (2020–2026) at Nadiya Reproductive Medicine Clinic following a prior NIPT result. In 122 cases, conventional amniotic fluid cell karyotyping was performed; in the remaining 2 cases, cytogenetic analysis was performed on products of conception following missed miscarriage, prior to planned amniocentesis. NIPT was conducted using the Sage Prenatal Screen kit (Yourgene Health) with sequencing on the Ion S5 Plus platform (Thermo Fisher Scientific) in 76 cases. The remaining 48 cases had undergone NIPT at external laboratories in Ukraine and the EU. Conventional cytogenetic analysis of amniotic fluid cells was performed using standard GTG-banding protocols. **Results.** Karyotype analysis confirmed fetal trisomy 21 in 90.1% of cases with a high-risk NIPT result. In contrast, none of the suspected cases of trisomy 16 ($n = 3$) or monosomy X ($n = 7$) were confirmed by invasive prenatal diagnosis, possibly reflecting confined placental mosaicism. Among the 122 pregnancies evaluated by amniotic fluid cell karyotyping, complete concordance with normal fetal karyotype was observed in low-risk NIPT cases, with two exceptions: one case of triploidy (69,XXY) associated with fetal hydrops detected by ultrasound, and one case of trisomy 21 in the absence of abnormal ultrasound findings. Separately, in 2 additional cases with a low-risk NIPT result, cytogenetic analysis of products of conception revealed trisomy 15; these cases were not included in the amniotic fluid concordance analysis. **Conclusions.** The highest concordance between NIPT and invasive cytogenetic findings was observed for normal fetal karyotypes and trisomy 21 (90.1%). In contrast, positive NIPT results for certain chromosomal abnormalities, particularly trisomy 16 and monosomy X, were not confirmed by invasive testing. These findings highlight the necessity of confirming all positive NIPT results by invasive prenatal diagnostic testing, underscoring the residual risk of false-negative NIPT results.

Keywords: NIPT; next-generation sequencing.

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