

# Discordance between karyotype from amniotic fluid and postnatal lymphocyte cultures

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## Summary

**Discordance between karyotype seen from amniocentesis and from neonatal blood is a very unusual condition with different possible causes.**

**We present a case of discordance between prenatal cytogenetic diagnosis from amniotic fluid and postnatal cytogenetic diagnosis from lymphocyte cultures.**

*Key words: amniocentesis, karyotype, array-CGH, lymphocyte cultures.*

## Introduction

Discordance between karyotype seen from amniocentesis and from neonatal blood is a very unusual condition reported in scientific literature (1-5).

## Case report

A 38 year old patient, at her second pregnancy undergone to genetic amniocentesis at 16th week of gestation because of high risk for fetal aneuploidies obtained at combined (nuchal translucency and bi-test) first trimester screening (risk of trisomy 21 1:45). Her past medical history was negative and her previous pregnancy was uneventful.

The cytogenetic analysis was carried out on cells withdrawn from amniotic fluid, evidencing a perfectly normal female karyotype (46 XX) throughout the entire examination.

The anomaly scan at 21 weeks and the growth scan at 31 weeks of gestation were normal and not morphological or auxological anomalies were noted. The baby was delivered at term of gestation.

After birth the child presented a series of congenital de-

fects (poor growth, polycythemia, hip dysplasia, laryngeal stridor) and a new karyotype study was performed on the peripheral blood of lymphocyte cultures at the pediatric hospital.

These examinations, contrary to those performed on amniotic fluid, revealed a duplication on the arm length of the 11 chromosome.

The anomaly was then confirmed by the genomic array-CGH technique, distributed in homogeneous form in the peripheral blood, extended to approximately 20 Mb. A blinded metaphasic evaluation, both of the amniotic fluid and that of the peripheral neonatal blood, was carried out by a third geneticist. In this case also, a difference was confirmed between the two karyotypes regarding to the anomaly verified.

All internal laboratorial controls have permitted the exclusion of two hypothesis of an exchange of samples and of contamination of the maternal cells.

## Discussion

The inconsistency between the results of the metaphasic examinations from amniocyte and lymphocyte cultures can only derive from a scarce visibility of the presence of abnormal chromosomes in the amniocytes.

As a conclusion, based on these differences, there are several possibilities that can be considered, as previously reported in scientific literature:

- Clonal selection in amniocyte cultures not permitting abnormal identification (1,4);
- Post-zygotic anomaly not visible in homogeneous form in all fetal tissues (2,3);
- Post-zygotic anomaly not visible in extra-embryonic material (5).

## Conclusion

This case confirms the possibility that chromosomal abnormalities cannot always be detected in amniotic fluid and therefore are not correctly diagnosed in the prenatal stage during the amniocentesis.

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