

The unprecedented recurrence of mosaic conjunction of tetraploidy and trisomy 18

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Objective

To report a rare case of mosaic tetraploidy (diploid/tetraploid mixoploidy).

Methods

Case report.

Results

A 32 years old third gravida attended our perinatology outpatient clinic at 14 weeks' gestation for further perinatal and genetic counseling due to previous history of fetus affected by chromosomal abnormality. The ultrasound examination showed single atrium and ventricle in the fetal heart, pes equinovarus, clenched hands and fetal hydrops, findings, which were similar with the findings of the previous medically aborted pregnancy. The parents opted for termination of the pregnancy. Karyotyping of the whole family was performed and was normal for the parents and their first healthy child. The postmortem fetal karyotype showed mosaic conjunction of tetraploidy and trisomy 18 ($4n+18/2n+18$), an uncommon type of mixoploidy and these findings were consistent with the genetical results of the previous performed chorionic villus sampling. The result was identical with the karyotype of the previous aborted fetus.

Conclusion

Mosaic tetraploidy (diploid/tetraploid mixoploidy) is a very rare clinical condition characterized with diminished survival rates and different congenital malformations. Mosaic conjunction of tetraploidy and trisomy 18 ($4n+18/2n+18$) was reported uniquely by Atnip et al in 1971. No similar case with recurrence has ever been reported until now.

