Spectrum of structural abnormalities in the first trimester diandric and digynic triploid pregnancies

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Objective
The objective of the study was to analyse spectrum of the structural abnormalities diagnosed in the first trimester triploid pregnancies in regard to the parental origin of triploidy (diandric or digynic).

Methods
A retrospective study of the triploid pregnancies diagnosed in the first trimester in a single ultrasound reference centre in the years 2017-2021. Analysis of the ultrasound results and results of the molecular testing, including testing for parental origin of triploidy by the use Quantitative Fluorescent Polymerase Chain Reaction (QF-PCR).

Results
Sixteen triploid pregnancies were included to the study at the mean gestational age at diagnosis of 13.2 weeks. Nine pregnancies had diandric origin (56.2%) and seven were digynic (43.8%). Cystic placenta occurred in all 9 diandric cases (in one of them in association with the giant chorioangiomas of the placenta) and non of 7 digynic ones, whereas asymmetrical fetal growth restriction was diagnosed in the first trimester in all digynic cases and non of diandric ones. In all diandric fetuses nuchal translucency (NT) was enlarged (beetwen 3.4 and 10 mm) and in 3 cases – fetal oedema was diagnosed (33.3%). NT in all digynic fetuses was within normal limits. Structural defects were detected in 12 of first trimester triploid pregnancies (75%; 12/16) (66.6% of diandric and 85.7% of digynic ones). The most frequent were heart abnormalities (56.2%; 9/16) and central nervous system abnormalities (37.5%; 6/16) including the posterior fossa defects - four cases (25%; 4/16) and holoprosencephaly – two cases (12.5%; 2/16). Omphalocele was detected in four diandric cases (25%; 4/16). Other defects included abnormalities of the extremities (18.7%; 3/16), cleft lip (6.2%; 1/16) and spina bifida (6.2%; 1/16).

Conclusion
Spectrum of the structural abnormalities diagnosed in the first trimester triploid pregnancies is very heterogenous and non-characteristic for the parental origin of triploidy, except for the omphalocele, which occurs in diandric cases, but not digynic ones. However, the phenotype of diandric and digynic triploidy is different and these differences may be visualized as early as in the first trimester of pregnancy. Enlarged NT and cystic placenta occur in diandric triploidy, whereas asymmetrical fetal growth restriction and NT within normal limits occur in digynic triploidy.