THE ROLE OF ULTRASOUND EXAMINATION IN THE DETECTION OF THE MOST COMMON TRISOMIES

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Objectives: The aim of this study was to retrospectively analyze the most often prenatally diagnosed autosomal trisomies, and to identify the role of ultrasound examination in the diagnostic workup.

Methods: Between 2011 and 2018, a total of 1706 diagnostic amniocenteses have been performed, the study included a group of 128 women with singleton pregnancies and confirmed fetal trisomy. The study group was divided into two subgroups, with abnormal ultrasound markers (N=105) and without (N=23).

Results: Based on the outcome of amniocentesis, 88 fetuses were diagnosed with trisomy 21, 29 with trisomy 18 and 11 with trisomy 13. Women with abnormal ultrasound markers and without differed significantly in terms of their age and gravidity (p<0.01). Abnormal ultrasound markers were present significantly more often in younger women and those with lower gravidity. Trisomy 21 was shown to be significantly more often associated with ultrasonographic evidence of increased NT and fetal hydrops, Trisomy 18 with omphalocele and CNS anomalies (ventriculomegaly, cerebellar abnormalities, holoprosencephaly); moreover, both Trisomy 13 and 18 significantly more often coexisted with other anomalies. Among the indications for prenatal testing, i.e. maternal age, obstetrical history and presence of abnormal ultrasound markers, only the latter was significantly associated with ultrasound an abnormal karyotype (p<0.01). Among the indications for invasive prenatal testing, significant associations with the presence of abnormal karyotype were found for an abnormal result of the first trimester combined test and the presence of abnormal ultrasound markers (p<0.01).

Conclusions: Based on the analysis of diagnosed cases of trisomy, it can be concluded unequivocally that ultrasound constitutes a vital component of the diagnostic workup, especially in younger women. Our study confirmed that the first trimester combined test still constitutes an essential element of screening for aneuploidy and should be recommended to all pregnant women, also those without increased risk of trisomy.