EP05.09  First trimester diagnosis of cystic hygroma by transvaginal ultrasound and cytogenetic evaluation

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Objective
The purpose of this article was to evaluate the association between fetal cystic hygroma detected in the first trimester of pregnancy and cytogenetic abnormalities, and the long-term prognosis.

Methods
We studied the outcome of fetuses in whom cystic hygroma was diagnosed in the first and early second-trimester of pregnancy using transabdominal and transvaginal ultrasonography (3D/4D ultrasonography). 357 consecutive fetuses between 8,0 and 14,0 weeks of gestation diagnosed as having a nuchal hygroma were evaluated ultrasonographically and karyotyped with transabdominal chorionic vilus sampling. Those with a normal chromosome complement were ultrasonographically monitored throughout the remainder of the pregnancy to document the resolution of the hygroma.

Results
168 (48%) fetuses were found to have a normal karyotype and 65 of these were aborted electively. The hygromas resolved in 78 of these karyotypically normal foetuses within four weeks of initial diagnosis and they were phenotypically normal at birth. 189 (52.0%) fetuses were karyotypically abnormal with Trisomy 21 being the most common abnormality. Out of 115 (32,27%) fetuses that had septated cystic hygroma, 105 (91,3%) of them were with pathological karyotype.

Conclusion
Prenatal cytogenetic analysis should be offered to women with fetal cystic hygroma diagnosed in the first trimester. A normal outcome is likely in those without chromosome abnormalities.