



## First trimester diagnosis of cystic hygromata 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 transvaginal ultrasonography (3D/4D ultrasonography). 303 consecutive fetuses between 8, 0 and 14, 0 weeks of gestation diagnosed as having a nuchal hygroma were evaluated ultrasonographically (3D/4D ultrasonography) 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

148 (48, 9%) of 303 fetuses were found to have a normal karyotype and fifty five of these were aborted electively. The hygromas resolved in 68 of these karyotypically normal fetuses within four weeks of initial diagnosis and they were phenotypically normal at birth. 155 (51, 9%) fetuses were karyotypically abnormal with trisomy twenty-one being the most common abnormality. 105 (34, 7%) fetuses had septated cystic hygroma, 84 (80, 1%) of them 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.