



First trimester diagnosis of cystic hygroma by transvaginal 3D/4D 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 205 fetuses in whom cystic hygroma was diagnosed in the first and early second-trimester of pregnancy using transvaginal sonography (3D/4D ultrasonography). Two hundred and five consecutive fetuses between 8.0 and 13.0 weeks of gestation diagnosed as having a nuchal hygroma were evaluated ultrasonographically and karyotyped. Those with a normal chromosome complement were ultrasonographically monitored thorough the remainder of the pregnancy to document the resolution of the hygroma.

Results

125 (60.9%) of the 205 fetuses were found to have a normal karyotype and twenty five of these were electively aborted. The hygromas resolved in 45 of these karyotypically normal fetuses within four weeks of initial diagnosis and they were phenotypically normal at birth. 80 (39.1%) fetuses were karyotypically abnormal with trisomy 21 being the most common abnormality. 52 fetuses had septated cystic hygroma, 45 of them with pathological karyotype.

Conclusion

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