Objective
To evaluate the association between fetal cystic hygroma detected in the first trimester of pregnancy, 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 ultrasound (3D/4D). 295 consecutive fetuses between 8 and 14 weeks of gestation diagnosed with nuchal hygroma were evaluated 3D/4D ultrasound and karyotyped with transabdominal chorionic villus sampling. Those with a normal chromosomal complement were scanned regularly monitored for the rest of the pregnancy to document the resolution of the hygroma.

Results
145 (48.5%) of 295 fetuses were found to have a normal karyotype and 45 of these were aborted electively. The hygromas resolved in 58 of these karyotypically normal fetuses within four weeks of initial diagnosis and they were phenotypically normal at birth. 150 (51.5%) fetuses were karyotypically abnormal with trisomy twentyone being the most common abnormality. 95 (63.3%) fetuses had septated cystic hygroma, 68 (71.6%) 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 chromosomal abnormalities.