Objective
To report a case of oligohydramnios where the fetal karyotype was carried from the cystic hygroma puncture.

Methods
To describe a case with literature review.

Results
The pregnant woman was a healthy female of 38 years who came to the fetal medicine unit at 12 weeks of gestation. She was in her third pregnancy and had a previous history of two pregnancies losses (all occurred in the first trimester of pregnancy). The current gestation had no history of vaginal bleeding. The patient denied smoking, alcohol intake or use of illegal drugs. Her husband was a 42-year-old healthy and non-consanguineous man. The patient had first trimester screening with increased nuchal translucency measurement (7 mm), presence of nasal bone, cervical cystic hygroma and subcutaneous edema of the head and trunk. Ascites was also evident later. The amniotic fluid was low (oligohydramnios), which prevented the execution of amniocentesis. Thus, at 18 weeks of pregnancy, in utero puncture of the cystic hygroma was performed for fetal karyotyping (it was removed 20 mL). Chromosomal analysis showed a X-chromosome monosomy (45, X), which was consistent with the diagnosis of Turner syndrome. On the sonographic evaluation, at 21 weeks of pregnancy, there was no evidence of fetal heartbeat. Spontaneous miscarriage of the fetus occurred two days later. The pathologic evaluation showed that it was female and had no major malformations. The fetal weight was 90 grams.

Conclusion
Pregnancies with fetal cystic hygroma are often associated with oligohydramnios and intrauterine fetal death, which can hamper the genetic evaluation through certain procedures, such as amniocentesis. Our case emphasizes the possibility of performing fetal karyotyping by puncturing the cyst hygroma in cases where procedures such as amniocentesis are not possible. It was essential for diagnostic definition of the fetus and hence correct genetic counselling for the family.