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CASE REPORT

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Fetal cystic hygroma with nonimmune hydrops fetalis and prenatal diagnosis of Turner syndrome

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Introduction

Fetal cystic hygroma is a manifestation of early lymphatic obstruction. It is mostly associated with nonimmune hydrops fetalis (NIHF). Since the advent of ultrasonography, more and more cases of cystic hygroma and fetal hydrops are being detected. Fetal hydrops and cystic hygroma can be diagnosed in the second trimester of gestation and is associated with a higher incidence of aneuploidy, and it has a high mortality. At times it becomes difficult to predict the outcome of such hydropic fetuses. The fetal outcome and the genetic counseling entirely depend upon the exact etiology of such hydrops. We report a case of cystic hygroma with NIHF where aneuploidy was detected on prenatal diagnosis.

Case report

A 29 year old second gravida was referred to our genetic center as a case of NIHF detected on routine obstetric sonography. There was no history of malformation, recurrent spontaneous abortion, bad obstetric outcome and hematological or genetic disorder in the family. High resolution ultrasonography for malformations was performed and the fetus was found to have a large cystic hygroma, bilateral pleural effusion, pericardial effusion, ascites, and polyhydramnios. Fetal prognosis and possibility of chromosomal disorder, single gene disorder, malformation, and infection were explained to the woman. Amniocentesis and karyotyping were done and the karyotype was 45 X, that is monosomy for X chromosome or Turner syndrome. Pregnancy was terminated and the fetus subjected to autopsy and karyotyping.

At autopsy a female fetus of 16 weeks gestational age with large multiloculated cystic hygroma was seen. Severe generalized edema with gross edema over hands and ankles was present. Upper and lower limb shortening with discrepancy of 2 weeks from gestational age was present. Facial dysmorphism in the form of low set ears, hypertelorism, depressed nasal bridge, and micrognathia was apparent (Figure 1).

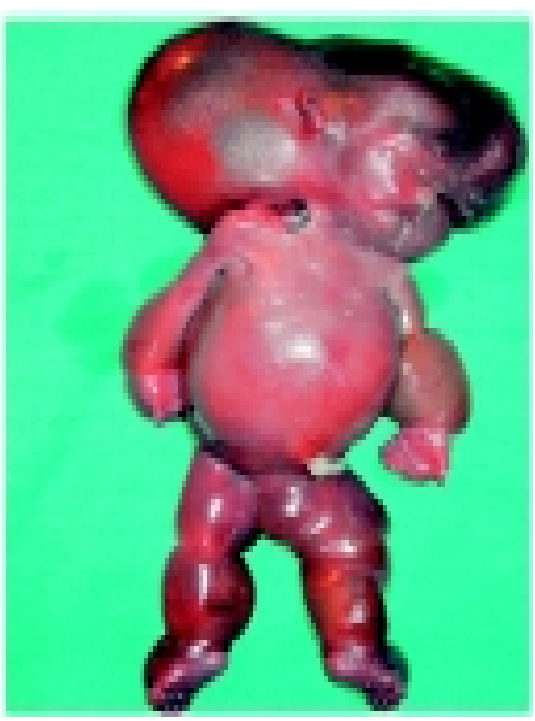

Figure 1. Fetus with Turner syndrome showing large cystic hygroma and severe generalized edema

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Internal examination revealed bilateral pleural effusion with hypoplasia of the lungs, pericardial effusion and severe ascites. Both kidneys were fused to form a horseshoe shaped multicystic kidney in the right side of the pelvis. There was no visceral displacement. Liver, spleen and heart were normal. TORCH test on cord blood was negative. Skiagram of fetus showed no evidence of skeletal dysplasia. Histopathological examination of the kidney showed renal dysplasia. Histopathological examination of placenta was normal.

The karyotype analysis was performed on cord blood, placenta, cystic hygroma fluid, and fetal skin. All the tissues showed 45 XO karyotype, which is monosomy of X chromosome (Figure 2). A final diagnosis of Turner's syndrome was made and counseling was done to explain the risk of recurrence and need for follow up in the next pregnancy.

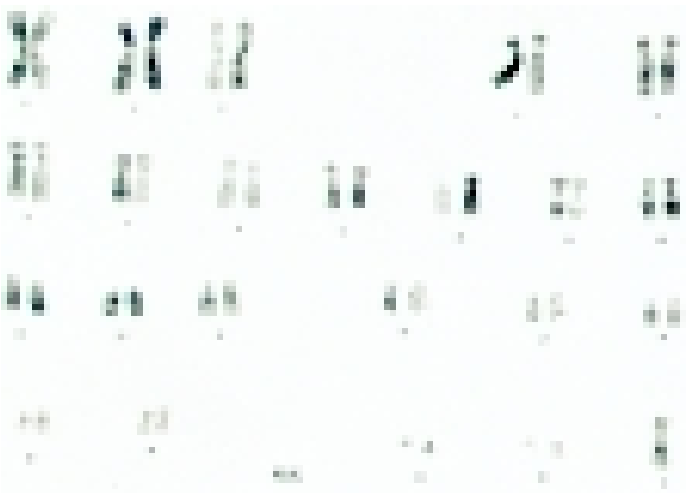

Figure 2. Karyotype showing 45 X, monosomy, in amniotic fluid.

Discussion

Cystic hygroma belongs to a group of disorders now recognized as lymphatic malformations. Cystic hygromas can range from increased nuchal translucency to thin-walled cystic masses that can become larger than the fetal head. The cysts may result from a lymphatic abnormality, possibly due to absent or inefficient connections between the lymphatic and venous systems. Cystic hygroma is strongly associated with Turner's syndrome (predominantly a 45 XO karyotype), trisomy 21 (Down syndrome), trisomy 18 (Edwards syndrome), and 45 X / 46 XY (Noonan syndrome). Fetal cystic hygroma associated with NIHF has a high incidence of aneuploidy, particularly Turner syndrome or trisomy 21. About 30 to 70% of fetuses with cystic hygroma have chromosomal aberrations [1]. A review of 900 karyotyped fetuses with cystic hygroma shows that 31.3A% have 45 X, 15% have trisomy 21, 7.3% have trisomy 18, 2.5% have trisomy 13 and 3.5% have other abnormal karyotypes [2]. Fetuses with cystic hygroma and NIHF in the second trimester have been associated with a poor prognosis. NIHF was also found to be associated with structural abnormalities in 83.3% cases, and chromosomal abnormalities in 47.3% [3].

Turner syndrome has a variable presentation. Cystic hygroma with NIHF during pregnancy is the most severe form. It can also present as short stature and primary amenorrhea at puberty, and premature ovarian failure at a later age. The severity of disorder is related to the extent of monosomy X / mosaicism in various tissues. Cytogenetic discrepancy between fetal tissue and body fluid in a fetus with cystic hygroma has also been reported [4]. In the present case chromosomal analysis from cord blood, placenta, cystic hygroma fluid, and fetal skin showed 45 XO karyotype, suggesting a case of pure monosomy X, leading to phenotype of Turner syndrome.

Since the incidence of the chromosomal anomalies in a fetus affected by cystic hygroma and NIHF is very high [4], it is necessary to carry out a routine karyotype test if the ultrasonogrpahy reveals such anomalies during pregnancy.

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