Non-immune Hydrops Fetalis Secondary to Nuchal Cystic Hygroma: Prenatal Detection and Postnatal Evaluation of 5 Cases

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Non-immune hydrops fetalis secondary to nuchal cystic hygroma is a lethal congenital malformation, resulting from the lack of communication between the jugular venous system and the cervical lymphatic vessels in the nuchal region. Identification of these abnormalities by sonography is not important for the management, but also for genetic counseling. In this paper, we report 5 cases of severe hydropic fetuses secondary to nuchal cystic hygroma, prospectively detected during sonographic examination between January 2002 and January 2003 at our hospital outpatient clinic.
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Key Words: Cystic hygroma, Non-immune hydrops, Prenatal diagnosis

Nuchal Cystic Hygroma (NCH) is a congenital malformation resulting from jugular lymphatic obstruction due to a lack of communication between the jugular venous system and the cervical lymphatic vessels in the nuchal area. On prenatal sonographic examination, diagnosis of the malformation is based on the visualization of a cystic structure located in the occipitocervical region.[1] This lethal anomaly is often associated with generalized hydrops secondary to the problem of lymphatic drainage. However, if a communication between the two systems occurs before fetal demise, the hygroma may regress leaving redundant skin folds in the nuchal area, which is called as "pterygium colli".[2]

Cystic Hygromas can be seen in association with or without chromosomal abnormalities. Turner syndrome is the most common chromosomal abnormality discovered in fetuses with hygromas; which is followed by trisomy 21, trisomi 18, trisomi 13, 13q-, 18q-, partial 11q/22q trisomy and trisomy 22 mosaicism, Noonan's syndrome, distichiasis lymphedema syndrome, Robert's syndrome, Cowchock syndrome, fetal alcohol syndrome, and aminopterin syndrome.[2,3]

Prenatal diagnosis and careful evaluation of these malformations is quite important for the management and genetic counseling of these affected parents.

In this paper, We report 5 cases of severe non-immune hydropic fetuses secondary to NCH, prospectively detected during sonographic examination between January 2002 and January 2003 at our outpatient clinic, and referred to the Genetic Department for prenatal karyotyping and genetic counseling. All of the pregnancies of these severely malformated fetuses were terminated by administration of Prostaglandin E.

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Case 1

A 20-year-old pregnant woman (gravida 1, para 0) with a consanguineous spouse applied to our antenatal out-patient clinic at 15th weeks of gestation, without any follow-up before. Ultrasonographic screening revealed , thin walled cystic mass projecting posteriorly from the occiput measuring 59x47mm in size, diffuse skin and scalp oedema, ascites, pericardial and pericardial effusion. Amniotic cell culture showed a normal 46, XY karyotype. The parents refused the autopsy. (figure 1)

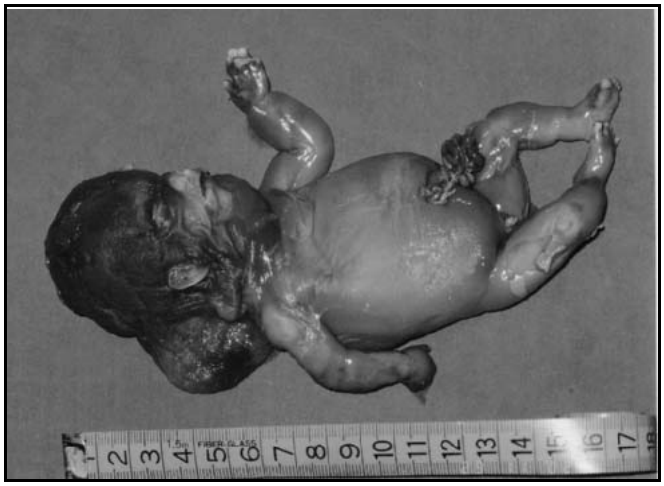

Figure 1. The appearance after termination. Note the generalized oedema, abdominal distention (ascites) and a large nuchal bleb.

Case 2

A 19-year-old pregnant woman (gravida 1, para 0) with a consanguineous spouse have referred to our hospital at 16th weeks of gestation. Ultrasonographic findings were as follows: Posterior cervical cyst as big as fetal head with multiple septaes, anhydramnios, thoracal hypoplasia, unilateral choroid plexus cyst in cranium, lemon sign deformity,

left ventricular hypoplasia in heart, ascites, pleural effusion, and unidentification of fetal bladder, kidneys and stomach. Because of the absence of amniotic fluid, cystic hygroma paracenthesis was performed, and cystic hygroma cell culture revealed a 45, XO karyotype. Parents were informed by genetic couselling. (Figure 2-3)

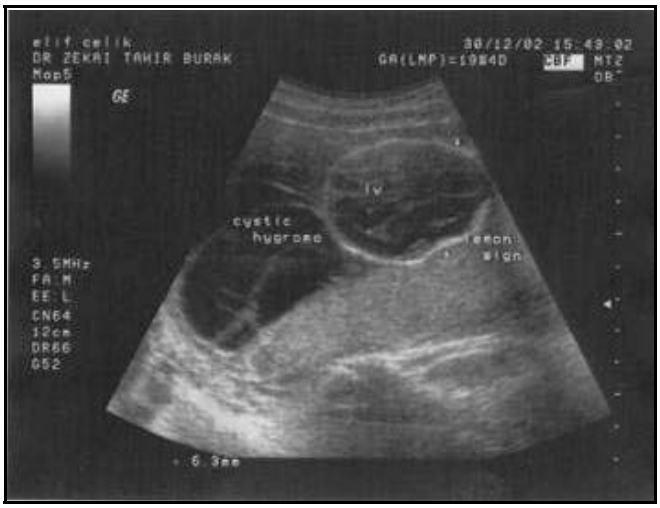

Figure 2. Sonographic appearance in transvers plan of case 2. Cystic hygroma with multiple septaes near the head, lemon sign, and hydrocephaly can be seen.

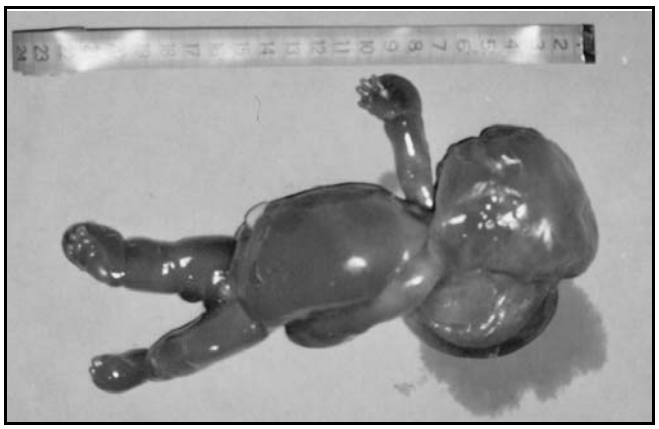

Figure 3. Case 2, after the termination.

Case 3

A 21-year-old pregnant woman (gravida 1, para 0) having Gilbert syndrome in her past was initially seen at 22nd weeks of gestation. In obstetrics history she only had been taking progesterone due to abortus imminens at 6th weeks of gestation. She has been married the son of her aunt. Sonographic examination demonstrated a singleton dead fetus with biometry consistent with 18th weeks' of gestation. The fetus had generalized oedema in the skin and in the scalp, and a multiseptate cystic mass measuring 104x36 mm around the neck. Cytogenetic analysis showed a normal distribution in karyotype, and necropsy showed no anomalies other than those detected by ultrasound. (Sonography in figure 4-5)

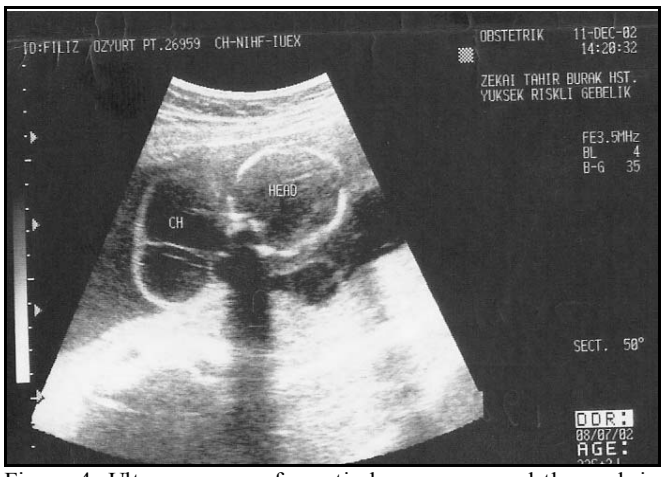

Figure 4. Ultrasonogram of cystic hygroma around the neck in case 3.

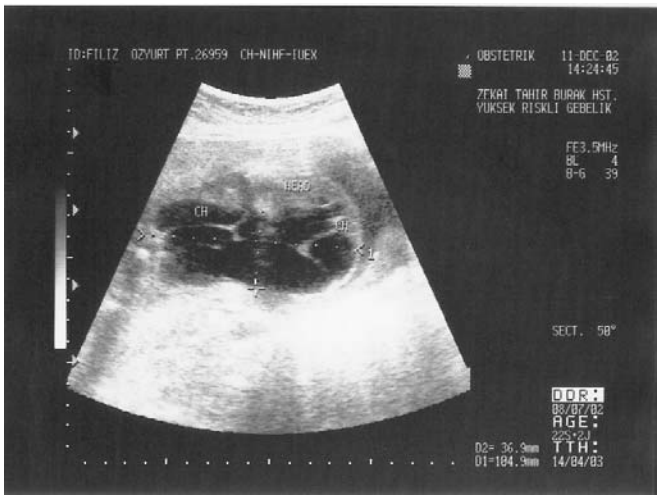

Figure 5. Transvers scan of the focused multiseptated cyst in case

Case 4

A 32-year-old pregnant woman (gravida 3, para 2) have applied to our antenatal out-patient clinic at 21st weeks of gestation. Her previous pregnancies had been normal. Sonographic examination demonstrated a dead fetus with biometry consistent with 16th weeks' gestation. The fetus showed diffuse oedema, and a large cystic hygroma colli 60x52 mm in size. Pregnancy was terminated, and postmortem examination demonstrated no anomalies different from sonographic findings.

Case 5

A 23-year-old pregnant woman (gravida 1, para 0), who has been married the son of her aunt have applied to our out-patient clinic for the routine ultrasonographic examination at 14th week of gestation. On sonographic evaluation, ascites, thickening of the skin and cystic masses divided by septa in the posterior nuchal region, and hydrocephaly (lateral ventricules=10mm in lenght) were found. Postmortem examination revealed micrognathia, low-set ears, exo-phtalmus, hypoplasic thorax and lungs, microcephaly, webs

located on shoulders and knees, suggesting "lethal multiple pterygium syndrome".

Discussion

NCH can be detected by routine ultrasonographic examinations. Depending on higher improvements in ultrasound technology specially in the last decade, it is possible to identify many fetal congenital malformations at the present time. For earlier diagnosis, It's been recommended to apply transvaginal sonograms by many authors.[1,3,4].

L. Kang et al have used three-dimensional (3-D) ultrasound to detect cystic hygromas in their study, and they have concluded that 3-D ultrasound might add novel visual depiction of the lesion in 3-D after reconstruction and, thus, assisted substantially in perinatal consultation.[5]

Despite using transvaginal or 3-D sonographies in modern obstetry, conventional sonography scans still dominate to identifying many lesions in routine.

On ultrasonography, it's important to differentiate these cysts from the other craniocervical masses such as encephalocele, meningocele, meningomyelocele, cytic teratoma, twin sac of a blighted ovum, or increased nuchal oedema.[2]

Cystic hygromas are congenital masses of the dorsal neck, resulting from failure of the lymph channels to communicate with the veins of the neck and upper thorax, and causing enlargement of the lymphatic spaces, which may then cause hydrops foetalis.[6-8] It presents an incidence of 1/6000 native. It is a very frequent relief in the precocious abortions. Singh and Carr recognized that the cystic hygroma was frequently seen in abortuses with a Turner's syndrome.[9]

Prognosis is generally poor in NCH. It's good only if the karyotype is normal and there are no hydrops or septations.[9]

Tanriverdi HA et al analyzed the prenatal management and prognostic factors of NCH by using cytogenetic tests and sonographic morphological features in their study. They reported that NCH, either septated or not, carried high risk of aneuploidies and advers fetal outcome.[10]

Fetal hydrops is associated in 33% of NCH cases 3. Only 9% of cases occur in healthy fetuses with normal karyotype whilst the remaining 91% have been observed in intrauterine exitus (89%) or in liveborn (2%) associating with abnormalities 3. In our study, all of the cases had hydrops fetalis. Also Rose et al reported 25 fetuses with cystic higroma. They had additional abnormalities in twenty(%80) fetuses; most common were non-immune hydrops(12/25,48%) and cardiac defects/5/25, 20%)11.

Familial recurrence can be occurred by a variety of genetic mechanisms, such as autosomal dominant with variable expression, autosomal dominant with germline mosaicism, or autosomal recessive.[6]

Consanguinity was documented in 4 of these 5 cases. Two of them were associated with many other malformations(case 2 and case 5).

The X choromosom aberration was observed just in one case in our study (case 2). This case was also carrying diagnostic findings of Turner syndrome in sonography, and the lesions were comfirmed by postnatal pathologic examination as well. In this case we performed cystic hygroma paracenthesis, and cystic hygroma cell culture prenatally. The result was 45, XO and then pregnancy was terminated.

In our 5th case we reported "Lethal Multipl Pterygium Syndrome", postnatally. Tolmie et al and Lockwood et al reported familial recurrence of prenatally diagnosed cystic hygroma, with postnatal findings consistent with this syndrome.[6]

Non-immune hydrops fetalis secondary to NCH is a lethal malformation. Prenatal diagnosis and careful evaluation of these malformations by sonography is quite important for the management and genetic counseling of these affected parents.

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