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Fetal cystic hygroma: the importance of natural history

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ABSTRACT

Objectives: To evaluate the natural history of cystic hygroma (CH) in fetal and neonatal periods to enrich parental counselling. Ultrasonographic characteristics, associated syndromes, chromosomal anomalies, fetal cardiac pathology and life after birth were considered.
Study design: From May 1985 to September 2010, 207 pregnancies were seen the authors' centre with suspected vascular–lymphatic fetal malformation: 156 of them had CH. Cases were followed up by telephone interview to determine fetal and neonatal outcomes. Chi-squared test was used for statistical analysis.
Results: Among the 156 cases of CH, the condition was septated in 75% of cases, associated with other pathologies in 74%, and retronuchal in 88%. Intrauterine regression was seen in 36% of cases, with complete disappearance in 77%. The karyotype was normal in 55% of cases. Follow-up was completed in 85 cases and revealed 54 spontaneous abortions (63%) and 31 live births (37%). Amongst these, 21 out of 31 children had a favourable outcome (68%). A negative embryo–fetal outcome was significantly associated with CH being associated with other pathologies, such as hydrops, retronuchal position and altered karyotype. Spontaneous regression or resolution of CH was associated with live births.
Conclusions: The management of pregnancies with a diagnosis of fetal CH requires knowledge of natural history of the malformation for appropriate parental counselling.

1. Introduction

Cystic hygroma (CH), first described by Redenbacher in 1828 [1], is a vascular–lymphatic malformation with an incidence of approximately 1:1000–6000 births [2] and 1:750 spontaneous abortions [3]. The malformation develops at the end of the sixth week of gestation [4]. It is anatomically characterized by dilated lymphatic ducts because of a lack of communication between the lymphatic and venous systems.

CH is located predominantly in the neck and represents 20–25% of cervical lymphatic tumours [5]. In 5% of cases, CH is located in the axillary region, the mediastinum, the abdomen, the retroperitoneum or the mesentery [1,6]. The lesion can expand over time, but there are often cases of lymphatic drainage that do not develop into actual cysts [7].

Ultrasonographic diagnosis of CH is usually obtained in the first trimester or at the beginning of the second trimester [8,9], becoming less frequent as the pregnancy progresses [10,11]. On ultrasonography, this lesion appears septated or non-septated by internal trabeculae.

Fetal CH may be associated with aneuploidy and other structural malformations. If associated with aneuploidy, CH is unlikely to present itself in an isolated form and has an incidence of 1:285. If CH is associated with other structural malformations has an incidence of 1:100 and the prognosis is more favourable [9,12].

Many published studies have suggested that CH is associated with poor fetal outcome, but the incidence of spontaneous intrauterine resolution is unknown. Moreover, the long-term prognosis of liveborn infants has been poorly documented.

The purpose of this study was to specify the natural history of CH in order to enrich parental counselling, with long-term follow up aimed at evaluating the well-being of children with a prenatal diagnosis of CH.

2. Materials and methods

This retrospective study was undertaken between May 1985 and September 2010. Two hundred and seven pregnant women with a diagnosis of suspected vascular–lymphatic fetal malformation were admitted to the Prenatal Diagnosis Unit, with three peaks in admission and diagnosis in 2001, 2004 and 2010.

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Ultrasonography was performed by maternal-fetal medicine physicians to increase the accuracy of each diagnosis. CH was defined as septated or non-septated, and the widest part was measured on transverse and sagittal sonographic views. The presence of hydrops fetalis and other fetal abnormalities was noted (Figs. 1–5).

The General Electric Medical System, Technos MP ESAOTE High Definition System and VOLUSON E8 with a 3.5 MHz transabdominal probe (transvaginal probe, when indicated) were used for ultrasonography over the study period.

Once CH was diagnosed, the parents received counselling to explain the key points of management of the individual case on the basis of natural history: the association of the malformation with chromosomopathies and heart disease; but also the probability of spontaneous resolution, especially if CH was isolated, as well as the high percentage of spontaneous abortions if CH was associated with other pathologies. The parents were offered genetic investigation by amniocentesis or cordocentesis according to gestational age, followed by programmed fetal echocardiography and possibly a consultation with a paediatric surgeon, where indicated. All ongoing pregnancies were followed up. Serial ultrasound checks were performed every 3–4 weeks to monitor the progress of the condition until delivery (vaginal or caesarean section, according to obstetric indications).

After evaluation of the clinical and sonographic findings, the cases were followed up by telephone interview to determine the outcome of the pregnancies, the medical condition and the impact this had on liveborn cases, taking into account the limits of such follow-up.

Verbal consent was obtained from each woman. Physicians maintained the study database, focusing on the protection of personal data.

Statistical analysis was performed using Chi-squared test, and significance was defined as $p < 0.05$.

3. Results

In total, 207 cases with suspected vascular–lymphatic malformations were examined, of which 156 were CH (75.4%, eight of which were in twin pregnancies), 41 were lost to follow-up and 30 pregnancies were terminated.

The mean maternal age at diagnosis was 32.3 (range 22–43) years, and the average gestational age at the time of diagnosis of the malformation was 15.1 (range 11–20) weeks.

The average weight at birth was 3122 (range 1040–4700) g. The average gestation period was approximately 37 (range 33–41) weeks for both vaginal and caesarean deliveries, while that of spontaneous abortion was 15 (range 10–20) weeks.

Ultrasonography found that CH was generally retronuchal (88%), septated (75%) and associated with other pathologies (74%, $n = 116$). Of the 116 cases, 59 had fetal non-immune hydrops (51%). Cases of non-septated CH were mainly associated with a normal karyotype and normal fetal echocardiography, in contrast to cases of septated CH.

Of the 156 pregnant women, 64% (100/156) opted for invasive tests to determine the fetal karyotype. Twelve women had chorionic villus sampling (in another hospital), 80 women underwent amniocentesis, and eight women underwent echo-guided cordocentesis.

The fetal karyotype was normal in 55% of cases (55/100) and abnormal in 45% of cases (45/100). The chromosomal abnormalities were: monosomy X (19%), trisomy 21 (14%), trisomy 18 (6%), trisomy 13 (3%), and other chromosomal abnormalities (3%).

Regarding the correlation between sonographic characterization and karyotype, 75% of CH were septated (117/156), and 52% (36/69) of those investigated had a normal karyotype. Twenty-five per cent (39/156) of CH were non-septated, and 61% (19/31) of those investigated had a normal karyotype (Figs. 1–3).

Cardiac pathologies were associated with CH: 49 of 156 (31%) women underwent echocardiography, and this was found to be

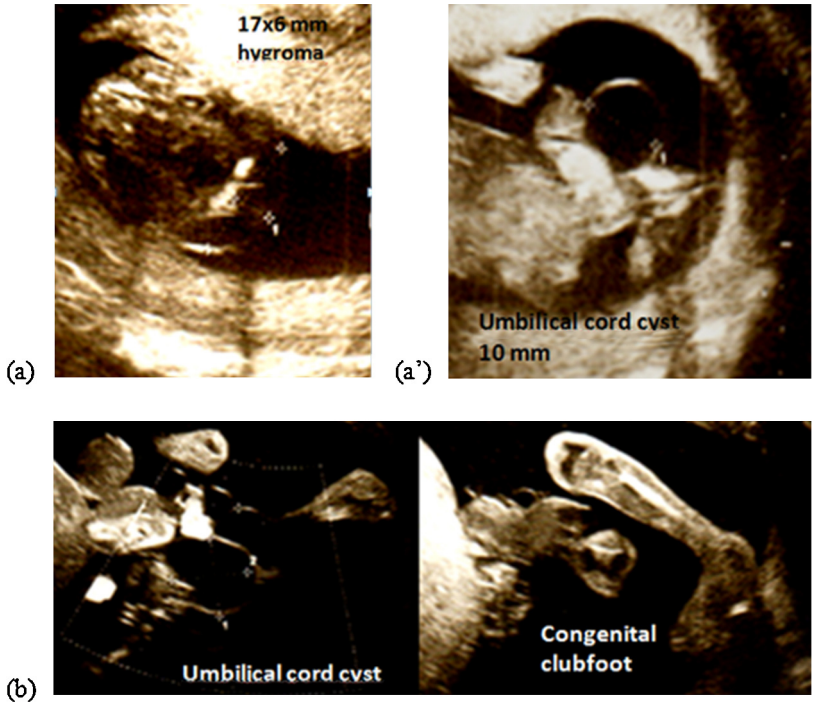

Fig. 1. Septated retronuchal associated hygroma; prenatal diagnosis at first trimester (2009); hygroma disappearance at 16 weeks of gestation. Anasarca, umbilical cord cyst (47, XY + 18). TOP at 22 weeks in another hospital. Ultrasound at (a and a′) 14 + 4 and (b) 17 + 5 weeks of gestation.