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Fetal lateral neck cysts: early second-trimester transvaginal diagnosis, natural history and clinical significance

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ABSTRACT

The objective of the study was to explore the natural history and clinical significance of lateral neck cysts during the early second trimester of pregnancy. A survey was conducted of pregnant women at 12–15 weeks' gestation who presented at the ultrasonographic unit between January 1991 and December 1994. During the 4-year period, of the 1500 fetuses scanned, 42 fetuses with lateral neck cysts were detected by high-resolution transvaginal ultrasonography. Twenty-six of the 42 fetuses were seen to have isolated lateral neck cysts, and 16 demonstrated a combination of nuchal thickness of ≥ 4 mm and lateral neck cysts. Natural history, fetal karyotype and pregnancy outcome were compared between these two groups. None of the fetuses with isolated neck cysts had an abnormal karyotype, and all cysts resolved spontaneously at 16–20 weeks. No congenital abnormalities were found among the 26 cases. Four chromosomal abnormalities (three trisomy 21 and one monosomy XO) were detected among the 16 fetuses with combined lesions, resulting in termination of pregnancy in all. Among the remaining 12 fetuses, spontaneous resolution of the neck findings occurred between 16 and 20 weeks' gestation. The finding of isolated lateral neck cysts in the early second trimester is not associated with increased risk of aneuploidy. However, the combination with nuchal thickness of ≥ 4 mm should prompt genetic counselling and consideration of karyotyping, since this delay in maturation of the cervical jugular lymphatic communication may be associated with chromosomal aberrations.

INTRODUCTION

With the advent of high-resolution transvaginal ultrasonography, small-part scanning of the developing fetus has become possible during the first and early second trimesters of pregnancy[1]. Various fetal structures that were not previously recognized during fetal embryogenesis have now become apparent, but their clinical significance is, as yet, undetermined. Although fetal nuchal translucency thickness and other nuchal cystic lesions have been largely reviewed in the recent literature[2–6], a relative paucity of data exists regarding the ultrasonographic findings of lateral neck cysts detected early in the second trimester[7]. Lateral neck cysts are small fluid accumulations situated within the anterolateral fetal neck. These findings have been rarely mentioned in the English literature, and are interchangeably described as 'nuchal blebs' or 'non-septated cystic hygroma'[8,9].

Although lateral neck cysts have been considered to be a transient normal variant, 5% have been associated with an abnormal fetal karyotype[9]. Since no factors have been reported in predicting whether lateral neck cysts are physiological or pathological[10], all patients who present with this ultrasonographic finding are counselled to have invasive karyotyping but in only 5% of cases is the karyotype abnormal[9].

In this report we present the experience of our institution with the management, pregnancy outcome and associated features of 42 cases with lateral neck cysts diagnosed in the early second trimester of pregnancy.

MATERIALS AND METHODS

From January 1991 to December 1994, 1500 fetuses, including 17 pairs of twins, were evaluated at 12–14.9 weeks' gestation (as determined by menstrual history and confirmed by ultrasound biometry) for the presence of lateral neck cysts and thickness of the nuchal region. In all cases, prenatal sonograms, medical records and clinical course were evaluated. Only fetuses with isolated

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lateral neck cysts (Figure 1) or lateral neck cysts in combination with nuchal thickness of ≥ 4 mm were included in the study (Figure 2), and fetuses with only increased nuchal translucency thickness (< 3 mm), nuchal hygromas or edema with or without hydrops formation were excluded.

A standardized technique for measuring the nuchal thickness was utilized. In an axial scan through the fetal neck at the level of the mandibular rami, the maximal thickness of the nuchal region was measured from the edge of the cervical spine. Lateral neck cysts were defined as any hypoechoic lucencies in the anterolateral aspects of the fetal cervical region. We preferred the term 'nuchal thickness' to describe the posterior aspect of the neck, in order to differentiate it from second-trimester, cystic fetal hygroma or first-trimester nuchal translucency[11]. To avoid any pitfalls from linear specular reflection as described by Hertzberg and associates[12], the criterion for increased nuchal translucency thickness was chosen as ≥ 4 mm. Sonograms were performed using a high-resolution transvaginal Doppler ultrasound machine (Elscint ESI 2000, Haifa, Israel) with 6.5–7.5-MHz probes. A complete anomaly survey was performed on each fetus. Specific sonographic observations included fetal echocardiography. Cases characterized by other organ abnormalities were excluded from this study.

Karyotyping was obtained by amniocentesis or chorionic villus sampling. In cases of continuing euploid pregnancies, the clinical course was evaluated by serial ultrasonographic examinations. Examinations of newborns were performed by certified neonatologists. Statistical evaluation to show the difference in fetal karyotype was performed by Fisher's exact test.

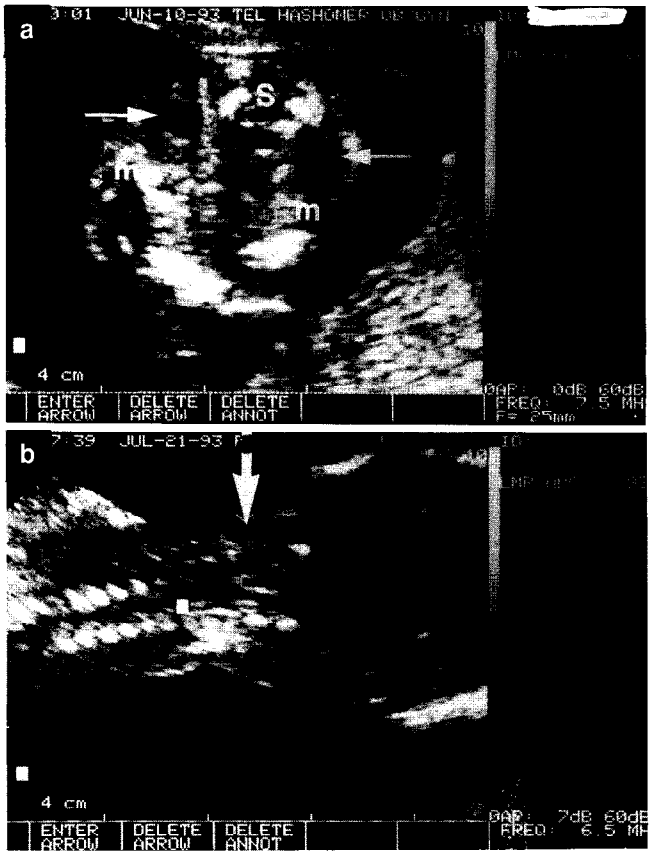

Figure 1 Transvaginal sonographic appearance of lateral neck cysts in eukaryotic fetuses. (a) Transverse view through fetal neck at 14 weeks' gestation, showing two lateral hypoechoic structures (arrows) situated posteriorly to the mandible ridges (m). S, spine. (b) Longitudinal section of another fetus, showing an anterolateral small cervical cyst (arrow). The head is to the right

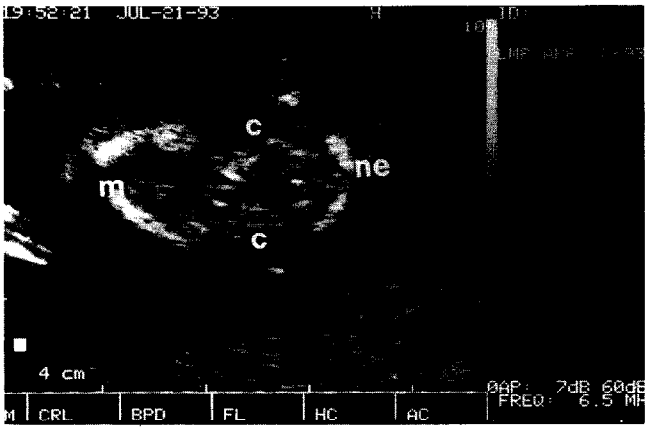

Figure 2 Transverse neck scan at the level of the mandible (m) in a fetus with trisomy 21, showing irregular lateral neck cysts (c) and nuchal edema (ne)

RESULTS

Forty-two consecutive cases with early second-trimester lateral neck cysts were detected between 12 and 14.9 weeks' gestation. In Israel today, many women choose to have an elective transvaginal scan in specialized centers for early fetal morphological evaluation. Patients in the current cohort were among 1500 women referred to our center for an early second-trimester ultrasonographic evaluation. The average maternal age was 25 years (range 21–35 years). Twenty-six fetuses had isolated lateral neck cysts, all with a nuchal thickness of < 4 mm, and 16 fetuses demonstrated a combination of lateral neck cysts and a nuchal thickness of ≥ 4 mm (Table 1). There was no difference in maternal age between these two groups. All fetuses with isolated lateral neck cysts proved to have a normal karyotype, and, at follow-up examinations between 16 and 20 weeks' gestation, all lesions had disappeared. The mean weight of the 26 cases with uncomplicated lateral neck cysts was 3350 g (range, 2900–3800 g), the mean gestational age at delivery was 39 weeks (range, 36–40 weeks) and there was no sex predominance. None were found to have any morphological abnormality after delivery. Among the 16 patients with combined lesions, an abnormal karyotype was obtained in four cases: one had monosomy XO, and the other three had trisomy 21. There were no additional sonographic abnormalities in the four aneuploid fetuses, except for one fetus with trisomy 21 who showed talipes equinovarus. An interesting finding in another case of trisomy 21 was the extension of the neck cysts into the axilar region, manifesting as lymphatic plexus (Figure 3). All pregnancies with abnormal fetal karyotype were

Table 1 Summary of 42 fetuses with either isolated lateral neck cysts or lateral cysts and nuchal thickening of ≥ 4 mm

		Karyotype				
Cervical findings	*n*	*Normal*	*Abnormal*	*Associated anomalies*	*TOP*	*Normal survivors*
Isolated lateral neck cysts	26	26	0*	0	0	26
Lateral cysts with nuchal thickness	16	12	4*†	2‡	4	12

TOP, termination of pregnancy; *p = 0.016 (Fisher's exact test); †trisomy 21 in three cases, monosomy XO in one case; ‡talipes equinovarus in trisomy 21, dilated axillary lymphatic plexuses in another trisomy 21

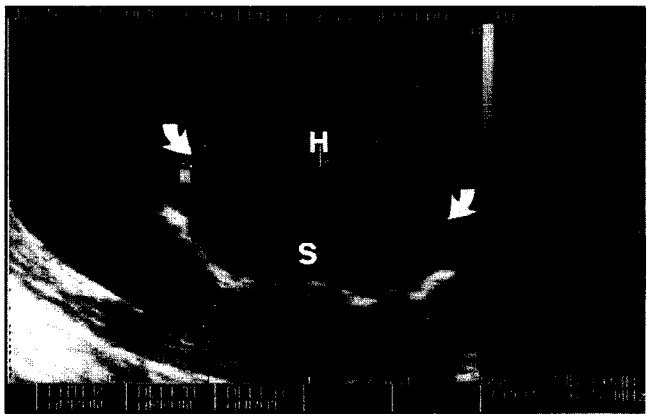

Figure 3 Transverse view of a fetal thorax at 15 weeks' gestation in a fetus with trisomy 21, showing bilateral axillary hypoechoic lesions (arrows) representing enlarged lymphatic lakes. H, heart; S, spine

terminated by dilatation and evacuation, and therefore pathological examination was not possible. In all the remaining 12 fetuses with normal karyotype, nuchal thickness and lateral cysts resolved between 16 and 20 weeks' gestation, and all were delivered as normal infants.

DISCUSSION

Our study confirms previous findings that cystic dilatations in the anterolateral aspects of the fetal cervical region may appear in the early second trimester of pregnancy[8,9]. The observed 2.8% incidence of lateral neck cysts in our population is higher than the 1.6% reported in the series by Bronshtein and co-workers[9]. This increased incidence is probably due to (1) our use of higher-frequency (7.5 MHz) transvaginal transducers that enable better resolution; and (2) the fact that our fetuses were scanned in earlier periods of gestation (12–15 weeks). Among the fetuses with the lateral neck cysts and nuchal thickness of ≥ 4 mm, we observed 25% abnormal karyotypes, which is lower than the 50–70% reported in previous studies[3,13]. This is explained by the exclusion of the cases with septated nuchal hygromas and fetal edema that are known to be associated with anomalies in other systems.

It is well known that fetal nuchal cystic hygroma results from failure to establish communication between the cervical lymphatic vessels and the jugular venous system[14]. Cystic hygroma colli is a well-defined, pathological accumulation of lymphatic fluid that protrudes from the skin surface, and is classically associated with monosomy XO[15]. Ville and colleagues[2] and Nicolaides and co-workers[11] recently recognized and described the distinction between first-trimester nuchal translucency and second-trimester nuchal edema or hygroma. Whereas a second-trimester, septated nuchal hygroma is almost always associated with underlying pathologies such as somatic chromosomal abnormalities and congenital heart defects[16,17], a first-trimester fetal nuchal translucency may be a physiological development. This is not surprising, if one remembers that physiological fluid collections or nuchal blebs are reported as being present in 40% of embryos between 10 and 12 weeks' gestation[18]. During embryogenesis, the fetal lymphatic channels drain into two large sacs lateral to the jugular veins. These lateral neck lymph sacs eventually form communication with the venous system and become the terminal portions of the right lymphatic and thoracic ducts. Failure or delay in development of this communication results in jugular lymph sac enlargement with statis of lymph fluid and cystic hygroma formation[14]. Since none of our 26 cases with isolated lateral neck cysts were associated with chromosomal aberrations or other fetal abnormalities and none developed nuchal hygromas, they can be considered as developmental events. In contrast to Bronshtein and colleagues[8,9] we preferred to use the term 'lateral neck cysts' rather than 'non-septated cystic hygroma' to describe these intracervical fetal translucencies. Furthermore, it is our opinion that the use of the term 'non-septated hygroma' by Bronshtein and colleagues[8,9] to describe the above early findings is misleading. However, when lateral neck cysts appeared with a nuchal thickening of ≥ 4 mm, their association with an abnormal karyotype was clear. Nevertheless, aneuploidy may be associated with the presence of increased nuchal thickening only, and the contribution of lateral neck cysts has yet to be defined. Although an association between nuchal thickness of ≥ 4 mm and chromosomal aberrations has been reported, resolution *in utero*[19,20] and normal fetal outcome is anticipated[4].

Following the theory of Jones[14], we hypothesize (Figure 4) that the manifestations of the jugular–lymphatic communication sequence may present a broad spectrum of events, ranging from pathological complete jugular lymphatic obstruction in cases with prominent septated hygromas accompanied by fetal ascites, to physiological delayed communication manifested only by the appearance of lateral neck cysts. Spanning these two broad categories is a short-term delay and partial occlusion represented by an appearance of the combined findings

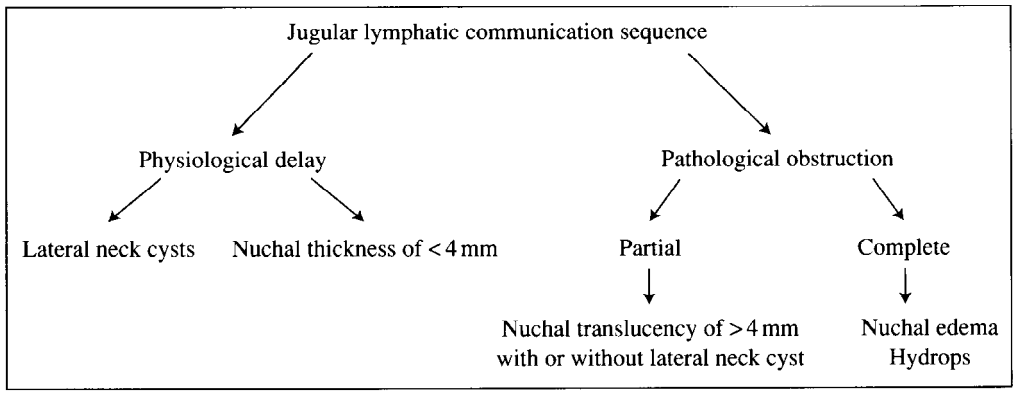

Figure 4 Schematic representation of the jugular lymphatic communication sequence in relation to ultrasonographic manifestations

of lateral neck cysts with nuchal translucency of ≥ 4 mm. With evolution in the time course, these findings may result in nuchal edema with septation and ascites typical of the second trimester, or regress spontaneously.

From the present series, we conclude that isolated lateral neck cysts during the early second trimester should not be considered as cystic hygromas, as they may represent physiologically delayed maturation of the jugular lymphatic communication system, rather than pathological lesions. When a combination with nuchal thickness of ≥ 4 mm is present, prompt genetic counselling and karyotyping are recommended, since this delay in maturation of the cervical jugular–lymphatic communications may be associated with chromosomal aberrations.

REFERENCES

1. Birnholz, J. C. (1992). Smaller parts scanning of the fetus. *Radiol. Clin. North Am.*, **30**, 977–91
2. Ville, Y., Lalondrelle, L., Doumerc, S., Daffos, F., Frydman, R., Dury, J. F. and Dumez, Y. (1992). First trimester diagnosis of nuchal anomalies: significance and fetal outcome. *Ultrasound Obstet. Gynecol.*, **2**, 314–16
3. Nicolaides, K. H., Brizot, M. L. and Snijders, R. J. M. (1994). Fetal nuchal translucency: ultrasound screening for fetal trisomy in the first trimester of pregnancy. *Br. J. Obstet. Gynaecol.*, **101**, 782–6
4. Shulman, L. P., Emerson, D. S., Grevengood, C., Felker, R. E., Gross, S. J., Phillips, O. P. and Elias, S. (1994). Clinical course and outcome of fetuses with isolated cystic nuchal lesions and normal karyotypes detected in the first trimester. *Am. J. Obstet. Gynecol.*, **171**, 1278–81
5. Pandya, P. P., Kondylios, A., Hilbert, L., Snijders, R. J. M. and Nicolaides, K. H. (1995). Chromosomal defects and outcome in 1015 fetuses with increased nuchal translucency. *Ultrasound Obstet. Gynecol.*, **5**, 15–19
6. Comas, C., Martinez, J. M., Ojuel, J., Casals, E., Puerto, B., Borell, A. and Fortuny, A. (1995). First-trimester nuchal edema as a marker of aneuploidy. *Ultrasound Obstet. Gynecol.*, **5**, 26–9
7. Moscoso, G. (1995). Fetal nuchal translucency: a need to understand the physiological basis (Editorial). *Ultrasound Obstet. Gynecol.*, **5**, 6–8
8. Bronshtein, M., Rottem, S., Yoffe, N. and Blumenfeld, Z. (1989). First trimester and early second trimester diagnosis of nuchal cystic hygroma by transvaginal sonography: diverse prognosis of the septated from the nonseptated lesions. *Am. J. Obstet. Gynecol.*, **161**, 78–82
9. Bronshtein, M., Bar-Hava, I., Blumenfeld, I., Bejar, J., Toder, V. and Blumenfeld, Z. (1993). The difference between septated and nonseptated nuchal cystic hygroma in the early second trimester. *Obstet. Gynecol.*, **81**, 683–7
10. Wilson, R. D., Venir, N. and Farquharson, D. F. (1992). Fetal nuchal fluid – physiological or pathological in pregnancies less than 17 menstrual weeks? *Prenat. Diagn.*, **12**, 755–63
11. Nicolaides, K. H., Azar, G. B., Byrne, D., Mansur, C. and Marks, K. (1992). Fetal nuchal translucency: ultrasound screening for chromosomal defects in the first trimester of pregnancy. *Br. Med. J.*, **82**, 43–8
12. Hertzberg, B. S., Bowie, J. D., Carroll, B. S., Killam, A. P. and Ruiz, P. (1989). Normal sonographic appearance of the fetal neck late in the first trimester: the pseudomembrane. *Radiology*, **171**, 427–9
13. Abramowicz, J. S., Warsof, S. L., Doyle, D. L., Smith, D. and Levy, D. L. (1989). Congenital cystic hygroma of the neck diagnosed prenatally: outcome with normal and abnormal karyotype. *Prenat. Diagn.*, **9**, 321–7
14. Jones, K. (1988). Jugular lymphatic obstruction sequence. In Smith, D. W. (ed.) *Recognizable Patterns of Human Malformations*, pp. 560–1. (Philadelphia: W. B. Saunders)
15. Chitayat, D., Kalousek, D. K. and Bammforth, J. S. (1989). Lymphatic abnormalities in fetuses with posterior cervical cystic hygroma. *Am. J. Med. Genet.*, **33**, 352–6
16. Bernstein, H. S., Filly, R. A., Golderg, J. D. and Golbus, M. S. (1991). Prognosis of fetuses with cystic hygroma. *Prenat. Diagn.*, **11**, 349–55
17. Achiron, R., Rotstein, Z., Lipitz, S., Mashiach, S. and Hegesh, J. (1994). First trimester diagnosis of fetal congenital heart disease by transvaginal ultrasonography. *Obstet. Gynecol.*, **84**, 69–72
18. Nishimura, H. and Okamoto, N. (1976). *Sequential Atlas of Human Congenital Malformations. Observations of Embryos, Fetuses and Newborns.* (Baltimore: University Park Press)
19. Darby, B. G. (1989). Resolution of cystic hygroma. *Prenat. Diagn.*, **9**, 447
20. Macken, M. B., Grantmyre, E. B. and Vincer, M. J. (1989). Regression of nuchal cystic hygroma *in utero*. *J. Ultrasound Med.*, **8**, 101–3