

# Fetal cystic hygroma colli

**Anton Galić**

<sup>1</sup>Private Obstetric Gynecological Practice "Venus" Kiseljak, <sup>2</sup>Department for Obstetrics and Gynecology, Croatian Hospital "Dr. Fra Mato Nikolić", Nova Bila, Bosnia and Herzegovina

### ABSTRACT

In this case report, using 2D and 3D transvaginal color ultrasound (TVUS) probe volume 5-8 MHz a 12-week gestational fetus with fetal cystic hygroma colli (FCHC) is presented, observed for eight weeks with thickening of the back neck presented as hypoechoic multilocular cyst. The changes of the back of the fetus neck at 8 weeks of pregnancy, which were presented as multilocular cysts and increased nuchal translucency (NT) with a value greater than 6 mm were clearly observed. Termination of pregnancy at 15 weeks of gestation was performed, and the autopsy showed additional structural anomalies (hydrops fetalis), and inflammations (pneumonia, hepatitis). The introduction of NT as a compulsory examination, ultrasound markers in standard ultrasound screening will facilitate the diagnosis of some congenital malformations.

**Key words:** fetal disease, prenatal diagnosis, ultrasound diagnostics, cardiac malformation, nuchal translucency, prenatal screening.

### Corresponding author:

Anton Galić  
Private Obstetric Gynecological Practice  
"Venus"  
Žrtava domovinskog rata b.b. 71250  
Kiseljak, Bosnia and Herzegovina  
Phone: +387 30 941 209;  
fax: +387 30 870 893;  
E-mail: venus.ordinacija@gmail.com

### Original submission:

20 November 2012;

### Revised submission:

28 January 2013;

### Accepted:

14 March 2013.

SEEHSJ 2013; 3(1):97-100

## INTRODUCTION

In searching for specific early ultrasound signs (markers) that could indicate an increased risk of inherited or acquired disorders, scientific research confirmed the exceptional value of ultrasound measurement of NT (nuchal translucency). Nuchal translucency ultrasound indicates a build-up of fluid (lymph) between the skin and subcutaneous fascia back on the neck of the embryo, which is revealed by ultrasound between 11 - 14 weeks of pregnancy, or if the distance crown rump (CRL) is between 45-84 mm (1). Normally tolerated thick folds is lower than 99th centile for CRL (1-4). Standard measurement of NT has been a legal obligation in the European Union since 2007. Standard measurement of NT is the method for detection of increasing risk of Down syndrome, as well as other congenital malformations (1, 5-7). Numerous studies have shown a connection between the findings of the ultrasound markers (nuchal translucency > 3 mm) with exactly defined chromosomal aberrations, especially with aneuploidy and Down syndrome (1,8). Association of this finding with Down syndrome is so significant that most authors studying this phenomenon have classified ultrasound cervical folds in screening for Down syndrome (6,9).

If the measurement of nuchal translucency and Triple test are performed at early stage of pregnancy, it is possible to detect almost 90% of all inherited diseases (1).

Ultrasound examination in the period between 11 - 14 weeks of pregnancy, especially among pregnant women who are classified as so-called moderate-risk group for Down syndrome, is included as well (1). "Additional" ultrasound makers measure the presence and length of the nasal bones, fronto-maxillary facial angle measuring flow during atrial contraction in the ductus venosus (colored Doppler) and tricuspid regurgitation findings (2). At this time, the nasal bone is not present in 60 to 70% of fetuses with trisomy 21, as well as 2% of chromosomally normal fetuses (3,4). It is similar to the maxillary nucleus, whose hypoplasia ("short maxilla") is located in 25% of fetuses with trisomy. The 21st fetal NT normally increases with gestation (crown-rump length) (3,5, 10,11). Fetuses with certain CRL, each of NT, which represent the probability factor, are multiplied with default risk based on ma-

ternal age and gestation in order to calculate a new risk (9). The higher the NT, the greater the probability factor and thus higher new risks. In contrast, the smaller the NT measures, the lower the risk factor and consequently the lower new risks (1,6,8). Fetal cystic hygroma is a manifestation of early lymphatic obstruction. It is mostly associated with nonimmune hydrops fetalis (11,12).

## CASE REPORT

Thirty five-year old woman reported problems with getting pregnant for the last ten years. Endometriosis was the main reason of her sterility. She underwent two laparoscopic surgeries and after that she could not get pregnant. During an ultrasound scan that found missing right adnexae, while left adnexae at uterus was good. Her medical documentation was not complete, especially pathology documentation (histopathological findings). Vaginitis test kit (Gima, Italy) was positive for *Chlamydia trachomatis*, *Gardnerella vaginalis* and *Trichomonas vaginalis*. Oral drug therapy was prescribed: azitromycin, metronidazole and tetracycline. Betadine vaginal tablets were applied. Finally, gynophilus vaginal capsules with *Lactobacillus vaginalis* were applied.

After 45 days, to our great surprise, the patient reported pregnancy. Ultrasound examination of uterus clearly showed gestational bubble with positive cardiac activity. The next ultrasound examination after one month, suspicious cystic thickening on the back neck of the embryo was found. At the ultrasound examination of the 12-week pregnancy, an increase of nuchal translucency (NT greater to 5 mm) and cystic thickening on the back neck of the fetus were found. The patient was informed that this finding indicated an increased risk for Down syndrome, Turner syndrome, etc. We recommended her to do Triple test including alpha-fetoprotein (AFP), human chorionic gonadotropin (hCG) and estriol concentration from blood, and amniocentesis. She rejected this proposal and requested termination of the pregnancy.

In the hospital after laboratory analyzes, the patient was treated with the prostaglandins in the form of gel (prepidil gel-karboprost) and intramuscular injections (prostin 15M-dinoprost). After five intramuscular injections she had an abortion. Complete material was sent to the Pathology Department. After three



Figure 1. 2D Presentation of nuchal translucency (Galić A, 2012)

weeks, the results of the autopsy showed positive cytomegalovirus (CMV) infection, hepatitis C positivity and general hydrops.

Nuchal translucency with more or less thickness is a phenomenon occurring during embryonic and fetal development (1,2). Cystic hygroma belongs to a group of disorders now recognized as lymphatic malformations (1). The cysts may result from a lymphatic abnormality, possibly due to absent or inefficient connections between the lymphatic and venous systems (1,2). Finding nuchal translucency in an embryo should not be a surprising or alarming event. The problem we face is a need to define the thickness and morphologic characteristics that are associated with pathologic fetal conditions, including chromosomal anomalies (4). Approximately 29% of cases of nuchal translucency seen in the first trimester are associated with chromosomal anomalies (3,4). The most frequent anomalies are trisomy 21 (13.5%), trisomy 18 (6.9%), Turner syndrome (3.8%), trisomy 13 (1.5%), and other anomalies (2.2%). (7,8). Chromosomal anomalies are associated almost exclusively with translucencies with a thickness greater than 3 mm (3,4,5). On the other hand, cystic

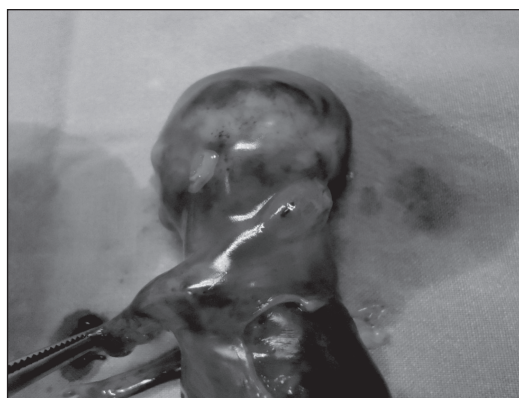


Figure 2. Fetal Cystic Hygroma Colli (Galić A, 2012)

hygromas of the neck are associated with chromosomal anomalies in 52% of cases when the hygroma colli is found as an isolated event in the first trimester and in 71% of cases if associated with other malformations (6). Other chromosomal anomalies occur in association with hygroma colli only sporadically (14). The present work confirmed previous statement suggesting that cystic hygroma colli diagnosed after 14 weeks gestation is almost universally lethal malformation (12,13). This result suggests that NT measurement in the first trimester is potentially useful for screening for fetal malformations.

Increasing NT, with the finding of FCHC and autopsy results confirmed the reliability of NT practices as US markers in detecting congenital malformations. The introduction of NT as a of compulsory examination, or ultrasound markers in standard ultrasound screening, facilitate the diagnosis of some congenital malformations.

## FUNDING

No specific funding was received for this study.

## TRANSPARENCY DECLARATIONS

## REFERENCE

1. Nicolaides KH eds. The 11-13<sup>+</sup>6 weeks scan. London: Fetal Medicine Foundation, 2004; 1-25.
2. Bindra R, Heath V, Liao A, Spencer K, Nicolaides KH. One stop clinic for assessment of risk for trisomy 21 at 11-14 weeks: A prospective study of 15,030 pregnancies. *Ultrasound Obstet Gynecol* 2002; 20:219-25.
3. Chasen ST, Skupski DW, McCullough LB, Chervenak FA. Prenatal informed consent for sonogram: the time for first-trimester nuchal translucency has come. *J Ultrasound Med* 2001; 20:1147-52.
4. Chervenak FA, Isaacson G, Blakemore KJ. Fetal cystic hygroma: cause and natural history. *NEJM* 1983; 309:822-5.
5. Nicolaides KH, Shawwa L, Brizot M, Snijders R. Ultrasonographically detectable marks of fetal chromosomal defects. *Ultrasound Obstet Gynecol* 1993; 3:56-69.
6. Bronshtein M, Rottem S, Yoffe N. First-trimester and early second-trimester diagnosis of nuchal cystic hygroma by transvaginal sonography: Diverse prognosis of the septated from the non-septated lesion. *Am J Obstet Gynecol* 1989; 161:78-82.
7. Michailides GD, Economides DL. Nuchal translucency measurement and pregnancy outcome in karyotypically normal fetuses. *Ultrasound Obstet Gynecol* 2001; 17:102-5.
8. Hyett J, Perdu M, Sharland G, Snijders R, Nicolaides KH. Using fetal nuchal translucency to screen for major congenital cardiac defects at 10-14 weeks of gestation: Population based cohort. *BMJ* 1999; 318:81-4.
9. Nicolaides KH, Azar G, Byrne D, Mansur C, Marks K. Fetal nuchal translucency ultrasound screening for chromosomal defects in first trimester of pregnancy. *BMJ* 1992; 304:867-9.
10. Nicolaides KH, Brizot ML, Snijders RJM. Fetal nuchal translucency: Ultrasound screening for fetal trisomy in the first trimester of pregnancy. *Br J Obstet Gynecol* 1994; 101:782-6.
11. Johnson MP, Johnson A, Holzgreve W, Isada NB, Wapner RJ, Treadwell MC, Heeger S, Evans MI. First-trimester simple hygroma: Cause and outcome. *Am J Obstet Gynecol* 1993; 168:156-61.
12. Boyd PA, Anthony MY, Manning N, Rodriguez CL, Wellesley DG, Chamberlain P. Antenatal diagnosis of cystic hygroma or nuchal pad - report of 92 cases with follow up of survivors. *Arch Dis Child* 1996; 74:38-42.
13. Bronshtein M, Bar-hava I, Blumenfeld I, Bejar J, Toder V, Blumenfeld Z. The difference between septated and nonseptated nuchal cystic hygroma in the early second trimester. *Obstet Gynecol* 1993; 81:683-7.
14. Chen C-P, Liu F-F, Jan S-W, Lee C-C, Town D-D, Lan C-C. Cytogenetic evaluation of cystic hygroma associated with hydrops fetalis, oligohydramnios or intrauterine fetal death: the roles of amniocentesis, postmortem chorionic villus sampling and cystic hygroma paracentesis. *Acta Obstet Gynecol Scand* 1996; 75:454-8.

Competing interests: none to declare.

## Fetalni cystic hygroma colli

Anton Galić

<sup>1</sup>Privatna ginekološko-opstetrička ordinacija "Venus", Kiseljak; <sup>2</sup>Hrvatska bolnica "Dr. fra Mato Nikolić", Nova Bila, Bosna i Hercegovina

## SAŽETAK

U radu je prikazan slučaj fetusa, gestacijske starosti 12 tjedana, kod koga je s osam tjedana primijećeno zadebljanje vrata sa stražnje strane, koje se očitovalo kao hipohogena multilokularna cista, pomoću 2D i 3D sonde TVUZ-a (transvaginalni ultrazvuk *Mindray CD-3*), jačine 5-8 Mghz. Jasno su uočene promjene na stražnjem dijelu vrata već u 8. tjednu trudnoće, koje su se očitovale kao multilokularne ciste, a koje su potvrđene i u 12. tjednu trudnoće, uz povećanje nuchalne translucijencije (NT) čija je vrijednost bila veća od 6 mm. U ovom slučaju, urađen je prekid trudnoće u 15. tjednu trudnoće, a obdukcijom su otkrivene i druge strukturalne anomalije (*hydrops fetalis*) i upale (upala pluća, jetre). Uvođenje mjerenja NT-a, kao obaveznog pregleda, odnosno ultrazvučnog markera kod standardnog ultrazvučnog skrininga, olakšat će dijagnozu nekih kongenitalnih malformacija.

**Ključne riječi:** fetalne bolesti, prenatalna dijagnostika, ultrazvučna dijagnostika.