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Cystic hygroma: a differential diagnosis for increased nuchal tranlucency

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ABSTRACT

Cystic hygroma is a congenital malformation of the lymphatics. Nuchal translucency(NT) is markedly increased in cystic hygroma. Cystic hygroma is associated with aneuploidies, adverse obstetrical and perinatal outcomes. First-trimester nuchal NT measurement with the help of ultrasound is a widely used technique to determine the risk of fetal aneuploidy. Diagnosis should be made early and thoroughly in order to carry out chorionic villus sampling to determine the karyotype early.

INTRODUCTION

First-trimester nuchal translucency (NT) measurement during ultrasonound is a widely used technique to determine the risk of fetal aneuploidy during antenatal period between 11-14 weeks of gestation. Cystic hygroma is another condition where the NT is markedly enlarged. Cystic hygroma has been associated with a high risk for aneuploidy. Thorough screening in first trimester is essential to detect early. These women need genetic councelling and prenatal diagnostic testing to determine the karyotype.

Case report

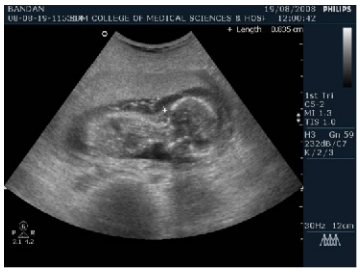
A 20-year old primigravida presented to obstetric out patient department on 10/8/08 with history of 3-months amenorrhea. Her last menstrual period was on 05/05/2008 with gestational age of 13 weeks 6 days of gestation at presentation. On clinical examination uterus was 14 week size. Ultrasound examination revealed Nuchal translucency(NT) of 6.5 mm. Soft tissue edema with fluid collection around the neck was noted and the edema extended from occipital region to lower thoracic region. Pleural effusion was noted on left side. There was no scalp edema. Patient was advised prenatal invasive testing with chorion villous biopsy but she refused. The risk of fetal aneuploidy and associated structural fetal malformations explained. Option of pregnancy termination was given to the patient. The patient opted for pregnancy termination. Pregnancy was terminated on 21/08/08. Diagnosis was confirmed at autopsy. Fetal tissue karyotype was normal.

DISCUSSION

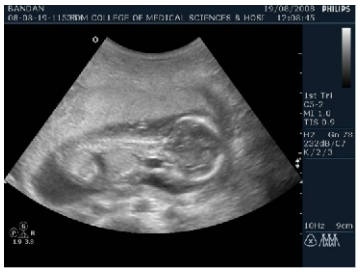
Cystic hygroma is a congenital malformation of the lymphatic system which results due to obstruction between the lymphatic and venous pathways in the fetal neck leading to lymph accumulation in the jugular lymphatic sacs of the nuchal region.[1]

Nuchal translucency is the hypoechoic region located between the skin and soft tissues behind the cervical spine. This hypoechoic space is presumed to represent mesenchymal edema and is often associated with distended jugular lymphatics. First-trimester nuchal translucency (NT) measurement is a widely used technique to determine the risk of fetal aneuploidy. Various causes of increased NT have been proposed. Fetal cardiac failure may cause NT enlargement. Various types of abnormalities are found in the extracellular matrix of the nuchal skin of fetuses with increased NT. Abnormal lymphatic development is demonstrated in fetuses with increased NT.[2]

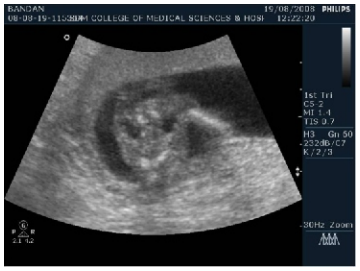
NT is markedly increased in cystic hygroma which extends along the entire length of the fetus, with or without septations clearly visible within the space. Cystic hygroma must be



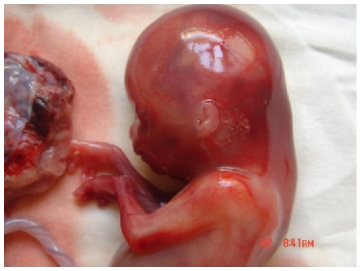
Picture 1: Ultrasound image showing cystic hygroma.



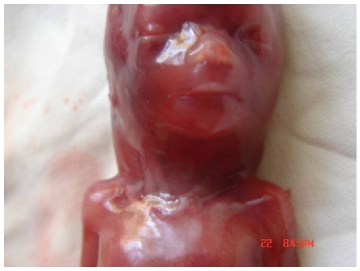
Picture 2: Ultrasound image showing pleural effusion and nuchal edema extending to anterior cervical region.



Picture 3: Axial view of fetal neck on Ultrasound showing nuchal edema extending to lateral aspect of cervical region.



Picture 4: Abortus showing cystic hygroma.



Picture 5: Abortus showing nuchal edema extending to lateral and anterior cervical region.

differentiated from a large NT. Cystic hygroma occurs in approximately 1 in 300 first-trimester ultrasounds. Cystic hygroma has been associated with a high risk for aneuploidy. The majority of cases are trisomy 21. Turner syndrome, trisomy 18, trisomy 13, and triploidy are the others. While many cases of cystic hygroma will have a normal karyotype, a large proportion of these cases will be complicated by a fetal malformation, such as a cardiac defect or a skeletal anomaly. Patients with a cystic hygroma on first-trimester ultrasound should be referred to genetic counseling, as only a small proportion of all cases of first-trimester cystic hygroma are associated with a normal live-born infant.[3]

Fetuses with septated cystic hygromas are more likely to be aneuploid and to develop hydrops, and thus are less likely to be survive than fetuses with nonseptated hygromas.[4]

Prognosis of fetal cystic hygroma detected during the first trimester is poor, and show that sonographic evaluation of fetal NT thickness in the first trimester is important. Outcome of pregnancy was unfavourable with increased risk of miscarriage, elective termination and serious structural abnormalities in 77.7% of cases. Chromosomal abnormalities were seen in 52.7% of cases.[5]

Cystic hygroma is one of the signs suggestive of chromosomal or congenital abnormalities that occur very early and are very specific. Good prognostic factors would be a normal karyotype and the spontaneous resolution of cystic hygroma in the second trimester of the pregnancy. Hydrops is a poor prognostic factor. Diagnosis should be made early and thoroughly in order to carry out chorionic villus sampling to determine the karyotype early, before the very important sign disappears as the gestation progresses.[6]

Survival rate progressively improves with normal karyotype, unilateral pleural effusion, atypical location, and resolution of cystic hygroma.[7]

Late-onset isolated cystic hygroma should be differentiated from the early-onset nuchal cystic hygroma. Late-onset isolated cystic hygroma does not require any prenatal intervention, but care has to be taken during labour and Caesarean section.[8]

Cystic hygroma is highly correlated with adverse perinatal outcome. Prenatal diagnosis and invasive procedures are to be discussed during counselling and stress the importance of close follow-up after delivery for appropriate medical support. A multidisciplinary approach is strictly recommended in live-born children.[9]

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