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"HYGROMA COLLI" SONOGRAPHIC INDICATION OF CHROMOSOMAL ABNORMALITY: A CASE REPORT

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ABSTRACT
This case report aims to highlight the significance of ultrasound imaging in the prenatal diagnosis of hygroma colli, a condition often associated with chromosomal abnormalities, particularly Turner Syndrome (45,XO). A 26-year-old primigravid woman at 17 weeks of gestation was referred for further evaluation of a fetal neck mass detected during routine ultrasound. The sonographic findings revealed a symmetrical, non-septated, hypoechoic cystic mass in the posterolateral region of the fetal neck, along with agenesis of one lung lobe. Based on these findings, pregnancy termination was planned and performed using misoprostol and laminaria insertion. The fetus was delivered with a birth weight of 150 g, a length of 14 cm, and Apgar Score 0/0. Postnatal examination confirmed the presence of hygroma colli, consistent with the ultrasound findings. This case reinforces the importance of early ultrasound screening in identifying fetal anomalies and guiding clinical decisions.

Keywords: hygroma colli; fetal anomaly; pregnancy termination; turner syndrome; ultrasound

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INTRODUCTION

Ultrasound is an useful tool to recognize and define structural defects in the fetus. Fetal hygromas as a manifestation of malformation of the lymphatic system are often the only sign of a severe fetal anomaly. About 62% of cystic hygroma cases are associated with Turner-Syndrome (45,XO).1 A cystic hygroma generally begins to develop between the 6th and 9th weeks of gestation due to failure of the jugular lymphatic sacs to drain into the internal jugular vein, which probably results in the dilatation of the lymphatic sacs into cystic spaces and may lead to the jugular lymphatic obstruction sequence and hydrops fetalis.2,3 We report a case in which ultrasound imaging was used to identify a cystic hygroma in a fetus at 17 weeks gestation and termination planned based on the ultrasound finding.

CASE ILLUSTRATION

A 26 years-old primigravid woman was referred to our hospital due to fetal neck mass detected during an ultrasound examination at 17 weeks gestation. A further ultrasound examination was performed, which revealed a single viable fetus with neck mass and agenesis one lobe of the fetal lung. The neck mass was symmetrical, thin walled, non septated, hypoechoic in the inside and occupied posteriolateral area of the neck. Based on this sonographic findings, patient was planned for pregnancy termination.

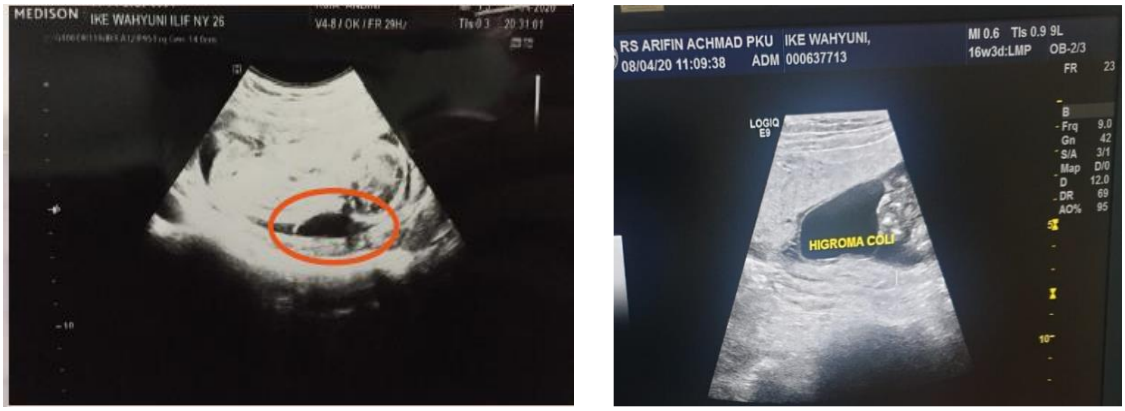

Figure 1. Sagittal view of fetal with hygroma colli (left), hypoechoic area in hygroma colli (right)

Figure 2. Axial vew of hygroma colli (left), another sagital view of hygroma colli (right)

Patient was given misoprostol 400 mcg every 3 hours per vaginam and then 2 pieces laminaria. The baby was born spontaneously with BW 150 gr, BH 14 cm, Apgar Score 0/0. There were soft bulge on the neck. Hygroma colli was seen behind the baby`s neck that correspond with USG examination.

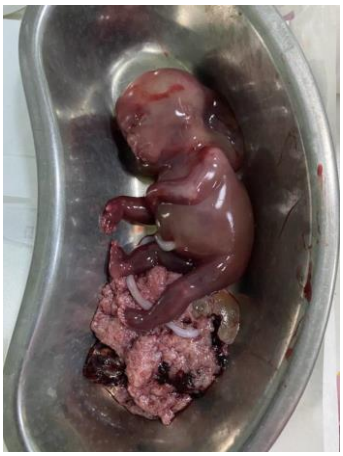

Figure 3. Hygroma colli was seen behind the baby`s neck that correspond with USG examination

DISCUSSION

Pregnancy termination in the patient was done because of the existence of multiple organ anomalies that found in ultrasound examination. So that the prognosis for subsequent fetal life is poor. Generally, survival rate in fetal with hygroma colli is 10%.4 It is also considered a possible cause of perinatal disability.5 Prenatal diagnosis of hygroma colli on ultrasound based on demonstation of a billateral, mostly symmetric, cystic structure located in the occipitocervical region with the lesion either septated or not.6 One study found nearly 47% of the pregnancies with cystic hygroma had multiple congenital anomalies, of which 58% had a chromosomal anomaly. Aneuploidies were major chromosomal defects.7 The risk of recurrence for aneuploidy is low, but cystic hygroma colli with normal karyotype may be inherited as an autosomal recessive trait with 25% recurrence.8 On this case, the karyotype examination was not done to the patient due to termination decision.

CONCLUSION

We want to emphasize that abdominal sonography has been the only instrumental in the prenatal diagnosis of cystic hygroma colli. Based on this sonographic finding the pregnancy has been terminated. We point to the value of early and subtle ultrasound examination as an excellent method in finding hints for chromosome abnormalities.

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