

# PRENATAL ULTRASOUND AND ABORTED FETAL CORRELATION IN RARE FETAL ANOMALIES

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## ABSTRACT

A significant improvement in the detection of rare congenital anomalies is possible with correlation of antenatal ultrasound and gross features of aborted fetus. Evaluation of aborted fetus following termination of pregnancy also enables diagnosis of pathologies undetected by prenatal ultrasound alone, leading to better preconceptional counseling for subsequent pregnancies.

Prenatal ultrasound and pathologic examination of the fetus are informative and complement each other. A very high level of agreement between prenatal ultrasound and pathological correlation was found in cases we studied in our hospital.

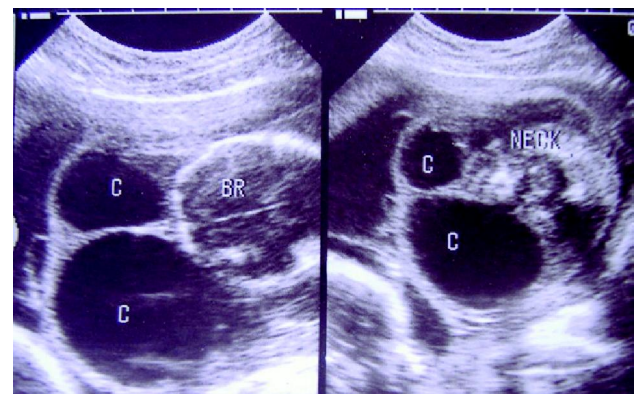
**KEY WORDS:** Prenatal ultrasound, Aborted fetus, Fetal anomalies

## Cystic Hygroma

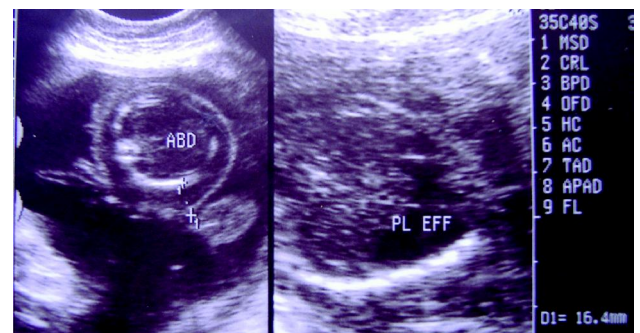
Cystic hygroma is a multiloculated cystic structure that is benign in nature. It may occur anywhere in the body, although it is most frequently encountered in the neck. Cystic hygroma frequently abut neurovascular structures. Lymphatic channels are formed around the sixth week of gestation. From these channels, sacs are formed that establish drainage with the venous system. Failure to establish venous drainage results in dilated disorganized lymph channels, which, in the largest form, present as cystic hygroma.<sup>1</sup> Posterior midline cervical cystic hygromas (PMC) are frequently found associated with chromosomal aberrations and such patients usually do not survive.<sup>2</sup>

We diagnosed two cases of cystic hygroma at 19 and 17 weeks of gestational ages. After therapeutic abortion our findings (Fig. 1 and 2) were consistent with aborted fetus.

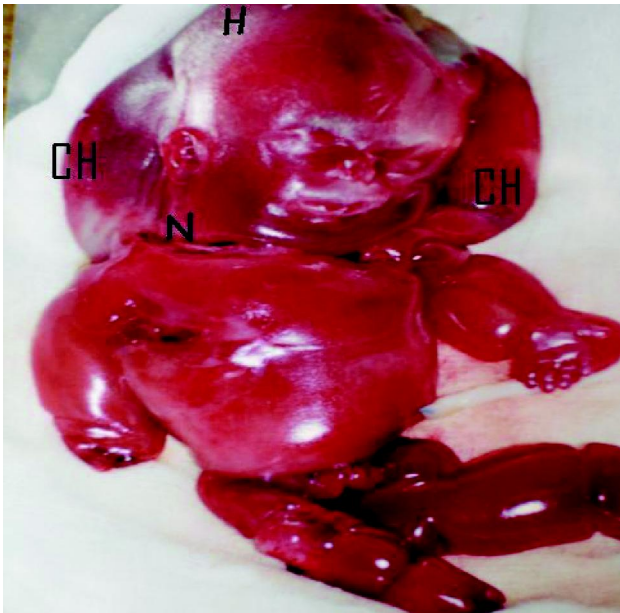
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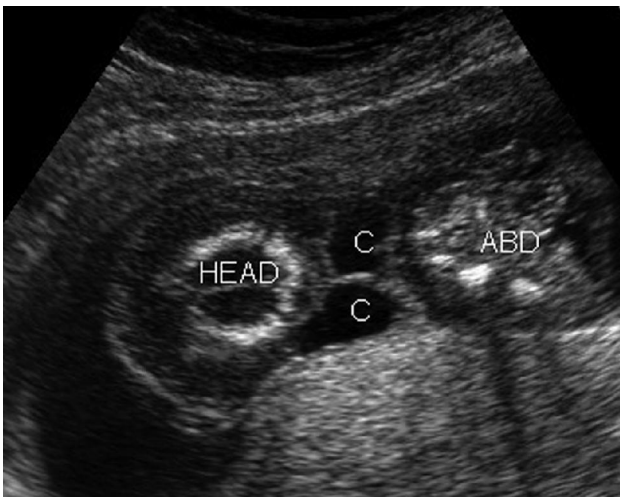
**Figure 1: (a).** Sonogram shows a posterior cystic hygroma (C) behind the fetal head (BR) and neck. Note the internal septum dividing cystic hygroma in midline in both images.



**Figure 1: (b).** Skin thickening and pleural effusion due to associated hydrops.



**Figure 1:** (c) Gross Specimen of aborted fetus shows cystic hygroma (CH) projecting bilaterally around neck (N). Fetal head (H).



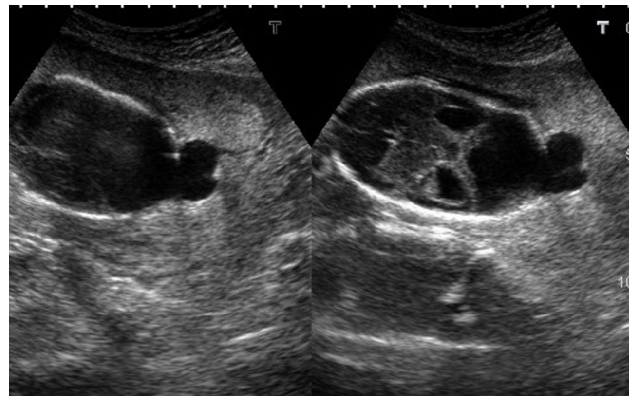
**Figure 2:** (a). Cystic hygroma with midline septum (coronal scan).



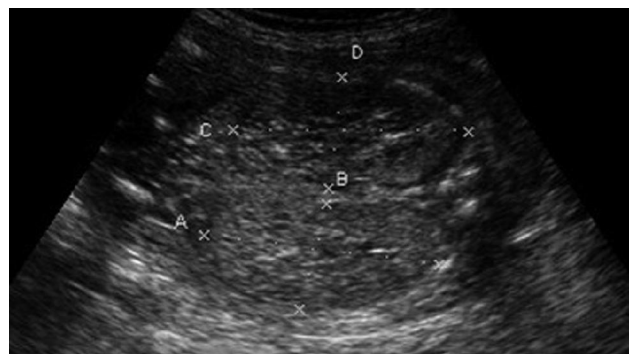
**Figure 2:** (b). Aborted fetus with cystic hygroma.

## Meckel-Gruber Syndrome

Meckel-Gruber syndrome is a rare and lethal autosomal recessive disorder characterized by occipital encephalocele, postaxial polydactyly and bilateral dysplastic cystic kidneys (Fig. 3). It can be associated with many other conditions.<sup>3</sup> Antenatal ultrasound examination can establish the correct diagnosis by identifying at least two of the major features described.<sup>4</sup>



**Figure 3:** (a). Occipital encephalocele corresponding to skull defect. Marked oligohydramnios and dilated ventricles.



**Figure 3:** (b). Enlarged polycystic kidneys.



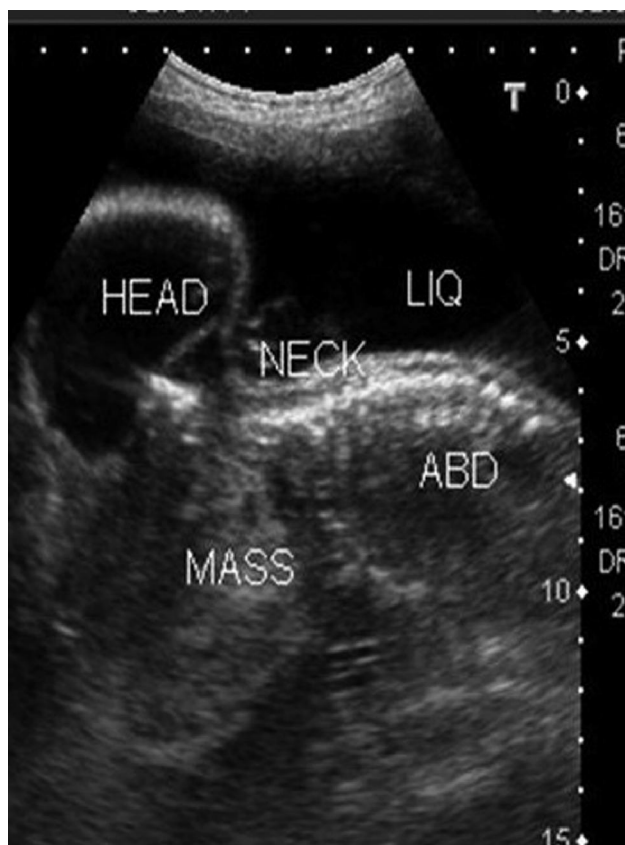
**Figure 3:** (c). Polydactyly was seen in the aborted fetus. This finding was missed on ultrasound.



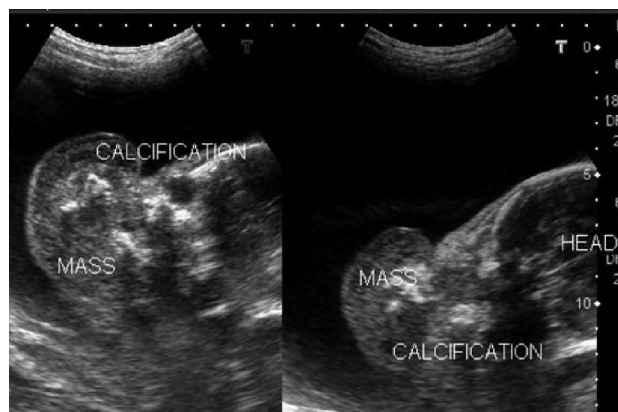
**Figure 3:** (d). Aborted fetus showing enlarged abdomen due to polycystic kidneys .Posterior skull defect is also seen .

## Fetal Facial Teratoma

Teratoma is the most common congenital neoplasm composed of tissues originating from all three germinal layers, and may occur in a variety of locations. Fetal intracranial teratomas generally are large solid/cystic tumors that often completely replace normal brain tissue. (Fig. 4) They may occur in sacrum, mediastinum or brain and face.<sup>5</sup>



**Figure 4:** (a). Long axis scan showing solid mass in the region of fetal neck and face.



**Figure 4:** (b). Calcifications seen within the mass.

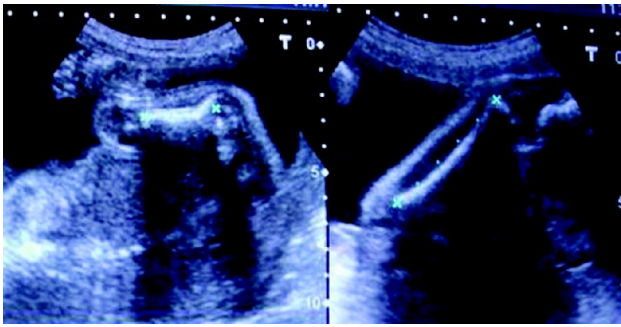
Associated congenital defects are frequently encountered, and if a fetal teratoma is suspected, a careful US survey should be performed.<sup>6</sup> Facial and cervical teratomas can also cause compression on trachea, mandible, carotid vessels and the hypoglossal nerve.<sup>7</sup> The cause of Polyhydramnios is unclear but may be related to pressure on upper gastrointestinal tract or transduction from the tumor.<sup>8,9</sup>



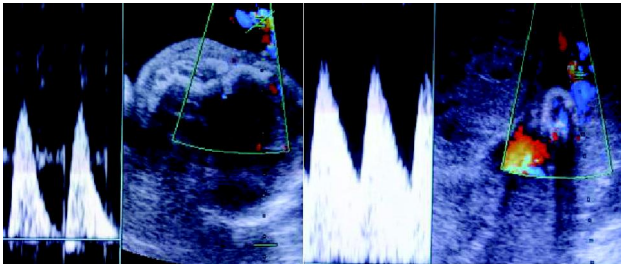
**Figure 4:** (c). Complex mass seen in the aborted fetus. Histopathology confirmed the diagnosis of teratoma.

## Thanatophoric Dwarf

Thanatophoric dysplasia is the most common type of neonatal lethal osteochondrodysplasias, with an estimated frequency of nearly of 1 in 20,000 births. It is a disorder characterized by extremely short ribs, tubular bones and macrocephaly.<sup>10</sup> The prenatal diagnosis of thanatophoric dysplasia has been well established by ultrasonography in the second trimester; however it is not always possible to differentiate the thanatophoric dysplasia fetuses from the others with skeletal dysplasias like fibrochondrogenesis or atelosteogenesis by ultrasonography. We established diagnosis of this condition in a twin pregnancy and correlated our findings with postnatal dead fetus with the help of pathological and radiological examination (Fig. 5).



**Figure 5:** (a). Short femur in the effected fetus. Normal femur in the twin also seen.



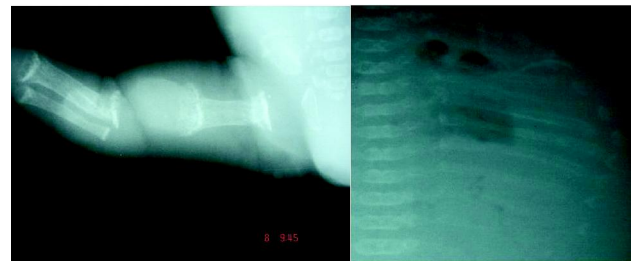
**Figure 5:** (b). Absent end diastolic flow seen in the umbilical artery of effected fetus. Note normal diastolic flow in the other twin.



**Figure 5:** (c). Notice dead fetus with shortening of long bones and normal alive twin.



**Figure 5:** (d). Placenta with both abnormally thin and normal umbilical cords.



**Figure 5:** (e). X- ray of aborted fetus showing metaphyseal flaring. Enlarged epiphysis of knee appearing to be inserting in metaphysis. Horizontal ribs with cupped anterior ends.

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