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Original Article

DIFFERENTIAL DIAGNOSIS OF A CASE OF BILATERAL CYSTIC SWELLING OF NECK IN AN ABORTED FETUS

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ABSTRACT

Objective: To analyse the anatomical and radiological observations in a fetus with bilateral cystic swelling of neck.

Materials and Methods: A total of 30 aborted fetuses were studied for academic purpose ranging from age 12 weeks to 36 weeks. A male aborted fetus of 24 weeks with huge swelling in cervicothoracic region was investigated by radiological and ultrasonographic examination. A detailed foetal autopsy was conducted for associated anomalies.

Observation: A huge cystic swelling was observed in the posterior region of neck and upper thorax with mandibulo facial defect. About 1.2cm defect was noted in occipital bone with herniation of echogenic brain contents and hypoechoic collection in to the occipital region.

Conclusion: Prenatal diagnosis of Cystic hygroma and Encephalocele can be made from 9th week onwards, which will be helpful for planning delivery or to deal with neonatal complications.

KEYWORDS: Cystic Hygroma; Encephalocele; Spina bifida; Multiple Pterygium.

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INTRODUCTION

Lymphangiomas are congenital lymphatic malformations that occur most commonly in the head, neck or axilla ; however , they may arise anywhere in the developing lymphatic system and constitute 5.6% of all benign lesions of infancy and childhood[1].

Encephalocele refers to a group of rare congenital anomalies of the central nervous system where brain tissue protrudes from a defect in the skull. Encephalocele can occur in isolation, as well as with other unrelated congenital defects or as part of a well described syndrome[2].

MATERIALS AND METHODS

After approval by the institutional ethical committee 30 aborted foetuses of 12 weeks to 36 weeks gestational age and both sexes were collected from Government Maternity Hospital with prior informed consent from parents or close family members. Among these a male aborted fetus of 24 weeks age weighing 1.25 kg presented a huge bilateral neck swelling with multiple pterygium-webbing.

OBSERVATIONS:

Following external features were observed in this case.(Figs.1&2)

1. Hypertelorism.
2. Saddle nose.
3. Micrognathia.
4. Uplifted auricle.
5. Low posterior hair line with unswept hair directional pattern.

6. Bilateral Cystic swellings with a midline septum.

7. Measurements of swellings : Right - 18 x 11 cm, Left – 17 x 5 cm

Fig. 1: Anterior view of neck swelling, facial features.

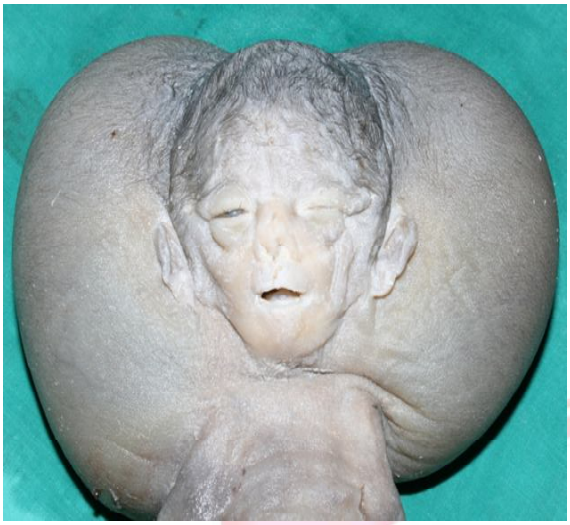
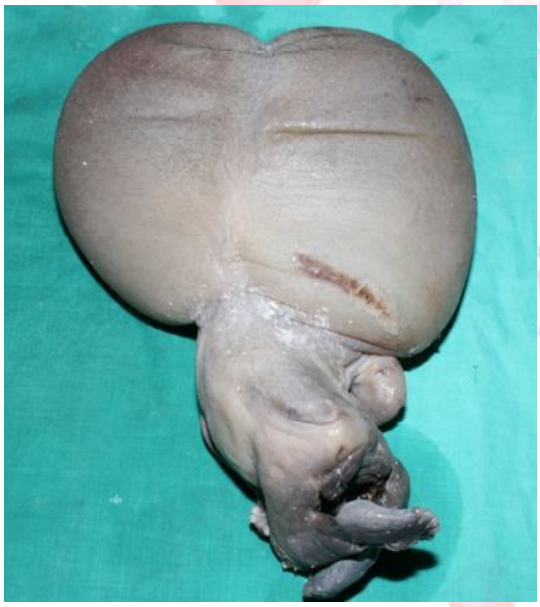


Fig. 2: Posterior view of neck swelling.



Radiological Investigations:

The aborted fetus was subjected to plain x-ray, neurosonogram and CT scan. The following observations were recorded.

Findings of plain x-ray (AP view) (Fig.3) were Bilateral well defined soft tissue density lesion in posterolateral aspect of neck, hyper flexion and rotation of dorso-lumbar spine, Non-visualization of posterior elements in the region of cervical and upper thoracic spine. Plain x-ray (lateral view) (Fig.4) revealed bony defect in occipital bone in continuity with the soft tissue density with no evidence of calcification.

Fig. 3: Plain x-ray (AP view).

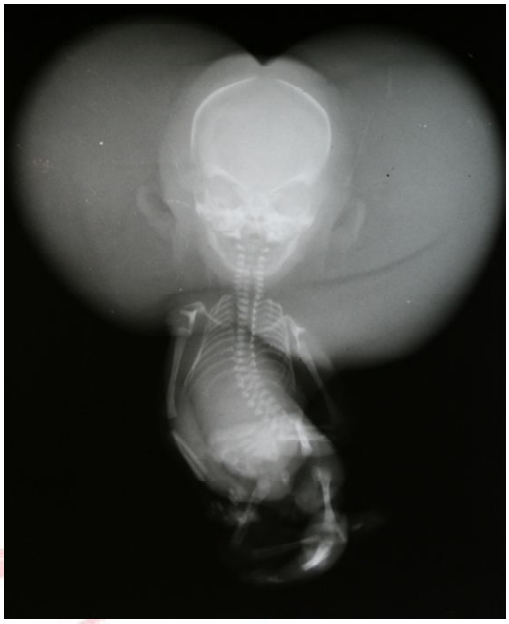
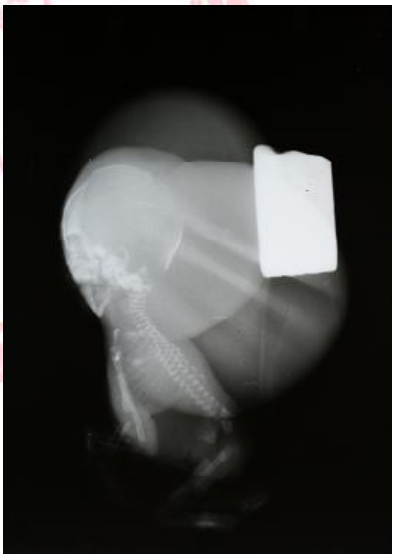


Fig. 4: Plain X-ray (lateral view).



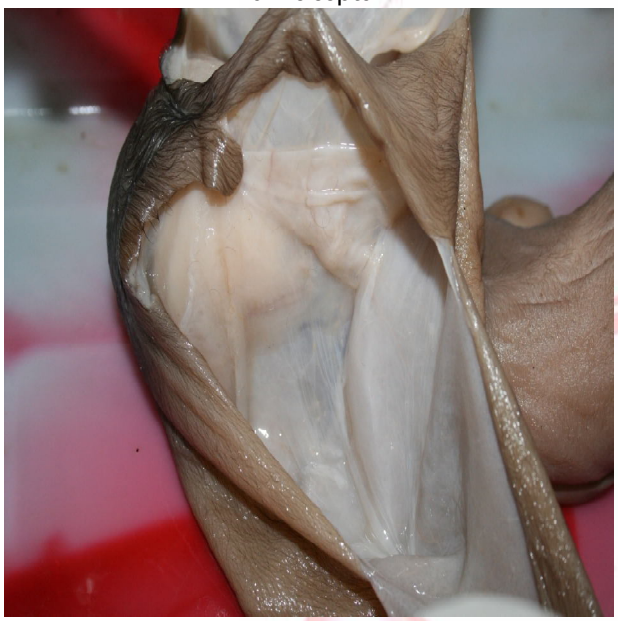
Findings of neurosonogram were 1.2 cm defect in occipital bone with herniation of echogenic brain contents and hypoechoic collection into occipital region. Brain parenchyma, ventricles and sulci appeared normal. Well defined cystic lesions with thick internal echoes noted on either side of head & neck in continuity with soft tissues of neck.

Findings of CT scan were 1.5 cm bony defect in occipital region with 5.5 x 3.8 cm well defined hypo dense lesion of 15-22 HU posterior to occipital bone. Herniation of soft tissue density of 37-40 HU noted through the defect in the occipital bone into the hypo dense lesion. Well defined soft tissue density lesion of 14-20 HU of head and neck measuring Right side 11.5x7cm,

Left side 10.4x6.7cm. The above swelling was in communication with soft tissue of neck.

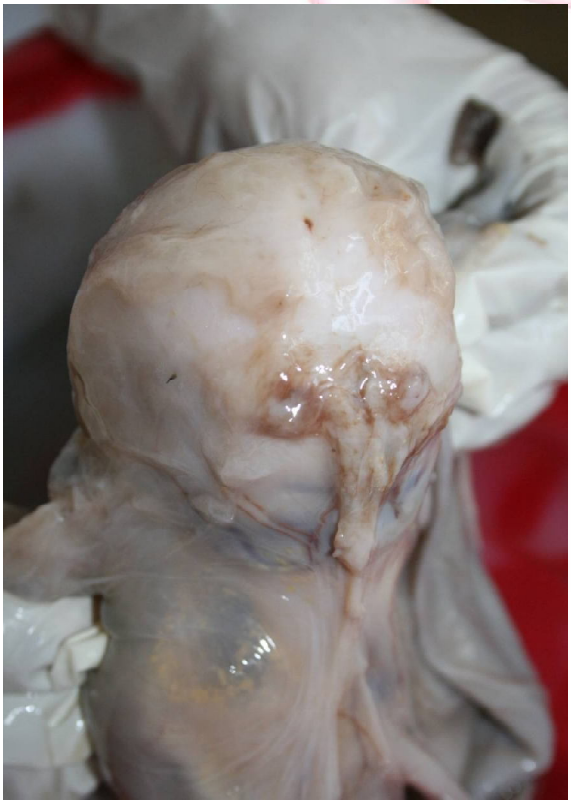
Later autopsy of the fetus was performed which revealed that the swelling had a midline septum and both right and left side the cysts were multilocular and more than 2 cms in size.(Fig. 5)

Fig. 5: Macro cystic Multilocular cyst – Left side with midline septum



After removal of the cyst, site of defect in occipital bone and the brain tissue protruding were visible.(Fig. 6)

Fig. 6: Brain tissue protruding through defect in occipital bone.



In the thorax hypoplasia of lungs and normal heart were observed(Fig.7). In the abdomen the liver was enlarged and caecum along with appendix were sub-hepatic in position(Fig.8). The kidneys and suprarenals were normal.

Fig. 7: Thorax -Hypoplasia of lungs, Normal Heart.

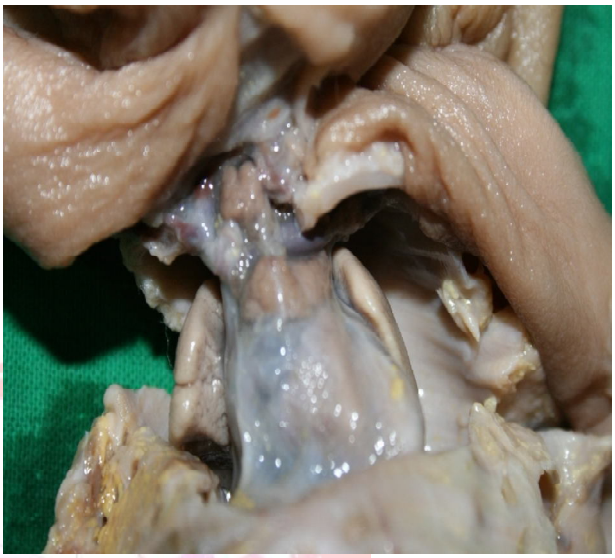
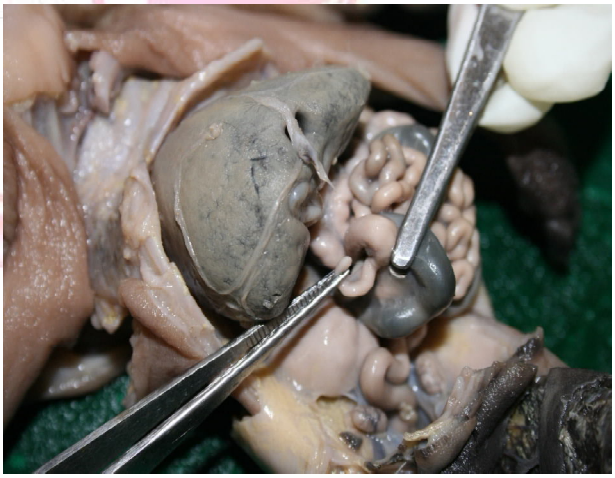


Fig. 8: Abdomen - Enlarged Liver, Sub-hepatic caecum and appendix.



DISCUSSION

Obseravations and investigations of present case suggest the following differential diagnosis.

1. Multiple pterygium.

2. Defect in occipital bone with associated spina bifida suggesting encephelocele.

3. Bilaterally symmetrical soft and cystic swelling with septation and extent of translucency suggesting cystic hygroma.

According to Sudha Sharma and Manudeep Singh [3] causes of cystic hygroma can be classified as -

1. Congenital:

Environmental factors - maternal infections or alcohol abuse.

Genetic factors:

a) Chromosomal anomalies - Trisomy 13,18,21 and Turner syndrome.

b) Non-chromosomal (Syndromic) - Noonan, Klinefelters, Fryns, multiple pterygium syndromes and achondroplasia.

2. Acquired:

Trauma(including surgery) Inflammation obstruction of a lymphatic drainage pathway.

Cystic hygroma, associated with a normal karyotype, can be inherited as an autosomal recessive trait [4].

Pathogenesis of cystic hygroma: includes 3 major theories

Failure of lymphatics to connect to the venous system[5].

Abnormal budding of lymphatic tissue[6].

Sequestered lymphatic rests that retain their embryonic growth potential[7].

Van der Putte[8] studied aborted fetuses with cervical cystic hygroma and Turner syndrome. He confirmed that no lymphatic venous communications were present near the jugulosubclavian junctions on either side.

Causes of Encephalocele may be[9]

- Neural tube defects – spina bifida in 7.3% of cases.
- 75% of cases there will be Occipital bone defect.
- Folate deficiency in early weeks of gestation.

Early maternal hyperthermia is one cause in the genesis of isolated occipital encephalocele[10].

It may be ultrasonographically difficult to differentiate occipital Encephalocele from cystic hygroma, teratoma, scalp oedema, haemangioma, brachial cleft cyst and clover leaf skull in the prenatal period[9].

Computerised Tomography is useful but Magnetic Resonance Imaging is best for demonstrating brain tissue in the Encephalocele[9].

CONCLUSION

Features of the present case were suggestive of a rare variant of lethal multiple pterygium syndrome with localized bilateral (septated) multilocular, macrocystic postero-lateral cystic hygroma of neck with occipital Encephalocele.

Prenatal diagnosis of cystic hygroma help obstetrician for planning delivery which reduces infant morbidity and mortality. It is even helpful to deal with Neonatal complication in consultation with pediatric surgeon.

Conflicts of Interests: None

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