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Cystic Hygroma: An Enigma

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Abstract: *Fetal cystic hygromas are congenital malformations of the lymphatic system appearing as single or multiloculated fluid-filled cavities, most often in the neck. They are thought to arise from failure of the lymphatic system to communicate with the venous system in the neck. They often progress to hydrops and cause fetal death. When a hygroma is detected during fetal life, careful sonographic examination of the entire fetus, determination of the fetal karyotype and an evaluation of the family history are indicated. The disease course of an infant with cystic hygroma is unpredictable. When diagnosed prenatally, the overall prognosis is poor. Cystic hygroma diagnosed after birth is usually associated with a good prognosis. This article reports a case of cystic hygroma detected at 28 weeks of gestation by ultrasonography. The diagnosis was confirmed by other corroborative findings and the pregnancy was terminated after taking the mother's consent. We also discuss the embryologic, genetic and pathologic correlates of these lymphatic system abnormalities as well as the clinical course and outcome of the fetus or newborn with a cystic hygroma. Management strategies are reviewed including newer nonsurgical therapies for the neonate with a cystic hygroma.*

Key words: *Chromosome aneuploidy, Cystic hygroma, Hydrops fetalis, Lymphangioma, Nuchal thickening*

I. Introduction:

Cystic hygroma is a congenital malformation of the lymphatic system in which obstruction between the lymphatic and venous pathways in the fetal neck leads to lymph accumulation in the jugular lymphatic sacs of the nuchal region. Cystic hygromas can be subclassified as septated or nonseptated (simple). In the first trimester, the overall prevalence of septated and nonseptated cystic hygromas is about 1 in 100 fetuses, and the prevalence of septated cystic hygromas is 1 in 285 fetuses [1, 2]. Cystic hygroma can affect any anatomic sub site in the human body. It usually affects the head and neck (~75%), with a predilection for the left side. Within the neck, the posterior triangle tends to be most frequently affected. Approximately 20% of CHs occur in the axilla; less common sub sites include the mediastinum, groin and retroperitoneum. CH is synonymous with cystic lymphangioma, which is also known as a macrocystic lymphatic malformation. Lymphangiomas are thought to arise from a combination of the following:

- Failure of lymphatics to connect to the venous system
- Abnormal budding of lymphatic tissue
- Sequestered lymphatic rests that retain their embryonic growth potential

These lymphatic rests can penetrate adjacent structures or dissect along fascial planes and eventually become canalized. These spaces retain their secretions and develop cystic components because of the lack of a venous outflow tract. The nature of the surrounding tissue determines whether the lymphangioma is capillary, cavernous or cystic.

CHs tend to form in loose areolar tissue, whereas capillary and cavernous forms of lymphangiomas tend to form in muscle. Studies using cell proliferation markers have demonstrated that lymphangioma enlargement is related more to engorgement than to actual cell proliferation. Molecular studies suggest that vascular endothelial growth factor C (VEGF-C) and its receptors may play an important role in the development of lymphatic malformations.

In addition to congenital development, lymphangiomas can be acquired. They can arise from trauma (including surgery), inflammation or obstruction of a lymphatic drainage pathway.

II. Case Presentation:

25 years old 2nd gravida at 28 weeks of pregnancy came for regular antenatal check up. She had delivered a full term, normal, male baby vaginally 3 years back. Her weight was 69 kg and height was 160 cm with BMI of 27. Her blood group was B Rhesus positive with no h/o Diabetes or Hypertension or consanguinity. She had no h/o smoking, alcohol or any other substance abuse.

2D ultrasound findings were:

A single living fetus with breech presentation and normal fetal movements was seen. The Heart Rate was 145/min. and weight was 1069 gm. The AFI was 23.09 cm suggesting polyhydramnios. The placenta was posterior, high up with grade I maturation. Cervix measured 3.05×3.16 cm and canal was closed. Rest all parameters in the fetal biometry was normal

Targeted imaging for fetal abnormalities with 4 D scan revealed:

Fetal skin: Present throughout, no breech in continuity

Cranium: Ventricular system, Cerebellum, Cisterna magna and Cervico-vertebral junction: normal. A multiseptate anechoic mass of 6.12×5.13 cm present in right lateral neck below the right pinna suggesting Cystic Hygroma (Fig. 1 & 2).

The spine, face & orbits, thorax and diaphragm were normal.

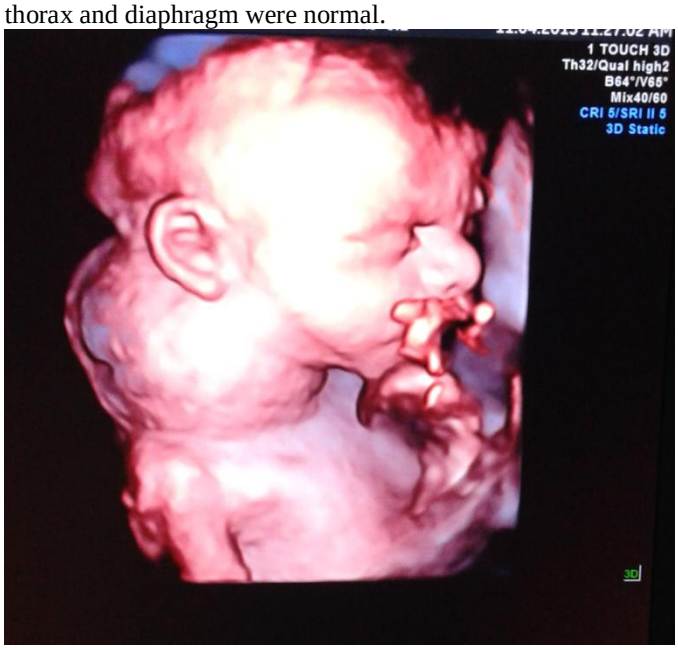

Fig. 1 shows the 3 D picture of the fetus.

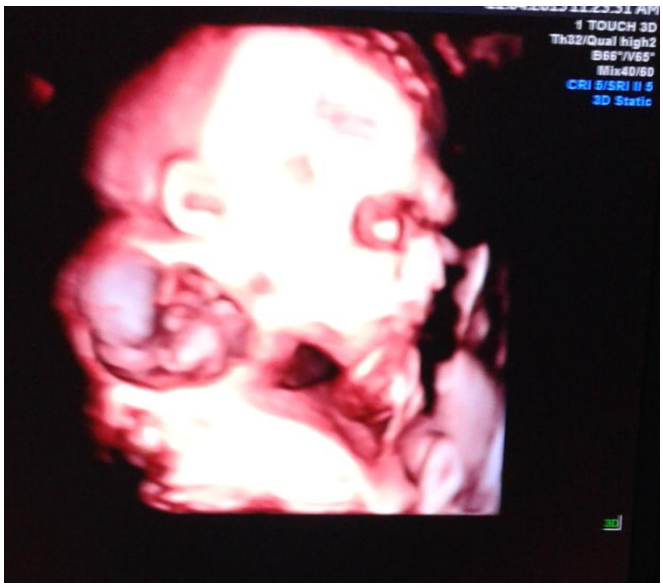

Fig. 2 shows the multiseptate anechoic mass of cystic Hygroma

Karyotype of the baby was 45, XO suggesting Turner's syndrome. The pregnancy was terminated after taking the patient's consent and proper counseling. A female baby was delivered who died immediately after birth. There was a visible swelling in the neck on the right side which showed multiple loculi on dissection (Fig. 3). The condition of the patient was stable after abortion and she was discharged after proper advice.

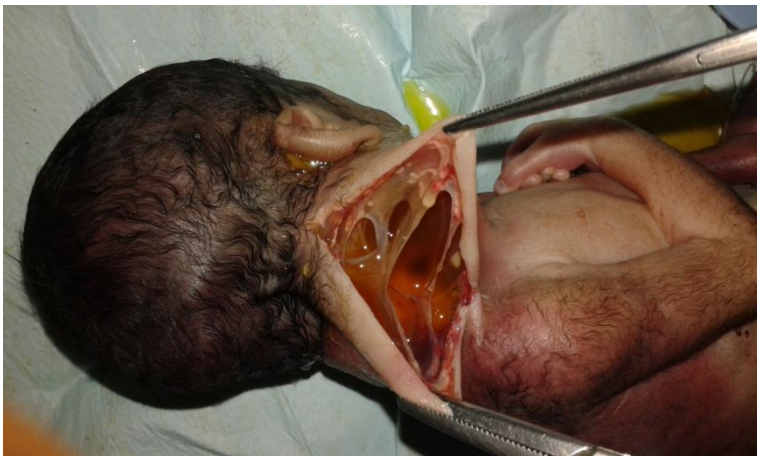

Fig. 3 shows the multiloculated fluid filled cavity of the Cystic Hygroma after delivery.

III. Discussion:

Cystic Hygromas are fluid filled sacculations in the fetal soft tissues that are the result of congenital dysplasia of the lymphatic system. Fetal posterior cervical Cystic Hygroma is a manifestation of a developmental abnormality in the jugular lymphatics which is most commonly seen in 45, X fetuses. Microscopically, it consists of dilated endothelium lined spaces filled with serous fluid and focal interstitial lymphoid aggregates [3].

Etiology:

Karyotypic abnormalities are present in 25-70% of children with CH. CH has been noted to be more common in persons with Turner syndrome, Down syndrome, Klinefelter syndrome and trisomy 18 and 13, though these are not considered to be causative. In addition, several nonchromosomal disorders including Noonan syndrome, Fryns syndrome, multiple pterygium syndrome and achondroplasia are associated with an increased incidence of CH. Intrauterine alcohol exposure has been associated with the development of lymphangiomas. Dissolution of bone caused by either lymphangiomas or hemangiomas is termed Gorham-Stout syndrome.

Embryological basis:

Florence Sabin (1901) has shown that the lymphatic system arises from 5 primitive sacs developed from the venous system [4]. These consist of paired jugular sacs, a single retroperitoneal sac and paired posterior sacs near the sciatic veins. Endothelial outbuddings from these sacs extend centrifugally to form the peripheral lymphatic system. The cisterna chyli, the thoracic duct and the subclavian lymph sacs develop as secondary structures. McLure and Sylvester (1909) first suggested that cystic hygroma probably arises from sequestrations of lymphatic tissue derived from portions of the primitive sacs during embryonic life. He believed that these rests retain their rapid and proliferative growth potential. These sequestrations may never achieve anastomoses with the larger lymphatic channels. Goetsch (1938) did some further histological work and found that sequestrated lymphoid tissue formed cysts: endothelial fibrillar membranes sprout from the walls of the cysts, penetrate into surrounding normal tissue and then canalize to produce more cysts filled with secretions from the fibrillae.

Prognosis:

In some situations, a cystic hygroma can be present in a healthy baby. If a chromosome abnormality is not found in the fetus, the outcome is generally better than for those who do have a chromosome abnormality. If a cystic hygroma is an isolated finding that resolves around 18-20 weeks gestation and the fetus has normal chromosomes, the outcome is good for 54-80% of these cases. In cases in which an isolated cystic hygroma does not resolve by 20 weeks gestation, 2-9% has a good outcome [5]. Overall, there is generally a poor prognosis associated with the prenatal finding of cystic hygroma. Studies have indicated that smaller cystic hygromas are more likely to resolve. Oligohydramnios (not enough amniotic fluid) or polyhydramnios (too much amniotic fluid) predicts a poor outcome. Hydrops occurs 22-76% of the time with a cystic hygroma and is almost always associated with miscarriage or fetal death.

Treatment:

A baby with a prenatally diagnosed cystic hygroma should be delivered in a major medical center equipped to deal with neonatal complications. An obstetrician usually decides the method of delivery. If the

cystic hygroma is large, a caesarean section may be performed. After birth, infants with persistent cystic hygroma must be monitored for airway obstruction. A thin needle may be used to reduce the volume of the cystic hygroma to prevent facial deformities and airway obstruction [5]. Close observation of the baby by a neonatalogist after birth is recommended. If resolution of the cystic hygroma does not occur before birth, a pediatric surgeon should be consulted. Cystic hygromas that develop in the third trimester (after thirty weeks gestation) or in the postnatal period are usually not associated with chromosome abnormalities. There is a chance of recurrence after surgical removal of the cystic hygroma. The chance of recurrence depends on the extent of the cystic hygroma and whether the wall of the cyst was able to be completely removed.

Bleomycin is an anti-neoplastic antibiotic, produced by the fermentation of streptomyces verticillus. Discovered in 1965, this drug was found to cause single- and double-strand DNA breaks and inhibition of DNA and RNA synthesis [6]. Since then it has been used for its anti-neoplastic property to treat malignancy. In the treatment of malignant pleural effusion, it was observed that bleomycin caused marked fibrosis and scarring. This sclerosing property was first put into use in the treatment of lymphatic malformation in 1977. Intralesional bleomycin injection is useful for the treatment of cystic hygroma. It should be used in patients with large cystic masses and extensive invasion to reduce the risk of injury to vital organs [7].

IV. Conclusion:

Hygroma cysticum coli or cystic hygroma remains a complex entity in terms of its development and management. Most recently, cystic hygroma has been categorized as part of a larger spectrum that includes lymphangiomas. The majority of lymph-angiomas occur in the head and neck as cystic hygromas with the posterior cervical region as the most common site. Since its original description, there have been many attempts at treatment modalities: Surgical excision remains the treatment of choice. Bleomycin intralesional sclerosant has response rates comparable to those of surgical removal but with the advantage of avoiding inadvertent nerve damage and scarring [8].

Fetuses with first-trimester simple nuchal hygromas are at high risk for aneuploidy and should be offered prenatal testing. Such fetuses with normal karyotypes will likely resolve their hygromas by 18 weeks gestation and most will be phenotypically normal at birth.

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