Posterior Midline Cervical Fetal Cystic Hygroma

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Abstract

Posterior midline cervical cystic hygromas (PMC) are frequently found associated with chromosomal aberrations and usually do not survive. The present report illustrates diagnosis of this condition by sonography in an 18 weeks old fetus and an amniocentesis revealed 45 XO karyotype and increased concentration of alpha- fetoproteins. Pregnancy was terminated in view of Turner's syndrome. The etiology and natural history of the condition is reviewed. ***(J Postgrad Med 1992; 38(2):93-96)***

Key Words: PMC Fetal cystic hygroma, antenatal sonography, karyotyping.

Cystic hygromas are anomalies of the lymphatic system characterised by single or multiple cysts within soft tissues, usually involving the neck[1]. In fetal life, cystic hygromas of the posterior triangle of neck are usually associated with various degrees of lymphoedema[2]. Singh and Carr recognised that this type of cystic hygroma was frequent in aboduses with a 45 X 0 karyotype[3] (Turner's Syndrome). Van der Putte has conducted studies of lymphatic systems to explore the morphogenesis of the defect (quoted by Chervenak et al[2]). Smith and Graham propose a mechanism of lymphatic maldevelopment and neck webbing and peripheral oederna in cases of Turner's Syndrome[4]. Prenatal ultrasound helps in the diagnosis of cystic hygromas and differentiate it from occipto-cervicalmeningomyelocele. It also allows the collection of amniotic fluid for chomosomal and biochemical study.

The present report substantiates the concept of "Jugular lymphatic - obstruction sequence"

Case Report

Diagnosis of PMC cystic hygroma was made in an 18 week fetus during the prenatal ultrasound. A thin-walled, multiseptate cystic structure posterior to the fetal head and neck eccentrically situated with respect to the long axis of the fetus without vertebral column defect (Fig 1) was diagnostic of hygroma. Gestational age was determined by biparietal diameter and it was corresponding with the menstrual history. A careful sonographic search for other associated congenital anomalies was done. Amniocentesis was performed; fetal karyotype was 45 X O and alpha-fetoprotein concentration was increased. Pregnancy was terminated in view of Turner's syndrome in fetus and a post-termination autopsy confirmed the cystic hygroma in the posterior aspect of the neck (Fig 2,3)

Discussion

Cystic hygromas probably develop from a defect in the formation of lymphatic vessels. In the embryo, the lymphatic system drains into the jugular lymphatic sacs. A communication between this primitive structure and the jugular vein is formed at 40 days of gestation. Failure of development of this communication results in lymphatic stasis[2]. Dilatation of the jugular lymphatic sac leads to the formation of cystic structures in the cervical region. If a connection between the lymphatic and the venous system does not occur at this point, a progressive peripheral lymphoedema and hydrops develops, leading to early intrauterine death. [2] Should the connection be formed, the sequence is interrupted, fluid collections resorbed; redundant skin gives rise to webbed neck (pterygium colli). Webbing of neck and puffiness of hands and feet are characteristic postnatal features of Turner's syndrome. Uplifting and anterior rotation of the ears and an abnormal hair pattern are other consequences of overdistension of jugular lymphatic sacs.

Lymphatic obstructive sequence[4]

Lack of jugular lymph sac
drainage into jugular vein
Accumulation of lymph fluid
within lymphatic system

Distended jugular lymph sac	Distended tertiary tributaries
1. Overgrowth of covering skin	1. Peripheral oedema
2. Rotated auricle	2. Deep-set narrow nails
3. Low posterior hairline	3. Puffy hands; feet

Cystic hygromas may be diagnosed reliably by ultrasound[5,7]. The hallmark of fetal cystic hygroma, as

seen sonographically in utero, is a thin-walled, multiseptate fluid-filled mass closely related to the fetal head and/or neck[8]. Sonographic features that differentiate cystic hygromas from other craniocervical masses are:

a) an intact skull and spinal column
b) lack of a solid component to the mass
c) constant position of mass relative to fetal head
d) presence of cysts and septae
e) tubular serpiginous or multilocular intradermal fluid collections are seen in the abdominal wall of the fetus[9].

The diagnosis of cystic hygromas should be made in the first trimester[10]. The differential diagnosis includes cervical meningomyelocele; encephalocele, neck tumours, twin sac of a blighted ovum or nuchal oedema. The distinction is often difficult[11]. In neural tube defects, the morphological abnormalities of the head and / or spine are frequently identifiable[12]. The presence of hydrocephalus increases the index of suspicion for encephalocele. Polyhydramnios has been noted in a large percentage of neural defects and was not present in our case of PMC fetal cystic hygroma. Furthermore, intrauterine cystic defect in fetal cystic hygroma is an asymmetrically distributed defect with respect to the long axis of the head and-spine in contradiction to neural tube defects, which are usually symmetrically situated as was our case, but without vertebral column defect appearance[13].

The concentration of alpha-fetoprotein in amniotic fluid in this case was elevated. The mechanism of this elevation in amniotic fluid when the cystic hygroma is intact is uncertain; but it may involve transudation through edematous skin or bowel mucosa[14].

Several other congenital anomalies have been reported in association with fetal cystic hygromas. These include Noonan's syndrome, fetal alcohol syndrome, distichiasis-lymphedema syndrome, familial pterygium colli and several chromosomal aneuploidies[15].

In the present case, due to abnormal karyotype (45 X -0) the pregnancy was terminated. In the presence of isolated cystic hygromas no modification of standard obstetrical management is required. On some occasions hygromas have been associated with prolongation of the second stage of labour[3]. A Cesarean section may be required if there are gigantic lesions. Infants need to be closely monitored for the risk of airway obstruction.

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