

dystrophy (ATD) described by Jeune *et al.* It is suggested that ATD may be a variant of Ellis van Creveld syndrome with lower incidence of ectodermal dysplasia and more severe thoracic dystrophy(4). There are no vertebral or skull abnormalities in both these conditions(2,4,5).

Saldino and Noonan in 1972(6) described in two still born siblings a malformation syndrome characterized by severe thoracic dystrophy, micromelia, hypoplastic long bones, multiple visceral anomalies and abnormal osseous maturation including that of spine. Our case also had similar skeletal feature with visceral malformations in the form of pulmonary hypoplasia, ventricular septal defect and urogenital abnormalities. The presence of vertebral changes indicate a generalised skeletal dysplasia. Short rib-polydactyly syndrome type II (Majewski) is also characterized by thoracic dystrophy and polydactyly; cleft lip and palate are frequently associated. However, the pelvic and long bones are not involved(7). The respiratory distress in most cases with short rib polydactyly syndromes I and II are not remediable and most cases die soon after birth.

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## Prenatal Diagnosis of Jugular Obstruction Sequence at 13 Weeks of Gestation

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Of all the major fetal anomalies reported in the first trimester of pregnancy, jugular lymphatic obstruction sequence is one which can be definitely diagnosed by ultrasound scanning. This case reported here at the thirteenth week of gestation is the first from our nation and the second in the world literature(1).

## Case Report

A 28-year-old third gravida was

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*Received for publication November 14, 1990;*

*Accepted March 28, 1991*



Fig. 1. Longitudinal ultrasonogram showing the cutaneous lymphangiectasis with occipito-cervical and axillary cysts.

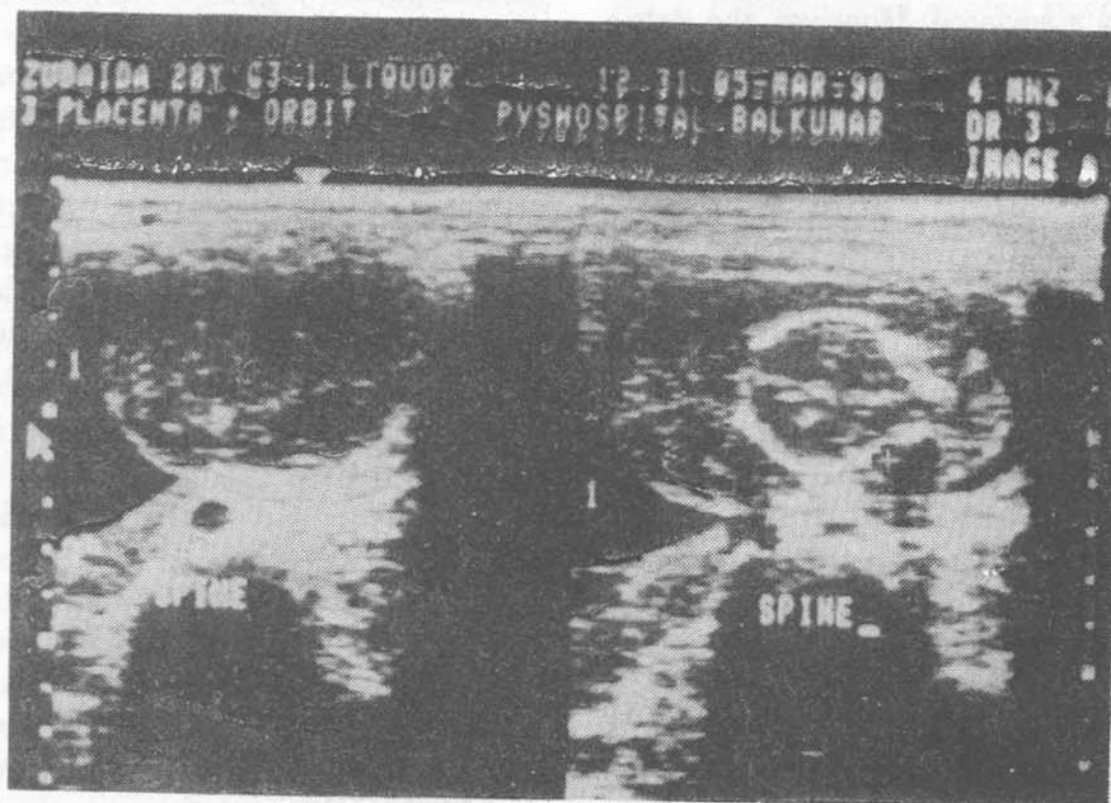
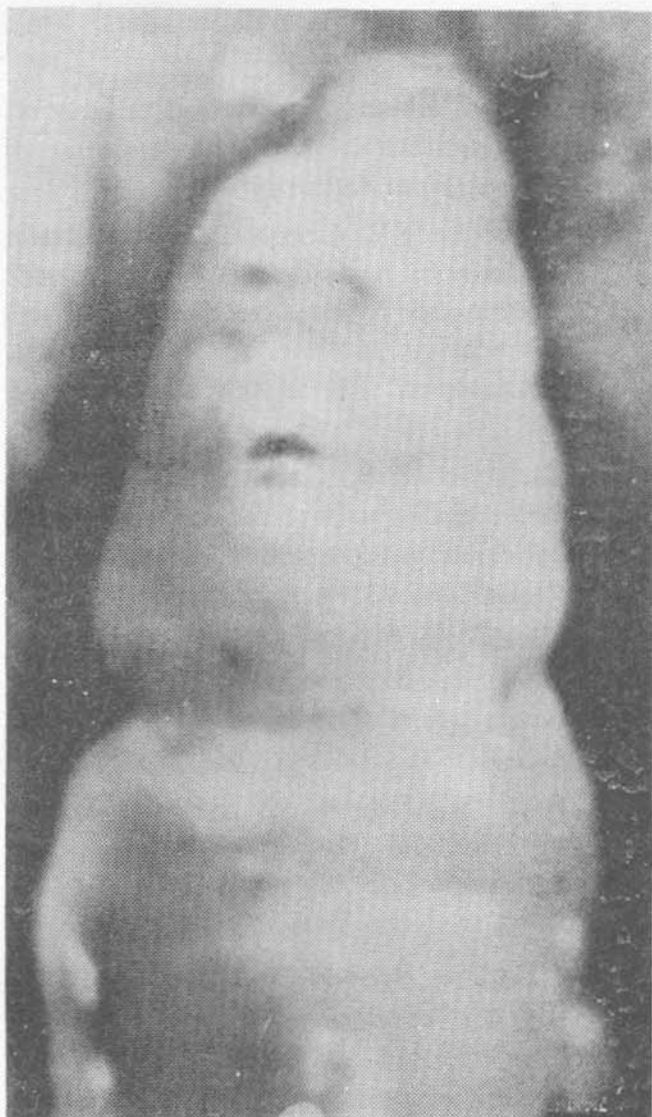


Fig. 2. (a) Axial scan of head above the cranium showing scalp lymphangiectasis.  
(b) Axial view of the smaller sized calvarium compared to the scalp lymphangiectasis.



*Fig. 3. The immediate post-abortion appearance of the fetus.*

scanned because of the previous history of severe degree oligohydramnios in her second pregnancy. The ultrasound scanning showed the biometry as that of 13 weeks of gestation. The fetal appearance was striking by the presence of massive cutaneous lymphangiectasis resulting in pronounced peripheral edema mainly involving the head, neck, upper limbs and the trunk (*Fig. 1*). There were two larger cysts in the suboccipital region extending anterolaterally around the neck and separated by the nuchal ligament. Smaller cysts

were seen in the axillary regions. The head was very massive due to surrounding extensive cutaneous lymph stasis (*Fig. 2*). No evidence of hydrops such as pericardial, pleural or peritoneal effusion were detected. The placental appearance was normal.

The pregnancy ended in spontaneous abortion after 4 days of the diagnosis (*Fig. 3*). The autopsy findings were consistent with the antenatal diagnosis.

### Discussion

The first accepted explanation regarding the embryology of the lymphatic system was postulated by Huntington in 1908(2); supported by Mc Clure and Butler in 1925(3). Sabin put forward another description in 1909(4). The primitive paired jugular lymph sacs lying lateral to the jugular veins usually establish its connection with the veins at 40 days of gestation. The failure of this process leads to the formation of cystic hygroma and other sequences of lymphatic obstruction(5). This case illustrates the gross changes that can be detected by ultrasound in the late first trimester.

The actual incidence of massive degree of jugular lymphatic obstruction sequence is not known whereas the incidence of cystic hygroma is reported to be 1 in 200 aborted fetuses with a crown-rump length of more than 30 mm(6) and 1 in 6000 of pregnancies(7). An autosomal recessive inheritance is reported in some(8). The lymphatic obstruction sequence can manifest in different sites(9) and with varying grades. The main site of involvement is the neck(10) which accounts for 75% of the cases. The axilla is involved in 20% of cases. In severe affection the whole body may be involved, as in this case. The cervical web seen in Turner's syndrome is sug-

gested to be as a result of the regressed cysts, though it is not specific(11). A number of other chromosomal aberrations are reported in association with this condition and the majority have nonimmune hydrops(12). The latter was absent in this case, probably due to the fact that the malformation was detected at an earlier stage.

The milder form of lymphatic obstruction sequence manifests as cystic hygroma which can be differentiated from encephalocele, meningo encephalocele and neck tumors by detailed scanning. In this case the gross obstructive cutaneous lymphangiectasis associated with the suboccipital and cervical cysts excludes the possibility of any other condition which can mimic this particular pathology(13).

In an analysis of 15 cases, the gestational age at which the diagnosis of cystic hygroma was made varied from 18 to 29 weeks(14). The mean period of gestation at the time of diagnosis in another review of 24 cases of cystic hygroma coli was 19.3 weeks(15) within a range of 14 to 26 weeks; whereas this was detected at 13 weeks.

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