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Ultrasonic Evaluation of Twin Pregnancies Associated With Raised Serum Alpha-Fetoprotein Levels

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Five cases of twin pregnancy associated with elevated maternal serum alpha-fetoprotein levels are presented. In each instance an abnormality was detected by ultrasound examination—in four cases this represented anomalous development of one or both fetuses: discordant anencephaly; a monozygotic heterokaryotypic twin pregnancy with coexistent Turner's syndrome and intrauterine death; concordant body-stalk syndrome; and fetal exomphalos together with an acardiac monster. In the remaining case, there had been an intrauterine death of one of the twins. Ultrasound examination proved valuable in all instances; three pregnancies were terminated and single healthy infants were delivered from each of the other two pregnancies.

Key words: Serum alpha-fetoprotein, Fetal abnormality, Twin pregnancy, Ultrasound

INTRODUCTION

Serum alpha-fetoprotein (AFP) screening at 16 to 20 weeks is effective in the detection of fetal neural tube defects in areas which harbor a high incidence of such abnormalities [6,13]. Other fetal abnormalities, including anterior abdominal wall defects [5], teratomata [10], missed abortion, threatened abortion, and multiple pregnancy [11,14] may also be associated with elevated serum AFP. Experience in the West of Scotland [8] has shown that the mean level of serum AFP in twin pregnancies corresponds closely to the 97th percentile [2.5 multiples of the median (MOM)] for singletons; this latter figure represents the intervention line in single pregnancy. When serum AFP levels are found to be elevated [11], a detailed ultrasound examination by an experienced operator is mandatory. The presence of a twin pregnancy is generally accepted as providing adequate explanation of raised values.

This paper describes five cases of abnormal twin pregnancy detected by ultrasound examination during the second trimester. All the pregnancies were associated with serum AFP levels that were elevated by singleton standards (greater than 2.5 MOM). In four cases there was an anomaly of one or both fetuses. Three pregnancies were terminated as a result of the ultrasound findings; single healthy infants were delivered from each of the other two pregnancies. Three of the patients were attending the antenatal clinic of

the Queen Mother's Hospital, Glasgow. All were examined during a two-year period during which there were 62 twin deliveries; the other two patients were referred from other hospitals. Cases 1–4 have been reported in detail elsewhere [9] and will, therefore, only be described briefly. Case 5 has not previously been reported.

CASE HISTORIES

Case 1

Serum AFP levels were 233 u/ml (6.8 MOM) and 231 u/ml (5.1 MOM) at 18 and 20 weeks, respectively. Ultrasound examination revealed discordant anencephaly. A normal healthy male infant, together with a stillborn female anencephalic, were delivered at 35 weeks.

Case 2

Twin pregnancy was diagnosed by ultrasound at routine first visit examination at 16 weeks. Serum AFP was 84 u/ml (3.0 MOM) one week later. Repeat ultrasound examination showed that one of the twins had died in utero. This became a fetus papyraceous and was delivered along with the healthy co-twin at term.

Case 3

Serum AFP was 92 u/ml (2.9 MOM) at 18 weeks. Ultrasound examination at 15 weeks had demonstrated the intrauterine death of one of the twins; repeat examination at 19 weeks showed a cystic structure related to the neck of the living fetus. This was thought to be compatible with a diagnosis of Turner's syndrome. The pregnancy was terminated; the larger fetus was shown to have a 45, XO karyotype, the smaller a 46, XX karyotype. The placenta was monochorionic, diamniotic.

Case 4

Serum AFP was grossly elevated: 695 u/ml (24.8 MOM) at 17 weeks. Ultrasound examination demonstrated a twin pregnancy with major structural disorganization of both fetuses, together with extreme oligohydramnios. The pregnancy was terminated, revealing concordant body-stalk anomaly.

Case 5

The patient, a 26-year-old primigravida, first presented for antenatal care at 14 weeks of gestational age. There was no prior history of illness or drug ingestion. Serum AFP levels were greatly elevated—135 u/ml (5.6 MOM) and 238 u/ml (8.5 MOM) at 16 and 17 weeks, respectively. When the patient was referred for ultrasound examination at 20 weeks, it showed one live but clearly abnormal fetus with marked scoliosis (but no evidence of spina bifida) and a large defect of the anterior trunk wall with herniation of most of the abdominal organs and much of the heart (Figs. 1a,b). In addition, there was much more extracorporeal tissue than could be accounted for by the albeit large defect. These appearances were thought to be explicable either by the presence of an acardiac monster or of a fetal tumor such as a teratoma. Only four limbs could be identified and there was polyhydramnios.

As a result of these findings, the pregnancy was terminated by intraamniotic injection of prostaglandin E_2. The ultrasound findings were largely confirmed: the fetus had a large exomphalos, the heart being displaced downward into the sac. The abdominal organs

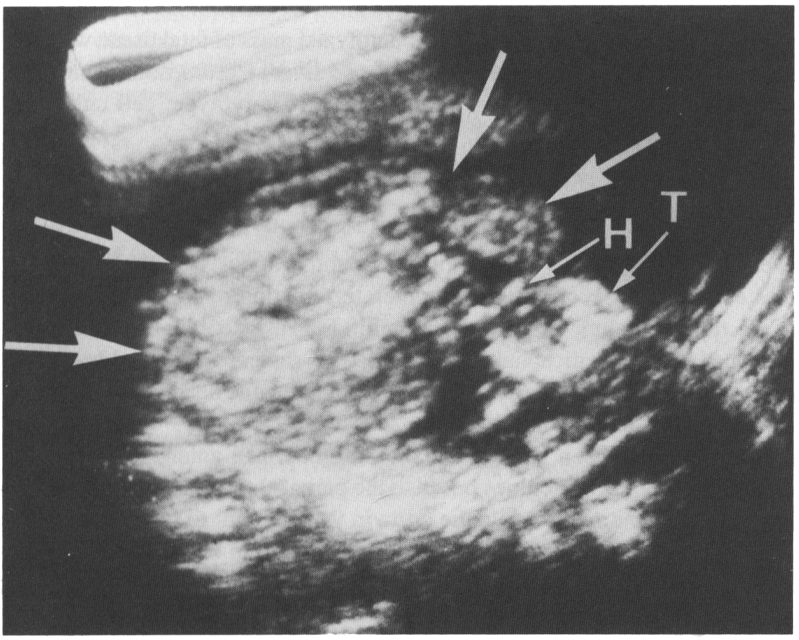

Fig. 1a. Ultrasonogram showing a transverse section through the lowermost part of the fetal thorax. The boundaries of the extracorporeal tissue are demarcated by four arrows. H, fetal heart; T, fetal thorax.

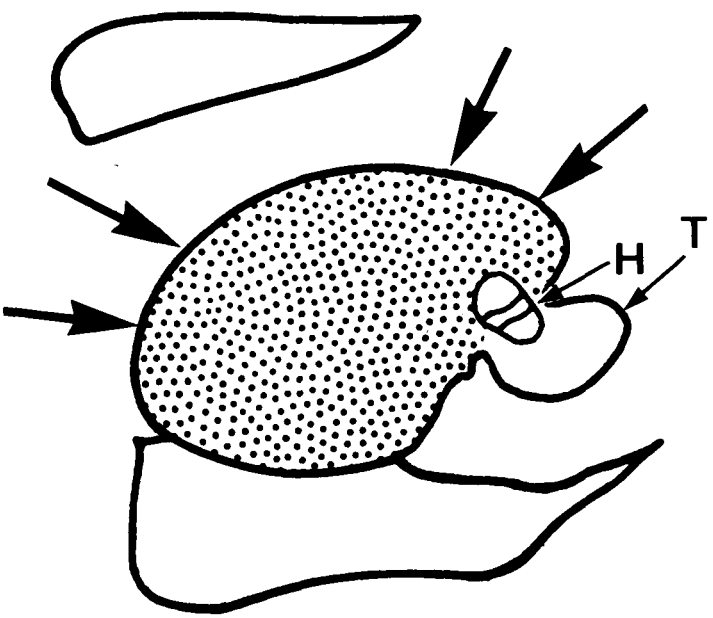

Fig. 1b. Line drawing of the ultrasonic image. The fetal thorax (T) is unshaded. The boundaries of the extracorporeal tissue are demarcated by four arrows (and the tissue is shaded). The fetal heart (H) is partly in the thorax and partly in the sac.

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were otherwise normal. The extra tissue represented an amorphous twin with two well-formed feet fused to a disorganized and partly autolyzed mass of fetal tissue which included cartilage, neural tissue, respiratory epithelium, and liver. Chromosome analysis of cells from the better formed twin showed a normal 46, XX karyotype; cell culture from the acardiac monster proved unsuccessful.

DISCUSSION

There have been relatively few reports of ultrasound diagnosis of abnormalities in twin pregnancies early in the second trimester when termination of pregnancy could be considered. Case reports include discordant spina bifida [4], discordant anencephaly [7,15], conjoined twins [12], and a dicephalus monster [9]. In the cases reported here, the finding of elevated levels of serum AFP proved useful in indicating the need for detailed ultrasound assessment to identify the abnormality.

In the past, there has been some reluctance to undertake antenatal diagnosis in twin pregnancies because of the difficulties posed to the parents by the finding of a handicapping abnormality in one of the twins while the co-twin was thought to be alive and apparently normal. While we were not confronted with this situation in our series, there have been reports of selective destruction of the affected twin. Destructive techniques have included: cardiac puncture [1], fetoscopically guided injection of air into the umbilical vein [3], and removal of the affected twin at hysterotomy [2]. Should such procedures prove safe and effective, they represent better solutions to this problem than does the sacrifice of both the abnormal and the normal fetus by termination of pregnancy.

CONCLUSIONS

In all cases of twin pregnancy associated with raised serum AFP values (by singleton standards), detailed ultrasound examination should be carried out to investigate the possibility of abnormality in one or both of the fetuses. The raised value should not be attributed simply to twinning without further investigation.

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