



**Figure 3** X-ray of the fetus presenting with two femora, two tibiae and two fibulae.

two fibulae, and 10 toes were present. X-ray confirmed the diagnosis (Figure 3). There was no autopsy due to complete preservation of the fetus.

### Discussion

The combined use of 2D and 3D sonography was of value in prenatal diagnosis of sirenomelia. To our knowledge only three reported cases of sirenomelia have been diagnosed prenatally before 19 weeks of gestation<sup>4,5</sup>. In two of the three pregnancies, amniotic fluid volume was only slightly reduced at 14 and 16 weeks of gestation. This was probably due to factors other than fetal urine production contributing to the production of amniotic fluid in the early second trimester. There is no report in the literature of prenatal diagnosis of sirenomelia by 3D or 'Live 3D' sonography. The additional information obtained from the 3D images was the overall view, the confirmation of 2D findings and especially the observation of abnormal fetal movements due to conjoined fetal legs. The diagnosis had been established by 2D ultrasound. However, 3D imaging could increase the physician's confidence and understanding of the fetal condition.

The etiology of sirenomelia is mostly unknown. Chromosomal abnormalities are rare and predominantly males are affected, with a sex ratio of 2.7 : 1. The male-to-female ratio is probably even higher due to the fact that when the external

genitalia were not visible and karyotyping was not performed, the fetus was considered to be female<sup>6</sup>. The present case had a male karyotype. Sirenomelia must be included as part of the differential diagnosis of oligohydramnios in the second trimester of pregnancy, together with premature rupture of the membranes, bilateral renal agenesis, polycystic kidney diseases, obstructive uropathies, and severe intrauterine growth restriction<sup>7</sup>. Since sirenomelia is uniformly lethal, early prenatal diagnosis is required in order to allow termination of pregnancy at an early stage, with minor risks and discomfort for the patient. Incorporation of 3D and four-dimensional sonography could provide a diagnostic feature that could assist in the prenatal diagnosis of sirenomelia.

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### Trisomy 7 following assisted conception treatment

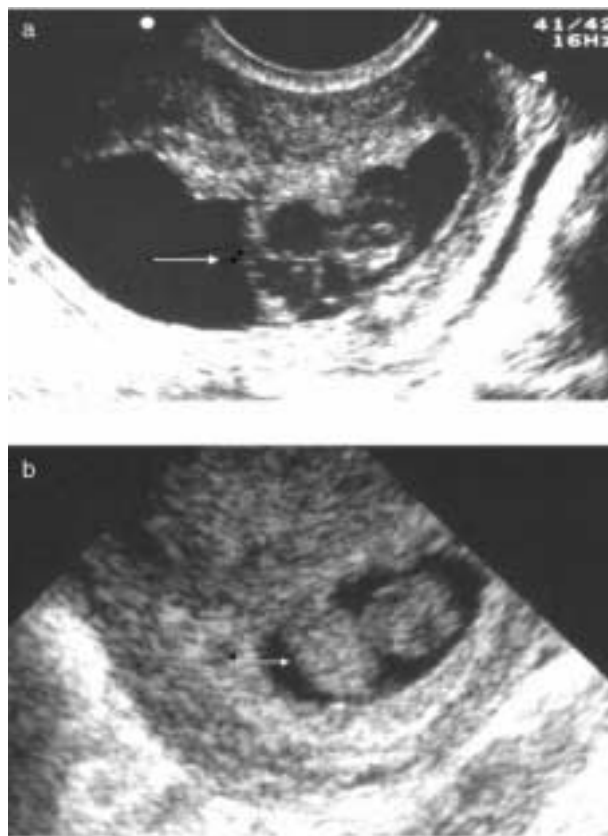
Advances in ultrasound technology have allowed the development of normograms for gestational sacs, embryonic pole length and embryonic heart rate from as early as 24–56 days from last menstrual period<sup>1,2</sup>. It has been demonstrated that smaller than expected gestational sacs or embryonic pole lengths, or delayed appearance of the fetal heart are predictive of early pregnancy loss<sup>3,4</sup>. Chorionic sac diameter is decreased in embryonic and anembryonic abortion<sup>5</sup>. However, abnormal ultrasound findings of pregnancies at an earlier gestation (5–9 weeks) that end in spontaneous abortion are not predictive of an abnormal karyotype<sup>6</sup>.

Trisomies 21 and 13 are the commonest types of chromosome abnormalities that end in spontaneous abortions. Trisomies such as 15 and 22 occur just as commonly but do not generate viable fetuses<sup>7</sup>. Trisomy 7 accounts for 4–10% of all trisomies<sup>7</sup>. The pathological features so far described in trisomy 7 range from empty gestational sacs with edematous

villi, fetus with cleft palate, right-sided aorta, malrotation of the gut, small kidneys and adrenals, and a single umbilical artery<sup>8</sup>. Trisomy 7p has been shown to be characterized by musculoskeletal, cardiovascular, neurological, genital and ocular abnormalities in decreasing frequency<sup>9</sup>. An unexpected case of trisomy 7 conceived after intrauterine donor insemination and resulting in a miscarriage is described here.

A 36-year-old woman and her 40-year-old husband were seen at the assisted conception unit. In 1986 the male partner underwent vasectomy and a reversal operation was attempted unsuccessfully in 1996. Ovulation was induced with 100 mg of clomiphene citrate from the 2nd day to the 6th day of the woman's cycle. The cycle was monitored with transvaginal ultrasound and two 18-mm follicles were noted on day 10. She received human chorionic gonadotrophin on day 11 of her cycle and underwent *in utero* insemination on day 13.

At 6 weeks 6 days of gestation ultrasound examination confirmed a single intrauterine gestational sac containing a yolk sac but no fetal pole was noted. A repeat ultrasound scan at 8 weeks 1 day of gestation showed a heterogeneous multicystic mass measuring 23 × 17 mm in size; the embryo was not identified separately (Figure 1). The chorion appeared normal. No fetal cardiac activity was noted. The ultrasound features were suggestive of a hydatidiform mole, a chorionic tumor or a single gestational sac surrounded by hematoma.



**Figure 1** Gestational sac at 8 + 1 weeks with the presence of a heterogeneous mass (arrow) measuring 23 × 17 mm. In one of the sections (a) the mass was seen to contain fluid-filled spaces. The embryo could not be identified separately. The chorion appeared normal.

A diagnosis of a non-viable pregnancy was made. On the same day the patient underwent evacuation of retained products of conception. Total hCG on the day of the scan was 61615 IU/L and dropped to 2 IU/L 2 weeks later excluding the diagnosis of molar pregnancy.

Samples from two opposite sites of the heterogeneous mass were taken and sent for karyotyping. Further samples were taken and processed for light microscopy. Chromosomal studies revealed trisomy 7.

## Discussion

Advances in image resolution provided by transvaginal probes have given us the opportunity to demonstrate normal and abnormal findings in the first trimester of pregnancy. There is growing evidence that a fetal anomaly scan performed at 11–14 weeks' gestation can detect most severe congenital defects, although a 22-week scan is recommended for congenital heart defects<sup>10</sup>. The above case shows that abnormal findings on transvaginal ultrasound at 7–9 weeks of gestation may help in selecting cases which may be referred for fetal karyotype.

Pregnancy loss at any gestation is devastating for the parents. Early fetal loss and the mental trauma associated with it is a neglected area. Couples are keen to know the cause of pregnancy failure and the possibility of recurrence. Identification of the cause of fetal loss reduces the feeling of self-blame<sup>11</sup>. Fifty percent of spontaneous abortions are associated with chromosomal abnormality<sup>7</sup>; 2–5% of reproducing couples have recurrent miscarriage. Chromosomal abnormalities have been reported in 6% of all recurrent spontaneous abortions<sup>7</sup>. When maternal age is taken into consideration, no association between spontaneous abortion with aneuploidy and the increased risk of a future live born infant with a chromosomal defect could be demonstrated<sup>8</sup>. In a study of maternal age and trisomies in spontaneous abortions, the prevalence of trisomy 7 was observed in the maternal age group with a mean age of 30.3 ± 0.8 years<sup>12</sup>.

Total hCG levels were found to be reduced in cases of trisomy 18 and 13<sup>13</sup>. Confined placental mosaicism (CPM), especially with trisomy 16, is associated with elevated levels of hCG. It is possible that abnormal hCG levels in pregnancies with CPM result from dysfunctional placenta, caused by abnormal chromosomal areas<sup>14</sup>.

In conclusion, with high resolution transvaginal probes it may be possible to select cases where further investigations like fetal karyotyping may be helpful in finding the cause of fetal loss at 7–9 weeks of gestation. An understanding of what caused their fetal loss may help couples with the grieving process.

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## Re: Is obsessional measurement taking worthwhile?

The exchange between Dr Michael Crane and Professor Stuart Campbell about the need for biparietal diameter (BPD), head circumference and other fetal measurements<sup>1</sup> compels me to respond. I agree that better and simpler methods of accumulating fetal measurements are needed and I, too, have imagined more user-friendly software for measurements and their analysis.

Sonography is operator-dependent, particularly for obtaining accurate measurements. Of course high accuracy for estimating delivery dates is important, but accurate estimation

of fetal weight and proportionality requires more measurements, not fewer. I am one who believes that better fetal monitoring results from many individual measurements to get an average, because not only is the average the most accurate estimate of age, but the distribution of the parameter ages around that average is important to the understanding of the parameters' relative proportions. Professor Campbell's statement that microcephaly or achondroplasia rely heavily on measurements is correct, and Dr Crane is correct that all measurements may not be absolutely necessary in every normal case. I agree with Professor Campbell that experience and good technique are necessary when the measurements are required. Obtaining routine measurements facilitates observation of the fetus and its activity in a systematic way that aids the gestalt. Indeed, I support many more empirical measurements, including the liver in cases with a decreased head circumference/abdominal circumference ratio.

As for the BPD and head circumference, I agree with Dr Crane that the BPD is a standard that is well understood, is fault tolerant and valid. If the head is molded enough to make the BPD inaccurate, then I propose that a simple head circumference is also insufficient. The head is a three-dimensional (3D) structure and 3D measurements are more accurate if 'eyeballing' axial and coronal sections reveals molding or suspicious head shape. (For specific discussion of 3D fetal head measurements and fetal size and age analysis I refer you to my textbook on the subject<sup>2</sup>).

While experienced sonologists and sonographers do 'trust their eye' and it is true that 'a significant abnormality diagnosis rarely hinges upon accurate measurements alone', the study of fetal size, growth and proportionality does require many accurate measurements. For those physicians who find the 'financial component' and time of recording of measurements too great and tedious I hope they will acknowledge and work collaboratively with those who have dedicated their careers to developing these observational skills. If poor technique in a rushed environment becomes our standard for diagnostic sonography, then it will never be recognized for the greater accuracy that can be achieved with deliberate technique in educated hands.

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