Prenatal Sonographic Diagnosis of Jeune Syndrome

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Prenatal Sonographic Diagnosis of Jeune Syndrome[1]

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Asphyxiating thoracic dysplasia (Jeune syndrome) is characterized by a narrow thoracic cage, which causes severe respiratory failure with frequent perinatal death; brachymelia, predominantly of the rhizomelic type; renal anomalies; and characteristic radiographic findings of ribs, pelvis, and long tubular bones. Inheritance is autosomal recessive. Prenatal sonographic examination was performed at 17 and 19 weeks of a fetus of parents whose first child had died of Jeune syndrome. The length of the humeri, femora, and tibiae was short (below the mean) for gestational age, and the thorax was abnormally flat and narrow. The iliac wings were square-shaped. We concluded that the fetus had Jeune syndrome. The characteristic skeletal changes of Jeune syndrome are distinct enough at 17 weeks of fetal age to permit sonographic diagnosis.

Index terms: Fetus, abnormalities, 856.1512 • Fetus, growth and development • Fetus, ultrasound studies, 856.1298

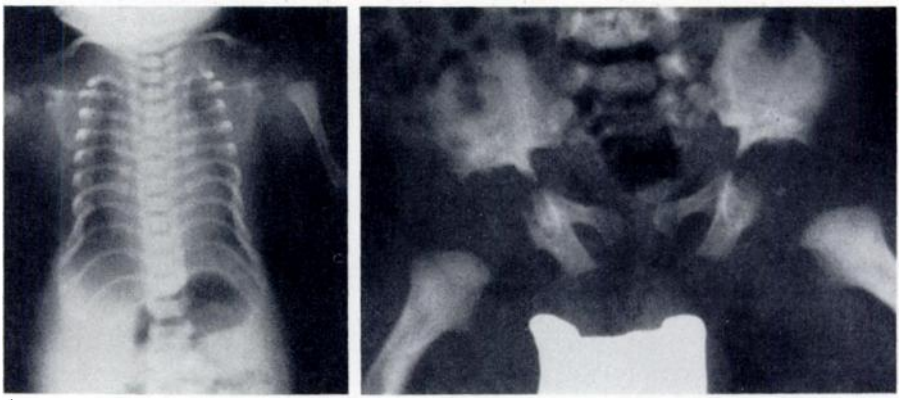
Radiology 1985; 154: 777-778

MODERN sonography permits prenatal diagnosis of hereditary disorders not amenable to biochemical tests, particularly syndromes that feature growth abnormalities of brain, kidneys, and skeleton. For most of these defects, embryologic development is not well known, and, without previous experience, one is often not sure whether skeletal and other defects are distinct enough to allow for an unequivocal prenatal sonographic distinction in time for termination of pregnancy. We report the prenatal sonographic skeletal abnormalities at 17 and 19 weeks of gestation in a fetus with Jeune syndrome

CASE REPORT

A healthy 29-year-old woman, gravida 3 para 2, was referred for genetic counseling and sonographic examination because a previous child had died at two days of age with the clinical and radiological findings of Jeune syndrome. Her husband was 26 years old, and the couple are unrelated. The pregnancy two years earlier was uncomplicated until spontaneous delivery at term. The male newborn weighed 3,300 g and measured 48 cm long with a head circumference (OFC) of 36.5 cm. He had an abnormally narrow, keel-shaped thorax and short upper arms and upper legs (Figs. 1 and 2). There was no hexadactyly. He was severely asphyxiated, with tachypnoea, and he died of respiratory failure at the age of 49 hours.

Figures 1 and 2



1. 2.

1. Chest radiograph of the deceased sibling.
2. Pelvic radiograph of the deceased sibling.

Autopsy disclosed broad costochondral junctions of the ribs, a patent ductus arterious and foramen ovale, a left hydroureter with stenosis above the junction with the bladder, and mild left hydronephrosis.

At genetic counseling a recurrence risk of 25% was given, and the patient was referred for sonographic examination of the current fetus for signs of Jeune syndrome.

At sonographic examination of the index fetus at 17 and 19 weeks following the onset of the last menstrual flow, measurements of biparietal and thoracic diameter, length of the humerus, ulna, femur, and tibia were determined. In addition, a thorough examination of the fetus for structural abnormalities of the skeleton, kidneys, and urinary bladder was performed at both examinations with special attention to the bony thoracic structures. Biparietal diameters corresponded to gestational ages at both examinations, while lengths of humerus, femur, and tibia were below the normal standards (1). At the first examination, all measurements were still within 2 SD below the mean values, whereas at the second examination (in the 19th week) humerus length (23 cm) was equal to −2 SD, and femur (22 cm) and tibia length (19 cm) were below −2 SD of the normal mean values.

Bilateral thoracic diameter at the lower thoracic aperture was normal for gestational age; however, on sagittal scans, the upper thorax was abnormally flat (Fig. 3). The sonographic picture of kidneys, ureters, and bladder was unremarkable. The results of sonographic examination suggested that the fetus had Jeune syndrome, and the parents decided to have the pregnancy terminated. The 21-week-old fetus had a flat and narrow thorax and short upper arms and legs (Fig. 4), and radiographic findings were in agreement (Figs. 5 and 6).

Autopsy confirmed the radiographic findings, demonstrated bilateral hypoplasia of the lungs, and revealed a few glomerular and tubular cysts in the kidneys.

DISCUSSION

Asphyxiating thoracic dysplasia (Jeune syndrome) can be suspected from clinical findings. (The combination of a narrow thoracic cage causing severe respiratory failure at birth with rhizomelic brachymelia and sometimes postaxial hexadactyly). The diagnosis can be unequivocally confirmed in most cases from radiographs of thorax and pelvis. Pathognomonic features at birth include short horizontal ribs with wide anterior portion, square-shaped iliac wings and horizontal acetabular roofs with spur-shaped projections on each side.

Radiologic differential diagnosis mainly includes the Ellis-van Creveld syndrome (chondroectodermal dysplasia); in this latter condition, however, polydactyly is a frequent feature and at least one half of the patients have a congenital heart defect. Both the Jeune syndrome and Ellis-van Creveld syndrome are recessively inherited with a 25% recurrence risk for siblings of an affected proband.

The embryologic development of the skeletal changes in Jeune syndrome is not well known, nor is the point of fetal development at which skeletal abnormalities are distinct enough to allow for an unambiguous prenatal sonographic diagnosis. Goodman and Gorlin (2) stated

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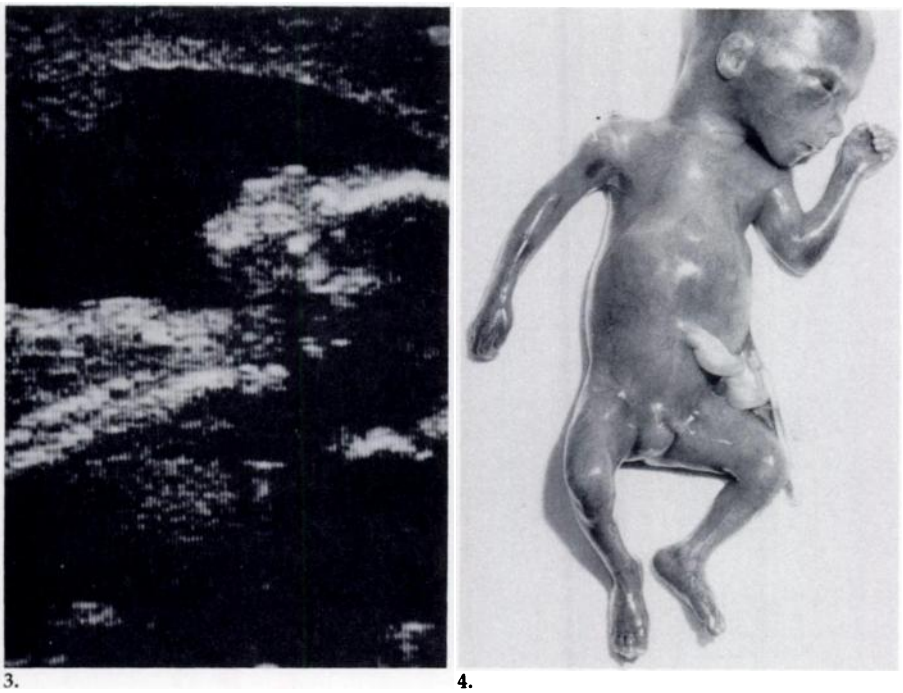
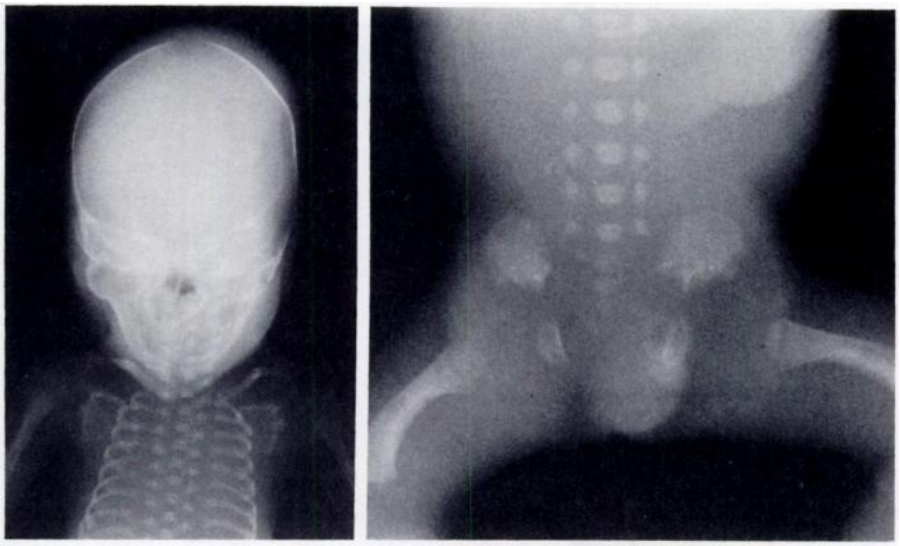
Figures 3 and 4



3. 4.

3. Sagittal scan through the thorax of the index fetus at 19 weeks of gestation.
4. The index fetus at 21 weeks of gestation.

Figures 5 and 6



5. 6.

5. Chest radiograph of the index fetus at 21 weeks of gestation.
6. Pelvic radiograph of the index fetus at 21 weeks of gestation.

that it is possible that the altered anatomy of the chest wall, polydactyly, and shortened extremities could be detected by US. Wladimiroff *et al.* (3) performed US studies on a fetus (between 16 and 22 weeks) from a mother who had already given birth to a child with Jeune syndrome. Findings were normal, and an unaffected baby was born. In a review article on prenatal ultrasonographic diagnosis of congenital malformations, Hansmann and Gembruch (4) listed a fetus with Jeune syndrome among the false negative results, but they did not present further information.

Our results show that prenatal diagnosis of Jeune syndrome is possible from the combination of a narrow thorax and shortening of the humerus, femur and tibia. The deviation in length of the large tubular bones (with exception of the forearm) was already distinct at 17 weeks and became more significant two weeks later. Autopsy radiographs of the fetus unequivocally confirmed the diagnosis. As the variability of clinical and radiologic findings in recessively inherited skeletal dysplasia syndromes is usually not high, it is likely that most, if not all fetuses affected with Jeune syndrome, could be detected by sonographic examination by the 20th week of gestation. As clinical and radiologic findings in thorax and limbs in the Ellis-van Creveld syndrome are very similar to those in Jeune syndrome, it is possible that fetuses affected with this latter syndrome are also amenable to prenatal US diagnosis during the second trimester (4).

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Addendum: After this paper went to press, a case of prenatal sonographic diagnosis of Jeune syndrome was reported in a fetus between 16 and 23 weeks of gestation (Lipson M, Waskey J, Rice J, et al. Prenatal diagnosis of asphyxiating thoracic dysplasia. Am J Med Genet 1984; 18:273–277). Progressive shortness of femoral length was observed at three subsequent examinations, but no further data on other skeletal findings were presented.

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