Antepartum Diagnosis of Potter Syndrome

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Abstract : *Potter syndrome (bilateral renal agenesis) is one of the rare lethal congenital malformations. This report describes a case of Potter syndrome diagnosed antenatally by ultrasonography and fetal pyelography. In cases of IUGR with oligohydramnios, the fetal well-being should be intensively assessed by ultrasonography, ultrasound-guided umbilical cord blood sampling and nonstress test. (Thai J Obstet Gynaecol 1991; 3:57-61.)*

Key words: Potter syndrome, antepartum diagnosis, fetal pyelography, oligohydramnios

The impact of the diagnostic application of ultrasound and its guided fetal cord blood sampling for the identification of various fetal anomalies has been momentous. Antenatal recognition of a lethal malformation would be helpful in deciding the most appropriate mode of delivery and in reducing the psychosomatic burden of parents for an unfavorable outcome.

Potter syndrome is a congenital malformation accompanied by bilateral renal agenesis, lung hypoplasia, characteristic facies and other abnormalities[1,2]. It is also often associated with oligohydramnios, intrauterine growth retardation and breech presentation. Since these infants die of renal or respiratory failure shortly after birth, the accurate antenatal diagnosis of Potter syndrome is of importance in order to avoid unnecessary caesarean section.

The present report shows a case of Potter syndrome diagnosed antenatally by ultrasonography, fetal pyelography and the analysis of umbilical cord blood.

Case Report

The patient was a 34-year-old, married woman, para 2-0-2-2, referred to the hospital because of retarded growth of the fetus at 29 weeks of

pregnancy. The family history was noncontributory. The prenatal course until then was uneventful. All blood analysis was normal except 75g GTT. GTT at 29 weeks of gestation showed 120, 245 and 231 mg/dl. An ultrasound examination revealed no detectable amount of amniotic fluid and an extremely growth-retarded fetus with scaphoid head, bell-shaped thorax and no demonstrable kidney (Figs. 1,2). The fetal bladder did not appear to fill during more than one hour of observation. Moreover, maternal administration of furosemide 20 mg failed to cause the filling of the fetal renal pelvis and the bladder with urine (Fig. 2), but nonstress testing was reactive (Fig. 3). Unconjugated estriol and hPL values were 5.8 ng/ml and 2.0 μU/ml, respectively, Repeated examination by ultrasonography did not demonstrate the fetal kidney. At 31 weeks, ultrasound-guided sampling of umbilical cord blood and fetal intravenous pyelography were performed to make an accurate diagnosis. The results of fetal blood analysis were as follows; WBCx 7800/mm^3, RBC 350 x 10^4/ mm^3, Hb 11.8 g/dl, Ht 36.0%, platelet count 178x10^3/mm^3, total protein 3.7g/dl, albumin 2.6 g/dl. Fetal anemia and hypoproteinemia were remarkable. Electrolytes and blood gas analysis were within normal range: Na 136 mEq/l, K 4.6 mEq/l, Cl 105 mEq/l, BUN 10 mg/dl, creatinine 0.6 mg/dl, pH 7.373, PCO_2 43.1 mmHg, PO_2 20.2 mmHg, HCO_3 25.0 mEq/l. Fetal nephrogram and pyelogram, administered with 5 ml of 60% urographin intravenously, did not contrast. On the other hand, maternal pyelogram was obtained in spite of fetal administration of the contrast medium.

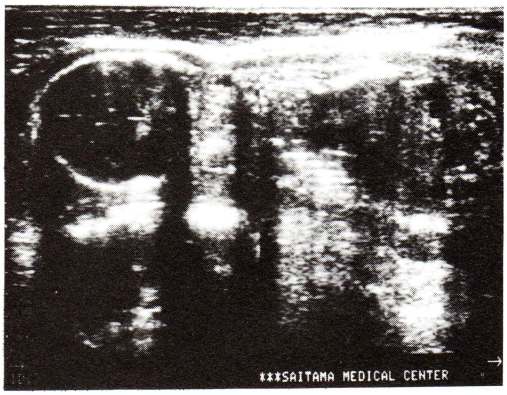

Fig. 1 Ultrasound picture of the fetus in utero (29th wk). Sagittal section of the fetus at the level of the head and thorax. Fetus is touched to the uterine wall, caused by anhydramnios. Bell-shaped thorax is characteristic.

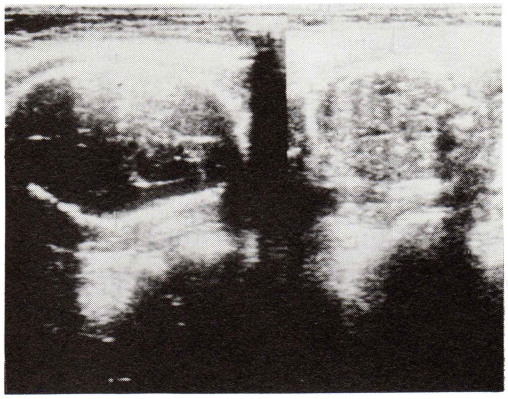

Fig. 2 Ultrasound pictures of the fetal head (left) and trunk (right). Transverse section of the fetus at the level of the kidney, which is not demonstrable after the maternal administration of furosemide. An extremely growth-retarded fetus with scaphoid head is observed.

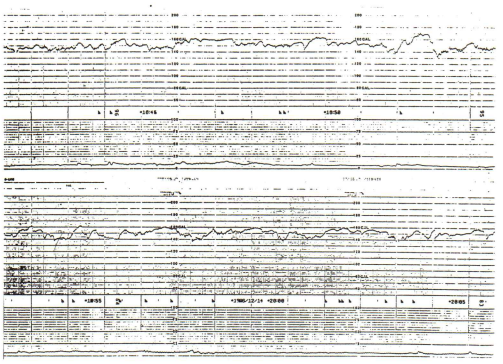

Fig. 3 Fetal cardiotocogram. Nonstress testing is reactive at 30 weeks of gestation.

A diagnosis of Potter syndrome was made by the findings of ultrasonography and fetal pyelography.

The patient went into spontaneous labour two days later and had a stillbirth in breech presentation. The infant weighed 1062 g. No amniotic fluid was recognized. The placenta weighed 200 g. The Potter facies with prominent epicanthic folds, flattened nose, low set ears and receding chin were noted. There were bowing legs with hypogenesis of the left foot, atresia ani and single umbilical artery. External genitalia was defective (Fig. 4). At autopsy, the infant was proved to be male with agenesis of bilateral kidneys. Hypoplastic lungs, malrotation of the intestine, mesenterica communis and retentio testis were also seen.

Discussion

In 1946, Potter[1] reported twenty instances of bilateral renal agenesis with pulmonary hypoplasia and a characteristic facial expression. Potter syndrome occurs predominantly in males and about once in 2500-3000 births[2,3]. Dry, loose skin, limb abnormalities such as bowed legs and clubbed feet, spade-like hands, ovoid adrenal glands, and urinary tract or genital abnormalities are also usually observed. Oligo- or anhydramnios, intrauterine growth retardation, prematurity and breech presentation are common. It is recognized that similar signs and symptoms are accompanied by the various types of urinary tract abnormalities and are also the outcome of fetal compression due to nonrenal oligohydramnios secondary to prolonged leakage of amniotic fluid, hence the term Potter sequence.

The aetiology is still unknown. The development of Potter syndrome may be due to the consequence of multiple early mesodermal defects which occur mainly sporadically or rarely in autosomal recessive inheritance. In Potter syndrome, limb deformities, flattened ears and nose, and hypoplastic lungs can be attributed to compression secondary to oligohydramnios but the epicanthic fold, the malformation of the tragus and antetragus, the abnormally low slanted position of ears, the changes in subcutaneous tissue, and the increased frequency among males cannot be explained merely due to oligohydramnios.

It has become much easier to detect various fetal malformations by monitoring the fetus by ultrasound equipment with high resolution. Antenatal diagnosis of urinary tract anoma-

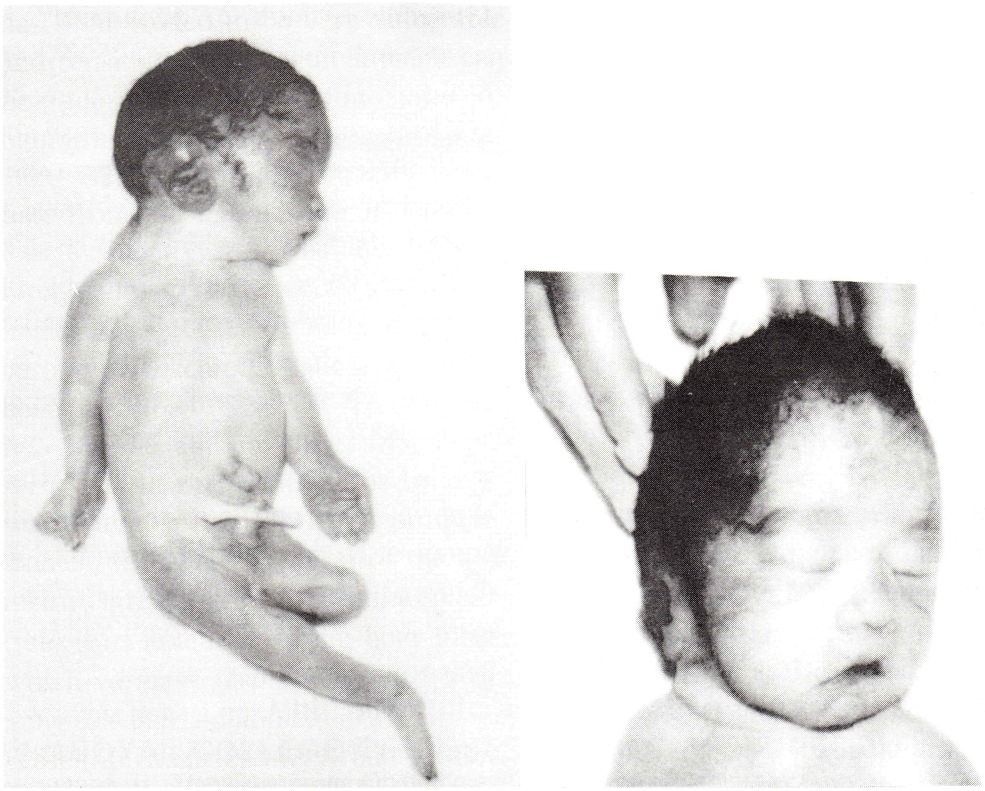

Fig. 4 Photographs of the fetus characteristic of the Potter face and bowing legs with hypogenesis of the left foot.

lies such as cystic kidney and obstuctive uropathies, has also been reported[3,6]. In the normal fetus the kidneys and urinary bladder can be easily identified after the 16th-20th week of gestation[5,7,8]. In addition to this, changes in bladder volume by urine can be observed to ascertain the functioning capacity of the urinary tract by repeated ultrasound examinations at 20 minutes intervals for 60-120 minutes observation periods[9,10,]. Furthermore, the stimulation by furosemide given to a normal pregnant woman causes the increased production of urine in the fetus and the filled bladder can be demonstrated within 60 minutes[11]. It was reported that intravenous pyelography, performed by injection of contrast material into maternal circulation was useful to examine the fetal kidney function[12]. In the present case, even direct injection of contrast material into fetal circulation produced no pyelogram of the fetal kidney and bladder. Therefore, bilateral renal agenesis of the fetus was confirmed. We also recognized anhydraminos, severe IUGR, breech presentation, scaphoid head, bell-shaped thorax

without breathing movement and a flexed-spine posture with bowed legs and feet.

Results of ultrasound-guided cord blood sampling at 31 weeks indicated severe hypoproteinemia and anemia of which the pathophysiology remains obscure. This malnutrition may be caused by placental dysfunction concluded from low values of unconjugated estriol and hPL. However, the values of electrolytes, BUN, creatinine, and blood gas were normal. It was postulated that the placenta in this case impaired the protein synthesis, whereas, the capacity of exchange or transfer of the compounds like electrolytes or oxygen was preserved.

Consequently, in cases of severe IUGR with oligohydramnios, congenital anomalies of the fetus should be intensively examined by ultrasonography as well as the ultrasound-guided sampling of umbilical cord blood in consideration of the possibility of Potter syndrome.

References

1. Potter EL. Bilateral renal agenesis. J Pediatr 1946; 29:68-76.
2. Potter EL. Bilateral absence of ureters and kidneys. A report of 50 cases. Obstet Gynecol 1965; 25:3-12.
3. Dicker D, Samuel N, Feldberg D, Goldman JA. The antenatal diagnosis of Potter syndrome (Potter sequence). A lethal and not-so-rare malformation. Eur J obstet Gynecol Reprod Biol 1984; 18:17-24.
4. Allen RW, Rehm NE, Scott JR, Kochenour NK. Antepartum diagnosis and intrapartum management of lethal renal defects. Obstet Gynecol 1981; 58:379-82.
5. Garrett WJ, Warren PS, Fisher CC. Ultrasound in the diagnosis and management of urinary tract disorders in the fetus. Ultrasound Med Biol 1984; 10:473-83.
6. Helin I, Persson P. Prenatal diagnosis of urinary tract abnormalities by ultrasound. Pediatr 1986; 78:879-83.
7. Campbell S, Wladimiroff JW, Dewhurst CW. The antenatal measurement of fetal urine production. Br J Obstet Gynaecol 1973; 80:680-6.
8. Lawson TL, Foley WD, Berland IL, Clark KE. Ultrasonic evaluation of fetal kidneys. Radiology 1981; 138:153-6.
9. Schmidt W, Kubli F. Early diagnosis of severe congenital malformations by ultrasonography. J Perinat Med 1982; 10: 233-41.
10. Wladimiroff JW, Campbell S. Fetal urine production in normal and complicated pregnancy. Lancet 1974; 1:151-4.
11. Wladimiroff JW. Effect of furosemide on fetal urine production. Br J Obstet Gynaecol 1975; 82:221-4.
12. Thomas CR, Lang EK, Lloyd FP. Fetal pyelography. A method for detecting fetal life. Obstet Gynecol 1963; 22:335-40.

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