

—Reports on Experiments and Clinical Cases—

## Prenatal diagnosis of autosomal recessive polycystic kidney disease

### A case report

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#### Abstract

We present a case of autosomal recessive polycystic kidney disease diagnosed at 28 weeks' gestation by ultrasonographic examination and magnetic resonance imaging (MRI). The fetal kidneys were symmetrically enlarged and highly echogenic by ultrasonographic examination and showed high-signal intensity on T<sub>2</sub>-weighted images by MRI. Cystic lesions were recognized by neither examination. In addition, the pulsatility index of the fetal renal artery was normal. These findings suggest a high water content in the renal parenchyma with tiny cysts and normal blood flow in autosomal recessive polycystic kidney disease. (J Nippon Med Sch 1999; 66: 188—190)

**Key words:** autosomal recessive polycystic kidney disease, ultrasonography, pulsed Doppler, magnetic resonance imaging

We present a case of autosomal recessive polycystic kidney disease (ARPKD) at 28 weeks' gestation diagnosed by magnetic resonance imaging (MRI) and ultrasonic examination. In infants<sup>1</sup>, some investigations concerning the renal artery Doppler waveforms in ARPKD have been reported. However, few investigations has been reported those in utero. In this study, thus, we performed pulsed Doppler study as an additional examination for ARPKD in utero.

#### Case Report

A 34-year-old woman, gravida 1, para 0, was referred to our hospital at 28 weeks' gestation for a high risk obstetric consultation due to oligohydramnios. The medical and genetic family and past histories of the patient and her husband were unmarked.

Ultrasonic examination was performed with a Toshiba UMCL 342 A using a 3.75 MHz transducer.

The amniotic index was 1.5 cm<sup>3</sup>. The fetal kidneys were symmetrically enlarged (right, 5.3 × 4.0 cm; left, 4.3 × 3.7 cm) and highly echogenic by ultrasonographic examination (**Fig. 1, left**). Multicystic echo patterns were not observed in the kidneys by transabdominal or transvaginal ultrasonographic examination (**Fig. 1, right**). A small bladder was recognized, but the fetal hourly urine production rate was not clearly detected. The estimated fetal weight was 1,200 gram (appropriate for gestational age), however the fetal chest was very narrow. Serial umbilical artery, fetal middle cerebral artery and the fetal renal artery Doppler waveforms were obtained when fetal breathing movement could not be detected, with a sample volume < 3 mm. The pulsatility indexes in the umbilical artery, the fetal middle cerebral artery and the fetal renal artery were 0.72, 2.11 and 2.76, respectively by pulsed Doppler studies (**Fig. 2**). The non stress test was reactive. MRI was performed at 28 weeks and 6 days' gestation

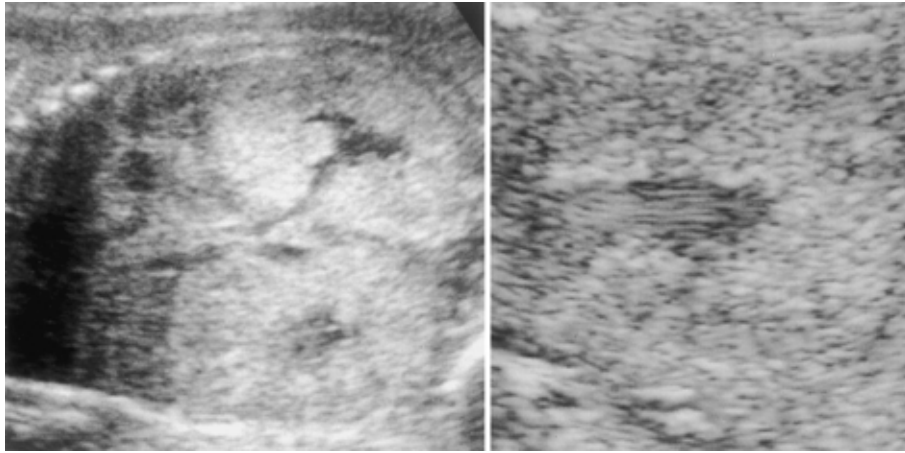


Fig. 1 The fetal kidneys at 28 weeks' gestation delete by longitudinal transabdominal (left) and semi-coronal transvaginal (right) ultrasonographic examination. The kidneys are hyperechoic. Multicystic echo patterns are not observed.

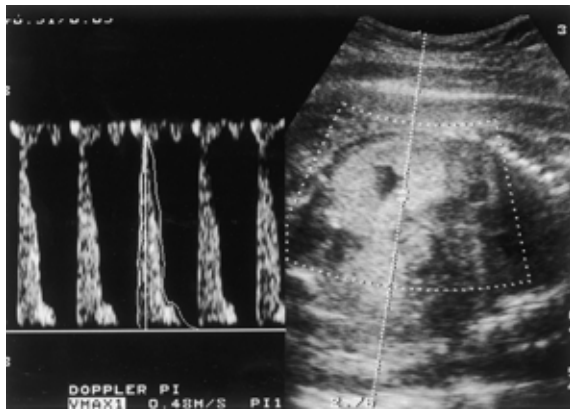


Fig. 2 Pulsed Doppler waveforms of the fetal renal artery at 28 weeks' gestation. The pulsatility index was 2.76 (normal).

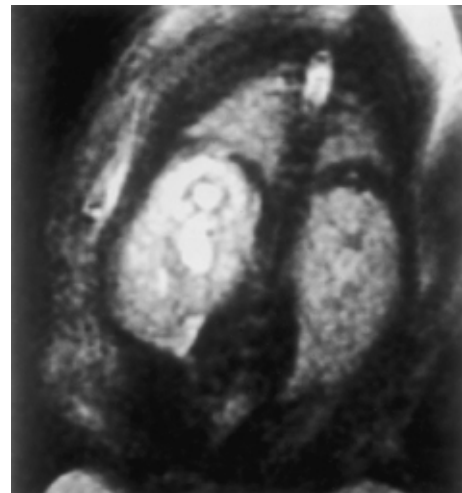


Fig. 3 Magnetic resonance imaging (single-shot-fast-spin-echo-method) of the fetal kidneys at 28 weeks' gestation. The kidneys show high-signal intensity on T<sub>2</sub>-weighted images.

using a Yokogawa Sigma Horizon, 1.5 Tesla (63.9 Mhz). The kidneys showed high-signal intensity on T<sub>2</sub>-weighted images, suggesting a high water content in the renal parenchyma (**Fig. 3**). Cystic lesions were not clearly observed in the kidneys by MRI.

Thus, we diagnosed this case as ARPKD, associated with tiny renal cysts. Informed consent was obtained to conduct this study. Possible interventions such as serial amnioinfusion during pregnancy were discussed with the patient and her husband, and they selected an early delivery. A male infant, weighing 1,530 gram, was delivered at 31 weeks and 1 days' gestation, and died during delivery.

### Discussion

Several delete studies have examined the prenatal

diagnosis of ARPKD by ultrasonography and MRI<sup>1,2</sup>. These sonographic features have consisted of bilateral enlarged hyperechogenic kidneys, because the cysts in the kidneys of ARPKD are usually too small to be recognized by ultrasonography<sup>2</sup>. In this case, MRI did not detect tiny cysts in the kidneys, and showed kidneys of high-signal intensity on T<sub>2</sub>-weighted images. In an earlier study by Nishi et al.<sup>3</sup>, a marked increase in signal intensity on T<sub>2</sub>-weighted images, suggesting a high water content in the renal parenchyma, and a change in size of kidney cysts was observed in ARPKD. These MRI findings suggest the presence of multiple, tiny renal cysts containing a watery substance. In this case, we did not observe

changes in the size of the cysts. Polycystic kidney diseases have been classified by the age of onset and degree of renal involvement, and the observation of tiny cysts alone as seen in our case indicates ARPKD with a poor prognosis<sup>3</sup>. In an earlier study by Romero et al.<sup>5</sup>, no cases of false positive diagnoses of (infantile) polycystic kidney disease were observed by these methods. Based on these studies, the current case may have had a poor prognosis.

In this case, we performed fetal pulsed Doppler study as an additional examination, and found normal PI levels in the umbilical artery, the middle cerebral artery and the renal artery. This may be the first study to report Doppler findings of the fetal renal artery in ARPKD. Placental insufficiency, causing fetal blood flow redistribution and resulting in decreased renal perfusion, has been proposed as a possible mechanism leading to oligohydramnios in small-for-gestational-age fetuses<sup>6</sup>. On the other hand, oligohydramnios associated with factors other than decreased fetal renal blood flow, such as in prolonged pregnancy, shows normal PI levels in the fetal renal artery<sup>7</sup>. In infants<sup>1</sup>, some investigations concerning the renal artery Doppler waveforms in ARPKD have been reported. Increased renal vascular resistance in patients with ARPKD has been reported to be correlated with renal function disturbance with the development of systemic arterial hypertension. Thus, Doppler study in utero may be a useful method to predict the prognosis in ARPKD. In this case, the normal PI level in the fetal renal artery may have indicated stable fetal blood circulation. The normal umbilical artery and fetal cerebral artery Doppler waveforms may also support this speculation. However, we could not examine the relationship between the Doppler waveforms in the fetal renal artery and the prognosis of the infant.

ARPKD is a rare hereditary disease involving cystic dilation of the renal collecting tubes<sup>3</sup>. Infants with

ARPKD often reveal Potter's facies and may develop respiratory distress as a consequence of pulmonary hypoplasia due to oligohydramnios. In this case, a narrow fetal chest was recognized by ultrasonography and MRI; thus serial amniocentesis was considered to prevent pulmonary hypoplasia. However, the patient and her husband selected an early delivery, because they were anxious about the indefinite predictive diagnosis of the prognosis of ARPKD. Therefore, further examinations are needed for the predictive diagnosis of the prognosis of ARPKD, such as Doppler studies in the fetal renal artery.

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