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Congenital Cystic Disease of the Kidney †

(SNUCH CPC-38)

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CASE PRESENTATION

This male newborn was admitted to SOHWA Children's Hospital on Aug. 27, 1987 because of abdominal distension and anuria. He was born by Cesarian section due to breech presentation on July 28, 1987 at KURO Hospital. The gestational age was 37^{+4} weeks and the membrane ruptured 2 hours before the delivery. At birth, his body weight was 2.370gm.

He had been admitted to KURO Hospital under the impression of hydronephrosis and acute renal failure until Aug. 18, 1987 (22nd day of life). At birth he had good Apgar scores. But weight gain was poor, and on the 15th hospital day an abdominal mass in the left upper quadrant was palpated. The abdominal sonography showed bilateral hydronephrosis. Serum BUN and creatinine were 188 mg% and 5.3 mg%. Serum Na was 132 mEq/L, K 6.5 mEq/L, Cl 97 mEq/L and CO_2 6 mEq/L. Peritoneal dialysis was done with peritosol from the 15th to 16th hospital day. On the 20th hospital day, serum BUN and creatinine became 118 mg% and 3.4 mg%. Blood electrolytes showed serum Na 116mEq/L, K 6.5 mEq/L, Cl 89 mEq/L and CO_2 17.5 mEq/L. IVP revealed non-visualization of both kidneys. At the 22nd hospital day, he was discharged against advice.

On admission to SOHWA Children's Hospital, the body weight was 2,700gm, height 46cm, head circumference 33.5cm and chest circumference 31cm. The body temperature was 36.8, °C pulse rate 140/min. and respiratory rate 60/min. The baby was flaccid and cried weakly. The anterior fontanel was open and flat. The neck was supple. The breathing sound was clear. Heart beats were regular and there was no murmur. The abdomen was markedly distended. The liver and the spleen were not palpated. The lower pole of both kidneys were palpated in both flanks. General activity, Moro reflex and sucking reflex were poor.

Laboratory data showed hemoglobin 9.8 gm%, hematocrit 30% and WBC 17,700/mm^3 (stab;2, seg:75, lym:23). Blood sugar was 52 mg%. Serum Ca and P were 7.0 mg% and 12.5 mg%. Serum Na was 120 mEq/L, K 7.5 mEq/L and Cl 67 mEq/L. The blood gas analysis revealed pH 7.168, PCO_2 17.2 mmHg, PO_2 50.9 mmHg, HCO_3 6.0 mmol/l, BE -19.7 mmol/l and O_2 sat. 70.7%. Serum total protein and albumin were 5.4 gm% and 3.2 gm%. Serum BUN and creatinine were 110 mg% and 5.2 mg%. Urinalysis failed due to anuria. CSF examination revealed no remarkable abnormality. Cultures of blood and CSF were negative.

The chest X-ray was normal. The abdominal sonography showed both multiple cystic

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lesions. Hemodialysis was recommended but refused. At the 2nd hospital day, serum BUN and creatinine were 68 mg% and 6.4 mg%. Serum Na was 108 mEq/L, K 8.3 mEq/L and Cl 65 mEq/L. Serum Ca and P were 5.5 mg% and 12.0 mg%. He had no urination at all. He was discharged against advice and expired at home on August 28, 1987.

DISCUSSION

In summary this infant had clinical manifestations and laboratory findings of progressive renal failure(Table 1) and radiologic evidence of bilateral cavitary abdominal masses since about 14 days after birth.

Renal failure may develop in various conditions in the neonatal period(Table 2), but the accompanying bilateral cavitary abdominal masses without any history suggesting prerenal factors are not so many. Bilateral cystic renal diseases such as polycystic kidney disease (PKD), bilateral congenital multicystic kidney (CMK), unilateral CMK with contralateral hydronephrosis
or cystic dysplasia, renal cyst in hereditary syndromes are a group of possible diagnoses, and bilateral hydronephrosis caused by obstructive anomaly or compressing mass of the urinary tract is another one.

Though autosomal dominant PKD can rarely cause clinical problems in infancy infrequently, it is not likely in this case because it manifests itself as masses or hematuria. Bilateral CMK is not appropriate in this case either, because the infant outlived a month. Because this infant didn't have any typical syndromal manifestations, renal cysts in hereditary syndromes like tuberous sclerosis, Meckel syndrome, Jeune syndrome, von Hippel Lindau syndrome, Zellweger syndrome or prune belly syndrome may be excluded (Bernstein 1990). So, the diagnostic possibility is much narrowed; 1) autosomal recessive PKD(ARPKD), 2) unilateral CMK with contralateral hydronephrosis or cystic dysplasia 3) bilateral hydronephrosis.

May I see the radiologic findings of this case at this point?

Dr. Yeon: The chest X-ray film shows unremarkable lung shadow. The bowel gas is also normally distributed. The abdomen shows bladder wall hypertrophy. The kidneys on ultrasonography show cystic lesions in both sides. The cysts are variable in size in the right, and the left are more concentrated in the periphery. The kidney size appears slightly larger than normal. The left kidney shows remaining renal parenchyma between the cysts.

Dr. Ha: Because the cavitary abdominal masses seem not to be hydronephrosis but bilaterally cystic kidneys on ultrasonography, there remains two possibilities; ARPKD and unilateral CMK with contralateral cystic dysplasia. There are several clinical and radiologic points of differentiation between the two conditions. In ARPKD, both kidneys are bilaterally enlarged with similar lesions, and in CMK with contralateral cystic dysplasia, renal sizes are variable and different in each other. ARPKD, especially infantile or juvenile type, is frequently accompanied by congenital hepatic fibrosis, but it is not usually evident in perinatal or neonatal type (Reeders 1990). Non-renal anomalies such as esophageal atresia, congenital heart diseases, meningomyelocele or cleft palate are often accompanied by CMK (Mir et al. 1983). The cysts of ARPKD are usually located in the renal medulla, while those of CMK are distributed over the cortex and medulla. Therefore considering the bilateral enlargement and similarity of both renal lesions and the lack of combined hepatic or other anomalies, I think that the infant had ARPKD of neonatal type.

Hypoxemia noted on 30th postnatal day may be the result of mild pulmonary hypoplasia because there is no evidence of pulmonary infection or cyanotic heart disease. The elevated BUN creatinine ratio indicated that the infant's nutritional state was very poor.

Clinical diagnosis(Dr. Ha):

1. Autosomal recessive polycystic kidney disease
2. Mild pulmonary hypoplasia

Table 1. Differential diagnosis of acute renal failure in neonate

I. Prerenal

1. Hypovolemia: dehydration, hemorrhage, shock
2. Sepsis
3. Hypoxia
4. Congestive heart failure
5. Hypokalemia

II. Renal

A. Congenital
1. Renal agenesis, hypoplasia
2. Renal dysplasia without cyst
3. Cystic renal diseases:
   a) Polycystic kidney disease-autosomal recessive type (ARPKD)
   b) Renal cystic dysplasia
      (1) bilateral congenital multicystic kidney
      (2) congenital multicystic kidney in a single kidney
      (3) congenital multicystic kidney with contralateral hydronephrosis
      (4) congenital multicystic kidney with contralateral dysplastic kidney
   c) Renal cyst in hereditary syndromes
      (1) prune belly syndrome
      (2) tuberous sclerosis
      (3) Meckel syndrome
      (4) Jeune syndrome
      (5) von Hippel Lindau syndrome
      (6) Zellweger syndrome
4. Congenital nephrotic syndrome

B. Vascular
1. Renal artery thrombosis
2. Renal vein thrombosis
3. Hemolytic uremic syndrome

C. Toxic
1. Drug related
2. Severe hemolysis
3. Rhabdomyolysis

D. Inflammatory, pyelonephritis

E. Prolonged prerenal
1. Acute tubular necrosis
2. Cortical necrosis
3. Medullary necrosis

III. Postrenal

A. Bilateral hydronephrosis
1. Obstructive anomalies of the urinary tract
   a) ureteropelvic junction obstruction
   b) ureterovesical junction obstruction
   c) ureterocele
   d) megaureter
   e) posterior urethral valve
   f) urethral diverticulum
   g) imperforated prepuce
2. Mass compressing urinary tract
3. Neurogenic bladder

B. Uric acid nephropathy

Table 2. Differential diagnosis between autosomal recessive polycystic kidney disease(ARPKD) and congenital multicystic kidney(CMK).

	ARPKD	CMK
Incidence	1/10,000 - 1/40,000	1/2,000
Heredity	autosomal recessive	sporadic
Sex ratio	M < F	M = F
Symptom onset	< 5 years	< 1 month
Renal involve	bilateral	mostly unilateral
Renal size	large	variable
Location of cyst	medulla	cortex and medulla
Associated findings	congenital hepatic fibrosis	ureteral abnormality contralateral hydronephrosis contralateral dysplasia esophageal atresia PDA, VSD meningomyelocele cleft palate

Pathological findings (Dr. Chi):

Postmortem examination confirmed the main findings located in the urinary system. In addition, there were patent ductus arteriosus, high arched palate and mild lung hypoplasia. The face of this patient showed prominent down-slanting epicanthic folds, beakedn nose

and large flabby ears indicative of Potter face. The kidneys were bilaterally cystic, but their sizes were different (Fig 1A & B). The right kidney was grossly cystic and cysts were found in the subcapsular portion as well as in the cortex and medulla. Fibrous bands were noted on cut sections. The right ureter was dilated because of the obstruction at ureterovesical junction (Fig 1A). The left kidney was somewhat enlarged and contained various-sized cysts predominantly in the cortex. Cut sections showed dense fibrosis and obliteration of the pelvocalyceal system (Fig 1B). The left ureter was not dilated and inserted normally into the dilated urinary bladder. Diverticular protrusion of the bladder was seen in the left lateral portion. The bladder was dilated and the wall was thickened and hypertrophied. The trigone area was preserved except for diverticular expansion. There was a posterior urethral valve, and so the proximal prostatic urethra was dilated and was contrasted to the stenotic anterior urethra. Microscopically, the right kidney showed preserved renal parenchyma that was scattered with dysplasific foci particularly in the subcapsular portion. Dense fibrosis and hyaline cartilage islands (Fig 2) were seen. The left kidney showed almost total absence of normal kidney tissue. Instead dense fibrous tissue, primitive ducts, and cartilage islands were seen (Fig 3).

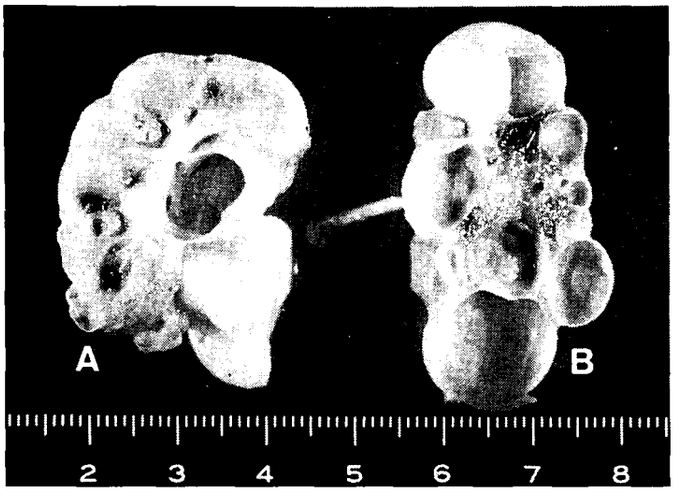

Fig 1. Gross appearance of the right (A) and left kidney (B), showing different pattern of lesion. The right kidney(A) shows a well developed pelvocalyceal system and minute subcapsular cysts. The remaining renal parenchyma is normal. The left kidney (B) on the other hand shows total obsolescence of normal renal parenchyma and replacement by large cysts and fibrosis. The pelvocalyceal system is not formed.

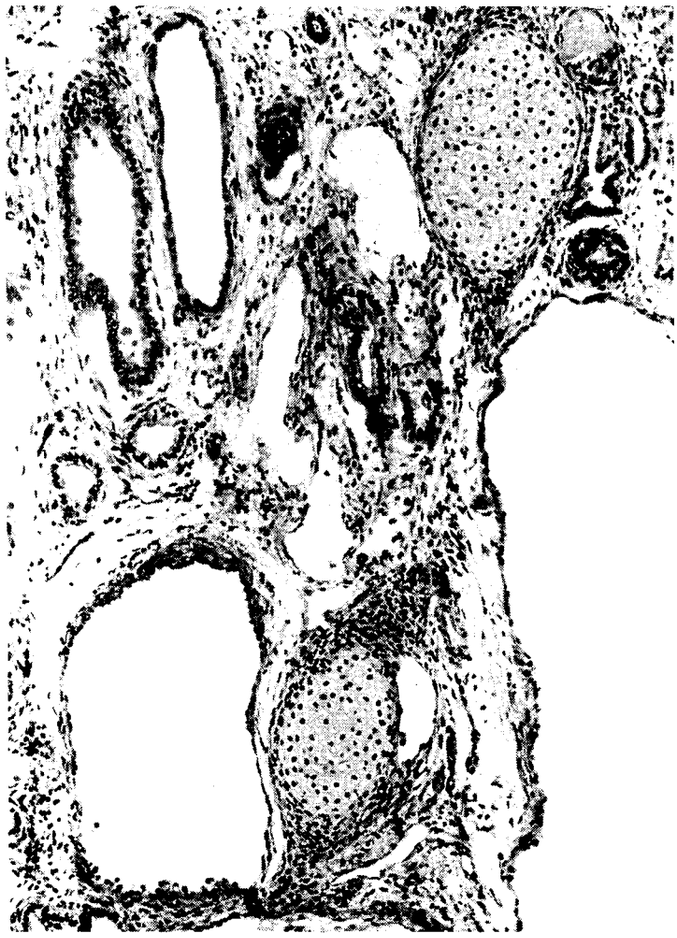

Fig 2. Photomicrograph of the right kidney, showing two islands of cartilage, fibrous tissue and cysts. These dysplastic foci were scattered subcapsularly. H&E X100

In summary, it seemed that two types of cystic disease were present in the kidney, in a same patient, namely type IV due to congenital urethral obstruction and independent type II disease which is characterized by dysplasia and arrest of ampullary activity during the kidney development. The direct cause of death was bronchopneumonia.

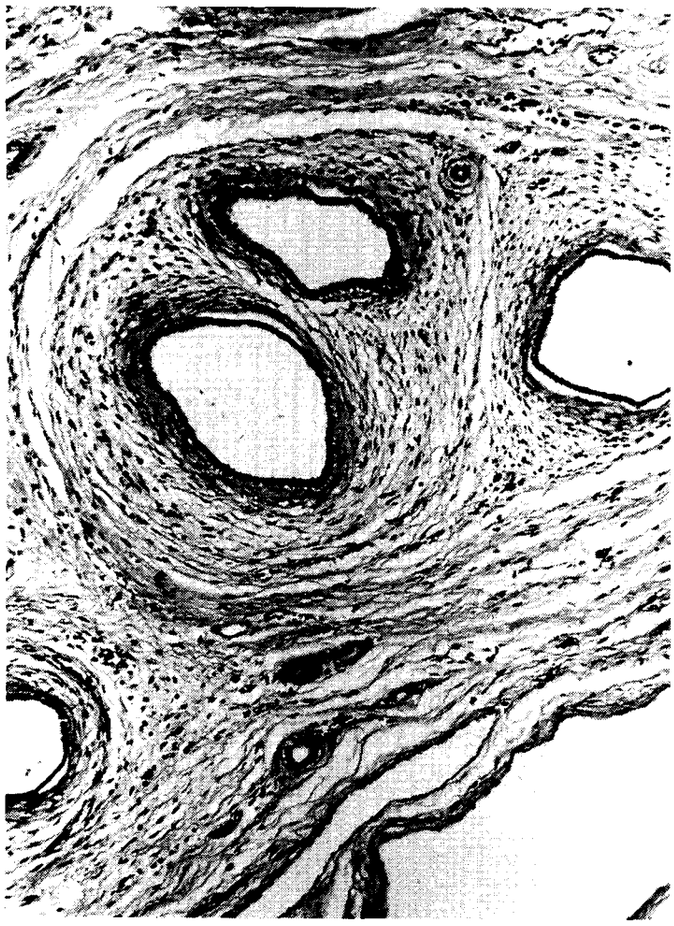

Fig 3. Photomicrograph of the left kidney, showing primitive ducts and fibrous tissue between multiple cysts. H&E X100

Pathological diagnosis:

Potter syndrome

- Facies renalis
- Pulmonary hypoplasia
- Cystic disease of the kidney, combined type IV and II, associated with posterior urethral valve, bladder diverticulum and right ureterovesical obstruction.
- High arched palate

Patent ductus arteriosus

Bronchopneumonia

Esophageal erosion

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