A Case with Nephrophthisis Complex or a Variant of the Disease

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Dear Sir,

Juvenile nephrophthisis (JN) is a chronic, hereditary, tubulointerstitial disease characterized by polyuria, polydipsia, decreased urinary concentration ability, anemia, growth retardation and progressive renal failure [1]. The age at the first clinical evaluation for the manifestations of JN is approximately 10 years, varying from 3 to 17 years, and the patients develop end stage renal failure with 1-10 years after the first presentation [1, 2]. We present a 5-year-old boy with JN who developed the symptoms of JN and renal failure at early infancy together with manifestations of nephrogenic diabetes insipidus.

The patient had polyuria since his birth and was admitted to the hospital with complaints of fever and irritability for 2 days. When he was 20 days old, he was treated in another hospital for 50 days as inpatient with the diagnosis of cystic renal disease and sepsis, and he received several antibiotics including vancomycin, cefotaxime, neutro-mycin and ampicillin. He had a history of asphyxia at birth. He had parental first-degree consanguinity, and his aunt received hemodialysis because of end stage renal failure of unknown origin. His physical examination revealed that his respiration was in acidotic pattern, and he had hepatomegaly of 3 cm below the last costal margin midclavicularly.

The laboratory examination showed iron deficiency anemia (hemoglobin: 7.4 g/dl), urinary pH: 6.5, density: 1.010, protein: + (700 mg/m²/day), glucose: – and normal sediment. Blood urea nitrogen (BUN) was

21 mg/dl (7.5 mmol/l), creatinine: 1.2 mg/dl (106.1 µmol/l), total protein: 7.5 g/dl, albumin: 5 g/dl, sodium: 152 mEq/l, potassium: 4.9 mEq/l, calcium: 9.5 mg/dl (2.4 mmol/l), phosphorus: 6 mg/dl (1.9 mmol/l). Arterial blood gases showed metabolic acidosis with pH: 7.1, pCO₂: 24 mm/Hg (3.2 kPa) and HCO₃: 12.9 mmol/l. His creatinine clearance was 25 ml/min/1.73 m².

Using abdominal ultrasonography, hepatic fibrosis and hy-perechogenic (grade 3) normal-sized kidneys with several cortical and medullary cysts showing the loss of corticomedullary differentiation were detected (fig. 1). CT of the abdomen showed multiple hypodense areas in

medullar and cortical regions (fig. 2) and the patient was diagnosed to have a cystic kidney disease and treated for acidosis. However, serum sodium levels were subsequently elevated and on the 4th day following of admission it was 184 mEq/l, serum potassium: 8.7 mEq/l, BUN: 52 mg/dl (18.6 mmol/l), creatinine: 1.7 mg/dl (150.3 µmol/l), osmolality: 384 mosm/kg while urinary sodium was 69 mEq/l, K: 8.4 mEq/l and osmolality 245 mosm/kg at the same time.

Serum vasopressin level was 35 pg/dl (normal < 8 pg/dl). Thus the patient was diagnosed to have nephrogenic diabetes insipidus. Development of nephrogenic diabetes insipidus together with the

laboratory data, pathognomonic ultrasonographic findings for JN and the family history led to the diagnosis of nephrophthisis [3-5]. Presence of hepatic fibrosis also supported the diagnosis [6]. Peritoneal dialysis was performed for the intractable acidosis and hyponatremia. Later on, the patient was discharged with the supportive therapy of hydrochlorothiazide, scholl solution, 1,25-dihydroxycholecalciferol and calcium carbonate. Iron was given for anemia.

The patient was followed up for 10 months after admission. During the last examination, his BUN level was 32 mg/dl (11.4 mmol/l) and creatinine: 3.2 mg/dl (282.9 µmol/l). He died because of acute bronchiolitis in another hospital when he was 15 months old.

The diagnosis of JN is generally based on genetic, clinical and pathological features [1, 3]. However, only a few patients show all three features simultaneously. As the disease is inherited in an autosomal recessive pattern, the presence of an affected member of the family facilitates the diagnosis [1, 2]. Since the renal cysts are not always present or are particularly hard to be detected in percutaneous needle biopsy specimens, as the material is rarely obtained from the medullary regions, the renal biopsy may not be specific for JN [2, 3]. The diagnosis of JN in this patient was based on family history and clinical and laboratory findings. However, he showed two unusual findings for JN. The first was the early onset of the symptoms and the second was the progression of renal insufficiency in early infancy. The asphyxia in the neonatal period or the administration of several tubulotoxic drugs in the first months of life may be the factors which caused the early onset of symptoms and the development of renal failure during infancy. The persistence of the decreased renal function

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Fig. 1. Abdominal ultrasonography shows multiple cortical and medullary cysts in normal-sized, hyperechogenic (grade III) kidneys with loss of corticomedullary differentiation.

Fig. 2. CT scan showing multiple hypodense areas in the upper poles of the kidneys.

1 year after discontinuation of tubulotoxic drugs suggests more destruction in renal parenchyma than expected, so this can be explained by the primary disease of the patient.

With the unusual presentation and with the factors facilitating renal failure early in infancy, our case may be part of the nephrophthisis complex, however it is also possible to be a new variant of the disease presenting with the early development of renal failure.

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