Glomerulocystic Kidney-Report of an Adult Case

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A 32-year-old adult case of glomerulocystic kidney disease (GCK) is reported. He had brain and muscle atrophy, probably due to congenital malformation. Progressive renal failure developed and he died. Autopsy disclosed multiple tiny cysts exclusively in the renal cortices. Microscopic study revealed that the cysts were dilated Bowman's spaces. This case is the 12th adult case reported in the world.

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Key words: cystic kidney, Bowman's space, brain atrophy

Introduction

Glomerulocystic kidney disease (GCK) which involves bilateral cortical polycystic renal changes due to uniform dilatation of Bowman's capsule was named by Taxy and Filmer in 1976 (1). Since then, there have been many case reports in newborns and infants. Eleven adult cases have been reported since the first report of a 25-year-old female by Dosa et al (2). We treated a 32-year-old male with renal dysfunction of unknown etiology associated with muscle and brain atrophy. GCK was disclosed by autopsy in the present case. This rare entity of cystic disease should be considered in the differential diagnosis of renal dysfunction of unknown cause.

Case Report

A 32-year-old male was referred to Tokai University Hospital in November 1989 because of advanced renal dysfunction (serum creatinine: 4.0 mg/dl). He had postbulbar optic neuritis and elevated intracranial pressure at the age of 13, but no tumor was noted by craniotomy at that time. He had experienced several episodes of disturbance of consciousness from epileptic seizures since the age of 14. Dementia of unknown etiology had also developed since that age. He was admitted to another hospital and was diagnosed as suspected Leigh encephalopathy (mitochondrial cerebromyopathy) and other metabolic disease of the neurons although no definite diagnosis was made. Advanced renal dysfunction was disclosed at the age of 32 by a local doctor. None of his family members had renal problems.

Physical examination on admission to our hospital revealed anemic conjunctiva and diffuse muscle atrophy. His blood pressure and body temperature were normal. No skin rash, lymphadenopathy or joint deformity was observed. There was

no peripheral edema. As depicted in Table 1, urinalysis disclosed no albuminuria, glucosuria or microhematuria except for high excretion of β_2 microglobulin (8.79 $\mu\text{g/ml}$; normal $<0.25 \mu\text{g/ml}$). Normocytic anemia ($205 \times 10^4/\mu\text{l}$ red blood cells) and renal dysfunction (4.7 mg/dl of serum creatinine) were demonstrated. Ultrasonography and CT scan showed bilateral atrophic kidneys with several small cysts. After admission, his general status gradually declined with occasional convulsions and disturbed consciousness; he died on February 24, 1990.

Autopsy disclosed marked brain, muscle and kidney atrophy. There was incomplete necrosis (diameter: 1.5 $\times$ 2.0 cm) in the bilateral putamen. Necrosis was also observed in the gray matter with growth of vessels. Dilations of the third ventricle of the cerebrum and aqueductus cerebri were noted, perhaps due to cerebral atrophy associated with slight necrosis in the peripheral area of the aqueductus cerebri. There was myelinoclasia in the optic nerve, and gliosis in the posterior lobe. Necrotic changes were also noted in the gray matter of the spinal cord, especially in the thoracic cord. These findings might explain the postbulbar optic neuritis, disturbance of consciousness and dementia.

Marked muscle atrophy was noted in the extremities. Muscle fibers were occasionally necrotic and no neurogenic change was observed. There was degeneration of mitochondria in the skeletal muscle. Both kidneys were atrophic (rt. 90 g, lt. 70 g) and tiny multiple cysts (max. dia. 4 mm) were located exclusively in the cortices (Fig. 1). A microscopic study showed normal to atrophic glomeruli in the markedly dilated Bowman's capsules (Fig. 2). About 20% of Bowman's capsules were normal in size microscopically. Tubuli showed a thyroid pattern but were not dilated. Intra-glomerular components were normal to slightly sclerotic although some of them showed poor expansion. Interstitial fibrosis and slight mononuclear cell

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Glomerulocystic Kidney

Table 1. Laboratory Data on Admission

Complete blood study		Serum study	
WBC	7,000/ μ l	IgG	1,730 mg/dl
RBC	205 $\times$ 10 ⁴ / μ l	IgA	291 mg/dl
Hgb	6.4 g/dl	IgM	246 mg/dl
Hct	16.8%	C3	71 mg/dl
Plt	17 $\times$ 10 ⁴ / μ l	C4	40 mg/dl
Chemical study		CH ₅₀	58.8 U/ml
UN	102 mg/dl	VLDL	(-)
Cr	4.7 mg/dl	HBsAg	(-)
Na	139 mEq/l	ANA	(-)
K	5.8 mEq/l	ESR	35/75 mm
C1	112 mEq/l	Coagulation study	
TP	6.9 g/dl	PT	9.8 sec (10.6 sec)
Alb	3.7 g/dl	PTT	34.7 sec (31.8 sec)
T-Bil	0.2 mg/dl	Fibrinogen	276 mg/dl
GOT	42 IU/l	Urinary analysis	
GPT	22 IU/l	Protein	(-) (0.16 g/day)
LDH	31 IU/l	Glucose	(-)
T-chol	105 mg/dl	OB	(-)
TG	52 mg/dl	β_2 microglob	
β_2 microglob	12.7 μ g/ml		8.79 μ g/ml (<0.25 μ g/ml)

ECG: posterior and lateral cardiac ischemia susp, EMG: lower motor neuron lesion susp, EEG: diffuse θ pattern, Abd US: atrophic kidney (Lt. 83 $\times$ 48 mm Rt. 62 $\times$ 38 mm), Abd CT: atrophic kidney and cystic lesion. (Footnote Hgb: hemoglobin, Hct: hematocrit, Plt: platelet, Cr: creatinine, T-Bil: total bilirubin, T-chol: total cholesterol, TG: triglyceride, β_2 microglob: β_2 microglobulin, sec: second, OB: occult blood)

Fig. 1. Tiny multiple cysts are shown exclusively in the cortices (Azan stain, $\times$ 5).

infiltration were observed in the tubulo-interstitial area (Fig. 3). There was no medullary atrophy and no urinary tract abnormalities.

These findings suggested that this patient had GCK and muscle and brain atrophy, probably due to Leigh encephalopathy or other metabolic abnormalities.

Discussion

Since the first case of GCK characterized by dilatation of

Fig. 2. Atrophic glomeruli are shown in markedly dilated Bowman's capsules (HE stain, $\times$ 400).

Fig. 3. Interstitial fibrosis and mononuclear cell infiltration are seen in the tubulo-interstitial area (Azan stain, $\times$ 200).

Bowman's capsule reported by Taxy and Filmer in 1976 (1), many infant and child cases have been reported. Many of the children with this condition died from progressive renal failure or other associated congenital anomalies. Eleven adult cases of GCK have been reported since the first report by Dosa et al in 1984 (Table 2). Most cases were sporadic except for those that reported by Carson et al (5) and Kaplan et al (7). Associated complications were heterogeneous and consisted of both acquired and congenital complications. The presented case had brain atrophy with mild necrosis and muscle atrophy, probably due to mitochondrial cerebromyopathy. However, the precise pathogenetic mechanism of GCK was not clear in this case. Adult cases of GCK have been reported to be associated with various disorders as shown in Table 2. Therefore, GCK might not be a distinct clinicopathological entity, but rather an associated pathological condition. Further accumulation of GCK cases is necessary to elucidate the pathogenesis of this rare renal

Table 2. Adult Cases of Glomerulocystic Kidney

Age (year)	Sex	Complications	Author	Reference No.	Year
(1) 25	F	Tbc (lung, abdominal lymph nodes, liver, spleen)	Dosa et al	2)	1984
(2) 23	M	Retinitis pigmentosa, Hearing loss, RTA type 1 Growth retardation	Kobayashi et al	9)	1985
(3) 29	M	Glomerulonephritis	Oh et al	10)	1986
(4) 18	F	SLE	Carson et al	3)	1987
(5) 60	M	None	Sessa et al	6)	1988
(6) 25	F	Dominant inheritance, Pyloric stenosis, Prognathism	Kaplan et al	4)	1989
(7) 63	M	Proliferation and cystic dilation of interlobular bile duct	Monma et al	7)	1990
(8) 32	M	Hypothyroidism	Kato et al	11)	1991
(9) 23	F	Hemolytic-uremic syndrome	Thompson and Morley	12)	1991
(10) 59	M	Hypertension	Egashira et al	5)	1991
(11) 69	M	Nephrotic syndrome	Uemasu et al	8)	1991
(12) 32	M	Brain and muscle atrophy	Present case		1993

involvement.

Some reports have suggested the usefulness of ultrasonography for diagnosis of GCK (3–5). However, the diameter of many cysts may be too small to obtain a definite diagnosis of GCK. Recently, Egashira et al reported MRI of GCK (5). They indicated uniform low intensity of the renal cortex on T1-weighted images and cortical high intensity similar to the medulla on T2-weighted images. GCK in the presented case was disclosed by autopsy. Ultrasonography and CT scans would not have revealed this condition. Therefore, it appears that tissue diagnosis by renal biopsy might have been the only procedure to obtain a definite diagnosis.

The prognosis of GCK is variable. Many infant and child cases have a poor prognosis because of progressive renal failure or associated congenital malformations. However, three of the 12 adult cases were over 60 (6–8). This fact indicates that there may be asymptomatic patients. GCK should be considered in the differential diagnosis of cystic disease in adults.

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