Dialysis Modality for Patients with Turner Syndrome and Renal Failure

KEY WORDS: Turner syndrome; renal failure; dialysis modality.

Editor:

Turner syndrome (TS) is a rare disease that occurs at a frequency of 1 in 2500 live female births (1). The classical karyotype is 45,X, but the 45,X cell line is seen in various forms of mosaicism (2). Turner syndrome has been associated with multiple organ abnormalities, including mental retardation, cardiovascular abnormality, renal abnormality, and gonadal dysgenesis (1). Certain dysmorphic features such as webbed neck, nail dysplasia, high palate, and short fourth metacarpal are also present (3). As advancements in current medical care improve quality of life and life span for patients with TS, a higher proportion of these patients are entering into adulthood, and the need for dialysis after renal failure develops in these patients is expected to rise (4).

Although many renal anomalies of TS are well known, including horseshoe kidney, duplication of collecting system, and poor development of renal blood vessels (5), the prevalence of end-stage renal disease (ESRD) in TS has never been reported—nor has the choice of dialysis modality for patients with TS. We collected cases with

features of TS under dialysis in a tertiary medical center for more than 15 years. We found 3 candidates, 1 of whom was excluded because of a non-TS gene mutation; and thus we present 2 patients with TS and ESRD.

CASE 1

A 48-year-old Chinese woman had a history of a variant type of TS (46,XXqi) with type 2 diabetes mellitus under insulin therapy for more than 10 years, but without adequate glycemic control. She had been admitted because of diabetic retinopathy and cataract at the age of 39. Because uremic symptoms developed after progressive deterioration of her renal function, she decided to receive renal replacement therapy at the age of 44.

Even after venous mapping by Doppler ultrasonography, her poor vascular condition was still not suitable for the creation of a vascular access for maintenance hemodialysis (HD). Thus, a continuous ambulatory peritoneal dialysis (CAPD) catheter was implanted, and she thereafter had an uneventful course of peritoneal dialysis (PD) with her family's assistance.

CASE 2

A Chinese woman had a history of TS (45,X), with mental retardation since childhood, and of ESRD with repeated episodes of catheter infection during the initial 3 months of HD at the age of 42. She was then transferred to our hospital for consideration of CAPD as the mode of renal replacement therapy.

A first episode of peritonitis was noted 2 years after initiation of CAPD, and she was switched to automated PD afterwards. She then had an uneventful course of dialysis for another 2 years with her brother's assistance. However, she had repeated episodes of peritonitis because of poor self-care after discontinuation of her brother's assistance, leading to removal of the PD catheter. Subsequently, a cuffed catheter was implanted over the right internal jugular vein for HD during her last admission in our hospital. She eventually expired of septic shock caused by severe peritonitis at the age of 47.

DISCUSSION

In case 1, the dialysis course was smooth under CAPD. In case 2, repeated catheter infection was noted during HD, and we decided to change the dialysis modality to CAPD in that patient. Her subsequent course was smooth at first, but discontinuation of assistance and poor self-care related to mental retardation caused by monosomy eventually led to mortality after the switch to HD.

Several renal diseases are associated with TS and may contribute to ESRD. First, patients with TS tend to have structural abnormalities of the kidneys such as horseshoe kidney, duplication of collecting system, and obstruction of the ureteropelvic junction, which may require surgery (6). Second, vascular abnormalities and hypertension are seen in TS. A report from Carvalho *et al.* revealed that, of patients with TS who underwent echocardiograms, 25.6% showed cardiovascular abnormalities (7). In 1997, Jelic and Marisavljevic reported a case of TS with renovascular hypertension (8). Third, some glomerulonephropathies are associated with TS, including membranoproliferative glomerulonephritis (9) and focal segmental glomerulosclerosis (10).

Because the national health insurance system in Taiwan allows all ESRD patients to receive dialysis therapy fully paid by governmental insurance despite their economic status, we can share our rare experience of choosing the dialysis modality for patients with TS. Because patients with TS usually have poor vascular condition, it may be difficult to create a vascular access for them, whether an arteriovenous fistula or a cuffed catheter. By contrast, with the adequate assistance of well-trained caregiver, CAPD or automated PD can provide patients with a relatively stable dialysis course. However, peritonitis and burnout in family support are still difficult issues to overcome during clinical care for PD patients. If family support is strong enough or if patients can take good care of themselves, CAPD or automated PD might be considered to be the treatment of choice for those patients.

This report is the first to address the dialysis modality of choice for TS patients with ESRD; more studies are needed to confirm our clinical experience. At this time, we do not know the type of ESRD care that would be preferable for TS patients. This article can serve as a launching point to survey patients having the comorbidities of TS and ESRD. We would be glad to have other medical professionals share their experiences with us. Such sharing can overcome the difficulties in gathering cases to reach general agreement on treatment guidelines for these patients.

DISCLOSURES

The authors have no conflicts of interest to declare.

W.S. Liu[1,2,3]
S.Y. Li[1,2]
W.C. Yang[1,2]
T.W. Chen[4,5]
C.C. Lin[1,2]*

Division of Nephrology[1]
Taipei Veterans General Hospital
School of Medicine[2]
National Yang-Ming University
Division of Nephrology[3]
Taipei City Hospital
Zhong-Xing Branch
Division of Nephrology[4]
Department of Medicine[5]
School of Medicine, College of Medicine
Taipei Medical University Hospital
Taipei, Taiwan

*email: lincc2@vghtpe.gov.tw

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