Renal Problems in Early Adult Patients with Turner Syndrome

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Introduction

Turner syndrome (TS) is caused by the haplo-insufficiency of some or all genes on the X chromosomes. This status results from a complete or partial absence of the second X chromosome or from structural anomalies of one X chromosome. For patients with TS who reach from adolescence to early adulthood, it is essential to make early diagnosis and effective management of the various accompanying problems in order to maintain quality of life. The most dangerous and life-threatening problems in adult patients with TS are definitely cardiovascular problems, and already many studies have been reported. However, the studies on renal problems are relatively limited, and there have been few follow-up studies on renal function in particular. Therefore, this study's investigators assessed renal function in patients with TS who had reached from adolescence to early adulthood.

Materials and Methods

Subjects

The study included 63 patients who were over 18 years old at the latest visit among patients who had been diagnosed with TS by a chromosome examination at Inje University, Busan Paik Hospital. The average patient age at the latest visit was 23.64 ± 4.51 years old, and the mean observation period was 6.09 ± 4.12 years. The mean age at the initial diagnosis was 10.49 ± 4.04 years old.

Methods

Urine and blood chemistry tests were performed in all patients for renal function and urine abnormality evaluation. Renal ultrasonography for radiological evaluation of the kidneys was conducted in all patients at the time of initial diagnosis, and it was repeated at least every three years in patients with renal malformation. For renal malformation suspected on the renal ultrasonography, intravenous pyelography was also performed. For some patients who appeared on the renal ultrasonography to require differential diagnosis with renal parenchyma abnormalities or tumors, DMSA renal scan and renal CT scan were conducted.

Results

Cytogenetic findings

Of the 63 patients, the karyotype showed 45,X in 32 (50.8%) patients, variant type in 31 (49.2%) patients. Of the 31 patients with variant type, 22 patients were mosaicism and 9 patients were structural aberration (Table 1).

Urinary abnormalities and renal function

The renal function at the latest visit was shown as normal in all patients; the serum BUN level was 9.72 ± 2.60, and the creatinine level was 0.64 ± 0.11. Nephrotic syndrome had developed in one patient; he was in remission after use of steroids and has been maintained in remission without relapse. Hematuria was accompanied in seven patients, and it accounted for 11.1% of the total. Six of those patients were found to have hematuria at the initial diagnosis; the other one developed it during follow-up. Three of the seven patients with hematuria had renal anomalies, while the other four did not. All cases of hematuria were microscopic hematuria and there was no patient showed gross hematuria. In addition, no patients had proteinuria except for one with prior nephrotic syndrome.

Incidence of renal anomalies according to the karyotype

Renal anomalies were observed in 20 of the 63 (31.7%) patients. Of the 32 patients with 45,X karyotype, 13 (40.6%) had renal anomalies, while these were found in 7 (22.6%) of the 31 patients with mosaicism/structural aberration. The incidence of renal anomalies of the patients with 45,X karyotype was higher than that of the patients with mosaicism/structural aberration, but there was no statistical significance (P>0.05).

Type of renal anomalies

The most frequent type of accompanying renal anomalies was horseshoe kidney, which occurred in 10 patients, it accounted for 50% of all cases of renal malformation. The other types of accompanying renal anomalies are provided in Table 2.

Table 1. Cytogenetic Findings in 63 Turner syndrome patients

Karyotype	No. of cases (%)	
Classic : 45,X	32 (50.8)	
Variant	31 (49.2)	
Mosaicism	22	
45,X/46,XX		5
45,X/46,X,i(Xq)		5
45,X/47,XXX		3
45,X/46,XY		2
45,X/46,X,+mar		1
45,X/47,XYY		1
45,X/46,XX/47,XXX		2
45,X/46,X,del(Xq)		2
45,X/46,X,r(X)		1
Structural aberration	9	
46,X,i(Xq)		7
46,X,del(Xp)		1
46,X,del(Xq)		1
Total	63 (100.0)	

Table 2. Type of renal anomalies

Karyotype	Number	Type of renal anomalies
45,X	7	Horseshoe kidney
	1	Renal agenesis, Rt
	1	pelvocalyces and ureter, Lt
	2	Bifid renal pelvis, Lt
	1	Axial deviation of both kidneys, Lt pyeloectasia
	1	Vertical axis deviation, Lt
Incidence of renal anomaly in 45,X=13/32 (40.6%)		
45,X/46,XX	3	Horseshoe kidney
45,X/46,X,del(Xq)	1	Horseshoe kidney
45,X/46,X,+mar	1	Hydronephrosis with incomplete UPJ obstruction, Lt
45,X/46,XX/47,XXX	1	Renal duplication, bilateral
46,X,del(Xp)	1	Renal duplication, Lt
Incidence of renal anomaly in mosaicism and structural aberration=7/31 (22.6%)		

Conclusion

The renal function in all patients with TS who had entered early adulthood presented as normal. This suggests that most patients with TS do not have any significant problems with renal function during childhood. However, accompanying renal malformation showed relatively high frequency, 31.7%, and hematuria events in 11% of all cases, careful attention should be in TS patients with hematuria to prevent progressing renal problems.

ESPE 2015 1241--P3

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DOI: 10.3252/pso.eu.54espe.2015

Poster presented at: 54th Annual Meeting 1-3 October ESPE 2015 BARCELONA European Society for Paediatric Endocrinology