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Renal Problems in Early Adult Patients with Turner Syndrome

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Introduction: The prevalence of renal anomalies in Turner syndrome (TS) has been reported to vary from 30 to 70%. However, the influence of renal anomalies on renal function and morbidity has been less well investigated. We evaluate the status of renal function and the presence of urinary abnormalities in early adult TS patients. **Patients and method:** Sixty-three girls with TS, who are attending Pediatric Endocrine Clinics in Busan Paik Hospital, were studied. The mean age of the patients at last follow-up was 23.64 ± 4.51 years and at diagnosis was 10.49 ± 4.04 years. The mean duration of follow-up was 6.09 ± 4.12 years. KUB sonography was performed in all TS patients and some of them also had IVP, renal DMSA scan, and renal CT. Renal function test with R-UA were examined in every visiting times. **Results:** Of the 63 patients, the karyotype showed 45,X in 32 (50.8%) patients, mosaicism in 22 (34.9%) and structural aberration in 9 (14.3%). Renal anomalies were observed in 20 of the 63 TS (31.7%). Of the 32 TS patients with 45,X karyotype, 13 (40.6%) had renal anomalies, while these were found in 7 (22.6%) of 31 TS patients with mosaicism/structural aberration. But there is no significant statistical differences between two karyotype groups. The renal anomalies included ten cases of horseshoe kidney, one case of renal agenesis, eight cases of abnormal renal collecting system, and one case of malrotation. At last follow-up time, the mean serum level of BUN and Cr level were 9.72 ± 2.60 and 0.64 ± 0.11 respectively. Hematuria was observed in 7 (11.1%) TS patients. Among them three TS patients have renal anomalies. One TS patient suffer from nephrotic syndrome for 2.5 years. **Conclusion:** The prevalence of renal anomalies in Korean TS patients was 31.7% and there is no significant differences between two different karyotypes. At last follow-up, all TS patients have normal renal function. Hematuria was observed 11% of TS patients. Although associated renal anomalies may not influence renal function in early adult, careful attention should be necessary in TS patients with hematuria to prevent progressing renal problems.

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To Predict Ovarian Function is a Single Determination of AMH Useful in Patients with Turner Syndrome?

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Background: Different studies have underlined the role of anti-Müllerian hormone (AMH) and inhibin B as markers of the

ovarian function in paediatric and adolescent patients with Turner syndrome (TS). **Objective and hypotheses:** Our study aims to verify the role of AMH in a cohort of patients affected by TS. **Method:** We analysed 23 TS patients, aged 2–34 years, describing their auxological parameters and the pubertal development, and evaluating their hormonal (AMH, FSH, LH, estradiol, and inhibin B) levels. **Results:** Twenty-one out of 23 (91.3%) were treated with GH. AMH resulted measurable only in two patients of 23 (8.7%), whereas inhibin B was measurable in 13 of 23 (56.5%) patients. Our results were highly heterogeneous. In particular, there are predictive factors neither for the response to GH treatment nor by the puberty. In fact, a good response of GH treatment both as final height and in relation to ΔTH (final height- mid-parental height) was independent from karyotype, from hormonal levels and from spontaneous puberty development. In addition, a karyotype 45X0 was not predictive of worst height gain or of not spontaneous pubertal development. **Conclusion:** There is not a predictive factor that allows to know in advance the evolution of puberty in patients with TS. A single determination of AMH is not informative; only repeated evaluations of this ovarian marker in childhood and adolescence may be useful to predict a spontaneous beginning of puberty and to suggest a possible fertility.

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Anthropometric Findings from Birth to Adulthood in Turkish Girls with Turner Syndrome and Association with Karyotype Distribution

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Background: Turner syndrome (TS) can manifest with various clinical features depending on the karyotype and the genetic background of affected subjects. **Objective and hypotheses:** The aim of this study was to evaluate growth parameters from birth to adulthood in girls with TS in a cross-

sectional study. **Method:** A total of 842 patients, with an age of diagnosis ranging from birth to 18 years followed-up between 1984 and 2014, from 35 different centers were included in this study. Patients who used growth hormone injections, estrogen or oxandrolone were excluded. **Results:** Fifty-one (8.8%) patients were born before 37 weeks. In this cohort small for gestational age was 33% and almost all of them were born termly. Mean birth length was 1.3 cm shorter and mean birth weight was 0.36 kg lower than normal population. At the mean of age of presentation was 10.1 ± 4.4 years, mean height, weight, and BMI-SDS were -3.1 ± 1.7 , -1.4 ± 1.5 , and 0.4 ± 1.7 respectively. There was no karyotype association with respect to birth length and weight or height and weight at presentation. Mid-parental height was the only parameter that had an effect on the prediction of height of children with TS. **Conclusion:** There was no effect of karyotype in height of girls with TS however weight was heavier in 46,X,i(Xq) and 45,X/46,X,i(Xq) karyotype groups.

tolerance or malignancies; four patients had transient increase in fasting glucose (>100 and <126 mg/dl); two patients developed hypothyroidism and were treated with levothyroxine. **Conclusions:** GH treatment is associated with highly significant changes in growth. In our study height velocity was maximum (8.53 cm/year) in the 1st year of GH treatment; the improvements in growth declined in the second (6.85 cm/year), 3rd year (4.11 cm/year), and 4th year (4.05 cm/year). GH therapy had a favourable safety profile. Delayed diagnosis of TS has a negative impact on growth outcomes.

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GH Therapy in Turner Syndrome

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Background: Turner syndrome (TS) is one of the most common causes of short stature in females. Adult height of patients with TS is 20 cm shorter than in general population. GH therapy improves height outcome in girls with TS; results depend on age at diagnosis, duration of therapy, and doses of GH. **Objective:** To evaluate growth and safety during the first 4 years of GH treatment in patients with TS. **Method:** Eight prepubertal girls with TS were included mean age of 11.54 years. They were treated with a mean dose of GH=0.037 mg/kg per day and followed for at least 4 years (mean 5.2 years). **Results:** The mean height SDS increased from -3.61 at baseline to -1.37 at 4 years. Main gain over 4 years was 23.55 cm. The mean weight SDS increased from -1.28 at baseline to -0.68 at 4 years. Bone age was delayed at diagnosis by a mean value of 1.17 years and after 4 years the delay decreased to 0.22 years (Table 1). **Safety profile:** There were no cases of diabetes mellitus, impaired glucose

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A Rare Variant of Turner Syndrome: First Clinical Report from Kuwait

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Background: Turner syndrome (TS) is characterised cytogenetically by X chromosome monosomy, the presence of an abnormal X chromosome, or mosaicism of a 45,X or have an abnormal sex chromosome rearrangement. Girls with variant TS show no features, fewer or milder features of TS. **Objective and hypotheses:** We are reporting on a clinical report of a girl with a rare variant of TS (46,X,i(X) (q10)). **Method:** This is a case report of a 12-year-old Kuwaiti girl who was referred for assessment of the short stature and hypothyroidism for which she has been already started on GH and thyroid replacement therapy. **Results:** The girl's height was at -4 s.d. Chromosomal analyses were revealed 46,X,i(X) (q10). Thyroid function test was normal on treatment with negative anti-TPO antibodies. Ultrasound of the abdomen and pelvis showed small uterus for her age and non-visualized ovaries with no renal anomalies. Echocardiography was normal. Genetic counselling was done for her and her family. She is currently under treatment with growth hormone and thyroid replacement with appropriate doses for her diagnosis. **Conclusion:** Our case demonstrated features similar to those in girls with this rare form of TS. However, our patient did not have oedema, cardiac or renal anomalies. This case demonstrates the importance of doing karyotype in such girl even without overt clinical features of TS to diagnose TS and its complications.

Table 1. (for abstract P3-1244)

Parameter	Baseline	1 year	2 years	3 years	4 years
Age (years)	11.54	12.54	13.54	14.54	15.54
Bone age (years)	10.37	10.95	12.48	14.05	15.32
Height SDS	-3.61	-3.05	-2.48	-1.92	-1.37
Height velocity (cm/year)	-	8.53	6.85	4.11	4.05
Weight SDS	-1.28	-1.08	-0.86	-0.80	-0.68