

**Table 1: Clinical and biochemical characteristics of Williams' Syndrome (WS) patients and controls.**

	<b>WS patients (n=29)</b>	<b>Controls (n=29)</b>	<b><i>p</i></b>
<b>Age (years)</b>	28.8±5.5 (19 – 39)	30.3±5.0 (22 – 40)	0.290
<b>Sex (males/females)</b>	55.2/44.8 (16/13)	55.2/44.8 (16/13)	1.000
<b>BMI (kg/m<sup>2</sup>)</b>	25.7±4.4 (19.0 – 35.1)	26.1±3.3 (21.2 – 32.0)	0.730
<b>Height (m)</b>	1.56±0.05 (1.46 – 1.66)	1.69±0.13 (1.51 – 1.93)	<b>0.001</b>
<b>Serum Calcium (mg/dL)</b>	9.5±0.4 (8.6 – 10.2)	9.4±0.3 (9.0 – 10.2)	0.070
<b>Ionized Calcium (mmol/L)</b>	1.21±0.04 (1.11 – 1.29)	1.18±0.04 (1.10 – 1.28)	<b>0.017</b>
<b>Urinary Calcium (mg/Kg/day)</b>	2.8±1.1 (1.0 – 5.3)	3.0±1.6 (0.1 – 5.1)	0.645
<b>Serum Phosphorus (mg/dL)</b>	3.1±0.7 (2.0 – 4.5)	3.8±0.5 (2.7 – 5.0)	<b>0.0001</b>
<b>Prevalence of serum Phosphorus &lt;2.7 mg/dL</b>	34.5 (10)	3.4 (1)	<b>0.005</b>
<b>Creatinine (mg/dL)</b>	0.7±0.1 (0.5 – 1.0)	0.8±0.2 (0.5 – 1.1)	0.060
<b>eGFR</b>	123.7±28.4 (88.0 – 190.)	132.7±23.4 (72.8-177.0)	0.197
<b>TmP/GFR (mmol/L)</b>	0.81±0.32 (0.20 – 1.39)	1.06±0.25 (0.10 – 1.50)	<b>0.001</b>
<b>Prevalence of TmP/GFR &lt;0.8 mmol/L</b>	37.9 (11)	3.4 (1)	<b>0.002</b>
<b>bALP (U/L)</b>	26.3±8.5 (12.0 – 50.0)	35.0±8.0 (20.5 – 49.0)	<b>0.0001</b>
<b>PTH (pg/mL)</b>	24.5±12.6 (7.8 – 55.7)	33.7±10.8 (17.3 – 48.3)	<b>0.004</b>
<b>25OHVitD (ng/mL)</b>	32.3±16.0 (12.5 – 76.0)	37.6±17.4 (18.0 – 75.0)	0.230
<b>Prevalence of 25OHVitD &lt;30 ng/mL</b>	51.7 (15)	37.9 (11)	0.429
<b>HbA1c (mmol/mol)</b>	32.5±3.7 (27-40)	-	-
<b>OC (ng/mL)</b>	19.4±5.3 (8.7 – 28.7)	24.5±8.7 (14.9 – 46.8)	<b>0.010</b>
<b>CTX (pg/mL)</b>	401.2±169.3 (206 – 802)	322.3±122.4 (115 – 539)	<b>0.046</b>
<b>FGF23 ( pg/mL)<sup>§</sup></b>	40.9±21.2 (10.1-87.7)	-	-
<b>BMD LS (Z-score)*</b>	-0.17±0.37 (-3.20 – 2.00)	0.16 ±0.37 (-1.26 – 1.86)	0.508
<b>BMD FN (Z-score)*</b>	-0.51±0.32 (-2.70 – 2.50)	0.36±0.32 (-0.62 – 1.80)	<b>0.009</b>
<b>Prevalence of BMD Z-score &lt; -2.0</b>	24.1 (7)	3.4 (1)	0.050
<b>Prevalence of vertebral fractures</b>	10.3 (3)	0 (0.0)	0.237

<b>Prevalence of hypothyroidism</b>	17.2 (5)	0 (0.0)	0.052
<b>Prevalence of diabetes mellitus type 2</b>	13.7 (4)	0 (0.0)	0.112

*Data are expressed as mean  $\pm$  SD or medians  $\pm$  SE or percentage with range or absolute number in parentheses, respectively. <sup>§</sup>FGF23 was measured in 19 patients. \*Adjusted for height by general linear modelling; BMI: Body Mass Index; Urinary calcium: calcium excretion in 24h urinary collection; eGFR: estimated glomerular filtration rate; TmP/GFR: tubular maximum reabsorption of phosphate to glomerular filtration rate; bALP: bone alkaline phosphatase; PTH: parathyroid hormone; 25OHVitD: 25-hydroxyvitamin D; OC: osteocalcin; CTX: carboxyterminal cross-linking telopeptide of type I collagen; BMD: bone mineral density.*

*SI conversion factors: calcium,  $\times$  0.25; urinary calcium,  $\times$  0.02; phosphorus,  $\times$  0.32; bALP,  $\times$  1; PTH,  $\times$  1.0; 25OHVitD,  $\times$  2.49; OC,  $\times$  1; CTX,  $\times$  1.*

**Table 2: Clinical and biochemical characteristics of Williams' Syndrome (WS) patients with low and with normal TmP/GFR.**

	<b>Low TmP/GFR (n=11)</b>	<b>Normal TmP/GFR (n=18)</b>	<b><i>p</i></b>
<b>Age (years)</b>	30.8±5.9 (24 – 38)	27.6±5.1 (19 – 39)	0.132
<b>Sex (males/females)</b>	45.5/54.5 (5/6)	61.1/38.9 (11/7)	0.46
<b>BMI (kg/m<sup>2</sup>)</b>	26.7±4.5 (19.0 – 35.1)	25.1±4.3 (19 – 35.0)	0.370
<b>Serum Calcium (mg/dL)</b>	9.5±0.5 (8.6 – 10.2)	9.5±0.3 (8.9– 10)	0.890
<b>Urinary Calcium (mg/Kg/day)</b>	2.9±1.3 (1.0 – 4.7)	2.8±1.0 (1.5 – 5.3)	0.825
<b>Serum Phosphorus (mg/dL)</b>	2.4±0.4 (2.0 – 3.1)	3.5±0.5 (3.0 – 4.5)	<b>0.0001</b>
<b>bALP (U/L)</b>	29.0±10.4 (12.0 – 50.0)	24.7±6.9 (15.0 – 42.0)	0.190
<b>PTH (pg/mL)</b>	29.2±14.9 (7.8 – 55.7)	21.6±10.3 (10.5 – 54.5)	0.119
<b>25OHVitD (ng/mL)</b>	28.5±13.6 (12.5 – 48.0)	34.6±17.2 (14.6 – 76.0)	0.321
<b>OC (ng/mL)</b>	18.4±6.5 (8.7 – 28.7)	20.1±4.5 (10.6 – 27.7)	0.419
<b>CTX (pg/mL)</b>	367.1±196.4 (217 – 802)	422.1±152.6 (206 – 741)	0.406
<b>FGF23 (pg/mL)<sup>§</sup></b>	37.3±17.3 <sup>§§</sup> (11.9 – 62.2)	43.1±23.6 <sup>§§§</sup> (10.1 – 87.7)	0.579
<b>BMD LS (Z-score)*</b>	0.21±1.10 (-2.00 – 1.50)	-0.39±1.50 (-3.20 – 2.00)	0.258
<b>BMD FN (Z-score)*</b>	-0.44±1.2 (-2.70 – 1.10)	-0.53±1.3 (-2.20 – 2.50)	0.852
<b>Prevalence of vertebral fractures</b>	9.1 (1)	11.1 (2)	1.000

*Data are expressed as mean ± SD or medians ± SE or percentage with range or absolute number in parentheses, respectively.*

*§FGF23 was measured in 19 patients. §§12 patients. §§§ 7 patients.*

*\* Adjusted for height by general linear modelling*

*BMI: Body Mass Index; Urinary calcium: calcium excretion in 24h urinary collection; TmP/GFR: tubular maximum reabsorption of phosphate to glomerular filtration rate; bALP: bone alkaline phosphatase; PTH: parathyroid hormone; 25OHVitD: 25-hydroxyvitamin D; OC: osteocalcin; CTX: carboxyterminal cross-linking telopeptide of type I collagen; BMD: bone mineral density.*

*SI conversion factors: calcium, × 0.25; urinary calcium, × 0.02; phosphorus, × 0.32; bALP, × 1; PTH, × 1.0; 25OHVitD, × 2.49; OC, × 1; CTX, × 1; FGF23, × 0.13*