

Prevalence of comorbid hyperparathyroidism and its association with renal dysfunction in Asian patients with X-linked hypophosphatemic rickets/osteomalacia

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Online Resource 1. Characteristics of patients with XLH by sex

	Men				Women			
	Overall	Non-hyperparathyroidism	Secondary hyperparathyroidism	Tertiary hyperparathyroidism	Overall	Non-hyperparathyroidism	Secondary hyperparathyroidism	Tertiary hyperparathyroidism
n (%)	24 (100)	9 (37.5)	13 (54.2)	2 (8.3)	45 (100)	23 (51.1)	20 (44.4)	2 (4.4)
Age, years, median [IQR]	30.00 [26.75, 44.00]	27.00 [18.00, 43.00]	36.00 [29.00, 47.00]	38.50 [32.25, 44.75]	35.00 [23.00, 49.00]	35.00 [22.50, 48.50]	33.50 [26.00, 48.25]	50.50 [44.75, 56.25]
Nationality, South Korea, n (%)	5 (20.8)	3 (33.3)	2 (15.4)	0 (0.0)	9 (20.0)	5 (21.7)	4 (20.0)	0 (0.0)
Height, cm, median [IQR]	156.50 [154.73, 163.33]	156.60 [154.80, 164.70]	155.70 [149.60, 160.40]	159.10 [157.55, 160.65]	146.95 [143.88, 152.35]	147.95 [145.18, 153.17]	145.85 [140.12, 150.33]	148.75 [142.08, 155.42]
Height, Z-score, median [IQR]	-2.45 [-2.75, -1.27]	-2.43 [-2.74, -1.04]	-2.58 [-3.63, -1.77]	-2.00 [-2.27, -1.73]	-2.12 [-2.71, -1.09]	-1.92 [-2.46, -0.94]	-2.33 [-3.42, -1.48]	-1.78 [-3.05, -0.50]
Weight, kg, median [IQR]	56.30 [52.73, 68.00]	61.60 [53.90, 64.30]	54.80 [53.00, 67.80]	66.70 [59.30, 74.10]	49.05 [44.60, 55.32]	48.50 [44.33, 54.42]	49.35 [46.00, 57.55]	44.85 [41.12, 48.57]
Weight, Z-score, median [IQR]	-0.59 [-1.14, 0.71]	0.09 [-0.94, 0.37]	-0.80 [-1.09, 0.69]	0.17 [-0.56, 0.89]	-0.50 [-1.31, 0.41]	-0.59 [-1.36, 0.29]	-0.44 [-1.04, 0.68]	-1.55 [-2.33, -0.78]

Body mass index, kg/m ² , median [IQR]	23.90 [21.15, 25.77]	24.10 [22.90, 25.70]	23.20 [21.20, 25.40]	26.60 [23.15, 30.05]	22.55 [20.48, 25.52]	21.85 [20.40, 24.95]	23.40 [21.32, 27.75]	20.15 [20.02, 20.27]
Body mass index, Z-score, median [IQR]	0.88 [0.06, 1.31]	0.93 [0.61, 1.29]	0.69 [0.09, 1.23]	0.96 [0.24, 1.69]	0.56 [-0.20, 1.39]	0.33 [-0.22, 1.25]	0.83 [0.15, 1.84]	-0.32 [-0.37, - 0.26]
<i>PHEX</i> mutation, n (%)	12 (50.0)	4 (44.4)	7 (53.8)	1 (50.0)	24 (53.3)	13 (56.5)	10 (50.0)	1 (50.0)
Age at diagnosis, years, median [IQR]	1.95 [1.27, 19.35]	6.82 [1.79, 19.14]	1.77 [1.34, 29.31]	0.19 [0.09, 0.28]	19.12 [2.10, 38.54]	13.72 [1.36, 43.93]	23.64 [6.64, 34.65]	2.49 [2.49, 2.49]
Time since diagnosis, years, median [IQR]	16.05 [4.43, 28.98]	13.18 [4.27, 19.28]	25.23 [7.19, 31.61]	38.36 [32.23, 44.49]	11.31 [1.33, 24.16]	13.36 [1.70, 20.60]	3.75 [0.86, 28.47]	59.51 [59.51, 59.51]
Ongoing treatment, n (%)								
Oral phosphate	18 (75.0)	6 (66.7)	10 (76.9)	2 (100.0)	30 (66.7)	15 (65.2)	13 (65.0)	2 (100.0)
Active vitamin D	21 (87.5)	9 (100.0)	10 (76.9)	2 (100.0)	37 (82.2)	21 (91.3)	15 (75.0)	1 (50.0)
History of calcimimetics								
Cinacalcet, n (%)	0 (0.0)	0 (0.0)	0 (0.0)	0 (0.0)	2 (4.4)	0 (0.0)	1 (5.0)	1 (50.0)
Evocalcet, n (%)	0 (0.0)	0 (0.0)	0 (0.0)	0 (0.0)	0 (0.0)	0 (0.0)	0 (0.0)	0 (0.0)
Parathyroidectomy	2 (8.3)	0.0 (0)	0.0 (0)	2 (100.0)	2 (4.4)	0.0 (0)	0.0 (0)	100.0 (2)
Serum phosphate, mg/dL, median [IQR]	2.00 [1.80, 2.20]	2.00 [1.90, 2.10]	2.00 [1.80, 2.10]	2.65 [2.58, 2.72]	2.30 [2.00, 2.50]	2.40 [2.10, 2.70]	2.10 [1.95, 2.50]	2.15 [2.08, 2.22]
Reference value: 2.4–4.3								
Serum calcium, mg/dL, median, [IQR]	9.40 [9.25, 9.75]	9.50 [9.30, 9.90]	9.40 [9.10, 9.70]	9.50 [9.45, 9.55]	9.30 [9.00, 9.50]	9.40 [9.10, 9.50]	9.20 [9.00, 9.40]	9.40 [9.30, 9.50]

Corrected Ca, mg/dL, median [IQR]	8.80 [8.50, 9.10]	9.00 [8.40, 9.10]	8.80 [8.50, 9.10]	8.95 [8.82, 9.07]	8.80 [8.60, 9.00]	8.80 [8.65, 9.05]	8.80 [8.60, 9.03]	8.80 [8.75, 8.85]
Reference value: 8.5–10.2								
Serum albumin, g/dL, median [IQR]	4.60 [4.50, 4.90]	4.60 [4.50, 4.90]	4.60 [4.20, 4.90]	4.55 [4.48, 4.62]	4.50 [4.30, 4.60]	4.40 [4.30, 4.60]	4.50 [4.18, 4.53]	4.60 [4.55, 4.65]
Reference value: 3.8–5.2								
Serum iPTH, pg/mL, median [IQR]	70.00 [45.00, 97.75]	35.70 [24.00, 48.30]	86.90 [73.00, 141.00]	94.50 [65.25, 123.75]	59.00 [48.00, 87.00]	48.00 [30.00, 53.00]	89.00 [76.55, 116.75]	154.50 [104.75, 204.25]
Reference value: 10–65								
TmP/GFR, mg/dL, median [IQR]	1.52 [1.25, 1.69]	1.69 [1.46, 1.93]	1.43 [1.14, 1.53]	1.70 [1.69, 1.71]	1.70 [1.38, 2.01]	1.79 [1.58, 2.27]	1.65 [1.25, 1.83]	1.31 [1.16, 1.47]
Reference value: 2.3–4.3								
Serum 25OHD, ng/mL, median [IQR]	17.50 [13.00, 21.75]	18.00 [13.00, 25.00]	16.00 [13.00, 19.00]	19.00 [16.50, 21.50]	16.00 [13.00, 20.00]	17.00 [15.00, 22.50]	14.50 [12.23, 16.38]	19.50 [17.75, 21.25]
Deficiency: <20								
Serum 1,25(OH)2D, pg/mL, median [IQR]	39.60 [23.91, 56.20]	51.50 [24.14, 61.50]	39.00 [26.90, 53.60]	45.20 [33.55, 56.85]	40.20 [26.00, 52.30]	37.00 [27.57, 53.05]	42.15 [25.40, 48.17]	44.10 [32.55, 55.65]
Reference value: 20–60								
U-Ca/Cr ratio, mg/gCr, median [IQR]	0.07 [0.04, 0.09]	0.10 [0.07, 0.16]	0.06 [0.03, 0.08]	0.04 [0.03, 0.04]	0.10 [0.06, 0.16]	0.12 [0.08, 0.16]	0.07 [0.04, 0.12]	0.16 [0.08, 0.23]
Reference value: <0.3								

Intact FGF23, pg/mL, median	288.00	231.00	208.00 [147.00,	2194.00	134.00	117.50 [87.10,	146.00 [102.62,	392.50 [370.25,
[IQR]	[146.50,	[139.00,	456.00]	[1441.00,	[88.97,	266.75]	229.50]	414.75]
FGF-related	596.50]	562.25]		2947.00]	268.00]			
hypophosphatemia: >30								
Serum creatinine, mg/dL,	0.69 [0.58,	0.75 [0.58,	0.64 [0.57, 0.73]	1.69 [1.36, 2.01]	0.48 [0.43,	0.48 [0.45,	0.50 [0.43, 0.57]	0.77 [0.75, 0.80]
median [IQR]	0.84]	0.81]			0.57]	0.54]		
Reference value: 0.61–1.04 for								
men, 0.47–0.79 for women								

Ca = calcium, eGFR = estimated glomerular filtration rate, FGF23 = fibroblast growth factor-23, iPTH = intact parathyroid hormone, IQR = interquartile range,

TmP/GFR = ratio of the maximum rate of tubular phosphate reabsorption to the glomerular filtration rate, U-Ca/Cr ratio = urinary calcium creatinine ratio, 1,25(OH)₂D = 1,25-dihydroxyvitamin D

Online Resource 2. Renal function and nephrocalcinosis by hyperparathyroidism classification and sex

[illegible]

Nephrocalcinosis on								
renal ultrasound								
n	11	4	5	2	16	10	5	1
Grade, n (%)								
Grade 1	2 (18)	2 (50)	0 (0)	0 (0)	6 (38)	4 (40)	3 (40)	0 (0)
Grade 2	1 (9.1)	0 (0)	1 (20)	0 (0)	4 (25)	2 (20)	2 (40)	0 (0)
Grade 3	4 (36)	1 (25)	3 (60)	0 (0)	3 (19)	2 (20)	0 (0)	1 (100)
Grade 4	3 (27)	1 (25)	1 (20)	1 (50)	3 (19)	2 (20)	1 (20)	0 (0)
Grade 5	1 (9.1)	0 (0)	0 (0)	1 (50)	0 (0)	1 (10)	0 (0)	0 (0)

Data are number (n) and percentage (%).

eGFR = estimated glomerular filtration rate, IQR = interquartile range

Online Resource 3. Association between eGFR at informed consent by hyperparathyroidism type and sex

