

Supplementary file

Table S1. Primer sequences used for *PHEX* gene amplification (obtained from Francis et al. [30]). Annealing temperature was modified. All primers pairs encompassed exon/intron sites.

Exon	Exon length (bp)	Forward Primer (5' -> 3')	Reverse Primer (5' -> 3')	Ta (°C)	PCR product (bp)
1	118	GCTCTTGAGACCAGCCACCA	ATAAAGCACAAAGGAACTTCTCG	58	249
2	69	TCTTGCCTATGTTTCCGAGGG	CTGTCTTCTCTCCACTTCCC	58	218
3	162	ATTCAGTGCTTGTCATTAATCC	TAAAGTGATCACCAAAACCCC	58	239
4	87	CAAATGACTTCCAACCTTGGCAC	GAACTCTAAAGGATAGGGATGG	58	208
5	227	CTAGTGTGCTGATCCAGTTTGC	GCAGCATGAGTCTCTTTCCC	58	370
6	69	CATCACTCTTGTAAACATGG	GGCCTTAGAACTAATGGGC	58	208
7	117	TCTTCCATGTCTCTCAAACATA	GGAAAAGAAAGAGATTTTCAGTG	58	219
8	84	GTAATCATACAGTAAGAAATGG	GAGATGAAAATCCAATCCCCTC	58	212
9	146	CATTCTGTTTGTCTCTCTCCCCT	TTTTCAAAGGATGTGAGAAGGGAAG	58	257

Exon	Exon length (bp)	Forward Primer (5'-> 3')	Reverse Primer (5'-> 3')	Ta (°C)	PCR product (bp)
10	94	TGTATGAGTAAGAGGTCCCCTCGATG	CTCCCCCTGTCTAATCCCTAAAGAT	58	321
11	129	TTCAGGTTGTTTGAATTGTTTTTCAG	GATCTGGCTAAATTGCCATTATTTT	58	255
12	102	AGCATGGAGTCAAGCTGAAAAGA	TGTCAAGCATGAACATCCATTAAA	58	306
13	78	AGATGAAAGGGCCATTTCACATA	TCACCAGTTGTAATTGCTAGGAC	58	261
14	104	AGAACAAATGATGTTGTGGTTTG	AAAGAGACTCCGCTTCTCACC	58	197
15	59	AGCCATGCTGTGTTTGTCTTTG	CTTACCCCTCCATCATAGTCATG	58	218
16	55	TGGGTCCTTGAAACCTCAGTG	TTCCTAATTGGTCAGTAACTGG	58	191
17	68	AGGATTATGCTCTGAGATTCATG	CATTATTATAAAAAGCAGCAGCTTAT	52	172
18	131	GGTGAGGGAAGGAAAGATG	AATGAACCACAAGGTGCCCC	58	245
19	66	TTCCCTTTTTCCTTCTGTAG	AACATGGCTATGGTATGAATTGAGG	52	174
20	105	TGAGCAAAGAGAAAAACCCACCGTT	GGAGCAAACCTCAAGTCCCTGCATCTC	52	219
21	77	TCCTCAGTATAATTGGAGCAG	CTGGTAGAGCCCCTTGGATGG	52	250

Exon	Exon length (bp)	Forward Primer (5'→3')	Reverse Primer (5'→3')	Ta (°C)	PCR product (bp)
22	103	ACAGAACCTGTTGATGTGCAAG	TGGCGGATGAAGGCTCAGTG	62	237

Table S2. Primer sequences used for *FGF23* gene amplification (obtained from Larsson et al. [31])

Exon	Exon length (bp)	Forward Primer (5'→3')	Reverse Primer (5'→3')	Ta (°C)	PCR product (bp)
1	211	AATCTCAGCACCCAGCCACTC	GATGGACAACAAGGGTGCTC	60	294
2	104	TTTCAGGAGGTGCTTGAAGG	TTGCAAAATGGTGACCAACAC	60	208
3	441	CTTCACGTGGTTCGCTCTTG	TGCTGAGGGATGGGTAAAG	60	510

Table S3. Biochemical characteristics of the patients

PATIENT No.	S-Ca (N 2.2–2.7 mmol/L)	S-P (N 4–7 mg/dL)	S-ALP (N 93–309 IU/L)	S-ALP Current (N 93–309 IU/L)	S-PTH (N 15–68.3 pg/mL)	S-25(OH)D3 (N 9.4–59.1; opt. 30–50 ng/mL)	U-Ca (N < 4 mg/kg/d)	U-P (N 16-20 mg/kg/d)	TRP 1% (N>80%)	TRP 2% During therapy (N>80%)
	2.44	↓3.13	↑522	312	21	39.6	0.4	↑23.28	84	↓69
	2.5	↓2.89	↑407	↑260	11.5	22.8	3.6	↓14.08	96	↓71
	2.59	↓2.46	↑730	↑356	30.7	26.4	0.2	↓10.6	84	↓68
	2.62	↓2.57	↑723	279	36.6	37.6	0.85	↓10.9	91	↓72
	2.57	↓2.52	↑1079	↑478	64.2	↑79.1	1.4	↑26.7	80.1	↓71
	2.50	↓3.00	↑482	200	37.6	20.0	0.59	↓13.9	85.4	-
7. (sister of Patient No. 6)	↓1.89	↓2.61	129	174	↓14.2	33.7	1.9	18.9	82	-
8.	2.33	↓2.45	↑488	↑469	47.7	14.5	0.9	↓13.34	80	↓65.2

PATIENT No.	S-Ca (N 2.2–2.7 mmol/L)	S-P (N 4–7 mg/dL)	S-ALP (N 93–309 IU/L)	S-ALP Current (N 93–309 IU/L)	S-PTH (N 15–68.3 pg/mL)	S-25(OH)D3 (N 9.4–59.1; opt. 30–50 ng/mL)	U-Ca (N < 4 mg/kg/d)	U-P (N 16-20 mg/kg/d)	TRP 1% (N>80%)	TRP 2% During therapy (N>80%)
9. (sister of Patient No. 8)	2.33	↓1.79	↑491	–	↑78.5	13.3	0.55	↓11.32	88	-
10.	2.38	↓2.48	↑592	↑321	↑72.5	45.0	1.2	17.98	88	↓72.7
11.	2.32	↓2.22	↑441	171	40.3	35.1	1.3	15.15	85	↓78

S — serum; U— urine; ALP — alkaline phosphatase; PTH — parathormone; TRP — tubular reabsorption of phosphate. The presented laboratory results were obtained at the time of HR diagnosis or during the first stay at the Department of Paediatric Endocrinology and Rheumatology, Poznan University of Medical Sciences. Additionally, the concentrations of ALP and TRP during pharmacologic therapy are given