

Supplementary Table 2. Patients' list; Clinical, biochemical, radiological and genetic data.

Family	Patient	Personal data		Presentation														Response to treatment	Type of Maintenance treatment	Genetic testing		
		Gender	Age of presentation	Clinical		Radiology			Biochemical													
				symptoms	Height z score	Ricket x-ray	DXA scan	Renal US	Before Treatment					After Treatment								
									25(OH)D nmol/L	Ca mmol/l	Po4 mmol/L	ALP U/L	PTH ng/L	25(OH)D nmol/L	Ca mmol/l	Po4 mmol/L	ALP U/L				PTH ng/L	
1	1	male	9	short stature limitation of activity bone pain bone deformity abnormal gait	-2.2	Bowing rachitic roseroy				6	2.1	1.5	399	181	6	2.2	1.5	95.6	53.7	no	Needs active form	Homozygous c.768 ins T
	2	female	6	short stature limitation of activity bone pain bone deformity abnormal gait	-2	rickets				6	2.3	1.6	331	128	7	2.3	1.5	135.7	28	no	Needs active form	Homozygous c.768 ins T
2	1	female	8	short stature limitation of activity bone pain bone deformity	-2.7	osteopenia bowing	mild osteopenia (-1.7)	no nephrocalcinosis		13	2.3	1.2	475	282	82	2.3	1	73	35	yes	50,000 U/2w	Homozygous c.367+1 G>A
	2	female	6	short stature limitation of activity bone pain bone deformity abnormal gait	-2.1	rickets	normal	no nephrocalcinosis		13	2.4	1.9	447	523	81	2.3	1.14	69.5	43.5	yes	50,000 U/w	Heterozygous c.367+1 G>A
3	1	female	5 years	short stature limitation of activity bone pain bone deformity numbness	153 cm	Osteopenia bowing	Osteopenia (-2.1)	no nephrocalcinosis		<5	1.7	1.6	356	312	86	2.2	1.2	55	35	yes	50,000 U/w	Homozygous c.367+1 G>A
	2	female	<10 years	bone pain		normal				34	2.3	1.1	53	46.5	52	2.3	1.3	53	31.9	yes	50,000 U/w	Heterozygous c.367+1 G>A

	3	male	6 years	bone pain	0.1	normal	normal		37	2.3	1.8	311	55	72	2.4	1.3	211	44.7	yes	50,000 U/m	Heterozygous c.367+1 G>A
	4	female	2 years	bone pain	0.4	normal		no nephrocalcinosis	39	2.4	1.5	186	31	99	2.4	1.4	160	22	yes	50,000 U/m	Heterozygous c.367+1 G>A
4	1	male	<10 years	bone pain	0.2	none	normal		14	2.3	1.8	363	81	31	2.2	1.9	336	43	no	Needs active form	Homozygous c.768 ins T
5	1	female	2 years	short stature limitation of activity bone pain bone deformity abnormal gait	129 cm	Osteopenia bowing	Osteopenia (-2.5)	no nephrocalcinosis	18	2	0.9	165	90	105	2.2	1.1	75	86.4	yes	50,000 U/w	Homozygous c.768 ins T
	2	female	<10 years	bone pain limitation of activity		normal			10	2.3	1	81	101	86	2.3	0.9	67	45	yes	50,000 U/m	Heterozygous c.768 ins T
	3	female	<10 years	short stature limitation of activity bone pain	-1.2	normal	normal	no nephrocalcinosis	19	2.3	1.5	380	122	55	2.4	1.5	139.4	30	yes	50,000 U/ 2w	Heterozygous c.768 ins T
6	1	male	<10 years	bone pain		none			21	2.2	1	102	65.8	52	2.2	1.25	109	45	yes	50,000 U/m	Heterozygous c.768 ins T
	2	male	4 years	bone pain	-0.5	normal			14	2.15	1.75	265	91	111	2.35	2	251	50	yes	50,000 U/w	Homozygous c.768 ins T
	3	male	3 years	short stature limitation of activity bone pain	-1.9	bowing		no nephrocalcinosis	6	2.2	1.2	215	45	161	2.3	1.58	168	30	yes	50,000 U/w	Homozygous c.768 ins T
7	1	male	<10 years	Limitation of activity bone pain muscle cramb	176cm	Rickets osteopenia	mild osteopenia (-1)	no nephrocalcinosis	8	2.3	1.4	655	79	47	2.4	1.1	200	36	no	Needs active form	Homozygous c.768 ins T
	2	male	10 years	Limitation of activity bone pain muscle cramb abnormal gait	167cm	Rickets osteopenia	mild osteopenia (-1)	no nephrocalcinosis	<5	1.6	1.4	1752	494	43	2.5	1.2	140	34	no	Needs active form	Homozygous c.768 ins T

	3	male	7 years	bone pain	-1.2	normal	normal		13	2.5	1.5	222	62.5	69	2.4	1.6	186	36	yes	50,000 U/w	Homozygous c.768 ins T
	4	male	5 years	bone pain	-1	normal	normal		25	2.5	1.4	260	44.6	58	2.5	1.4	216	28	yes	50,000 U/ 2w	Homozygous c.768 ins T
	5	male	<10 years	short stature limitation of activity	165cm	rickets			19	2.6	1.7	178		74	2.3	1.3	27		yes	50,000 U/m	Heterozygous c.768 ins T
	6	female	<10 years	Limitation of activity		none			18	2.5	1.4	98	101	67	2.3	1.2	56	68	yes	50,000 U/m	Heterozygous c.768 ins T
8	1	female	6 years	Limitation of activity bone pain bone deformity abnormal gait	-0.66	Rickets osteopenia			16	2.2	1.1	817	154	86	2.2	1.9	81	42	yes	50,000 U/ 2w	Homozygous c.367+1 G>A
9	1	female	<10 years	bone pain	-1.7	none			13	2.2	1.1	645	161	130	2.6	1.5	282	40	yes	50,000 U/ m	Homozygous c.367+1 G>A
	2	female	3 years	Limitation of activity bone pain bone deformity muscle cramps abnormal gait	-0.8	Rickets osteopenia			9	1.8	1	753	339	59	2.3	1.3	120	69	yes	50,000 U/ m	Homozygous c.367+1 G>A
	3	female	5 years	short stature limitation of activity bone pain	-1.7	none			<5	1.5	1.2	488	316	98	2.1	1.7	210	46	yes	50,000 U/ m	Homozygous c.367+1 G>A
	4	female	5 years	short stature limitation of activity bone pain	-1.7	none			<5	1.9	0.85	2963	542	70	2.2	1.4	382	94	yes	50,000 U/ m	Homozygous c.367+1 G>A
	5	female	<10 years	bone pain short stature limitation of activity	148cm	osteopenia			13	2.2	0.55	134	104	49	2.3	1.1	118	80	yes	50,000 U/w	Homozygous c.367+1 G>A