

**Supplement Table 1.** Clinical and molecular findings and pamidronate treatment results of the patients

Patient	Sex	Age (years)	Parental consanguinity	Eye phenotype	Bone phenotype	During pamidronate therapy			Homozygous mutations in <i>LRP 5</i> gene
					Radiological findings	Fracture	Z score (initial)	Z score (final)	
1	F	10.4	First cousin once removed	Bilateral microphthalmia, retinal detachment, intraocular hemorrhage and shallow anterior chamber. Anteriorly convex and atrophic irises, hazy posterior part of the lens and vitreous, non-homogenous increased vitreous density and left corneal leukoma. Orbital USG revealed hyperechogenous findings on right ocular fundus.	Generalized demineralization, increased dorsal kyphosis and lumbar lordosis, mild lumbosacral scoliosis, platyspondyly with biconcave vertebral bodies, thin long and tubular bones and narrow diaphyses.	+	-5.33	-3.0 (after 7 years)	R1002X
2	F	7.7	First cousin	Bilateral microphthalmia, left band keratopathy, left aphakia, right persistent hyperplastic primary vitreous and right cataract.	Generalized demineralization, increased dorsal kyphosis and lumbar lordosis, minimal thoracal scoliosis, severe platyspondyly with biconcave vertebral bodies, thin long and tubular bones, narrow diaphyses, and bilateral acetabular protrusion.	-	-4.19	-1.3 (after 5 years)	V336M
3	M	5.6	First cousin	Bilateral microphthalmia, cataract, band keratopathy, adhesion between iris and lens and calcification of the right globe. Orbital USG also confirmed the calcified rudimentary right globe and non-discriminative intraocular formations.	Generalized demineralization, bones, increased dorsal kyphosis and lumbar lordosis, severe platyspondyly, fractures on vertebrae, thin long and tubular bones, narrow diaphyses and bent left tibia.	+	-6.19	-2.8 (after 3.5 year)	G507S mutation and possible a large deletion in one allele

**Supplement Table 2.** The biochemical parameters of the patients

	Patient 1	Patient 2	Patient 3	Normal values
Calcium	9.7	9.6	9.8	8.4-10.8 mg/dl
Phosphate	4.3	4.6	4.1	2.7-5.5 mg/dl
ALP	52	97	142	60-525 IU/L
25-OHD	28.8	<b>20.5</b>	36.1	20-120 ug/L
PTH	47.8	24.8	25.1	12-72 pg/ml
Fasting glucose	89	86	89	60-105 mg/dl
Fasting insulin	6.3	<b>12.6</b>	6.7	2.6-24.9 $\mu$ U/ml
2 <sup>nd</sup> hour glucose in OGTT	131	<b>156</b>	93	<140 mg/dl
LDL	93	108	66	63-130 mg/dl
HDL	72	53	63	30-75 mg/dl
Total cholesterol	174	182	144	114-200 mg/dl
Triglyceride	61	NA	76	30-135 mg/dl

ALP:alkaline phosphatase, 25OHD: 25-OH vitamin D, PTH:parathormone,  
OGTT:oral glucose tolerance test, LDL: low density lipoprotein, HDL:high density lipoprotein, NA:not available