



Table S1: Comparison of OLS parameters between OI Type I quantitative and qualitative mutations:

OLS Parameters	OI qualitative mutation (n=6)	OI quantitative mutation (n=13)	p-value	test
Cortical bone				
density (number/mm²)	433.44 (58.27)	404.72 (97.27)	0.516	t-test
porosity (%)	1.03 (0.16)	0.81 (0.17)	0.019	t-test
area (μm²)	23.41 [21.13-25.48]	18.67 [17.99-23.65]	0.087	MWU
perimeter (μm)	20.47 [19.99-22.60]	19.63 [18.11-22.32]	0.244	MWU
aspect-ratio	2.86 (0.28)	3.04 (0.45)	0.401	t-test
Trabecular bone				
density (number/mm²)	345.59 (66.74)	375.55 (68.84)	0.393	t-test
porosity (%)	0.74 [0.62-0.83]	0.77 [0.65-0.82]	0.682	MWU
area (μm²)	21.49 (2.47)	21.05 (3.55)	0.789	t-test
perimeter (μm)	19.59 (1.38)	19.67 (1.63)	0.922	t-test
aspect-ratio	2.50 [2.47-2.60]	2.63 [2.55-2.82]	0.102	MWU

Table S2: Comparison of Bone histomorphometric parameters between Controls and OI Type I:

Bone histomorphometry variables	Controls (n=24)	OI type I (n=19)	P value	Difference %	test
Structural parameters					
CORE Width	7.25 (2.03)	3.94 (1.61)	<0.001	-45.66	t-test
Ct.Wi	769.00 [643.2-1081.5]	539.00 [427.0; 765.0]	0.002	-29.91%	MWU
BV/TV	22.14 (4.24)	12.17 (6.28)	<0.001	-45.03%	t-test
Tb.Th	130.91 (24.38)	109.79 (29.42)	0.014	-16.13%	t-test
Tb.N	1.70 (0.21)	1.07 (0.35)	<0.001	-37.06%	t-test
BS/BV	16.75 (3.14)	18.99 (4.91)	0.08	+13.37%	t-test
BS/TV	3.61 (0.45)	2.12 (0.74)	<0.001	-41.27%	t-test
Static parameters of bone formation					
O.Th	6.32 (1.57)	5.13 (1.33)	0.006	-18.83%	MWU
OS/BS	27.81 [21.30; 34.26]	39.87 [27.31; 61.69]	0.004	+43.37%	MWU
OV/BV	2.89 (1.20)	4.40 (2.56)	0.025	+52.25%	t-test
Ob.S/BS	8.15 [5.19; 11.91]	17.76 [13.84; 24.58]	<0.001	+117.91%	MWU
Ob.S/OS	29.19 (14.30)	46.31 (11.84)	<0.001	+58.65%	t-test
Dynamic parameters of bone formation					
MS/BS	13.50 (4.52)	24.39 (8.74)	<0.001	+80.67%	t-test
MS/OS	48.10 (17.90)	58.57 (18.05)	0.077	+21.77%	t-test
MAR	0.95 (0.11)	0.66 (0.12)	<0.001	-30.53%	t-test
Aj.AR	0.45 (0.15)	0.39 (0.12)	0.162	-13.33%	t-test
Mlt	15.14 (4.64)	14.46 (5.36)	0.678	-4.49%	t-test
Omt	6.78 (1.72)	7.82 (1.88)	0.078	+15.34%	t-test
BFR/BS	47.19 (17.74)	59.16 (23.78)	0.082	+25.37%	t-test
BFR/BV	76.04 (36.24)	108.58 (44.66)	0.017	+42.79%	t-test
BFR/ObS	531.28 [409.53; 905.89]	310.53 [240.97; 354.62]	<0.001	-41.55%	MWU
Bone resorption parameters					
ES/BS	15.93 [12.78; 17.98]	18.18 [15.27; 20.25]	0.163	+14.12%	MWU
Oc.S/BS	0.88 [0.58; 1.52]	1.19 [0.98-1.52]	0.123	+35.23%	MWU
N.Oc/B.Pm	0.31 [0.15; 0.41]	0.39 [0.30-0.43]	0.047	+25.81%	MWU