DOI: 10.37190/ABB-02610-2025-02

Balancing Challenges: Exploring Postural Stability in Osteogenesis Imperfecta

Krzysztof Graff[1], Katarzyna Kaczmarczyk[2*], Małgorzata Syczewska[1]

[1]Pediatric Rehabilitation Clinic, Children's Memorial Health Institute, Warsaw, Poland
[2]Józef Piłsudski Academy of Physical Education in Warsaw, Warsaw, Poland
[*]Corresponding author: Katarzyna Kaczmarczyk, Józef Piłsudski Academy of Physical Education in Warsaw, Warsaw, Poland, e-mail address: katarzyna.kaczmarczyk@awf.edu.pl

Submitted: 12th March 2025
Accepted: 17th April 2025

Abstract

Purpose

Abnormalities in bone structure impact motor functions, including the ability to maintain stable posture. This study assessed static and dynamic balance in patients with osteogenesis imperfecta (OI) across different disease types, as compared to a healthy population.

Methods

The study group included 87 patients with OI: Type I (n = 45), Type III (n = 28), and Type IV (n = 14). Balance was assessed using the AMTI (Advanced Mechanical Technology, Inc.) platform. Measurements in standing (ST) position during anterior-posterior (AP) and medial-lateral (ML) tilts, with eyes open, for 30 seconds.

Results

Significant differences in balance parameters under static conditions were found between OI types (61.5% between Types I and III, 38.5% between Types I and IV, and 30.7% between Types III and IV). Across all OI types, maintaining balance predominantly involved displacement of the COP (Centre of Pressure) in the sagittal plane, observed in 84.7% of Type II OI, 75.2% of Type III OI, and 74.5% of Type IV OI cases. Under dynamic conditions, significant differences in balance parameters were noted in 84.6% of comparisons between Types I and III, 46.1% between Types I and IV, and 69.2% between Types III and IV.

Conclusions

Balance assessment in individuals with OI is essential for injury prevention, improving mobility and daily function, and monitoring therapy effectiveness. Development of more preventive strategies aimed at reducing fracture risk and enhancing the quality of life for these patients. The relatively small number of patients with type III and the wide age range represent limitations of our study.

Keywords

balance, osteogenesis imperfecta, pediatric, AMTI

Introduction

Osteogenesis imperfecta (OI) is a rare, genetically inherited disorder of the connective tissue. It is primarily characterized by mutations that impair collagen biosynthesis, leading to abnormal bone mineralization and altered mechanical properties compared to healthy bone tissue [8],[13],[16] The condition occurs in approximately 1 in 15,000–20,000 births [1],[29].

The classic Sillence classification divided OI into four primary types [28], whereas currently, based on patients' clinical symptoms, OI is categorized into one of five types

[1],[20],[32]. Type I OI patients are often ambulatory, with slightly reduced height and a low number of fractures, remaining independent in daily activities. Type II is an extremely severe form, usually associated with perinatal mortality; survivors experience significant deformities in the long bones, chest, and spine, have very limited independence, and require close caregiver support. Type III OI is the most severe, with long bone deformities, spinal curvature, prenatal fractures, and very short stature. Patients experience a high frequency of fractures and mostly require assistive orthopedic devices or a wheelchair for mobility. Type IV ranges from moderate to moderately severe, with mild to significant long bone deformities; patients move independently for short distances but may use a wheelchair for longer distances. Type V is characterized by radial head dislocations and ossification of the interosseous membrane.

These clinical symptoms, along with frequent fractures and recurrent immobilization periods, often lead OI patients to avoid physical activity. Bone structure abnormalities and weakened musculoskeletal systems affect motor functions, including the ability to maintain stable posture [3],[8].

Balance assessment helps evaluate the degree of functional impairments that can impact daily functioning. Conventional biomechanical assessments of postural stability predominantly rely on linear mathematical models. Commonly employed center of pressure (COP) displacement metrics, derived from force platform measurements, serve as fundamental indicators for quantifying postural stability. Several studies have implemented standardized, reliable and valid test of balance impairements involving quiet standing tasks, with participants performing two-legged or single-leg stances, both with eyes open and closed [14],[23],[6]. However, static balance assessments fail to capture the intricate complexity of postural control mechanisms. Dynamic assessment conducted on unstable (movable) platforms offers insights into the biomechanics and neuromuscular control strategies involved in maintaining equilibrium [15]. Nevertheless, despite the utility of both static and dynamic balance assessments, they do not fully characterize the behavior of human physiological systems, which are inherently nonlinear. Consequently, nonlinear analytical approaches have gained increasing prominence in contemporary research [30],[36].

Adolescents with Type I OI, for example, show poorer postural balance as compared to healthy peers [5], [25]. Studies have also found significantly lower muscle mass and strength in individuals with Type III OI as compared to Types I and IV, as well as age-matched references [5],[34]. Early identification of balance issues allows for interventions to strengthen posture-stabilizing muscles, apply suitable orthopedic devices, and reduce fall risks and fracture rates in children and adolescents with OI. Improved balance can also enhance a sense of security, increase independence, and improve daily functioning. However, there is a notable lack of

comprehensive literature assessing balance across different OI types, particularly in pediatric patients.

The aim of the study, therefore, was to evaluate the static and dynamic balance in patients with OI across different types of the disease in comparison to a healthy population.

Materials and methods

The study group included 87 patients diagnosed with OI: Type I (*n*=45), Type III (*n*=28), and Type IV (*n*=14), aged between 5 and 62 years. No patients with Type II and V OI were included. A summary of patient characteristics is presented in Table 1.

The inclusion criteria were consented to participate, the ability to stand independently, and no fractures in the previous six months. The exclusion criteria were refusal to participate due to fracture risk, inability to stand independently, recent lower limb fractures (within the past six months), or incomplete bone healing after a fracture.

The adult participants with OI were parents of the children in the study, whereas the pediatric patients were recruited from the Pediatric Rehabilitation Clinic of the Children's Memorial Health Institute in Warsaw, Poland, between 2000 and 2022. Over the study period, 288 balance measurements were conducted, with intervals of at least one year. Ethical approval was granted by the IP-CZD Bioethics Committee (Approval No. W4/2017, Consent 35/KBE/2017). The patients' medical records were reviewed to determine the total number of fractures and fractures involving the lower limbs.

Balance was assessed using the AMTI AcuSway platform, which samples at 100 Hz. Measurements included static balance in standing position (ST) and dynamic balance while performing anterior-posterior (AP) and medial-lateral (ML) tilts, performed with eyes open for 30 seconds. Patients stood barefoot in a comfortable position, with their feet hip-width apart, arms relaxed at their sides, and gaze fixed on a point at eye level.

The analysis focused on the following parameters: maximum and minimum displacements along the anterior-posterior axis (*AP uXmax*, *AP uXmin*) and the medial-lateral axis (*AP uYmax*, *AP uYmin*), total displacements (*AP uSumX*, *AP uSumY*), symmetry in anterior-posterior movements (*AP uSymX*), the total path length of the Center of Pressure (COP) (*AP uPath Length*), average COP velocity (*AP uVAvg*), and the 95th percentile ellipse area of COP displacements (*AP uArea95*). The percentage displacement of COP in the sagittal and frontal planes was calculated using the following formula proposed by Mraz [18]:

$$WCOP = 100 \times \frac{AP\ length - LR\ length}{AP\ length + LR\ length}$$

Where:

- *WCOP* = COP displacement in %;
- *AP length* = stabilogram length in the sagittal plane;
- *LR length* = stabilogram length in the frontal plane.

WCOP indices were interpreted as follows:

- WCOP = 0: no dominant displacement in either plane;
- WCOP > 0: dominance in the sagittal plane;
- WCOP < 0: dominance in the frontal plane.

Pediatric results were compared with reference data from healthy children [10], while adult results were compared with data obtained from healthy adults (parents, caregivers, or older siblings of OI patients). The subjects from the control group were asked to fill the questionnaire regarding their health and functional status. This questionnaire was prepared by the experienced physiotherapist (one of the authors) to exclude subjects with potential health biases The questionnaire asked about BMI, neurological and orthopaedic diseases, injuries during last 5 years, medications taken for chronic diseases, which could compromise balance abilities, surgeries in the last 2 years.

Statistical analysis was performed using Statistica 13. The Shapiro-Wilk test was used to assess the normality of balance parameter distributions. Differences in balance parameters between genders within OI types were analyzed using the Mann-Whitney U test. Static and dynamic balance parameters were compared across OI types using the Kruskal-Wallis ANOVA test, adopting a significance level of $\alpha = 0.05$.

Table 1. Characteristics of the Study Group with OI

OI Type	Sex	Age	Body weight	Body height	Fractures of lower limbs
			Median min/max	Median min/max	Median min/max
Type I	Girls	14.46 (4.8–54.2)	43.5 (11–78)	152.4 (100–180)	5 (0–11)
	Boys	14.1 (5.2–62.9)	48 (14–92)	158.5 (101–176)	3 (0–16)
Type III	Girls	16.8 (4.9–49.2)	26 (9–56)	108.3 (73.7–150)	19 (1–28)
	Boys	15.4 (4.8–54.3)	35 (9.6–85.0)	117 (77.4–150)	17 (2–29)
Type IV	Girls	15.4 (8.8–41.8)	43.5 (27–53)	130 (114–158)	9 (2–15)
	Boys	15.3 (7.0–23.9)	47 (18–93)	137.5 (105–153)	6 (4–7)

Results

The total number of fractures was highest in patients with Type III OI (median: 28 fractures), followed by Type IV OI (median: 13 fractures) and Type I OI (median: 7 fractures). Significant differences in the average number of fractures were observed between Types I and III OI, and Types I and IV OI ($p < 0.001$), as well as between Types III and IV OI ($p < 0.01$). The number of lower limb fractures was highest in Types III and IV OI (median: 8 fractures) and lower in Type I OI (median: 4 fractures). Significant differences were found between Types I and III OI and between Types I and IV OI ($p<0.001$). Detailed results of the fracture analysis are presented in Table 2.

Table 2. Fractures across different OI types

Fractures	Type I OI	Type III OI	Type IV OI
	x±SD min÷max	x±SD min÷max	x±SD min÷max
Average number of fractures per patient	11.34±9.46*** 1÷33	23.6±13.1** 3÷50	14.65±7.79 5÷41
Total number of all fractures	1611	2209	878
Average number of lower limb fractures per patient	5.09± 4.69$^{###}$ 0÷16	11.4±10.5 2÷45	9.32±6.14 1÷25
Total number of all lower limb fractures	726	1167	622

***type I-III $p < 0.001$, ***type I-IV $p < 0.001$, **type III-IV $p < 0.01$
$^{###}$ type I-III $p < 0.001$, $^{###}$type I-IV $p < 0.001$

Analysis of normalized static balance parameters revealed statistically significant differences in 61.5% of the parameters assessed, including *ST uXmax*, *ST uXmin*, *ST uSumX*, *ST uWCOP*, *ST uArea Circ*, *ST uPath Length*, *ST uVAvg*, and *ST uArea95*. Significant differences were observed in 61.5% of parameters between OI Types I and III, 38.5% between OI Types I and IV, and 30.7% between OI Types III and IV.

Of the statistically significant balance parameters, 87.5% of patients with Type III OI and Type IV showed the highest COP displacement values in static conditions, whereas patients with Type I OI displayed values closest to normal. The WCOP index for maintaining static balance showed a different pattern. In all OI groups, COP displacement predominantly occurred in the sagittal plane, affecting 84.7% of patients with Type I OI, 75.2% with Type III OI, and 74.5% with Type IV OI. Displacement dominance in the frontal plane was observed in 11.9% of Type I OI patients, 22.6% of Type III OI patients, and 20% of Type IV OI patients.

Detailed statistical analysis of normalized balance parameters in static conditions (ST) is presented in Table 3.

Table 3. Statistical analysis of normalized static balance parameters in patients with OI (ST Standing)

OI Type	*n*	Med	Min	Max	Lower quartile	Upper quartile
	ST uXmax					
type I$^{***, ###}$	149	110.31	35.05	481.08	82.47	150.46
type III	95	181.65	47.95	608.11	126.32	240.0
type IV	54	143.88	41.89	522.11	101.35	225.26
	ST uXmin					
type I$^{***,###}$	149	114.55	25.45	377.78	83.33	160.63
type III$^{\wedge}$	95	188.41	65.28	1058.8	151.18	252.94
type IV	54	163.07	45.67	443.14	102.36	229.41
	ST uYmax					
type I	149	129.14	46.32	396.57	103.53	177.65
type III	95	147.47	61.62	422.22	112	182.86
type IV	54	146.3	52.32	480.25	114.81	183.94
	ST uYmin					
type I	149	133.33	49.06	519.5	100.94	188.68
type III	95	157.55	54.22	537.78	116.98	212.05
type IV	55	146.23	58.49	441.51	117.78	193.71
	ST uSymX					
type I	149	97.27	26.29	306.75	76.87	123.45
type III	95	91.64	34.98	297.59	76.52	126.45
type IV	55	99.44	39.42	273.76	75.93	130.12
	ST uSymY					
type I	149	102.56	47.64	253.02	83.02	127.53
type III	95	99.3	37.45	479.13	76.75	120.02
type IV	55	94.6	46.73	223.75	78.58	133.17
	ST uSumX					
type I$^{***,###}$	149	95.24	17.01	295.69	61.17	126.52
type III	95	177.78	30.04	775.25	112.88	239.6
type IV	54	119.28	43.96	375.25	86.36	233.33
	ST uSumY					
type I	149	126.42	55.97	447.83	96.89	167.25
type III	95	150.31	65.63	456.14	115.2	191.71
type IV	54	143.99	55.97	335.67	114.91	187.5
	ST uWCOP					
type I$^{***,###}$	149	137.14	-191.6	486.66	58.0	203.44
type III	95	44.57	-240.8	283.96	-5.05	93.88
type IV	55	77.6	-90.38	381.32	5.93	148.85
	ST uAreaCirc					
type I$^{***,###}$	149	119.05	28.13	544.69	88.02	167.8
type III$^{\wedge}$	92	248.53	60.11	968.23	168.75	319.96
type IV	53	179.57	62.87	565.64	115.02	241.67
	ST uPathLength					
type I$^{***,###}$	149	119.27	18.14	385.66	87.92	150.5
type III$^{\wedge\wedge\wedge}$	94	249.69	57.66	637.27	177.7	302.12
type IV	55	143.56	44.66	372.62	94.25	191.19

	ST uVAvg					
type I***	149	101.87	18.13	244.14	60.54	141.01
type III^^^	94	213.31	57.7	634.23	155.06	293.31
type IV	55	125.75	44.75	371.17	67.56	154.68
	ST uArea95					
type I***	140	148.42	19.94	1462.8	90.19	268.88
type III	89	226.02	54.13	1093.6	146.75	356.69
type IV	47	179.58	45.67	939.02	130.06	388.89

***difference between type I-III OI; $p < 0.001$, ### difference between type I-IV OI; $p < 0.001$, ^ difference between type III-IV OI; $p < 0.05$

Dynamic balance analysis during anterior-posterior (AP) tilts revealed statistically significant differences in 85% of the measured parameters, including *AP uXmax*, *AP uXmin*, *AP uYmax*, *AP uMin*, *AP uSymX*, *AP uSumY*, *AP uWCOP*, *AP uAreaCirc*, *AP uPathLength*, *AP uVAvg*, and *AP uArea95*. Significant differences were observed in 84.6% of parameters between Type I OI and Type III OI, in 46.1% of parameters between Type I and Type IV OI, and in 69.2% of parameters between Type III and Type IV OI. Among the measured parameters, patients with Type III OI demonstrated significantly lower balance parameter values, indicating reduced anterior-posterior tilt dynamics. Patients with Type I OI, followed by those with Type IV OI, showed the least deviation from normal values. A detailed statistical analysis is presented in Table 4.

Table 4. Statistical analysis of normalized dynamic balance parameters in patients with OI while performing anterior-posterior (AP) tilts

OI Type	***n***	**Med**	**Min**	**Max**	**Lower quartile**	**Upper quartile**
	AP uXmax					
type I**	146	108.63	31.7	230.15	79.09	130.84
type III^^^	90	89.46	22.75	269.67	66.39	111.6
type IV	51	111.37	49.06	282.9	86.0	150.11
	AP uXmin					
type I**	146	103.14	35.26	250.57	85.97	130.84
type III^^^	90	88.23	25.69	190.95	68.01	115.95
type IV	53	114.36	69.68	355.31	91.68	135.01
	AP uYmax					
type I***, ###	146	88.33	23.81	120.36	78.38	101.61
type III^^^	90	40.35	11.6	98.63	30.14	49.84
type IV	53	68.82	23.54	99.37	55.57	83.42
	AP uYmin					
type I***,###	146	90.04	45.33	123.42	78.62	100.96
type III^^^	90	43.74	12.03	96.47	30.73	55.14
type IV	53	69.58	43.33	101.33	60.29	78.99
	AP uSymX					
type I	146	102.62	27.79	418.57	82.87	123.63
type III	90	100.71	31.48	328.57	82.96	125.71

type IV	53	102.89	31.81	169.75	84.89	119.29
	AP uSymY					
type I	146	101.23	44.48	149.32	89.76	110.57
type III	90	94.25	37.97	153.02	74.6	114.8
type IV	53	100.54	30.54	129.24	89.29	109.63
	AP uSumX					
type I***	146	103.87	52.8	199.04	86.51	123.05
type III$^{\wedge\wedge\wedge}$	90	88.58	23.55	232.32	68.39	110.52
type IV	53	109.05	64.41	313.69	87.21	135.83
	AP uSumY					
type I$^{***,###}$	146	88.85	34.24	110.78	78.97	98.93
type III$^{\wedge\wedge\wedge}$	90	43.63	13.99	92.47	3264	52.98
type IV	53	67.07	39.38	100.48	58.55	79.74
	AP uWCOP					
type I$^{***,###}$	146	90.39	-1.02	136.79	81.95	101.25
type III$^{\wedge\wedge\wedge}$	90	54.71	-1.32	94.34	36.84	68.43
type IV	53	70.14	5.89	104.94	57.41	85.82
	AP uAreaCirc					
type I***	146	84.5	29.46	179.63	66.9	104.05
type III$^{\wedge\wedge\wedge}$	90	39.06	4.51	125.51	26.92	54.93
type IV	53	72.43	33.73	230.47	51.93	94.54
	AP uPathLength					
type I$^{***,\#}$	146	91.53	41.69	238.72	76.26	109.59
type III	90	71.76	9.55	151.68	59.95	95.18
type IV	51	81.18	47.88	140.59	65.62	99.8
	AP uVAvg					
type I***	146	82.16	33.89	135.71	66.53	101.91
type III	90	63.64	35.77	155.53	51.84	82.05
type IV	53	70.57	23.47	144.7	57.71	91.61
	ST uArea95					
type I$^{***,\#\#}$	136	86.91	26.77	180.15	62.14	107.78
type III$^{\wedge\wedge\wedge}$	85	25.85	1.53	81.64	14.36	38.98
type IV	45	65.71	2.68	230.69	44.89	74.36

***difference between type I-III OI; $p < 0.001$, **difference between type I-III OI; $p < 0.01$, $^{###}$ difference between type I-IV OI; $p < 0.001$, $^{\#\#}$ difference between type I-IV OI; $p < 0.01$, , $^{\#}$difference between type I-IV OI; $p < 0.05$, $\wedge\wedge\wedge$ difference between type III-IV OI; $p < 0.001$, $\wedge$ difference between type III-IV OI; $p < 0.05$

The analysis of dynamic balance during medial-lateral (ML) tilts revealed statistically significant differences across OI types for 92% of the measured parameters. These parameters included *ML uXmax*, *ML uXmin*, *ML uYmax*, *ML uYmin*, *ML uSymX*, *ML uSumX*, *ML uSumY*, *ML uWCOP*, *ML uAreaCirc*, *ML uPathLength*, *ML uVAvg*, and *ML uArea95*. Significant differences in balance parameters were observed in 84.6% of parameters between Type I and Type III OI, 53.8% of parameters between Type I and Type IV OI, and 84.6% of parameters between Type III and Type IV OI. A detailed statistical analysis is presented in Table 5.

Table 5. Statistical analysis of normalized dynamic balance parameters in patients with OI while performing medial-lateral (ML) tilts

OI Type	*n*	Med	Min	Max	Lower quartile	Upper quartile
ML uXmax						
type I*** ###	144	88.77	48.44	125.48	79.20	99.92
type III^^^	90	40.0	6.58	84.38	26.39	50.35
type IV	51	71.14	29.7	109.95	57.27	82.14
ML uXmin						
type I*###	144	90.32	44.36	121.06	80.86	99.28
type III^^^	90	40.39	10.57	91.7	30.26	56.62
type IV	51	67.96	33.93	97.14	54.94	76.52
ML uYmax						
type I***	144	91.9	41.08	172.66	74.66	114.56
type III^^^	90	68.0	29.29	150.31	53.35	87.5
type IV	51	83.55	42.61	213.42	67.83	108.03
ML uYmin						
type I***	144	99.04	28.91	277.89	74.04	121.26
type III^^^	89	73.55	19.17	175.98	54.58	94.56
type IV	47	98.93	36.53	201.07	66.3	135.46
ML uSymX						
type I	144	100.74	79.71	136.99	94.78	107.16
type III^	90	97.16	38.36	187.86	84.8	106.96
type IV	51	103.01	74.61	143.35	96.8	114.5
ML uSymY						
type I	144	95.29	30.74	175.25	77.12	114.32
type III	90	96.5	25.53	189.76	76.41	120.25
type IV	51	91.69	39.05	193.74	73.82	110.03
ML uSumaX						
type I***###	144	90.32	46.17	123.71	82.45	100.36
type III^^^	90	40.24	8.54	86.23	29.33	52.19
type IV	51	69.84	31.85	101.75	58.22	78.48
ML uSumaY						
type I***	144	99.15	42.95	174.74	84.45	116.23
type III^^^	90	71.82	25.6	147.61	57.21	86.77
type IV	51	95.01	46.38	199.47	77.31	131.95
ML uWCOP						
type I***	144	-91.68	-122.54	197.34	-102.43	-67.47
type III^	90	-25.14	-105.13	95.01	-62.52	62.09
type IV	51	-74.89	-111.29	105.98	-89.38	43.61
ML uAreaCirc						
type I***###	144	83.92	26.45	191.18	68.15	105.61
type III^^^	90	27.73	4.08	90.92	17.44	41.87
type IV	51	63.07	16.64	119.12	44.51	73.76
ML uPathLength						
type I***###	144	92.78	46.55	171.08	75.99	109.73
type III^^	90	58.02	35.73	121.56	47.74	80.02
type IV	51	77.12	32.93	127.45	60.03	95.86
ML uVAvg						

type I***##	144	82.37	32.73	153.27	68.94	102.19
type III	90	53.17	27.87	123.0	42.34	70.09
type IV	51	66.6	26.78	119.93	51.65	89.06
ML uArea95						
type I***###	135	88.9	25.11	224.06	64.92	116.47
type III^^^	85	19.14	1.90	94.75	10.11	34.74
type IV	42	58.8	11.94	149.47	37.95	79.92

***difference between type I-III OI; $p < 0.001$, *difference between type I-III OI; $p < 0.05$, ### difference between type I-IV OI; $p < 0.001$, ## difference between type I-IV OI; $p < 0.01$, ^^^ difference between type III-IV OI; $p < 0.001$, ^^ difference between type III-IV OI; $p < 0.01$, ^ difference between type III-IV OI; $p < 0.05$

Discussion

~~There is no data about the balance problems in patients with OI, therefore it is not possible to discuss our results against other studies.~~ There is limited data about the balance problems in patients with OI, they mainly concern the type I of OI [11], therefore the possibility to compare the results from this study with others is meagre. Our assessment of balance in patients with OI revealed variations across the different types of the condition. In Type III OI patients, a large amplitude of sway was observed under static conditions, but sway amplitudes were relatively smaller in dynamic evaluations. To maintain balance during static standing, patients with Type III OI performed rapid compensatory movements involving their lower limbs, torso, and upper limbs to counterbalance their functional deficits. This resulted in the center of pressure (COP) path length and velocity being twice as high compared to Type I OI patients and 1.7 times higher compared to Type IV OI patients.

In dynamic balance tests, such as anterior-posterior (AP) and medial-lateral (ML) tilts, patients with Type III OI exhibited significantly smaller ranges of sway. For AP tilts, the COP path length and velocity were 1.3 times lower than in Type I OI and 1.1 times lower than in Type IV OI. For ML tilts, the COP path length and velocity were 1.6 times lower than in Type I OI and 1.3 times lower than in Type IV OI. The greatest challenges for Type III OI patients were maintaining balance during free-standing (ST), followed by ML tilts, with the least difficulty observed in AP tilts. This suggests poorer muscular stabilization, asymmetrical limb deformities, and weakened anterior, posterior, and lateral muscular chains, which are essential for maintaining posture during tilting movements.

Compensatory factors related to the defective type I collagen in osteogenesis imperfecta (OI) likely affect proprioceptive sensory information [25]. Mechanoreceptors, located in the muscles, tendons, ligaments, and skin, are primarily composed of type I collagen and play a crucial role in the sensory inputs necessary for postural balance [19].

Previous studies have shown that both symmetrical and asymmetrical joint contractures disrupt overall body balance [24]. Other factors, such as the innate predisposition of OI patients

to low muscle mass [35], weakened lower limb muscle strength [33], and periods of enforced immobility [21], have been linked to selective atrophy of antigravity muscles, including the soleus, back extensors, and quadriceps. While most pathophysiological effects of immobility improve after mobilization, skeletal muscle recovery is slower. These combined factors – muscular weakness, limb axis abnormalities, osteopenia, and sarcopenia – further impair motor function and balance [12]. Muscle atrophy, in cases of critical illness or chronic conditions like OI, results in what is termed *acquired weakness* [21].

Deficits in lower leg muscle mass [13] and weakened foot extensors [3] have been shown to significantly impair balance control. Additionally, pQCT studies suggest that reduced bone and muscle mass in Type III OI patients contribute to balance impairments [34]. Conversely, balance parameters in patients with Type I OI, and to a lesser extent Type IV OI, were closest to those of healthy individuals. This conforms with findings by Pouliot-Laforte et al. [25], who observed only slightly poorer postural balance in children and adolescents with Type I OI compared to healthy peers. In older patients, vestibular changes, dizziness, or compensatory head movements to maintain balance may also influence balance performance [16].

There is a common perception that physical activity and exercise should be limited in patients with OI due to bone fragility and the associated risk of fractures. Children and young adults with OI often experience a repetitive cycle of fractures, immobility, and muscle deconditioning, which leads to functional limitations [2]. However, the ability to maintain balance during standing or walking is not a static function in OI patients. Some individuals lose mobility and transition to wheelchair use, while others regain mobility and begin to walk. Understanding disease-specific symptoms and their impact on balance is critical for setting short- and long-term rehabilitation goals.

Although many children with OI would stand to benefit from breaking the repetitive cycle of fracture, immobilization, demineralization, and fracture through physical therapy [31], improving balance and reducing fall risk requires therapies that enhance the mechanical and metabolic properties of bone. Pharmacological treatments, such as pamidronate and related drugs, are now standard for OI management, reducing pain, improving growth, and enhancing functional mobility [4],[7],[22]. Surgical interventions [27] play a key role in correcting limb deformities, strengthening bones, and enabling weight-bearing and balance maintenance [26]. Gradual improvements in muscle strength, pelvic stability, and upper and lower limb control can significantly enhance postural balance.

Conclusions

This study has provided a comprehensive assessment of static and dynamic balance in patients

with osteogenesis imperfecta (OI), highlighting significant differences across OI types. Patients with Type III OI exhibit the most profound balance deficits, characterized by high sway amplitude during static standing and reduced stability during dynamic tilts. These challenges are compounded by muscular weakness, limb deformities, and frequent fractures, which impair functional mobility and increase fall risk. In contrast, patients with Type I OI demonstrate balance parameters closest to healthy individuals, followed by those with Type IV OI, reflecting milder impairments.

The findings underscore the critical need for tailored interventions that address the unique challenges associated with each OI type. Rehabilitation strategies should focus on strengthening postural muscles, correcting limb deformities, and improving proprioception to enhance balance and reduce the risk of falls and fractures. Pharmacological treatments and surgical interventions to correct deformities can further support functional improvements. Additionally, promoting safe physical activity can mitigate the cycle of immobility and deconditioning that exacerbates balance deficits in OI patients, especially children.

This study has also emphasized the importance of balance assessments as a tool for monitoring therapeutic progress and guiding individualized treatment plans. Future research should explore the integration of advanced balance training programs and the long-term impact of emerging treatments on functional outcomes in OI. By addressing the specific needs of each patient, clinicians can enhance mobility, independence, and quality of life for individuals with OI. We acknowledge that the relatively small number of patients, particulary type III and the wide age range represent limitations of our study.

Acknowledgements

The data necessary to reproduce the analyses presented here are not publicly accessible. Data are available from the first author upon reasonable request.

Relevant conflicts of interest/financial disclosures

The authors received no specific funding for this study and have no conflict of interest to declare concerning this work.

Author Roles:

(1) Research project: A. Conception, B. Organization, C. Execution;

(2) Statistical Analysis: A. Design, B. Execution, C. Review and Critique;

(3) Manuscript: A. Writing of the first draft, B. Review and Critique.

KG - 1A, 1B, 1C, 2B, 3A, 3B

KK – 1C, 2C, 3B

MS - 1A, 2A, 2C, 3B

References:

1. BOTOR M., FUS-KUJAWA A., UROCZYNSKA M., AT AL. Osteogenesis Imperfecta: Current and Prospective Therapies, Biomolecules, 2021, 11(10):1493, DOI: 10.3390/biom11101493.
2. CARVALHO P.A.F., REGIS T.S., FAIÇAL A.V.B., et al. Functional status of individuals with osteogenesis imperfecta: data from a reference center, J Pediatr (Rio J), 2023, 99(1): 94-98, DOI: 10.1016/j.jped.2022.07.002
3. CAUDILL A., FLANAGAN A., HASSANI S., ET AL. Ankle Strength and Functional Limitations in Children and Adolescents with Type I Osteogenesis Imperfecta, Pediatric Physical Therapy, 2010, 22(3): 288-295, DOI: 10.1097/PEP.0b013e3181ea8b8d..
4. CHO T.J., KO J.M., KIM H., ET AL. Management of Osteogenesis Imperfecta: A Multidisciplinary Comprehensive Approach, Clin Orthop Surg, 2020, 12(4): 417-429, DOI: 10.4055/cios20060.
5. COÊLHO G., COCATO L., CASTRO L.C., ET AL. Postural balance, handgrip strength and mobility in Brazilian children and adolescents with osteogenesis imperfecta, Jornal de Pediatria, 2021, 97(30): 315-320, DOI: 10.1016/j.jped.2020.05.003.
6. DA SILVA R.D., BILODEAU M., PARREIRA R., TEIXEIRA D., AMORIM C. Age-related differences in time-limit performance and force platform-based balance measures during one-leg stance. *Journal of Electromyography & Kinesiology*, 2013, 23(3): 634–639. DOI: 10.1016/j.jelekin.2012.11.011
7. DWAN K., PHILLIPI C.A., STEINER R.D., ET AL. Bisphosphonate therapy for osteogenesis imperfecta, Cochrane Database of Systematic Reviews, 2016, 10: CD005088. DOI: 10.1002/14651858.CD005088.pub4.
8. ENGELBERT R.H., UITERWAAL C.S., GERVER W.J., ET AL. Osteogenesis imperfecta in childhood: impairment and disability. A prospective study with 4-year follow-up, Archives of Physical Medicine and Rehabilitation, 2004, 85(5): 772-778.
9. FORLINO A. AND MARINI J.C., Osteogenesis imperfecta, Lancet, 2016, 16: 1657-71. DOI: 10.1016/S0140-6736(15)00728-X.
10. GRAFF K., SZCZERBIK E., KALINOWSKA M., ET AL. Balance assessment in healthy children and adolescents aged 6-18 years based on six tests collected on AMTI AccuSway force platform, Acta of Bioengineering and Biomechanics, 2020, 22(2): 121-130.

11. GRAFF K., KALINOWSKA M., SZCZERBIK E., SYCZEWSKA M. Changes in balance parameters in group of osteogenesis imperfecta. *Gait & Posture*, 2018, 65(Suppl 1): 351–352. https://doi.org/10.1016/j.gaitpost.2018.07
12. GREMMINGER V.L. AND PHILLIPS C.L., Impact of Intrinsic Muscle Weakness on Muscle-Bone Crosstalk in Osteogenesis Imperfecta, Int J Mol Sci, 2021, 22(9): 4963. DOI: 10.3390/ijms22094963.
13. HARIRI C., MCKECHNIE J., MASON A., ET AL. Impairment of muscle mass and muscle function in osteogenesis imperfecta: A systematic review, Endocrine Abstracts, 2023, 12, | DOI: 10.1530/endoabs.95.P12
14. HARRO C., MARQUIS A., PIPER N., BURDIS C. Reliability and validity of force platform measures of balance impairment in individuals with Parkinson disease, *Phys Ther*, 2016, 96(10): 1540–1550. DOI: 10.2522/ptj.20150301
15. HARRO C., GARASCIA C. Reliability and validity of computerized force platform measures of balance function in healthy older adults, *J Geriatr Phys Ther*, 2019, 42(1): 20–26. DOI: 10.1519/JPT.0000000000000131
16. KUURILA K., KENTALA E., KARJALAINEN S., ET AL. Vestibular dysfunction in adult patients with osteogenesis imperfecta, Am. J. Med. Genet, 2023, 120A: 350-358. DOI: 10.1002/ajmg.a.20088.
17. MARINI J.C., REICH A. AND SMITH S.M., Osteogenesis imperfecta due to mutations in non-collagenous genes: lessons in the biology of bone formation, Curr Opin Pediatr, 2014, 26(4): 500-507.
18. MRAZ M., CURZYTEK M., MRAZ M.A., ET AL. Body balance in patients with systemic vertigo after rehabilitation exercise, J Physiol Pharmacol, 2007, 58(5): 427-36.
19. NARDONE A. AND TURCATO A.M., An overview of the physiology and pathophysiology of postural control, Biosyst Biorobotics, 2018, 19: 3-28.
20. NIJHUIS W., VERHOEF M., VAN BERGEN C., ET AL. Fractures in Osteogenesis Imperfecta: Pathogenesis, Treatment, Rehabilitation and Prevention, Children, 2022, 9(2): 268. DOI: 10.3390/children9020268.
21. PARRY S.M. AND PUTHUCHEARY Z.A., The impact of extended bed rest on the musculoskeletal system in the critical care environment, Extrem Physiol Med, 2015, 9(4): 16. DOI: 10.1186/s13728-015-0036-7.
22. PINHEIRO B., ZAMBRANO M.B., VANZ A.P., ET AL. Cyclic pamidronate treatment for osteogenesis imperfecta: Report from a Brazilian reference center, Genet Mol Biol, 2019, 42(1 suppl 1): 252-260. DOI: 10.1590/1678-4685-GMB-2018-0097.

23. PONCE-GONZÁLEZ J., SANCHIS-MOYSI J., GONZÁLEZ-HENRÍQUEZ J.J., ARTEAGA-ORTIZ R., CALBET J., DORADO C. A reliable unipedal stance test for the assessment of balance using a force platform, *J Sports Med Phys Fitness*, 2014, 54(1): 108–117. PMID: 24346036
24. POTTER P.J., KIRBY R.L., MACLEOD D.A., The effects of simulated knee-flexion contractures on standing balance, Am J Phys Med Rehabil, 1990, 69(3): 144-7. DOI: 10.1097/00002060-199006000-00009.
25. POULIOT-LAFORTE A, LEMAY M., RAUCH F.., ET AL. Static Postural Control in Youth With Osteogenesis Imperfecta Type I, Archives of Physical Medicine and Rehabilitation, 2017, 98(10): 1948-1954.
26. RALSTON S.H. AND GASTON M.S. Management of Osteogenesis Imperfecta, Front. Endocrinol, 2020, 10: 924. DOI: 10.3389/fendo.2019.00924.
27. ROSEMBERG D.L., GOIANO E.O., AKKARI M., ET AL. Effects of a telescopic intramedullary rod for treating patients with osteogenesis imperfecta of the femur, J Child Orthop, 2018, 12(1): 97-103. DOI: 10.1302/1863-2548.12.170009.
28. SILLENCE D., Osteogenesis imperfecta: an expanding panorama of variants, Clin Orthop, 1981, 159: 11-25.
29. SUBRAMANIAN S., ANASTASOPOULOU C. AND VISWANATHAN V.K., Osteogenesis Imperfecta. In: StatPearls [Internet]. Treasure Island (FL): StatPearls Publishing, 2024. Available from: https://www.ncbi.nlm.nih.gov/books/NBK536957/
30. STIRLING J.R., ZAKYNTHINAKI M.S. Stability and the maintenance of balance following a perturbation from quiet stance, *Chaos*, 2004, 14(4): 1012–1021. DOI: 10.1063/1.1804972
31. VAN BRUSSEL M., TAKKEN T., CUNO S.P.M., ET AL. Physical Training in Children with Osteogenesis Imperfecta, The Journal of Pediatrics, 2008, 152(1): 111-116. DOI: 10.1016/j.jpeds.2007.06.029.
32. VAN DIJK F.S. AND SILLENCE D.O., Osteogenesis imperfecta: clinical diagnosis, nomenclature and severity assessment, American Journal of Medical Genetics, 2014, 164A(6): 1470-1481. DOI: 10.1002/ajmg.a.36545.
33. VEILLEUX L.N., LEMAY M., POULIOT-LAFORTE A., ET AL. Muscle Anatomy and Dynamic Muscle Function in Osteogenesis Imperfecta Type I, The Journal of Clinical Endocrinology & Metabolism, 2014, 99(2): E356-E362, DOI:10.1210/jc.2013-3209.
34. VEILLEUX L.N., TREJO P. AND RAUCH F., Muscle abnormalities in osteogenesis imperfecta, Journal of musculoskeletal & neuronal interactions, 2017, 17(2): 1.

35. YUAN Y., XU Y.F., FENG C., ET AL. Low muscle density in children with osteogenesis imperfecta using opportunistic low-dose chest CT: a case-control study, BMC Musculoskelet Disord, 2024, (25): 478, DOI: 10.1186/s12891-024-07596-7.
36. ZAKYNTHINAKI M.S., MADERA MILLA J., LÓPEZ DIAZ DE DURANA A., CORDENTE MARTÍNEZ C.A., RODRÍGUEZ ROMO G., SILLERO QUINTANA M., SAMPEDRO MOLINUEVO J. Rotated balance in humans due to repetitive rotational movement, *Chaos*, 2010, 20(1): 013121. DOI: 10.1063/1.3308616