

Osteogenesis Imperfecta in Childhood: Impairment and Disability

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ABSTRACT. *Objective.* To determine clinical characteristics in children with osteogenesis imperfecta (OI) regarding impairment (range of joint motion and muscle strength) and disability (functional skills) in relation to the different types of the disease, and to study the correlation between characteristics of impairment and disability.

Methods. In a cross-sectional study 54 children with OI (OI type I: 24; OI type III: 15; OI type IV: 15), the range of joint motion, muscle strength, and functional ability were measured in a standardized way and analyzed statistically.

Results. The range of joint motion in almost all joints differed significantly with respect to the different disease types. In OI type I patients, generalized hypermobility of the joints was present, without decrease in joint motion. In OI type III the extremities were severely maligned, especially the lower limbs. In type IV the upper and lower extremities were equally maligned. Muscle strength differed significantly with respect to the different types of OI. In type I patients, muscle strength was normal except for the periarticular hip muscles. In type III, especially in the lower extremities, muscle strength was severely decreased, with a muscular imbalance around the hip joint. In type IV, muscle strength was mainly decreased in the proximal muscles of the upper and lower extremities. In children 7.5 years of age, significant differences existed among the different disease types in functional skills regarding mobility. No significant difference was observed in self-care and social function, although the most severely affected children showed a tendency to score better with social function. Older children differed significantly concerning mobility and self-care items. In children 7.5 years old, a correlation was sometimes observed between impairment and disability items, although in older children a moderate to good correlation was always present ($r > .6$).

Conclusion. In OI, severity-related profiles exist, within the different subtypes of the disease, regarding range of joint motion, muscle strength, and functional skills. In younger children, impairment parameters do not sufficiently correlate for disability. Rehabilitation strategies in younger children should therefore focus on improvement of functional skills and not only on impairment parameters. *Pediatrics* 1997;99(2). URL: <http://www.pediatrics.org/cgi/content/full/99/2/e3>; *osteogenesis im-*

perfecta, pediatrics, muscle strength, range of joint motion, impairment, disability, functional outcome.

ABBREVIATIONS. OI, osteogenesis imperfecta; JAM, alignment and motion (scale); PEDI, Pediatric Evaluation of Disability Inventory; MMT, manual muscle testing.

Osteogenesis imperfecta (OI) is a congenital connective tissue disorder. In all its forms OI probably affects 1 in 5000 to 10 000 individuals without racial or ethnic preference.¹ Major clinical characteristics of OI include fragility of bone, osteopenia, variable degrees of short stature, and progressive skeletal deformities. Additional clinical manifestations are blue sclerae, dentinogenesis imperfecta, joint laxity, and maturity-onset deafness.² The nonlethal OI syndromes are subdivided into six types (IA, IB, II, III, IVA, and IVB).^{3,4} The most common type, OI type I, is characterized by osteopenia leading to fractures, distinctly blue sclerae, and a high incidence of adult-onset conductive hearing loss. Within type I are two subgroups, IA characterized by normal teeth, and IB with dentinogenesis imperfecta. OI type II is usually lethal in the perinatal period. OI type III is characterized by severe osteopenia leading to multiple fractures, progressive deformity of bones and spine, and severely decreased height.

OI type IV is rare and is characterized by osteopenia leading to fractures. Sclerae are normal. Short stature and deformity of the long bones and spine tend to be more marked in type IV than in type I. Some patients have normal teeth (type IVA). Others have dentinogenesis imperfecta (type IVB).

Although these clinical features provide a basis for classification, a significant proportion of patients cannot be classified in this way.⁴ Studies on collagen I genes and their mutations in OI are numerous. Smith⁵ states that within the last two decades biochemical advances have been so rapid that at one time it seemed that OI could be explained entirely in molecular terms. Current work suggests that this is not the case and that we still have much to learn about the relationship between phenotype and genotype.⁵

In 1965, Nagi⁶ introduced a scheme that delineated the major pathways from disease or active pathology to various functional implications. This scheme consisted of four domains: active pathology, impairment, functional limitation, and disability. In 1980 the World Health Organization introduced the International Classification of Impairments, Disabilities

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and Handicaps,⁷ a system that classified the consequences of disease in three levels. Both schemes have guided disability research during the past 25 years and have undergone various modifications through the years.

Studies on impairment and disability issues in OI patients are scarce. Rehabilitation approaches focus on improving range of motion of the joints as well as muscle strength of trunk and extremity muscles. Improvement of functional abilities is mainly focused on sitting, standing, and ambulation.^{8,9}

Our purpose was to investigate impairment patterns in OI, such as range of joint motion and muscle strength, as well as disability or functional limitation items, such as functional skills and the amount of parental assistance, in correlation to the different disease types. We also investigated correlations between impairment and disability items.

METHODS

In this cross-sectional study, we examined 54 patients (27 boys and 27 girls) with OI who were treated in our hospital, a national referral center for children with OI. Children were included in this study only if the diagnosis of OI was definite. Children who had recent fractures needing immobilization and children who had intramedullary stabilization of the long bones within the previous half year were excluded from the protocol because of the possible influence on range of motion, muscle strength, and functional ability. All measurements were performed by the first author (R.H.H.E.).

Range of joint motion was measured in a standardized way with a standard two-legged goniometer, using the anatomical landmark method. This method was found to be highly reproducible in healthy subjects.¹⁰ Range of motion was compared to reference values for children.¹¹ In the upper extremities range of motion of the shoulder joint (flexion and abduction), elbow joint (flexion and extension), and wrist joint (dorsal and palmar flexion), as well as flexion and extension in the second metacarpophalangeal joint, were measured. In the lower extremities, flexion, extension, abduction, and external and internal rotation were measured in the hip joint. In the knee joint, flexion and extension were measured as well as plantar and dorsal flexion in the ankle joint. Children <2 years of age were excluded from range of joint motion measurements because difficulties in positioning anatomical landmarks existed. To measure the total range of motion in the upper and lower extremities, the Joint Alignment and Motion (JAM) scale was used.¹² The JAM scale consists of a 5-point scale (0–4). Each individual joint is scored according to an estimate of the percentage of normal motion and joint alignment. These estimates are determined as an examiner assists the patient in an active-assisted range of motion of each joint.

Based on a knowledge of a joint's normal range of motion the examiner visually estimates whether a joint's range of motion is normal or limited by quartiles. Hypermobility of the joints was scored according to the criteria of Bulbena et al.¹³

Muscle strength was scored according to the manual muscle

testing criteria of the Medical Research Council using a 6-point scale (range: 0–5).¹⁴ Children < 5 years of age were excluded from muscle strength measurements because no reliable measurement could be performed. Muscle strength of the anteflexors and abductors of the shoulder, flexors and extensors of the elbow, flexors and extensors of the wrist, and flexors of the fingers were tested in the upper extremities. In the lower extremities, flexors, abductors, and extensors of the hip joint, flexors and extensors of the knee joint, and dorsal flexors and plantar flexors of the ankle joint were tested.

Functional abilities regarding ambulation were scored according to the criteria of Bleck.¹⁵ He classified the possibility for ambulation in a nonwalker, exercise walker, household walker, neighborhood walker, and community walker.

Functional abilities and the amount of parental assistance were scored using a translated version of the Pediatric Evaluation of Disability Inventory (PEDI).¹⁶ The PEDI consists of two major dimensions (functional skills scales and caregiver assistance scales) in the content domains of self-care, mobility, and social function. The performance of functional tasks and activities is measured by the functional skills scale, which samples a set of behaviors that are believed to be important for daily functioning. The amount of help required to accomplish daily tasks is measured by the caregiver assistance scales. Reference values are provided for children between 0.5 and 7.5 years. Normal values are defined in the range of 2 SD. Results of children >7.5 years are calculated as a scaled score. In healthy children >7.5 years of age, the normal score is 100%.

STATISTICS

The total score for muscle strength in the upper and lower extremities was calculated by adding the strength of all tested muscles and dividing by the number of tested muscles. In this way, the mean ratio for muscle strength in the upper and lower extremities was calculated. Regarding range of joint motion and muscle strength, no significant differences between the right and left extremities were observed. Data are presented as the median, range, and 25th and 75th percentiles, because of the skewed distribution of the parameters in our population. In Tables 2, 4, 5, 6, and 7 we analyzed the relationship between the different subtypes and the subsequent tests. Statistical comparison of different groups of patients was performed with the Kruskal-Wallis test. Correlations were calculated with the Spearman rank correlation (r_s). $P < .05$ was considered significant. For statistical analysis, Epi Info version 6 was used.¹⁷

RESULTS

Data regarding the different disease types, age, gender, and number of children are presented in Table 1. In the OI type IV group, a 2-year-old boy was seen without primary dentition, so the presence

TABLE 1. Patient Characteristics in Osteogenesis Imperfecta

Type	n	Mean Age (y-mo)	SD	Range	Boys (n)	Girls (n)
I	24	6.4	3.9	1.0–15.0	10	14
IA	18					
IB	6					
III	15	6.7	2.8	2.0–12.0	7	8
IV	15	9.2	3.9	2.1–15.1	10	5
IVA	9					
IVB	5					
Total group	54	7.3	3.8	1.0–15.1	27	27

of dentinogenesis imperfecta could not be classified in this patient.

Range of Joint Motion

Data regarding the median range of motion of the aforementioned joints are presented in Table 2. Joint motion was different for the various OI types. Especially in type III children, range of motion was severely decreased in the shoulder, hip, knee, and ankle joint. Alignment in the extremities of these patients, especially in the lower extremities, was severely disturbed. All patients had extremely externally rotated hips with a severe decrease in internal rotation.

Data regarding the JAM score, indicating the amount of malalignment of the extremities, and the hypermobility score according to Bulbena are presented in Table 3. Malalignment of the extremities was found mainly in OI types III and IV, and generalized hypermobility of the joints was observed only in type I children. In type I children a significant relation regarding generalized hypermobility of the joints was found, compared to the other types ($P = .0001$).

Muscle Strength

In all muscle groups a significant difference existed concerning the different types of the disease (Table 4). In children with OI type I, a decrease in muscle strength was noted only in the periarticular hip muscles (Medical Research Council grade 4). In this type, the mean ratio in muscle strength regarding the upper extremity was 4.8 and regarding the lower extremity 4.4. In children with OI type III, muscle strength was generally decreased. In the periarticular hip muscles, a muscular imbalance was observed. Hip extensor and abductor muscles scored less than hip flexor muscles. In this type, the mean ratio in muscle strength regarding the upper and

lower extremities was 3.8 and 3.3, respectively. In children with OI type IV, muscle strength was mainly decreased in the proximal muscles of the extremities. Around the hip and knee joint, muscular imbalance was observed. In this type, the mean ratio in muscle strength regarding the upper and lower extremity was 4.4 and 3.6, respectively.

Functional Abilities

Data are presented in Table 5. In OI type I children, basic conditions for standing and walking were present. All but one child, age 1.0 year, were able to walk. Walking distance in this group differed, but almost 70% were able to walk in the neighborhood or even further. Six of 15 patients with OI type III managed to stand or walk, whereas 9 children were not able to bear weight on their feet. A 12-year-old boy was not able to sit without support. All children with OI type III moved around their neighborhood using an electrical wheelchair. In OI type IV, all children were able to bear weight on their feet, but only eight children were able to walk in a household situation or farther.

Functional abilities according to the PEDI are presented in Tables 6 and 7. In children ≤ 7.5 years, a significant difference between the disease types existed only for mobility. In self-care and social function almost all children scored between 1 to 2 SD from the normal population. Although a significant difference in social function was absent, a tendency was observed among OI type III children to score better than the other types.

With regard to care-giver assistance in children ≤ 7.5 years, the same tendency was observed. Only for mobility a significant difference was found. In children > 7.5 years, a significant difference between the three disease types was noted for self-care and mobility. The more severe the disease, the less func-

TABLE 2. Median and Interquartile Range of Joint Motion Related to Type of Osteogenesis Imperfecta*

Joint and Reference Value	Type I			Type III			Type IV			Kruskal-Wallis Test <i>p</i>
	n	Median	P25-P75	n	Median	P25-P75	n	Median	P25-P75	
Shoulder anteflexion/180	24	180	180-180	15	110	90-170	15	160	150-180	.0001†
Shoulder abduction/180	24	180	180-180	15	110	90-170	15	160	150-180	.0001†
Elbow flexion/150	24	150	150-150	15	130	115-150	15	150	130-150	.0007†
Elbow extension/0	24	0	0-15	15	0	-25-0	15	0	0-10	.002†
Wrist palmar flexion/45	24	90	80-90	15	70	60-90	15	60	60-70	.003†
Wrist dorsal flexion/45	24	90	70-90	15	60	50-70	15	60	60-70	.001†
Metacarpophalangeal II flexion/90	24	90	90-90	15	90	90-90	15	90	70-90	.2
Metacarpophalangeal II extension/45	24	45	45-45	15	45	40-45	15	45	45-45	.04†
Hip flexion/130	23	140	140-140	14	120	120-130	15	130	130-140	.0001†
Hip extension/0-10	23	10	0-10	14	-30	-30--20	15	-10	-10-0	.0001†
Hip abduction/30-45	23	50	45-50	14	30	20-30	15	35	30-45	.0001†
Hip left external rotation/40-50	21	90	85-100	14	90	80-100	15	75	55-90	.03†
Hip right external rotation/40-50	21	90	85-100	14	90	80-100	15	70	60-80	.004†
Hip left internal rotation/30-45	20	10	-10-20	14	-10	-20-0	15	10	0-30	.06
Hip right internal rotation/30-45	20	20	0-30	14	-10	-20-0	15	10	0-20	.03†
Knee flexion/120-150	23	150	140-150	15	120	110-130	15	140	130-150	.0001†
Knee extension/0-10	23	15	10-15	15	-15	-30-0	15	-5	-20-0	.0001†
Ankle dorsal flexion/20	23	25	20-30	15	0	-30-10	15	15	10-20	.0001†
Ankle plantar flexion/50	23	50	50-60	15	50	50-70	15	50	50-50	.1

* Median range of joint motion in degrees, with P25 representing the 25th and P75 the 75th. In all joints a symmetrical range of motion was observed, except for the hip joint (external rotation-internal rotation). Reference values for range joint motion from Bernbeck and Dahmen.¹¹

† $P < 0.05$.

TABLE 3. Alignment of the Extremities and Presence of Generalized Hypermobility of the Joints for Types of Osteogenesis Imperfecta

	Type I		Type III		Type IV	
	Median	P25–P75	Median	P25–P75	Median	P25–P75
JAM score for upper extremity*	0	0–0.5	1.75	1–2	1	0.75–1.25
JAM score for lower extremity*	0	0–0	3	1.5–3.25	0.5	0.25–2.25
Generalized hypermobility†	7.5	4.5–8.5	0	0–0	0	0–1

* According to the Joint Alignment and Motion (JAM) scale,¹² The higher the JAM score, the more malalignment is observed.

† According to Bulbena et al.¹³ Generalized hypermobility of the joints is present if the score is >4 in girls and >3 in boys.

TABLE 4. Muscle Strength* Related to Type of Osteogenesis Imperfecta

	Type I			Type III			Type IV			Kruskall-Wallis Test <i>P</i>
	n	Median	P25–P75	n	Median	P25–P75	n	Median	P25–P75	
Shoulder anteflexors	15	5	5–5	12	4	3.5–4	11	4	4–5	.0002+
Shoulder abductors	15	5	5–5	12	4	3.5–4	11	4	4–5	.0002+
Elbow flexors	15	5	5–5	12	4	4–4	11	4	4–5	.0002+
Elbow extensors	15	5	5–5	12	4	4–4	11	4	4–5	.0001+
Wrist dorsal flexors	15	5	5–5	12	4	4–4	11	5	4–5	.0001+
Wrist palmar flexors	15	5	5–5	12	4	4–4	11	5	4–5	.0001+
Finger flexors	15	5	5–5	12	4	4–4	11	5	4–5	.0001+
Hip flexors	14	4	4–5	12	4	3–4	11	4	3–4	.0001+
Hip extensors	14	4	4–4	12	3	2–3	11	3	2–3	.0003+
Hip abductors	14	4	4–5	12	3	2–3	11	3	2–3	.001+
Knee flexors	14	5	4–5	12	4	4–4	11	4	4–4	.007+
Knee extensors	14	5	4–5	12	4	3–4	11	4	3–4	.002+
Ankle plantar flexors	14	5	5–5	12	4	3–4	11	4	4–5	.0001+
Ankle dorsal flexors	14	5	5–5	12	4	4–4	11	4.5	4–5	.0001+

* According to Medical Research Council criteria.¹⁴ Median muscle strength is presented, with P25 representing the 25th percentile and P75 the 75th percentile.

† $P < .05$.

TABLE 5. Functional Abilities Related to Type of Osteogenesis Imperfecta*

Functional Ability	Type I (n = 24)		Type III (n = 15)		Type IV (n = 15)		χ^2	<i>P</i>
	No	%	No	%	No	%		
Sitting unsupported	0	0	1	6.7	0	0	2.6	.3
Bottom-shuffling	0	0	3	20	0	0	8.3	.02
Non-standing	1	4.2	5	33.3	0	0	10.5	.005
Weight bearing on feet	0	0	1	6.7	4	26.7	8.0	.02
Exercise walker	2	8.3	3	20	3	20	1.4	.5
Household walker	4	16.7	2	13.3	5	33.3	2.2	.3
Neighborhood walker	7	29.1	0	0	2	13.3	5.8	.05
Community walker	10	41.7	0	0	1	6.7	12.3	.002

* Data of children are presented when the highest level of functional ability has been mastered.

tional capability was observed. The tendency observed in younger children, that social function is more developed in the OI type III group, could not be confirmed in this age group. With regard to the amount of care-giver assistance, we observed that parental assistance increased when functional abilities were less developed.

Correlations Between Impairment and Disability

The correlations between impairment and disability items are presented in Table 8. In children ≤ 7.5 years, range of motion and muscle strength in the upper extremities were correlated with self-care in the PEDI, whereas range of motion and muscle strength in the lower extremities were correlated with mobility. A negative correlation existed between range of motion (JAM scale) and muscle strength (muscle strength of all muscles divided by the number of muscles) ($r = -0.8$; $P = .004$). No correlation was observed between range of motion

and self-care ($r = -0.3$; $P = .1$) and between muscle strength and self-care ($r = .1$; $P = .7$). In children ≤ 7.5 years, a moderate to good correlation existed between range of motion (JAM scale) and muscle strength ($r = -0.6$; $P = .002$). A moderate to good correlation was observed between range of motion and mobility ($r = -0.6$; $P = .003$) and a moderate correlation was observed between muscle strength and mobility ($r = .4$; $P = .1$).

In children > 7.5 years, in the upper extremities a negative correlation was observed between range of motion and muscle strength ($r = -0.6$; $P = .002$). A moderate correlation was found between range of motion and self-care ($r = -0.5$; $P = .01$), but a high correlation existed between muscle strength and self-care ($r = .7$; $P = .0001$). Regarding the lower extremities a correlation existed between range of motion and muscle strength ($r = -0.6$; $P = .001$) as well as range of motion and mobility ($r = -0.7$; $P = .0001$) and muscle strength and mobility ($r = .9$; $P = .0001$).

TABLE 6. Functional Abilities in Children ≤ 7.5 Years of Age According to Type of Osteogenesis Imperfecta*

	Type I (n = 12)		Type III (n = 6)		Type IV (n = 3)		Kruskall-Wallis Test P
	Median	P25–P75	Median	P25–P75	Median	P25–P75	
Self-care	39.3	31.7–59.5	33.4	31–37.7	30.4	12.6–33.3	.0
Mobility	25.0	14.8–41.0	10	10–10	10	10–10	.003†
Social function	39.9	34.9–42.3	52.3	40–54.4	44.1	36.1–44.1	.2
Self-care Care giver assistance	36.8	27.4–57.5	37.7	27.9–41.9	26.1	10–27	.1
Mobility Care giver assistance	33.8	25.3–51.3	10	10–13.7	14.9	10–18.3	.003†
Social function Care giver assistance	52.9	41.2–63.0	60.34	53–66.3	49.5	22.7–63.7	.2

* Measured with the Pediatric Evaluation of Disability Inventory.¹⁶ Reference score in healthy children < 7.5 years is 50 ± 20 (2 SD).
† $P < .05$.

TABLE 7. Functional Abilities in Children > 7.5 Years of Age According to Type of Osteogenesis Imperfecta*

	Type I (n = 12)		Type III (n = 6)		Type IV (n = 9)		Kruskall-Wallis Test P
	Median	P25–P75	Median	P25–P75	Median	P25–P75	
Self-care	72.8	53.7–87.2	65.1	46.7–71.7	85.1	75.9–93.0	.003†
Mobility	79.8	63.7–91.7	47.4	20.9–53.1	58.2	53.9–75.2	.001†
Social function	66.25	55.9–75.4	79.8	49.1–89.1	89.1	89.1–100	.6
Self-care Care giver assistance	71.8	47.3–86.5	64.1	37.2–68.1	83.2	69.6–89.7	.05
Mobility Care giver assistance	78.9	61.1–86.0	47.1	11.7–49.8	72.7	68.5–100	.002†
Social function Care giver assistance	86.4	55.3–94.9	94.9	61.3–100	100	100–100	.2

* Measured with the Pediatric Evaluation of Disability Inventory.¹⁶ Healthy children > 7.5 years have mastered all items. Their score in all domains is 100.
† $P < .05$.

DISCUSSION

In this study, we defined type-related profiles in OI and investigated whether impairment items are prerequisites for functional skills.

Regarding range of joint motion, type-related profiles do exist in OI. Generalized hypermobility of the joints was present in almost all children with OI type I, as proved by the hypermobility scale of Bulbena. In this study we preferred the Bulbena scale over the Beighton scale.¹⁸ Although the Beighton scale has been used for ages all over the world, the reliability of this hypermobility assessment has not been tested properly. The Bulbena scale showed a high internal consistency reliability (Cronbach's α -coefficient: 0.9).¹³ Beighton assessed the spine and four joints bilaterally (thumb, little finger, elbow, and knee). A score ≥ 4 on this 9-point scale indicated generalized hypermobility. In our study, we frequently observed hypermobile thumbs and little fingers without the presence of other hypermobile joints. According to Beighton's criteria, generalized hypermobility was present, but in our opinion, hypermobility located only in the thumb and fingers must be defined as local, not generalized, hypermobility.

In almost all OI type III children, bowing of the long bones was observed mainly in the diaphysis

(tibiae: antecurvature; femora: antecurvature/lateral deviation), and we noted little influence of bowing on range of joint motion. After correction of bowing by intramedullary rodding, loss of joint motion was caused by articular and soft tissues contractures.

In the wrist and fingers no limitation in range of motion was measured, except for two girls with severely decreased flexion of the metacarpophalangeal joints. This condition had been present since birth without a known etiology. The same observations have been reported by Binder et al.⁸

Manual muscle strength testing is hypothesized as a valuable tool for the clinical assessment of patients with neuromuscular problems. Barr et al¹⁹ studied the inter- and intratester reliability in Duchenne and Becker muscular dystrophy and found high reliability (intertester correlation coefficient: 0.90; intratester correlation coefficient: 0.80). Schwartz et al²⁰ compared manual muscle testing (MMT) and hand-held myometry in the upper extremity and concluded that the range of myometry measurement for a particular MMT grade appeared to be most specific for MMT scores < 4 and less specific for MMT scores ≥ 4 . We used manual muscle strength measurements in all patients. We preferred to use a hand-held myometer as reported by Bäckman et al,²¹ but were afraid of

TABLE 8. Correlation Between Impairment (Range of Motion and Muscle Strength) and Disability (PEDI) Items in the Upper and Lower Extremities*

Upper Extremity/Age ≤ 7.5 yr	Muscle Strength		Self-care (n = 21)	
	r _s	P	r _s	P
Range of motion (n = 21)	-0.8	.004†	-0.3	.1
Muscle strength (n = 12)			0.1	.7
Upper Extremity/Age > 7.5 yr	Muscle Strength		Self-care (n = 27)	
	r _s	P	r _s	P
Range of Motion (n = 33)	-0.6	.002†	-0.5	.01†
Muscle strength (n = 25)			0.7	.0001†
Lower Extremity/Age ≤ 7.5 yr	Muscle Strength		Mobility (n = 21)	
	r _s	P	r _s	P
Range of motion (n = 21)	-0.6	.01	-0.6	.003†
Muscle strength (n = 16)			0.4	.1
Lower Extremity/Age > 7.5 yr	Muscle Strength		Mobility (n = 27)	
	r _s	P	r _s	P
Range of motion (n = 32)	-0.6	.001†	-0.7	.0001†
Muscle strength (n = 30)			0.9	.0001†

* PEDI indicates Pediatric Evaluation of Disability Inventory.¹⁶ r_s indicates Spearman rank correlation.

† P < .05.

producing fractures and therefore did not use this method even though it might be more quantitative.

Muscle strength was also related to OI disease type. It was remarkable that all OI types demonstrated a muscular imbalance between flexor and extensor muscles in the hip joint. Range of motion and muscle strength were highly correlated. Consequently, the more malalignment existed, the less muscle strength was observed. A possible explanation for this observation might be the presence of biomechanical changes in the joints. Children with OI type III are born with severely bowed legs, placing their legs in semiflexion, abduction, and external rotation. In this position abduction and extension muscles of the hip joint cannot develop properly. This possible biomechanical explanation, combined with the different development of motor milestones, in which extension activity in the prone position is severely retarded,²² might be the reason for the underdevelopment of extensor muscles. In all children the mean ratio for muscle strength in the upper extremities was higher than in the lower extremities. This might have been caused by the fracture frequency, which seemed higher in the legs. Thus, frequent immobilization, disuse, and diminished functional capacity of the legs are thought to be responsible for decreased muscle strength in the lower extremities.

Children who had recent fractures needing immobilization, and children who had intramedullary stabilization of the long bones in the previous half year were excluded from this study. Therefore, eight children could not participate in this study. Because these children were almost equally divided among the OI types, no potential selection bias resulted from these exclusions. The wide range of ages of the children in the OI subgroups is likely to produce a range of findings related to the duration of the disability. The Medical Research Council grading system for

measuring muscle strength does not take into account differences in gender, age, and weight. Because age and gender in our study are quite different across types, it makes the outcome of measurements less quantitative. Therefore, all measurements were performed by a single experienced person and all measurements were performed in a standardized manner. Despite these precautions, a potential bias in judgment of muscle strength grade 4 (good) and grade 5 (normal) cannot be excluded.

We defined high correlation as $\rho \geq 0.9$ and moderate to good correlation as ≥ 0.6 .

In recent studies, the relationship between impairment and disability in childhood was found to be disputable.²³ No studies were found regarding the functional (dis)ability in OI patients, other than ambulation. In this study we found that children ≤7.5 years, despite the severity of the disease, were able to perform almost normal activities regarding their self-care. Concerning mobility, children with the severest type of OI scored the lowest in functional skills and obtained the most parental assistance. Because almost all children with OI type III were wheelchair-bound, the problems with mobility were not surprising. We observed a tendency, although not significant, for younger children with OI type III to score higher in social function than other types, perhaps to compensate for their disability. This tendency was not observed in older children. The slight discrepancies between the different OI types in self-care and social function observed in the younger children became more pronounced when children grew older. This might have been caused by the progressive disproportion of the habitus in the more severe types and the increased fracture incidence, leading to less activity.

In younger children, range of motion and muscle strength in the upper extremities did not predict the arm and hand function in the self-care items because

correlations between these items were low. Muscle strength in the lower extremities did not predict mobility because correlations were moderate. We observed that severely handicapped children were able to fulfill many items regarding self-care despite severe contractures in the shoulder joint and severely decreased strength in the arm muscles. Therefore, in this age group, optimal range of motion and maximal muscle strength are not always basics for maximal functional skills. This has consequences for the approach to rehabilitation. Training of functional skills must be one of the major goals in rehabilitation strategies.

In children >7.5 years, muscle strength has a more significant role within impairment and disability items. From our observations it seemed that muscle strength is a prerequisite for all functional skills, especially in the mobility domain, where items such as climbing a staircase are requested.

It seems that the functional skills in younger children are not solely dependent on impairment items. When children grow older and more functional skills are required, muscle strength and, to a lesser extent, range of motion are conditions for maximal functional skills.

In conclusion, we found that in OI, type-related profiles exist, concerning impairment items, such as range of motion and muscle strength. In children ≤7.5 years, functional skills such as self-care and social function were not significantly different among the three OI types, as mobility differed significantly. In this age group, we observed that social function was better developed in children with the most severe type (OI type III), possibly to compensate for their disability. In this age group, impairment items were sometimes conditions for the achievement of functional skills.

In children >7.5 years, differences between the different types of the disease became more pronounced, evidenced by functional skills such as self-care and mobility. In this age group, impairment items were correlated with disability items. These observations have consequences for rehabilitation strategies. The early correction of bowing of the long bones, resulting in the early restoration of alignment and biomechanical possibilities, may influence functional abilities other than ambulation.

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