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Outcomes of modified Sofield procedure in children with osteogenesis imperfecta in Oman

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ABSTRACT

Objectives: Osteogenesis imperfecta (OI) is the most commonly inherited bone disease, with a wide variety of clinical features, most commonly recurrent fractures and bone deformities. The management is multidisciplinary, and surgical management usually aims to correct deformities or prevent further fractures.

Methods: This study aimed to assess the outcomes of Omani OI patients who underwent a modified Sofield procedure. This was a descriptive retrospective study. All patients who underwent a modified Sofield procedure from 2013 to 2019 in one center in Oman were included in the study. Demographic data were collected, as well as surgical details. The level of mobility pre operatively and postoperatively was also included in the study. A survey to assess the quality of life was sent to all patients to assess the impact of the procedure on their lives.

Results: The procedure was done on a total of 37 bones, 66.7% of them were male. The complication rate was found to be 13.5%. The mean quality-of-life score was found to be 82.3 after the surgeries compared to 77.4 before the procedure (P = 0.097).

Conclusion: We report a lower complication rate in our cohort of OI patients compared to what has been observed in other studies. The level of mobility improved in one-third of the patients, and in most of the remaining two-thirds, the level that was achieved was maintained. Interestingly, although patients reported reduced fractures postoperatively, they still feared getting new fractures, which restricted their daily activities.

Keywords: Mobility, Modified Sofield procedure, Oman, Osteogenesis imperfecta, Quality of life

INTRODUCTION

Osteogenesis imperfecta (OI) is a genetic bone disease that is inherited in an autosomal dominant fashion. It is the most commonly inherited bone disease, with a prevalence of 1 in 10,000–20,000 births.[1] Most OI cases are caused by autosomal dominant defects in the genes that encode Type I collagen, COL1A1, or COL1A2, causing bone fragility, reduced bone mass, and growth deficiency.[2] Due to the different genotypes, this disease has various clinical features, such as macrocephaly, blue sclerae, and dentinogenesis imperfecta. Therefore, eight types of OI were described. This classification was initiated in 1979 by Sillence who suggested four classes of OI: Type I mild non-deforming; Type II perinatal lethal; Type III severely deforming; and Type IV moderately deforming. In 2004 and 2007, four more types were added: Type V

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moderate-to-severe disease often causing deformity and short stature, with normal teeth and sclera; Type VI, a moderate disease with severe vertebral body involvement with compression, white or blue sclera, normal teeth, and no Wormian bones; Type VII that clinically is similar to Type II although with smaller head and white or faintly blue sclera; and Type VIII that is similar to Type III although with a round face, normal sclera, and a barrel-shaped chest.[3]

The management approach for OI patients is multidisciplinary and involves a variety of healthcare providers from different specialties such as physiotherapists, pediatricians, orthopedists, geneticists, and dentists. The specific treatment plan depends on the severity of OI. In mild forms of the disease, patients might lead a near-normal life with no medical interventions or only with Vitamin D and calcium supplements. However, severe forms of OI warrant medical management with bisphosphonates, or surgical interventions to correct deformities or prevent further fractures in certain instances.

Surgical treatment of OI is based on the Sofield-Millar technique, described in 1959. It consists of multiple osteotomies of the bowed long bones followed by intramedullary rodding using, at that time, Kuntscher or Rush rods, which serve to realign and stabilize deformed and weakened bones.[4] Since then, this procedure has undergone many modifications and innovative changes, and it has reported positive outcomes on mobility and risk of fractures. However, it does not come without complications. This study aimed to examine the outcomes of Omani OI patients who underwent a modified Sofield procedure in one major institute. There are few similar studies done in the Arabian Gulf countries published in the literature, but this is the first of its kind in Oman.

MATERIALS AND METHODS

The study was a retrospective study that included OI patients who underwent the modified Sofield procedure and followed up in Armed Forces Hospital (AFH), Muscat, between 2013 and 2019. The data were collected from the hospital's information system, and missing data were obtained by calling the patient's phone number registered in the hospital system. Inclusion criteria were all patients who were diagnosed with OI and underwent the modified Sofield procedure in AFH.

The data collected included demographic characteristics, clinical manifestations of OI, medical management, surgical management, and outcomes. Demographic characteristics included the age at diagnosis, gender, and disease duration. Clinical features of the disease included fractures, reduced bone mass, level of mobility, short stature, skeletal deformities, blue sclerae, dentinogenesis imperfecta, joint laxity, and adult-onset deafness. Outcomes were measured by reduction of the number of fractures, need for reoperation, improvement of mobility, and improvement of the quality of life.

The number of fractures preoperatively and postoperatively was reported by the parents as the patients are not treated exclusively in our hospital, and most often seek help in the regional hospitals in case of fractures. The level of mobility was categorized by the authors into six categories according to the best level of movement the patient achieved: No movement/bedridden, wheelchair mobilization, crawling, stand only, walk with support, and walk without support. Quality of life was measured by the OI Quality-of-Life Questionnaire, which is a validated questionnaire developed specifically for pediatric patients with OI.[5] The questionnaire assesses the quality of life in different integral domains of function: Being safe and careful, reduced function, fatigue, pain, fear, and life skills. Two questionnaires were sent to the patient after taking their permission to participate in the study from one of the parents. One copy was for the quality of life before the operation, and the other was for the quality of life after the operation. This was to seek any subjective differences in the quality of life after the procedure. The license to use the OI quality-of-life questionnaire was obtained from the authors.

Surgical technique

Under image guidance, the center of rotation of angulation is identified and a skin incision is made at that level. Corrective osteotomy is done, and the number of osteotomies depends on the severity of the deformity. In general, 1–2 osteotomies are done, although three osteotomies are required in certain cases. This is followed by retrograde reaming of the proximal shaft and antegrade reaming of the distal shaft. The nail is then inserted in a retrograde manner in the proximal part and antegrade in the distal part. Finally, distal locking is done with Kirschner's wires.

Statistical analysis

Means and standard deviations were calculated for continuous variables, while frequencies and percentages were calculated for categorized variables. The quality-of-life score was compared preoperatively and postoperatively, with Student's *t*-test. $P < 0.05$ was considered statistically significant. These statistical values were analyzed using the IBM SPSS statistics version 23 program. The data analysis was done with the help of an experienced statistician.

RESULTS

Twelve patients underwent the Sofield procedure in the study period, eight were male and four were female (66.7% and 33.3%, respectively). We had a total of 37 bones that

underwent the same procedure by the same surgeon. This included 18 tibias, nine femurs, eight humeri, one radius, and one ulna, as shown in [Table 1]. [Figures 1 and 2] show pre- and post-operative radiographs of two patients in the study. The mean age of the patients at the time of surgery was 8 years, with a median of 7 years. The average duration of follow-up was 2.2 years. The type of OI could not be obtained as most of these patients were following up with a pediatrician in another hospital. However, we categorized them according to the time of onset of the first fracture into before or during birth (nine patients, 75%), before walking (two patients, 16.7%), and after walking (one patient, 8.3%), indicating a higher number of severe types of OI in our cohort.

The frequency of the fractures pre- and post-operative was obtained, as shown in [Table 2]. One patient out of the study cohort reported <10 fractures per year before the surgery, while the majority had more than 20 fractures per year (seven patients, 58.3%). In addition, four patients reported 10–19 fractures per year (33.3%). On the other hand, when the number of fractures per year was compared after surgery, 50% (six patients) of the patients reported no fractures, 33.3% (four patients) had <10 fractures per year, and 8.3% (one patient) had 10–19 fractures, and only 8.3% (one patient) had more than 20 fractures.

The level of mobility of the patients was categorized into six groups and we assessed the post-operative change, as demonstrated in [Table 3]. Preoperatively, one patient was bedridden, one was crawling, three mobilized by wheelchair, five patients could stand but not walk, and two patients could walk without support. After the surgery, the level of mobility was found to be as follows; one patient was bedridden, three mobilized by wheelchair, four patients could stand but not walk, and four could walk without support.

Table 1: Details of the 12 patients who underwent Sofield-Millar operations.

Case	Gender	Age at 1st surgery (year+month)	Age at the final assessment (year+month)	Duration of follow-up (year+month)	Number and sites of operation								Number of operated limbs
					Femur		Tibia		Humerus		Radius and Ulna		
					R	L	R	L	R	L	R	L	
1	M	3+11	5+11	2+2	1	1	-	-	-	-	-	-	2
2	M	1+10	3+2	1+6	1	1	-	-	-	-	-	-	2
3	F	8+0	10+6	2+5	1	-	1	1	1	-	-	-	4
4	M	4+11	7+7	2+10	2	1	-	-	-	-	-	-	3
5	M	4+3	6+9	2+7	-	-	1	1	1	1	-	-	4
6	M	2+6	4+5	2+0	1	1	1	1	-	-	-	-	4
7	F	13+9	16+3	2+6	-	-	1	1	-	-	-	-	2
8	F	4+7	7+5	2+10	1	1	1	1	-	-	-	-	4
9	F	3+10	6+8	2+10	-	-	1	1	1	1	-	-	4
10	M	4+0	6+2	2+2	1	1	1	1	-	-	-	-	4
11	M	14+9	16+5	1+8	-	-	-	-	1	1	1	-	3
12	M	10+7	12+0	1+5	-	-	-	-	1	-	-	-	1
Mean (years)		6.4	8.4	2.2									

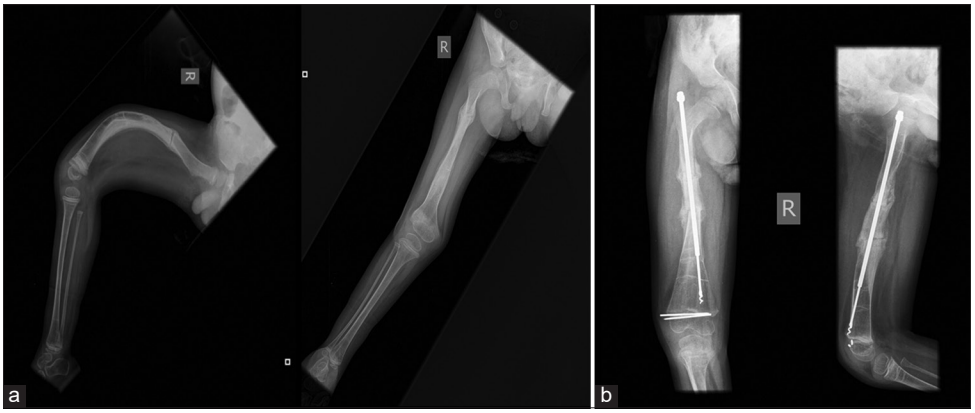

Figure 1: AP and lateral view of the right femur of one of the patients, (a) pre-operative and (b) post-operative.

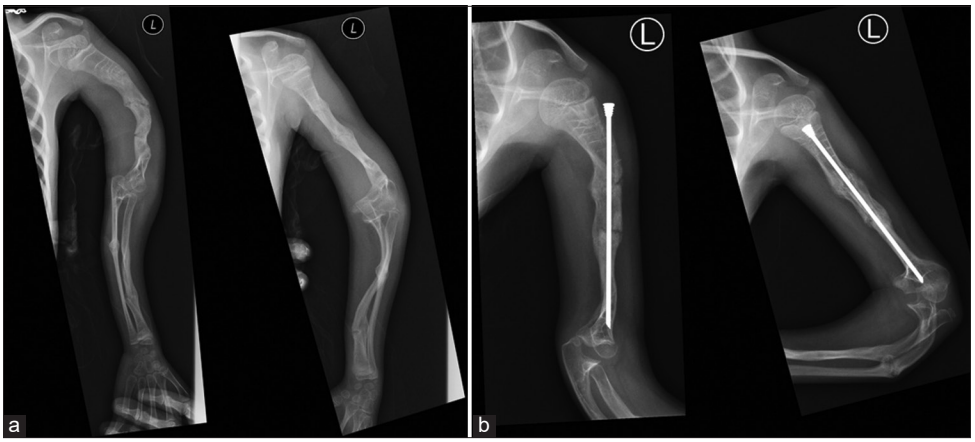

Figure 2: AP and lateral view of the left humerus of one of the patients, (a) pre-operative and (b) post-operative.

Table 2: The number of fractures pre- and post-operative per year.

Fractures/year	Pre-operative, *n* (%)	Post-operative, *n* (%)
No fractures	-	6 (50%)
<10	1 (8.3%)	4 (33.3%)
10–19	4 (33.3%)	1 (8.3%)
>20	7 (58.3%)	1 (8.3%)

Three of the 37 operated bones required revision, which included bilateral tibia and one femur. Five patients with a total of seven bones faced complications postoperatively, including the need for revision, removal of protruding Kirschner wire, and nerve palsy, mainly radial nerve palsy, in surgeries of the humerus (18.9%). Three patients had radial nerve palsy postoperatively. However, they all recovered on follow-up.

The mean quality-of-life score was 77.4 preoperatively compared to 82.3 postoperatively (P = 0.097). This did not show a statistically significant difference due to the small number of patients in the study. The scores of some of the domains are represented in bar charts, as shown in the appendix. When looking at the quality of life of patients who underwent any upper extremity bone surgery to those who did not, it is noticed that the average score of quality of life before surgery was lower than the other patients (64.8 vs. 87.6 in patients who did not require upper extremity surgery). Their quality of life increased postoperatively, similar to those who only had lower extremity surgery. However, the average remained lower (70.3 vs. 92.0).

To analyze patients' answers in individual domain scores, we compared the numbers of patients preoperatively and postoperatively who answered always and most of the time, giving them lower scores. Starting with being safe and careful domain, we found that a smaller number of patients postoperatively received extra help to keep them safe (seven patients preoperatively vs. four patients postoperatively), tried themselves to keep safe from breaking a bone (nine vs. five patients), or thought before starting to play sport to avoid breaking a bone (seven vs. five patients, and three patients answered never postoperatively). However, they more frequently stayed away from some activities to avoid fractures postoperatively (six vs. eight answered always and most of the time preoperatively and postoperatively, respectively). In the reduced activity domain, a smaller number of patients required equipment to move around postoperatively as four patients answered always and most of the time, and three answered never postoperatively compared to five always and most of the time and one never preoperatively. However, their answers to other questions in the same domain reflected that OI stopped them from doing things (four answered always and most of the time preoperatively compared to seven patients postoperatively). They needed to do things differently due to their disease (six patients answered always and most of the time, and three patients answered never preoperatively compared to seven patients answered always and most of the time and one patient answered never postoperatively). In the pain domain, fewer patients postoperatively had pain in the legs and arms (five patients preoperatively vs. four patients postoperatively). They took pain medications (six vs. three patients), giving them better scores. In the fear domain, fewer patients were always or most of the time worried about breaking a bone (seven vs. five patients) and were scared to do something that might cause a fracture (eight vs. seven patients, with fewer patients answering always postoperatively) or scared from needles (six vs. three patients). An equal number of patients answered always and most of the time to being worried about new people handling them preoperatively and postoperatively (five patients each). However, postoperatively, more patients were worried about coming to the hospital (four vs. five patients). The last domain is assessing life skills. Patients postoperatively had more freedom as their friends (five patients answered never

Table 3: Age at which the surgeries were done, plus the mobility level pre- and post-operative.

Case	Operation	Age	Mobility pre-operative	Mobility post-operative
1	Both femurs were operated on the same day.	3 years 11 months.	Stand, not walking.	Walking without aids.
2	Left femur.	1 year 10 months.	Walking without aids.	Walking without aids.
	Right femur.	2 years 2 months.		
3	Right humerus.	8 years 0 month.	Wheelchair.	Stand, not walking.
	Right femur.	9 years 0 month		
	Bilateral tibia.	10 years 0 month		
4	Right femur.	4 years 11 months.	Stand, not walking.	Stand. Not walking.
	Left femur and revision of right femur on the same day.	5 years 8 months.		
5	Left humerus.	4 years 3 months.	Walking without aids.	Walking without aids.
	Bilateral tibia.	4 years 7 months.		
	Right humerus.	5 years 0 month.		
6	Both femurs.	2 years 6 months.	Wheelchair.	Wheelchair.
	Right tibia.	2 years 11 months.		
	Left tibia.	3 years 6 months.		
7	Both tibias were done on the same day.	13 years 9 months.	Wheelchair.	Wheelchair.
8	Both femurs.	4 years 7 months.	Stand, not walking.	Walking without aids.
	Both tibias.	5 years 1 month.		
9	Both tibias.	3 years 10 months.	No movement	Stand, not walking.
	Left humerus.	4 years 4 months.		
	Right humerus.	4 years 8 months.		
10	Both femurs operated on the same day.	4 years 0 month.	Stand, not walking.	Wheelchair.
	Both tibias were operated on the same day.	4 years 7 months.		
11	Left humerus.	14 years 9 months.	Crawling.	No movement.
	Right humerus, radius and ulna.	15 years 3 months.		
12	Right humerus	10 years 7 months.	Stand, not walking	Stand, not walking

preoperatively compared to three patients postoperatively, giving them better scores). An equal number of patients had their families always and most of the time, let them decide what was safe for them (seven patients each). However, patients, postoperatively, were less likely to have their family let them choose their own activities, with eight patients answering always and most of the time preoperatively compared to six patients postoperatively. They also felt more different than their peers because they needed to be more careful as eight patients answered always and most of the time to feeling different postoperatively compared to six patients preoperatively.

DISCUSSION

The management of OI is multidisciplinary and requires addressing various aspects of the patient to improve the overall quality of life. Fractures, pain, and deformities are the most troubling factors of this disease. Medical treatment with bisphosphonates and calcium with Vitamin D are important for treating osteoporosis. Surgical management is directed to treat deformities, prevent further fractures, and improve the overall patient's function and quality of life. There are multiple options for the surgical fixation in OI patients, the most famously used one is intramedullary rod fixation.

Sofield and Miller described their technique of fixation in 1959.[6] Later, Bailey and Dubow described another technique using telescoping rods, which achieved the best results in growing bones.[7]

Bone plating used to be an option; however, studies showed that it is unfavorable due to the higher complication rates, shorter length of time to revision, and unknown effect on longitudinal growth.[8] A modified version of the Sofield procedure was developed by Li *et al*. in 2000. The exposure is minimal, the periosteum is preserved, and the number of osteotomies is kept to a minimum. This is all to reduce the rate of avascular necrosis, bone atrophy, and intraoperative bleeding.[9]

In our study, the incidence of fractures was reduced, compared to before surgery, which agrees with what is described in two previous studies.[6,10] Although the numbers indicate a sound reduction in the number of fractures, we could not run statistical significance tests due to the small sample size. The overall outcome of the surgery is better than what was described in other similar studies.[10,11] We had less reoperation and overall complications rate with only 8.1% rate of reoperation and 18.9% rate of complications in our study compared to 48% reported in Batur and Demir[10] and an 88.2% femoral revision rate and 91.7% tibial revision rate

reported in another study.[11] The outcomes of the surgery can be attributed to many factors and one of which is the distribution of disease severity in the cohort. Although we did not classify the severity using Sillence classification, we identified the onset of fracture as a marker of severity. The majority of our patients had fractures before or during birth indicating more severe disease.

The mobility status of the patients was not significantly affected by the surgery. This was also described by Batur and Demir,[10] wherein there was not much improvement in the level of mobility. However, the level was maintained. This reinforces what was published by Khoshhal and Ellis that this procedure is important for rehabilitation but does not improve ambulation.[12] An article published by the same authors on the upper limb OI surgeries concluded that this procedure indeed improves patients' function.[13] This is also reflected in our study with the increase in the average of patients' quality-of-life score after the procedure. The lower quality-of-life score in those who had upper extremity surgery when compared to those who had lower extremity surgery alone reflects the amount of disability those patients experience when their upper extremity is involved in their disease. Other than the type of OI, a prospective study in 2004 identified muscle mass as a significant predictor of the level of ambulation,[14] which was not accounted for in our study.

What is striking is that the overall calculated quality of life did not increase significantly. On looking at the questionnaire's specific domains, the patient reported better pain control. However, in some areas, the fear, mobility, and overall social activities remained grossly the same, if not lower than pre-operative states. There are no prior studies that investigated the effect of this surgical procedure on quality of life. Patients postoperatively still feared fractures when mobilizing or doing certain activities, although they reported fewer fractures. Our reasoning behind this observation is that family overprotectiveness and past experiences with fractures made them restrict the child's activity to preserve the operated limb. This might warrant addressing parents' and patients' fears in follow-up visits. Reassurance and parents' education regarding the proven decrease in fractures postoperatively as well as a structured psychosocial assessment program may change and improve the overall quality of life and patient satisfaction.

Limitations

This was a retrospective descriptive study; thus, no statistical significance could be calculated. The limited number of patients in the study may not represent the population. Although questionnaire instructions were explained to the caregiver, it might not represent the patient's true state as many of our patients are still young and could not answer the questions themselves. Further larger prospective studies regarding the patients' quality of life are required to know the effects of the surgery on the quality of life.

CONCLUSION

Our study supports the previous evidence that this procedure is important in reducing the number of fractures and treating deformities in patients with OI. However, it is important to note that it does not improve the level of mobility, but it does maintain the level, the child was able to achieve. Therefore, it is important to address the patient and the family's expectations, to improve the quality of life.

AUTHORS' CONTRIBUTIONS

AR and YM: Conceptualization. AR: Initial draft preparation. AR, YM, and MO: Writing, review, and editing. All authors have critically reviewed and approved the final draft and are responsible for the manuscript's content and similarity index.

ETHICAL APPROVAL

Ethical approval code AFMS MREC005/2022 was obtained from the Forces of Medical Services Ethics Committee on 4th July 2022, in the Armed Forces Hospital, Muscat.

DECLARATION OF PATIENTS' CONSENT

The authors certify that they have obtained all appropriate patients consent forms. In the form, the patients' parents have given their consent for their images and other clinical information to be reported in the journal. The patients' parents understand that the patients' names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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CONFLICTS OF INTEREST

There are no conflicting relationships or activities.

REFERENCES

1. Monti E, Mottes M, Fraschini P, Brunelli P, Forlino A, Venturi G, *et al.* Current and emerging treatments for the management of osteogenesis imperfecta. Ther Clin Risk Manag 2010;6:367-81.
2. Marini JC, Forlino A, Cabral WA, Barnes AM, San Antonio JD, Milgrom S, *et al.* Consortium for osteogenesis imperfecta mutations in the helical domain of Type I collagen: Regions rich in lethal mutations align with collagen binding sites for

integrins and proteoglycans. Hum Mutat 2007;28:209-21.
3. Van Dijk FS, Pals G, Van Rijn RR, Nikkels PG, Cobben JM. Classification of osteogenesis imperfecta revisited. Eur J Med Genet 2010;53:1-5.
4. Georgescu VC, Gavriliu TŞ, Dan D, Pârvan AA. Surgical treatment in osteogenesis imperfecta 10 years experience. J Med Life 2013;6:205-13.
5. Hill CL, Baird WO, Walters SJ. Quality of life in children and adolescents with osteogenesis imperfecta: A qualitative interview based study. Health Qual Life Outcomes 2014;12:54.
6. Sofield HA, Miller E. Fragmentation, realignment, and intramedullary rod fixation of deformities of the long bones in children: A ten-year appraisal. Bone Joint J 1959;41:1371-91.
7. Rodriguez RP, Bailey RW. Internal fixation of the femur in patients with osteogenesis imperfecta. Clin Orthop Relat Res 1981;159:126-33.
8. Enright WJ, Noonan KJ. Bone plating in patients with Type III osteogenesis imperfecta: Results and complications. Iowa Orthop J 2006;26:37-40.
9. Li YH, Chow W, Leong JC. The Sofield-Millar operation in osteogenesis imperfecta. A modified technique. J Bone Joint Surg Br 2000;82:11-6.
10. Batur OC, Demir S. Our results of modified sofeild procedure in lower extremity deformities with osteogenesis imperfecta. J Bone Res 2017;5:182.
11. Abulsaad M, Abdelrahman A. Modified Sofield-Millar operation: Less invasive surgery of lower limbs in osteogenesis imperfecta. Int Orthop 2009;33:527-32.
12. Khoshhal KI, Ellis RD. Effect of lower limb sofield procedure on ambulation in osteogenesis imperfecta. J Pediatr Orthop 2001;21:233-5.
13. Khoshhal KI, Ellis RD. Functional outcome of sofield procedure in the upper limb in osteogenesis imperfecta. J Pediatr Orthop 2001;21:236-7.
14. Engelbert RH, Uiterwaal CS, Gerver WJ, van der Net JJ, Pruijs HE, Helders PJ. Osteogenesis imperfecta in childhood: impairment and disability. A prospective study with 4-year follow-up. Arch Phys Med Rehabil 2004;85:772–8.