CASE REPORT

Interdisciplinary Care Improves Functional Mobility in an Individual with Type IX Osteogenesis Imperfecta

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Introduction

Osteogenesis imperfecta (OI), in its typical form, is a genetic disorder of type I collagen. OI may be mild causing a few fractures to severe with hundreds of fractures in a lifetime. Characteristics in individuals with moderate to severe OI include short stature, multiple fractures, muscle weakness, joint laxity, skeletal deformities, hearing loss, curvature of the spine, blue sclera, and/or diffuse osteopenia which challenges their overall mobility [4, 9]. The traditional classification of OI by Sillence is a four-type subdivision [13]. Types I–IV are a heterogeneous group due to type I collagen gene mutations. Type I, the most prevalent with the lowest fracture risk, are community walkers. Type II is lethal in the perinatal period. Type III is moderate to severe with multiple fractures and progressive bone deformity, therefore typically limited to household ambulation. Type IV is between type I and III with moderate risk of bone deformities and fractures, but those affected are typically community walkers.

The revised OI classifications now identify at least eight types of OI, and research continues to identify more recessive forms [16,

17]. Types V through VIII are recessive forms, with mutations in genes other than COL1A1 or COL1A2; these types present with moderate to severe characteristics and are approximately less than 10% of all OI cases [7]. Type IX OI was recently identified as an autosomal recessive form due to a homozygous mutation in the PPIB gene [1]. Individuals with type IX present with clinical features similar to individuals with type III or IV but have radiographic features of a recessive form of OI including wide and/or flat bones and popcorn lesions in the growth plates [1].

Over the last decade, the interdisciplinary care of bisphosphonate treatment, surgical rodding, genetic testing, and rehabilitation has resulted in improved functional abilities in children with moderate to severe OI [4, 5, 7, 18]. Optimal care occurs with an interdisciplinary care approach working toward the goal of independence in self-care and mobility. Surgical osteotomy and intramedullary (IM) rod placement may be indicated for children with multiple fractures and long bone deformities. The goal of IM rodding is to provide internal stability, correct deformity, reduce fractures, and promote proper alignment throughout growth for weight bearing [4]. The Fassier and Duval telescopic growing rod technique has resulted in fewer surgeries, lower complication rates, less rod migration, shorter recovery, and improved mobility [4, 5, 10, 18].

General rehabilitation guidelines have been developed by the OI Foundation [9], Shriners Hospital for Children in Canada [4], Campbell [2], and Gerber [6]. Physical therapy (PT) is essential in facilitating family education, safe weight bearing, aerobic conditioning, and strengthening in a safe environment while maximizing gait and functional independence. Literature review yielded minimal published rehabilitation studies on individuals with OI. Caudill et al. [3] reported decreased ankle strength and functional limitation in individuals with type I versus healthy peers. Children with type I and IV OI also have reduced exercise capacity, muscle force, and overall cardiopulmonary function compared to typically developing peers [14, 15]. There are no case studies focused on rehabilitation and none specific to type IX OI. The purpose of this case report is to illustrate rehabilitation and functional mobility progression in an individual with type IX OI following bilateral femoral and tibia osteotomies with IM rodding over the course of the first two post-operative years.

Work performed at Hospital for Special Surgery.

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Case Report

The individual is a 6-year-old African-American female born full-term cesarean delivery with multiple fractures and diagnosed with phenotypic type III OI at birth. Over 35 fractures occurred from birth to 4 years of age with no other significant past medical history.

The child presented with no blue sclera, an older sibling with phenotypic IV OI, West Africa family origin, popcorn lesions, and wide flat bones on radiographic films. These clinical features suggested that the child had a recessive form of OI due to a different clinical presentation than a typical individual with type III OI. Therefore, family genetic testing occurred and concluded this individual and sibling to have type IX OI [1].

Parents reported that the child achieved developmental milestones of rolling at 6 months, sitting at 12 months, and crawling at 2 years of age. Early intervention PT services occurred in home from birth to 3 years. The child received intravenous pamidronate treatment from 2 to 4 years of age; however, once diagnosed with type IX OI, pamidronate treatment ended due to not meeting the treatment criteria. Radiographic films at 3.5 years showed lateral bowing of bilateral femurs (Fig. 1). She presented with $>30^\circ$ anterior tibial bowing bilaterally (right tibia/fibula shown in Figs. 2 and 3). Radiographic films of her spine, at 51 month, showed mild convex left mid-thoracic scoliosis, loss in lumbar vertebral body height, and osseous structures diffusely osteoporotic consistent with history of OI. At 3.5 years, her height was 89 cm (third percentile) and weight of 13.2 kg (eighth percentile) with dexa scan Z score of lumbar spine -3.9 and overall body -2.9 indicating osteoporosis.

Tests and Measures

The individual in this case report was followed for 2 years post-op. Changes in mobility were measured at preoperative,

Fig. 1. Preoperative AP FROG lateral of pelvis and hips at 3.5 years of age. Radiographic films demonstrate that osseous structures are diffusely osteoporotic and the long bones are diffusely gracile consistent with diagnosis of OI. There are healed fractures of the proximal shafts of bilateral femurs with lateral bowing of the femurs: $\sim 60^\circ$ on left and 30° on right.

Fig. 2. Preoperative lateral right tibia radiographic film with mid-shaft-healed fracture and anterior bowing of 30° .

initial post-op PT evaluation, 6 months, and 2 years post-op by the Gross Motor Function Measure-88 (GMFM-88) [11, 12]. The GMFM has excellent interrater reliability in children with OI [11] and was chosen due to the ability to test functional mobility progression in five dimensions (rolling, sitting, crawling, standing, and walking). Gait progression was also evaluated by the Gillette Functional Assessment Questionnaire Ambulation Scale (FAQ) [8]. The FAQ is a parent-reported 10-level scale used to describe walking ability from unable to take steps (0) to ambulating on various surfaces and running (10). All measurements throughout course of PT were tested by the same therapist.

Initial PT Evaluation

The child was referred to outpatient PT, at 3.5 years of age. PT orders included AAROM/AROM, strengthening, and a standing program to progress to gait with hip-knee-ankle-foot orthoses (HKAFO) with an anterior tibia shell. At the PT evaluation, the child was nonambulatory, dependent for transfers and self care, side scooted on bottom in home, and used a stroller in the community as her mode of mobility. The parent's goal was for their daughter to walk. After

Fig. 3. Preoperative anteroposterior right tibia radiographic film view.

4 months of PT, she was walking 30 ft with HKAFOs unlocked at hips and locked at knees with a rolling walker (RW) and contact guard assist (CGA) in therapy only. She needed maximum assistance (max A) for transfers to/from stroller. Her primary mode of mobility in the home was hands and knees crawling and a stroller in the community. Her total GMFM score was 54%. At 46 months of age, a left femur fracture occurred, and 6 weeks after cast removal, she fractured her right femur which was treated with closed reduction and spica cast prior to orthopedic surgical intervention.

Surgical Intervention

At 4 years of age, the child underwent a two-staged surgery for bilateral femoral and tibia bowing deformities and fractures. Surgical intervention included right femur and tibia osteotomies with placement of Fassier-Duval telescoping IM rod. She was placed in a spica cast following surgery with right hip flexion for fracture healing. Two weeks later, the child underwent left femur and tibia osteotomies with rodding. Following the second surgery, bilateral long-leg casts were chosen over a spica cast to allow use of upper extremities (UE's) and trunk for mobility. The child received 2 days of inpatient PT for family education on positioning and transfer training following each surgery. The individual was in casts and nonweight bearing for a total of 8 weeks.

Postoperative PT

Outpatient PT resumed at 8 weeks post-op, and orders included AAROM, strengthening, and standing weight bearing as tolerated in bilateral knee-ankle-foot orthoses (KAFOs). PT evaluation found diminished AAROM of bilateral knee flexion and right hip abduction from preoperative ROM. Overall strength was fair in upper extremities (UE's) and poor in trunk and lower extremities (LE). Following surgery, she was nonambulatory and dependent with self-care and transfers (Table 1). GMFM total score was 35% (Table 2).

PT Intervention

Physical therapy occurred two to three times a week for 45 min for one and a half years post-op through Medicaid insurance coverage. Once the child started school, PT was decreased to once a week. Precautions included no PROM, distal resistance, or torsional forces due to fracture risk. Child required CGA to close supervision for safety during all sessions. The family and child were involved in setting PT goals, educated on safe transfers and mobility, fall prevention, and home exercise program on a regular basis. The child had age-appropriate cognition, language, and social

Table 1 Functional mobility changes

	Preoperative	Postoperative 8-week PT evaluation	Postoperative 6 months	Postoperative 2 years
Age (years)	3.9	4	4.5	6
Ambulation	30 ft with HKAF0, RW, and supervision	Nonambulatory	≥150 ft with KAFO unlocked, RW, and supervision	100 ft independent; >300 ft with HAFO, RW and supervision
Gillette functional assessment questionnaire walking scale [8]	4	1	6	6
Sit to stand from 12" chair	Minimum assist	Maximum assist	CGA with UEs	Independent
Stairs	Seated scooting up/down two stairs on bottom with UEs, CGA	Dependent	Four stairs nonreciprocal, with two rails and minimum assist	One flight nonreciprocal, with one rail and CGA

HKAF0 hip-knee-ankle-foot orthoses, KAFO knee-ankle-foot orthoses, HAFO hinged-ankle-foot orthoses, RW rolling walker, UEs upper extremities, CGA contact guard assist

Table 2 Gross motor functional mobility scores (GMFM)

GMFM categories	Preoperative with HKAFOs	PT evaluation postoperative	6 months postoperative with KAFO	2 years postoperative with solid AFO
Lying/rolling	100%	96%	100%	100%
Sitting	97%	80%	97%	100%
Crawling/kneeling	64%	0%	93%	100%
Standing	8%	0%	26%	82%
Walking, running, jumping	3%	0%	19%	53%
Total score of five categories out of 100%	54%	35%	67%	87%

HKAFOs hip-knee-ankle-foot orthoses, *KAFO* knee-ankle-foot orthoses

Table 3 Postoperative rehabilitation mobility progression

Time from postoperative PT evaluation	Functional mobility/therapeutic exercise
0–2 weeks	LE AAROM; quadruped over pillow or bolster with assist
2 weeks	Static bench stance with KAFOs locked at knee, minimum assist, UEs
4 weeks	Against gravity LE AROM; tricycle; prone to quadruped transitions; static stance at bench; gait 5–10 ft, RW, KAFOs, and minimum assist
6 weeks	Gait 75 ft with RW, KAFO, CGA; static stance with unilateral UE reaching at bench; floor long sit to 6" bench transfers with UEs and minimum assist
8 weeks	Crawling forward independent; backward crawling for strengthening; standing and gait activities (forward/backward) with RW or at bench with KAFOs and minimum assist
10 weeks	Good bilateral quadriceps strength progressed to stance and cruising with intermittent KAFOs unlocked: transitioning quadruped to tall kneeling independent
3–4 months	Gait 150 ft with RW, KAFOs unlocked and contact guard assist
4–5 months	Independent floor to 6–12" bench transfers; sit to stand from 12" surface with UEs; gait with RW as primary mode of mobility home and therapy
5–6 months	Four stairs nonreciprocal with two rails and minimum assist side stepping; gait 150 ft and stepping over 1 to 2 in. objects with RW, KAFOs, and close supervision
6–9 months	Gait 150–300 ft with RW, solid AFOs, and supervision; sit to/from stand independent
9 months–1 year	Standing independent 1–2 min; gait 300 ft RW; squat to/from stand independent
1–1.5 years	Static stance >5 min independent; dynamic stance for catch and kicking with minimum assist or hand held assist; gait with posterior RW >300 ft, gait in home independent without assistive device
1.5–2 years	Gait independent at home, PT, and classroom without assistive devices; gait with RW in school hallways; stroller for community; independent with: floor to stand transfers with UEs, squat to stand, static catch and kicking, step over 2–4 in. objects hand-held assist

AAROM active assistive range of motion, *KAFO* knee-ankle-foot orthosis, *UE* upper extremity, *LE* lower extremity, *RW* rolling walker, *CGA* contact guard assist, *PRW* posterior rolling walker, *RW* rolling walker

skills. Therefore, verbal cues were used during rehab for proper alignment, and she was able to express her level of fatigue. As a preschooler, she participated in educating others on assisting with braces, transfers, and gait to prepare for school. Activities were advanced as tolerated by the child's feedback and visual compensation patterns observed by her PT.

Functional Mobility Progression

The PT progression of functional mobility is briefly outlined in this section and in more detail in Table 3. Initial PT focused on parent transfer training and AAROM of LEs. A standing program was initiated at 2 weeks and full bilateral LE AAROM was achieved at 4 weeks. Rehabilitation during 4 to 8 weeks focused on weight shifting in quadruped/crawling, prone UE bolster walk outs, and gait with KAFOs locked. The child was referred to occupational therapy for additional UE strengthening, ADL/self-care, and school preparation activities. At 8 weeks, crawling became her primary mode of mobility in the home, and she was walking 100 ft with RW, KAFOs locked, and CGA. At 10 weeks, gait progressed to an intermittent unlocked knee in the KAFOs with improved quadriceps strength. After 4 months of PT, she achieved floor to 6" bench and sit to stand transfers with use of UEs independent and tolerated 30 to 45 min of standing/walking. Stairs were introduced by the child's request at 5 months due to living in a walk up apartment. By 6 months, the child was walking ≥150 ft with supervision, an RW, and KAFOs unlocked. She transitioned to solid AFOs at 9 months post-op. Between 9 months and 1.5 years post-op, the child's PT included LE and trunk strengthening, gait on uneven surfaces, balance, and stair training.

The child continued to improve her walking endurance, strength, and balance by 1.5 years post-op, and the child was walking in the home independently and up to 300 ft with an RW, AFOs, and supervision at school and outdoor level surfaces. She transitioned to hinged AFOs with an anterior shell at 2 years post-op. A posterior rolling walker was issued for the child to use at school and community to promote upright posture and less reliance on UEs during gait. Throughout the course of PT, functional mobility and gait improved by strengthening of UEs, LEs, and trunk musculature through developmental play. Muscles of focus were included but were not limited to hip extensors, hip

abductors, trunk, and knee extensors to achieve independent stance and gait. See Table 4 for therapeutic exercises and treatment techniques used during rehabilitation.

Outcomes

The child in this case report, a 6 year old with type IX OI, demonstrated significant improvements in strength, ambulation, and functional mobility with fewer incidents of fractures over 2 years post-op bilateral femoral and tibia rodding with rehabilitation (Tables 1, 2, and 3). The child's overall total body DEXA scan Z score improved from -2.9 to -0.7 post-op. Her mobility status improved from only walking in PT with HKAFOs to walking independent in her home and school. On the GMFM, she demonstrated the following improvements: 36% in crawling/kneeling, 74% in standing, and 50% in walking categories with an overall total GMFM score change of 33% from preoperative status (Table 2). Standing hip to ankle AP radiographic films demonstrates improved femora and tibia alignment bilaterally post-op (Fig. 4).

Table 4 Developmental strengthening activities utilized

Trunk Strengthening:

- Long sit scooting forward and backward on bottom (without UEs) for weight shifts to activate abdominals
- Supine bilateral hand to feet ball pass
- Quadruped (initially over bolster), progress to UE unilateral reaching, and prone walk outs over bolster with assist at trunk
- Straddle sitting on bolster or firm playground ball 180–360° walk around with feet on floor and knees at 90° to encourage leg/trunk dissociation and trunk stabilization
- Sitting on various surfaces for postural reactions and activate abdominals during activities such as catching at ball or elevating legs or arms to catch beads or ring

Knee Extension Strengthening:

- Quad sets with progress to short arc quads over bolster, straight leg raises, and sitting terminal knee extension exercises while playing game/touching a target or ball
- Tricycle for strength and endurance
- Platform swing/scooter board knee flexion/extension activities
- Sit to stand from various heights and squat to stand activities

Hip Strengthening:

- Hip adduction, abduction, extension, and internal rotation were initially performed by antigravity AAROM exercises with progression to AROM against gravity
- Hip abduction strengthening progressed to against gravity in side lying; side stepping in quadruped, side scooting while seated on bolster, tall kneeling activities, and side stepping with RW or along a wall
- Hip extension strengthening included gluteal squeezes, prone over inclined ramp with bilateral UE reaching over head for gluteal co-activation, crawling and walking backward, and quadruped leg extension

Upper Extremity (UE) Strengthening and Endurance:

- UE strengthening and endurance was initially performed by AROM reaching against gravity during games in all planes of motion
- Endurance promoted by maintaining shoulder flexion during games with bilateral UE reaching overhead such as throwing bean bags at target, dress up, and ADLs
- Weight bearing activities in prone, quadruped, and walkouts over bolster with assist at trunk and/or verbal cues for proper alignment
- Once walking with RW progressed to open chain UE work such as pushing chair or shopping cart with UEs
- Prone net swing with UE reaching overhead and pulling rope

Fig. 4. Post-operative 9 months standing hip to ankle anteroposterior radiographic film view demonstrates healing fractures and improved alignment after bilateral femora and tibia osteotomy and intramedullary rod placement.

By 2 years post-op, the child achieved independent functional mobility in rolling, crawling, sitting, quadruped, sit to stand, static standing balance, and one half kneel to stand through right LE. Her LE functional strength improved allowing her independence with the ability to stand without use of UEs to play catch, kick a ball, walk backward ten steps, and pick up objects from floor through a squat to stand transition. She also achieved her goal of climbing a flight of stairs into her home. However, her primary source of mobility in the community remained an adaptive stroller due to fatigue and safety in her urban environment. The GMFM total score increased 33% over 2 years from 54 to 87% (Table 2). The most dramatic changes occurred between 6 months and 2 years post-op with a 56% improvement in standing and 34% in walking categories. The child's level of walking, evaluated by the FAQ changed from a level 1 to level 6 (walks more than 15–50 ft outside the home, stroller in community) throughout the course of rehabilitation.

Discussion

This case report demonstrates positive functional mobility outcomes with the current interdisciplinary team approach in

a 6 year-old child with type IX OI. Similar findings of improved gait and functional mobility GMFM scores have been reported by Ruck et al. in individuals with type I, III, and IV OI after IM rodding [10]. The child in this case report continues to be followed by her interdisciplinary team, including PT, for optimum care. Although this child made vast progress in her overall mobility, she presents with impaired gross motor skills, quality of movement, gait impairments, and functional strength compared to healthy peers. Due to her diagnosis of OI, this individual continues to be at risk for fractures and will likely re-fracture multiple times in her lifespan. Over the 2 years post-op, she had four fractures from falls (radius, ulna, clavicle, and tibia). In the future, she will likely require a rod revision surgery and will benefit from continued scoliosis monitoring. She also will benefit from PT throughout her lifespan to encourage a safe exercise program, promote aerobic and muscle strengthening, and achieve her highest level of functional mobility [4]. In the future, a wheelchair may also be beneficial to allow more independence in her school and urban environment.

Additional case reports and clinical trials are needed to advance the field of rehabilitation for individuals with OI. This is the first documented case report on an individual with type IX OI and the first case report to focus on rehabilitation in an individual with OI. Therefore, a follow-up case report on this child could be beneficial to document long-term function mobility outcomes in individuals with type IX OI. This case report provides an example of post-op rehabilitation activities and mobility progression for other clinicians. Each individual's rehabilitation program and outcomes will vary due to their severity of OI; therefore, an interdisciplinary team approach is ideal for safe and optimal mobility progression.

Disclosures

Conflict of Interest: Lisa C. Drefus, PT, DPT, Sandra Cassady, PT, PhD, FAACVPR, and Cathleen L. Raggio, MD, have declared that they have no conflict of interest.

Human/Animal Rights: All procedures followed were in accordance with the ethical standards of the responsible committee on human experimentation (institutional and national) and with the Helsinki Declaration of 1975, as revised in 2008 (5).

Informed Consent: Informed consent was waived from all patients for being included in the study.

Required Author Forms Disclosure forms provided by the authors are available with the online version of this article.

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