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Effect of physical therapy regimen on the recovery of individual with Osteogenesis Imperfecta: A Case Report

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Abstract

Brittle bone disease otherwise known as Osteogenesis Imperfecta(OI).Reduced bone quality following excessive bone fragility is involved in the genetic disorders like Osteogenesis Imperfecta(OI).Osteopenia, recurrent fractures, progressive deformity, loss of mobility, and chronic bone pain are some characteristics of severe osteogenesis imperfecta. In patients with OI Post operative fracture treatment is challenging and disappointing fixation of implants due to altered bone quality is resulted. In this case report we are presenting a case of 17year old boy with femur shaft fractured secondary to osteogenesis imperfecta who came to Dr. D.Y.Patil College of Physiotherapy (DYPCPT) for physiotherapy treatment. Patient came with the complaint of difficulty in standing, walking since 2years which was aggravated last week. Patient also had weakness in lower limbs and pain in both knee joints since last 2years. The patient had a past medical history of osteogenesis imperfecta. Proper assessment of range of motion (ROM), manual muscle testing (MMT) and pain (VAS) was taken before and after the treatment. The treatment was focused on managing pain, correcting the deformity, strengthening of the muscles and improvement of patient's quality of life. This concludes that the patient's mobility and muscle strength in the lower limb muscles were enhanced with physiotherapy.

Keywords: Brittle bone disease, Physical Therapy, Rehabilitation, Case Report.

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Introduction

Reduced bone quality following excessive bone fragility is involved in the genetic disorders like Osteogenesis Imperfecta(OI).Formation and strength of the bone also the structure of the tissue is affected by the gene mutation which causes OI[1]. The clinical classification of Sillence et al. well-founded about prognosis and genetic counselling. There are four different catagories of OI: Type I: mild, Type II: perinatal lethal, Type III: progressively deforming, and Type IV: moderate deforming. Therefore, occurrence of fracture is greater in OI patients as compared to healthy individuals. In type I, sometimes fractures occur before puberty and have small amount of deformity with normal stature. In type II, perinatal death caused by pulmonary insufficiency due to fractures before delivery. In type III OI, more severe deformity with short stature while in last type the disorder is progressive, with increasing deformity of the limbs and spine, dependence on others for help in walking, and chronic pain[2]. The prevalence of OI has increased over the year now it is 1 in 10,000 to 1 in 20,000, early it was 1 in 40,000[3–5].

In this case report we are presenting a case of 17 year old boy with both the knees fractured secondary to osteogenesis imperfecta who came to Dr. D.Y.Patil College of Physiotherapy (DYPCPT) for physiotherapy treatment.

Patient and observation

The boy was coming for physiotherapy treatment at DYPCPT with the complaint of difficulty in standing, walking since 2 years which was aggravated last week. Patient also had weakness in lower limbs and pain in both knee joints since last 2 years. The individual is known case of osteogenesis imperfecta.

Clinical findings: The patient was assessed in the supine lying position. The patient was vitally stable. On observation, the patient had an ectomorphic build. The patient had more pain in the morning as compared to evening and the pain was graded on Numerical Pain Rating Scale (NPRS) as 8/10 on activity and 6/10 at rest. On observation flexion deformity in both the knees can be easily marked. On palpation grade 2 tenderness and swelling was present over right knee joint.

Table 1: Postural assessment before physiotherapy treatment

	Anterior view	Lateral view	Posterior view
Head	Tilted to the right, left earlobe elevated	Forward head posture	Tilted to the right
shoulders	Right shoulder elevated	protracted	Right shoulder elevated
spine	-	-	-
hip	Right hip elevated	-	Right hip elevated
knees	Genu varum	flexed	Genu varum
ankle	everted	-	everted

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Table 2: Rang of motion before physiotherapy treatment:

Movement	Pre	
	right	left
Hip flexion	0-60	0-60
Hip extension	0-20	0-20
Hip abduction/adduction	Can not perform	Can not perform
Knee flexion	20-140	16-145
Knee extension	140-20	145-16
Dorsiflexion	0-15	0-15
Plantarflexion	0-30	0-30

Table 3: Manual muscle testing before physiotherapy treatment:

Hip flexors	3/5
Hip extensors	3/5
Knee flexors	2/5
Knee extensors	2/5
Ankle dorsiflexors	3/5
Ankle plantarflexors	3/5

Table 4: Limb length assessment table:

	right	Left
Greater trochanter to lateral femoral condyle	45cm	41cm
Lateral femoral condyle to lateral malleolus	37cm	36.5cm

Timeline

The patient was normal till 2018 when he started experiencing pain and weakness in lower limbs while walking and stair climbing. In June 2019 he had a fall from stairs so went to hospital and diagnosed with hairline fracture of left 1/3rd shaft of femur. It was managed conservatively. Again in October 2019 he had fall from stairs and got fractured with right femur shaft which was operated by k-wire fixation. Further in January 2021 k-wire was removed as it was painful during knee movements. In December 2021 he was diagnosed with osteogenesis imperfecta, for which he had four doses of injection Zoldonat with the break of four months in between. From 4th November 2022 he is coming to DYPCPT for physiotherapy treatment.

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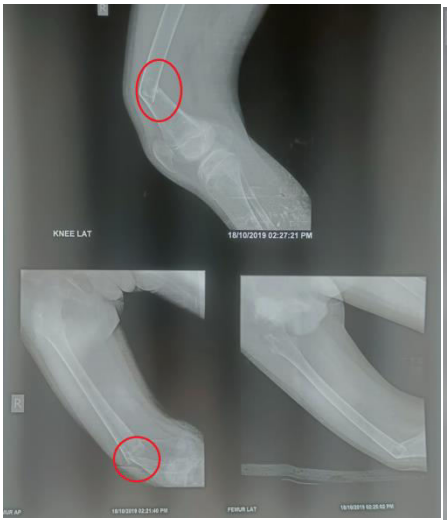

Fig:1 Pre operative x-ray of fracture of right femur shaft

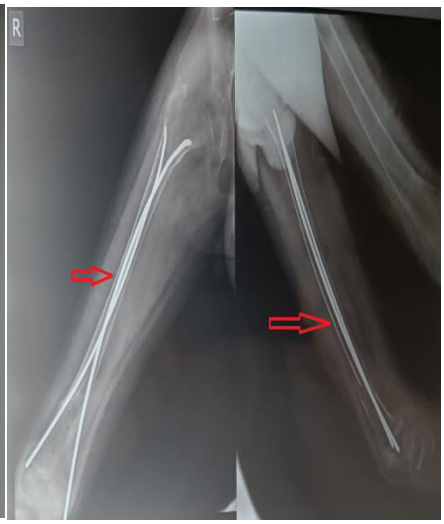

fig:2 Post operative x-ray of fracture of right femur shaft

Diagnosis:
Right femur shaft fracture secondary to osteogenesis imperfecta.

Physiotherapy management:
The patient and relatives were educated about the condition and physiotherapy on better and fast recovery. The treatment will be focused on managing pain, correcting the deformity, strengthening of the muscles and improvement of patient's quality of life. The detailed management protocol is mentioned in the table below.

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Table 4: detailed physiotherapy management protocol[6,7].

Intervention	Dosage	Rationale
Phase-I: Non Weight Bearing Phase		
Cryotherapy	10 minutes, thrice daily	To improve blood flow to the affected part and reduce pain
Active Range of Motion for Bilateral Upper and Lower limb	10 repetition, twice daily for each joint	To maintain and improve the range of motion.
Isometric exercises for quadriceps hamstrings and glutei	10 repetitions, 5 seconds hold, twice daily	To prevent muscle atrophy and maintain contractility of muscle
Pre crutch training	10 repetition, twice daily	To prepare individuals for ambulation using walker
Phase - II: Weight bearing phase		
Weight Bearing as tolerated for affected lower limb	Thrice daily 6 minutes daily with 1 minute interval after every 2 minute	To promote fracture healing, proprioceptive training to improve joint position sense and balance.
Ambulation using walker	Thrice daily 100 meters per round	To prevent side effect of bed rest, to improve cardiovascular endurance.

Follow up and outcome measures of intervention:

After proper one month of rehabilitation the patient's range of motions i.e both active and passive were improved at the time of discharge. By values of pre and post manual muscle testing an increase in muscle power was observed. After the physiotherapy treatment patient had pain 3/10 on activity and 0/10 at rest on NPRS.

Table 5: comparison of pre and post Range of motion

	Pre Treatment		Post Treatment	
	right	Left	right	left
Hip flexion	0-60	0-60	0-67	0-70
Hip extension	0-20	0-20	0-25	0-27
Hip abduction	Can not perform	Can not perform	0-10	0-10
Knee flexion	20-140	16-145	15-140	10-145
Knee extension	140-20	145-16	140-15	145-10
Dorsiflexion	0-15	0-15	0-15	0-15
Plantarflexion	0-30	0-30	0-30	0-30

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Table 6:Comparison of Pre ans post manual muscle testing.

Muscles	Pre	Post
Hip flexors	3/5	4/5
Hip extensors	3/5	4/5
Knee flexors	2/5	3/5
Knee extensors	2/5	3/5
Ankle dorsiflexors	3/5	4/5
Ankle plantarflexors	3/5	4/5

Patient perspective

After successful surgery we have initiated physical therapy and medical therapy, a well-planned physical therapy was initiated once a day which improved mobility of hip, knee and ankle with improved strength in the lower limbs.

Informed consent

Informed consent was taken from patient and his parents.

Discussion

Children accompanied by OI are studied by distinct authors to compare outcomes between different treatments.Outcomes between surgical and conservative treatments of 29 children with long bone fragility fractures is compared by Chiarello et al.. They have described a slightly lower non-union and delayed-union rates under surgical treatment[8,9]. Enright and Noonan reported high complication rates resulted by femoral and tibial fractures with bone plating in four children with type 3 OI[10]. Agarwal and Joseph and Gamble et al. have found over a 10-14 years period, 15%-20% prevalence on non-union fracture in a heterogeneous group of children with OI[11–13]. Although, the number of patients is small, and the results could not be extrapolated to adults, whereas it is 7.3 times higher non-union rate compared with adults with non-OI than in children with non-OI[14–16].

In this study we have studied a case of 17 year old boy with hairline fracture of left 1/3rd shaft of femur which was managed conservatively and right femur shaft fracture which was operated by k-wire fixation secondary to osteogenesis imperfecta. After the surgery proper physiotherapy was given. Rehabilitation resulted in improved functional activities of the patient.

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Conclusion

Physiotherapy improved the patient's mobility and muscle strength in the lower limb muscles. The prognosis had eased at the end of the session. At the time of the discharge the patient as well as the doctor were pleased with the outcome at the time of discharge. If the patient or his family had been enlightened about the need for physiotherapy before any surgery the patient would have been more better served.

Competing interest

All Author Declare no Competing Interests

Authors Contribution:

All Authors Contributed Equally in drafting the manuscript

Figures:

Fig 3: patient in standing position

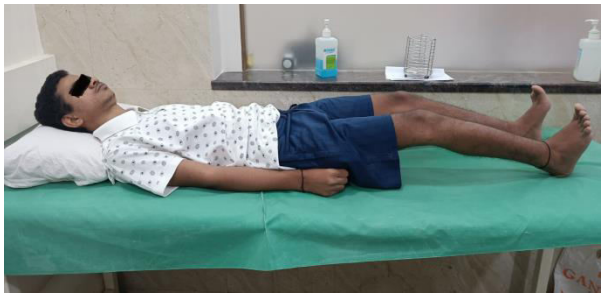

fig 4: patient in lying position

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