

## Effectiveness of Physical Therapy and Rehabilitation on Specific and Rare Form of Osteogenesis Imperfecta: A Case Report for Type VI

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**Abstract:** A child patient who was 14 years old was admitted to our clinic with the diagnosis of type VI Osteogenesis Imperfecta (OI). Although different treatments have been defined for OI, there are few studies related to effectiveness of physical therapy and rehabilitation on type VI. In this case, we presented that physical therapy and rehabilitation may be an integral part of treatment for patient with type VI OI.

### 1. INTRODUCTION

Osteogenesis imperfecta (OI) is a genetic disorder of increased bone fragility and low bone mass.<sup>1</sup> There are some gene alterations such as COL1A1 and COL1A2 in patients with OI.

Because the etiopathogenesis of OI is based on alterations in the metabolism of type I collagen, these alterations related to COL1A1 and COL1A2 aggravate disorders of type I collagen. This condition causes more susceptibility to bone fracture, skeletal deformity and various growth problems.<sup>2</sup>

Despite the various classifications of OI, the most frequently used classification scheme for OI was developed by Silience et al.<sup>3</sup> There are four main types of OI according to this scheme: I to IV.<sup>3</sup> This classification system is continually modified according to new phenotypes and ranges from 4 to 11 types.<sup>3</sup>

In all these types, type VI OI was described as a separate entity in 2002 according to clinical, histological and molecular grounds.<sup>4</sup> Its unique bone histological feature of increased unmineralized osteoid indicating a mineralization defect is a major factor in diagnosis of Type VI. This condition causes mass bone fractures compared to other types. Bone fractures of type VI significantly affect the functionality of the individual. For this reason, keeping functionality of the individual at the top level is important for the daily living activities of the patient. In this case, functional and muscle strength changes of a patient with type VI OI after physical therapy and rehabilitation were presented.

### 2. CASE PRESENTATION AND METHODS

A child patient who was 14 years old was admitted to our clinic with the diagnosis of type VI OI. Written and

verbal consent was obtained from the patient and his family for the study. Short height according to his peers, right thoracal left lumbar S scoliosis, malformations in the femur's corpus and humerus's crista and tight iliopsoas muscle were detected on physical assessment of patient. The patient's ambulation was providing with wheelchair by the family. He received 56 points from the Functional Independence Measure (FIM) scale, which was used for the determination of the functional status, with the motor score subscale was 21 and the cognitive score subscale was 35. He received 21 points from the 36-Item Short Form Survey (SF-36) scale which was used for assessment of daily living activities. The grip strength was determined with hand-held dynamometer. It was found that dominant right hand grip strength was 1,1 kg. and left hand grip strength was 1,0 kg. He received 0 point from the Functional Ambulation Classification (FAC) used for assessment of ambulation. In our physical therapy and rehabilitation programme, we used pulsed electromagnetic field treatment of 100 Gauss intensity and 50 Hz. frequency once per a week for six weeks.<sup>5</sup> It was thought that electromagnetic field treatment can be effective for the facilitation of bone regeneration process.<sup>5</sup> Isometric knee extension exercises were preferred for malformations of the femur's corpus. Because concentric or eccentric contractions of muscles can lead to fractures of bone as a result of overloading. Isometric knee extension exercises were repeated 100 times per session. For femur head dysplasia, femur head centralization applications were performed manually. Neuromuscular electrical stimulation (NMES) was used to increase quadriceps femoris muscle strength. Since it was thought that scoliosis might restrict respiratory activities, therapeutic positional kinesiotaping was applied to the patient for providing upright posture in per session. In addition, torsional and diaphragmatic respiratory exercises for scoliosis were performed and repeated frequently in daily living activities. The physical assessments were made with three-month intervals in 1 year treatment. Baseline and follow-up scores of patient were shown in Table 1.

**Table 1.** Baseline and follow-up scores of patient.

Outcome Measures	Baseline	Third month	Sixth month	Ninth month	Twelfth month
Motor FIM	21	33	33	34	35
Cognitive FIM	35	35	35	35	35
SF-36	21	22	24	25	26
FAC	0	0	0	0	0
Right HHD	1.1	1.4	1.6	1.9	2.2
Left HHD	1	1,2	1.5	1.8	2

FIM: Functional Independence Measure , SF-36: 36-Item Short Form Survey, FAC: Functional Ambulation Classification, HHD: Hand-Held Dynamometer

### 3. DISCUSSION

It has been determined that the scores of motor FIM, SF-36, right and left hand-held dynamometer can increase although changes of this outcome measures are not high according to results of this study. No increase in cognitive FIM and FAC of the patient is consistent with the clinical features of type VI OI.

Three types of treatment are available for OI in nowadays. Non-surgical management is composed of physical therapy and rehabilitation, bracing and splinting. Surgical management is consisted of intramedullary rod positioning, spinal and basilar impression surgery. Medical therapy, such as bisphosphonates or growth hormone alternatives, is used to increase the strength of bone and to decrease the number of fractures.<sup>6</sup> Although different treatments are defined, it has been highlighted that multidisciplinary treatment is important for reaching maximum function.<sup>6</sup>

As for physical therapy and rehabilitation, it can be used to increase motor function for patient with OI.<sup>7</sup> Studies on physical therapy in OI are few. A 4-year follow-up study of rehabilitation in OI showed an increase in self-care and social function with all types of OI but mobility level plateaued in moderate-severe OI.<sup>8</sup> The aim for a child patient with severe type of OI is to reach independence in daily living activities as possible as.<sup>9</sup>

Although multidisciplinary treatment is highlighted, medical therapy have been performed for patients with type VI OI. In medical therapy, denosumab may be an effective method for type VI.<sup>10</sup> Intravenous pamidronate seems to be less effective in type VI OI.<sup>11</sup> In contrast to intravenous pamidronate, patients with type VI OI who received intravenous bisphosphonate treatment during growth may have an increase in lumbar spine areal bone mineral density, a higher final height z-score and presented some reshaping of vertebral bodies.<sup>12</sup>

In addition to medical treatment, we think that physical therapy and rehabilitation should be highlighted. Our study may be important about this topic. Further studies are required to investigate efficiency of physical therapy and rehabilitation for type VI OI.

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