CASE REPORTS

OSTEOGENESIS IMPERFECTA IN A WEIGHTLIFTER

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ABSTRACT

Objective: To discuss the case of a 42-yr-old weightlifter with osteogenesis imperfecta.

Clinical Features: The patient had bilateral acute elbow pain and a previous history of more than 35 fractures of the spine and extremities.

Intervention: There is no current treatment for osteogenesis imperfecta. Treatment objectives were designed to minimize pain, improve range of motion, and decrease stress on the elbow joints. Nutritional supplementation was used to help maintain bone density. The elbow pain improved with treatment, and the patient has had no new fractures in the last 6 years.

Conclusion: Although most patients with osteogenesis imperfecta are physically inactive because of the high risk of fracture, some patients with milder forms of the condition may be involved in some athletic activities. Although manipulation is contraindicated in patients with osteogenesis imperfecta, chiropractors may be of service by offering pain relief and rehabilitation, in addition to advice regarding nutrition and supplements. (J Manipulative Physiol Ther 2002;25:334-39)

Key Indexing Terms: *Osteogenesis Imperfecta; Wheelchair Athletics; Weightlifting; Chiropractic*

INTRODUCTION

Osteogenesis imperfecta is a genetic mutation affecting the quality and quantity of collagen production. A hallmark finding in patients with osteogenesis imperfecta is an abnormally fragile skeleton susceptible to fracture with minimal trauma. The following is a case report of a 42-year-old man with osteogenesis imperfecta who has had more than 35 fractures of his spine and extremities. Despite his disability, he has accomplished extraordinary achievements in the field of weightlifting. In 1991, he won the National Wheelchair Athletic Association Championship in Oklahoma; in 1993, he was the Stoke-Mandeville Games Champion in England; and in 1994 and 1995, he was the USA National Wheelchair Athletic Association Champion in Boston. In 1991, at a body weight of 119 lbs, he set a national record at the US National Wheelchair Athletic Association Championships by bench pressing 3 repetitions at 270 lbs.

CASE REPORT

A 42-year-old African American man previously diagnosed with osteogenesis imperfecta tarda (type I) was seen as a patient by one of the authors (RES). No family history (including both parents, 4 brothers and 1 sister) of osteogenesis imperfecta or other congenital anomalies was present. He was able to stand and walk with assistance devices or supports; however, his ambulation was limited by intense hip and knee pain, which developed after several minutes of weight-bearing (Fig 1 , *A* and *B*). He used a wheelchair and was able to drive a car by using hand controls. His history included at least 35 fractures of both the spine and extremities. His chief complaint was acute elbow pain, bilaterally.

Physical Examination

The patient was 46 inches tall and weighed 118 lbs. His skull was conical-shaped, characteristic of the deformity

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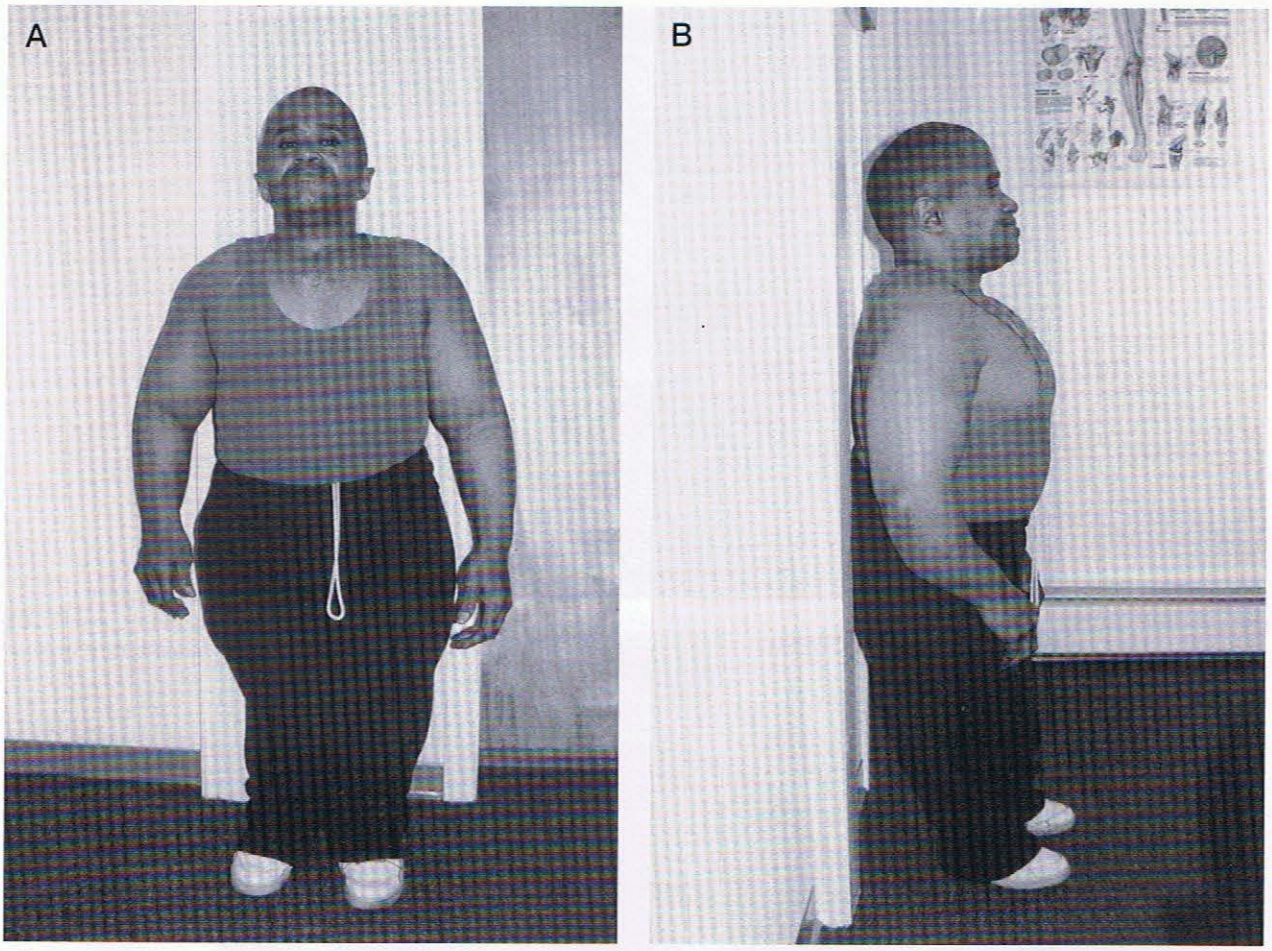

Fig 1. A, *Front and* **B,** *side view of patient with osteogenesis imperfecta, showing short stature and significant muscular development of the patient's thorax and upper extremeties.*

found in osteogenesis imperfecta. His eyes showed the classic blue sclera, and he required corrective contact lenses for vision. He had abnormal dentition and normal hearing. He was unable to perform heel to toe walk as a result of deformation of the foot from the underlying bone disease. Examination of the elbows showed the inability to fully extend and lock the elbows.

Radiographic Examination

Radiographs of both elbows showed osseous deformity and evidence of previous multiple fractures. The elbow articulation at both the ulnar and radial side showed deformity and disorganization, which was consistent with the patient's inability to fully extend both his elbows (Fig 2 , *A* and *B*). Radiographic assessment of the knees showed osteoporosis and severe bowing deformities of both femurs. Stabilizing pins were placed in both femurs (removed in 1985) and in the tibia, bilaterally (still in place and noted on radiographic examination, Fig 3). There was severe deformity of the pelvis, with bilateral protrusio acetabulae. Both iliac wings showed contour deformity. The proximal femora were bowed, with bilateral coxa vara deformity of the femoral necks (Fig 4).

Treatment

With the understanding that there is no treatment for osteogenesis imperfecta, an attempt to help the patient with his current symptoms was initiated. A course of therapy, consisting of cross-friction massage, ice to reduce pain and swelling, and a change in his training routine to reduce pressure on the elbows, was instituted.

Seated pressing exercises were added to balance and complement his development. Nutritional supplementation with microcrystalline hydroxyapatite concentrate, calcium, magnesium, and protein was instituted to help support his bony structures.

Prognosis

Aside from making excellent gains in his competitiveness, there has been a reduction in musculoskeletal pain and injury (such as strain-sprain) and no new fractures during the

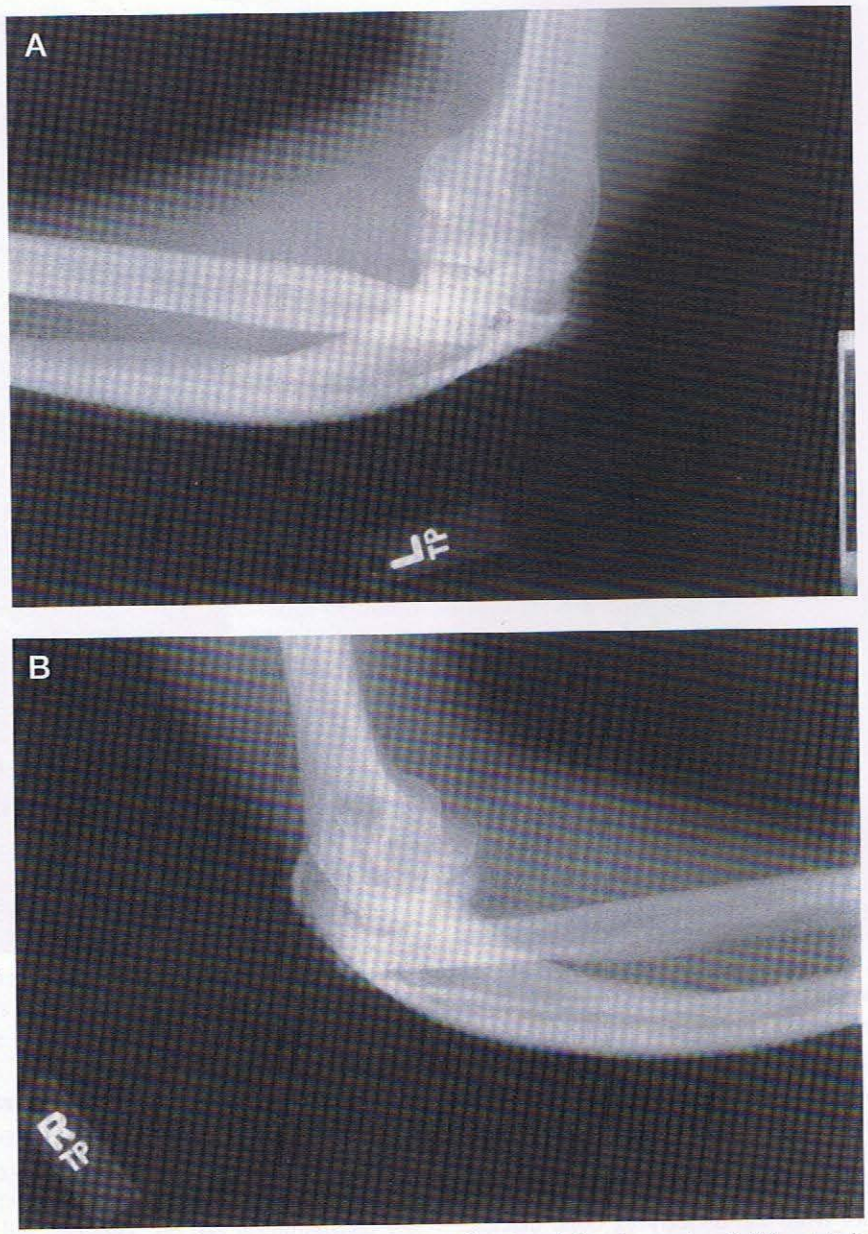

Fig 2. *Radiograph of A, left and B, right elbow, showing bowing deformity of the ulna and radial head dislocation, bilaterally.*

past 6 years, whereas in the past the fractures were commonplace, occurring every few months. His prognosis is somewhat guarded because of his predisposition to fractures.

Discussion

Osteogenesis imperfecta is an inherited mutation in one of two genetic loci coding for type I collagen—COL1A1 and COL1A2. Type I collagen is normally found within bone, tendons, ligaments, teeth, and the sclera of the eye. There are 4 major clinical considerations when diagnosing osteogenesis imperfecta. The criteria are osteoporosis with an abnormally fragile skeleton, blue sclera, blue-grey to yellowish-brown opalescent teeth (dentinogenesis imperfecta), and otosclerosis leading to presenile hearing loss. At least 2 of these features must be present for a positive diagnosis. Other common features include ligamentous laxity, abnormal temperature regulation with episodic diaphoresis, easy bruisability of the skin, constipation, premature vascular calcification, and inappropriate euphoria.[5,8] The defective collagen can also affect the cardiovascular system, leading to complications of aortic valve incompetence and dilatation of the aortic root.[6]

Traditionally, this disorder has been subdivided into 2 types (congenita and tarda). The congenita form is associated with the development of severe osseous fractures and deformity in utero. The skull is paper thin and soft. This type

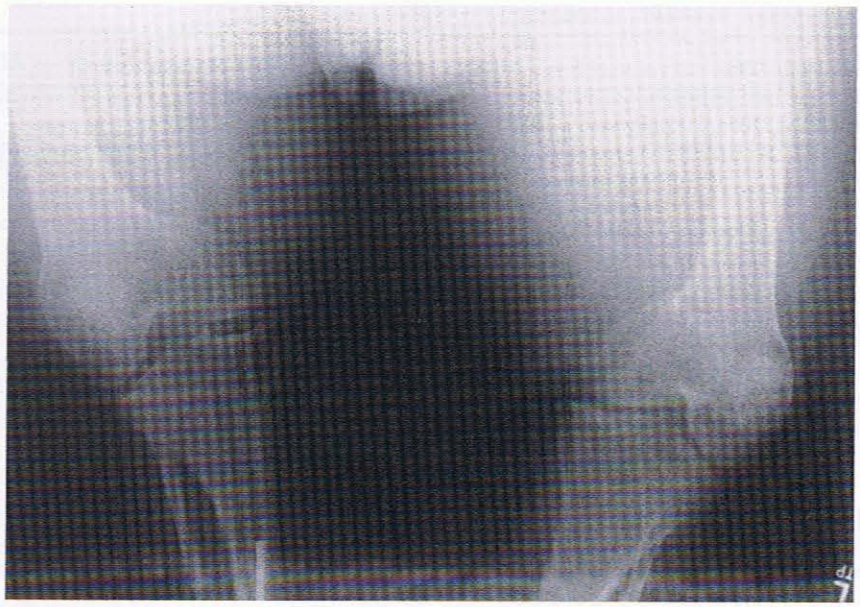

Fig 3. *Severe bowing deformity of the distal femur and proximal tibia, bilaterally. The pencil-thin cortices and overall osteoporosis are consistent with osteogenesis imperfecta. There is an intramedullary rod in the right tibia.*

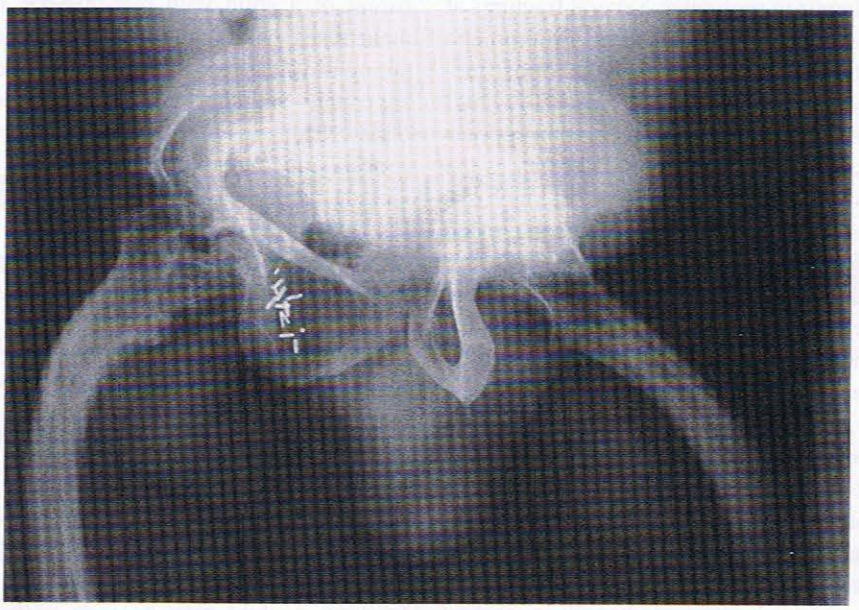

Fig 4. *Severe bowing deformity affecting the entire pelvis and proximal femora. There is bilateral protrusio acetabulae. Gross osteoporosis is identified.*

is associated with a high rate of stillbirths and infant mortality. The tarda form has a later onset of manifestations, which include fractures, dentinogenesis imperfecta, and blue sclera. They may or may not have bowing deformities. Bone fragility commonly decreases after puberty but does not completely disappear.[8] However, subdividing patients with osteogenesis imperfecta into congenita and tarda forms fails to accommodate the wide variation in expression of this condition.

More recently, osteogenesis imperfecta has been classified into 4 types by Sillence et al.[7] Table 1 outlines these classifications. Although type I is the most common form, most presentations are atypical and do not readily fit into any of the categories.

The radiographic appearance of osteogenesis is highly variable. The primary finding is one of diffuse osteopenia with pencil-thin cortices. The degree to which the bone density is decreased is proportional to the severity of the condition, and in mild cases may appear completely normal. The bones most commonly appear thin and gracile; however a short and thin or cystic appearance may be evident.[5,8]

Table 1. *The 4 types of osteogenesis imperfecta as determined by Sillence et al*[5,7,8]

Type	Inheritance pattern	Sclera	Otosclerosis	Miscellaneous
I	Autosomal dominant	Blue	Present	Most common type Variant type IB form is characterized by the presence of dentinogenesis imperfecta Similar to the tarda form Most are ambulatory
II	Autosomal dominant or recessive	Blue	Absent	Commonly lethal at birth Severe osseous fragility Similar to the congenita form Those who survive are confined to a wheelchair
III	Autosomal recessive	White (may be blue at birth)	Absent	Rare, lethal early in life Death is common before third decade due to pulmonary complications Severe osseous fragility Prominent ligamentous laxity Severe dwarfism and progressive kyphoscoliosis Wormian bones Those who survive are confined to a wheelchair
IV	Autosomal dominant	White (may be blue at birth)	Present	Varies from normal height to severe dwarfism Variable expression, may be ambulatory or confined to a wheelchair

Single or multiple fractures may be evident. They are typically transverse in nature and are more common in the lower extremity, the femur in particular. Several fractures at different stages of healing may be visualized. Micromelia and bowing deformities are common as a sequela of fractures in utero or in early childhood.[5,8]

Other findings include a severe kyphoscoliosis in approximately 40% of affected individuals. This deformity is the result of a combination of ligamentous laxity, osteoporosis, compression fractures, and posttraumatic damage to the end plates. Early onset of degenerative joint disease is common as a result of ligamentous laxity and joint incongruity produced by fracture deformity. Skull radiographs may show multiple wormian bones, enlarged frontal and mastoid sinuses, and platybasia with or without basilar invagination.[5,8]

Currently, there is no adequate therapy aimed at treating osteogenesis imperfecta. Although there is hope that genetic therapy may benefit patients with this condition, current therapeutic goals must be directed at maintaining the patient's bone density and providing relief from pain. Growth hormone administered to patients with moderate forms of osteogenesis imperfecta has shown some benefit in increasing bone mineral density.[10] Biphosphonates are currently being evaluated as a treatment option and appear to improve bone density while reducing the risk of fracture and pain.[10,11] Intramedullary rodding of long bones, especially of the femurs and the tibia, helps to reduce the risk of fracture and has been shown to improve and maintain the patient's ability to ambulate.[9,10] Surgery is also indicated in patients with progressive spinal deformity or basilar impression.[10] Swimming may be beneficial to patients with osteogenesis imperfecta because it offers a supported environment that should reduce the risk for fracture.

The high risk for fracture with even the most routine daily activities and the unavailability of adequate treatment options suggest the need for a highly restrictive lifestyle. Shea-Landry and Cole[6] suggest that one of the roles of a parent of a child with osteogenesis imperfecta is to help them resist participation in sports and prevent them from taking physical risks. Patients with the mild type IV forms are usually ambulatory[1] and may be more physically active.

Few examples of physically active patients with osteogenesis imperfecta tarda exist in the literature. Case reports include individuals with avulsion fracture bilaterally at the insertion of the triceps tendon, patellar tendon ruptures after skiing injuries, a ruptured patellar tendon during squash, and an Achilles tendon rupture in a soccer player.[3,4] Each of these individuals returned to athletic participation after their injuries healed. Bone softening disorders, including osteogenesis imperfecta, must be considered in any patient with tendinous ruptures or avulsions in atypical locations, especially if the injury is bilateral.

The diagnosis of osteogenesis imperfecta is an important one for several reasons. Early diagnosis will make the parents and child aware of the risks and manifestations of this disorder. In addition, because of the high potential for fracture, a child may be seen with multiple fractures at different stages of healing, which is one of the warning signs of child abuse. Osteogenesis imperfecta can be further differentiated from nonaccidental injury (NAI), as 80% of fractures related to NAI occur before the age of 18 months and are rare after the age of 2 years.[6] Physicians are required to report all cases of suspected child abuse or NAI. Although laws exist which absolve doctors from criminal liability in child abuse cases, they may be vulnerable to potential civil suits as a result of the emotional trauma inflicted on the child and parents of a wrongful accusation.[2] A detailed history, thorough examination, and appropriate radiographs will assist in defining the classic manifestations of this condition (ie, diffuse osteoporosis).

Conclusion

Patients with mild to moderate forms of osteogenesis imperfecta may be physically active and involved in athletic activities. Although these patients will have fractures as a result of the osseous fragility, they are also likely to be seen for additional musculoskeletal complaints. Although manipulation is contraindicated, this case report demonstrates that chiropractic care including pain management, rehabilitation, and nutritional counseling can benefit a patient with osteogenesis imperfecta.

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