Traumatic hand fracture in a patient with osteogenesis imperfecta

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Abstract

Objective: This article presents a case of osteogenesis imperfecta and reviews the clinical and radiographic features of this condition.

Clinical Features: A 27-year-old woman presented with pain in her left hand after a fall while dancing. Plain films revealed multiple fractures in the digits of her left hand.

Intervention and Outcome: The patient was referred to an orthopedic specialist for treatment.

Conclusion: Osteogenesis imperfecta is a heritable disorder of bone, which predisposes patients to fractures after trivial trauma, as was demonstrated in this case.

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Introduction

Osteogenesis imperfecta, or brittle bone syndrome, is a serious inherited disorder of connective tissue.¹ One of the most common connective tissue disorders, osteogenesis imperfecta affects 1 in 30 000 of the population across all demographics. Four types of osteogenesis imperfecta exist ranging from mild to lethal. It is important that health care providers, especially those using manual therapy, understand the implications that such a condition has on the management of their patients. The following is a discussion of

1 particular case of osteogenesis imperfecta. The patient's history is reviewed, followed by a discussion of the disorder, its clinical features, and the radiographic findings.

Case report

After a traumatic fall, a 27-year-old woman presented with comminuted fractures of the third and fourth proximal phalanges, medial displacement of the distal fracture fragment of the third metacarpal, and volar angulation of both fractures. The injury occurred while dancing. The patient's partner spun her; she slipped; and as she began to fall, he instinctively grabbed her hand. Radiographs were obtained the

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Fig 1. Lateral (A) and oblique (B) views of the phalanges demonstrate volar angulation across both fracture sites, more prominent in the fourth digit. Note the spiral fracture of the third proximal phalanx on the posteroanterior (PA) view (C). There is medial offset of the distal fracture fragment with slight varus angulation. There is a comminuted fracture of the fourth proximal phalanx.

following morning, and the patient was referred to an orthopedic specialist (Figs 1-3).

With a prior history of osteogenesis imperfecta, the complicated injury after minor trauma came as no surprise to the patient. She experienced bilateral femur fractures at birth, with a subsequent femur fracture at 2 years of age while merely standing up in her crib (Fig 4). Her parents were accused of child abuse on several occasions. Nearly every long bone of her body was fractured at some point in her childhood (Figs 5 and 6), but a period of 10 years followed where no fractures were reported. At the age of 24 years, the patient had a fracture to the surgical neck of the humerus while she was a passenger involved in a motor vehicle accident. She has also experienced fractures of her first tarsal, rib, and patella, each after otherwise trivial trauma. Other clinical features of this patient include blue sclerae and a history of tooth fractures, both characteristic of this condition. Family history revealed osteogenesis imperfecta in 1 of the patient's 5 siblings.

Discussion

Clinical features

The clinical features commonly observed in patients with osteogenesis imperfecta include abnormal bone formation, growth deficiency, bone fragility, blue sclerae, hearing loss, skin thinness, joint laxity

and hypermobility, and dentinogenesis imperfecta.¹ The basic defect is abnormal synthesis of type I collagen molecules. There are 4 main classes of osteogenesis imperfecta designated I to IV with subclasses.¹ Each class is named by reference to the genetic abnormality, clinical features, or both. Type I is known as *dominant with blue sclerae*, type II as *lethal perinatal*, type III as *progressive deforming*, and type IV as *dominant with normal sclerae*.² There are some reports that alterations can occur in bone matrix components other than type I collagen, and these have led to 3 discrete types of identification of histomorphometric evaluation (types V and VI) and linkage analysis (rhizomelic osteogenesis imperfecta) that remain poorly understood.³

Type I (nondeforming osteogenesis imperfecta) is a dominantly inherited disease characterized by blue sclerae and a tendency to sustain fractures from minimal trauma.³ Most of the patients with osteogenesis imperfecta (60%) belong to this group.³ Fractures are rare in the perinatal period and chiefly occur from childhood to puberty. The long bones of the arms and legs and the small bones of the hands and feet are most frequently broken. There is significant interfamilial and intrafamilial heterogeneity. In mild cases, few fractures and/or mild osteopenia can be observed, whereas severe cases can experience more than 50 fractures. Fractures typically heal with exuberant callus formation, and only 15% of patients develop deformities of the long bones. The number of fractures

Fig 2. Postsurgical PA (A) and oblique (B) views show intramedullary rods in the third and fourth proximal phalanges. There is remarkably good alignment of the fracture fragments.

decreases throughout adulthood and then often increases in menopause in women and after the sixth decade in men. Patients with osteogenesis imperfecta type I usually have a normal life span and die of unrelated illnesses.²

Type II is the most severe form of the disease and suggests a very poor prognosis.² Premature birth and intrauterine growth retardation are often present.³ Newborns have soft calvarial bones, a distinctive triangular face, blue sclerae, and a beaked nose. A narrow thorax, short and deformed extremities with multiple fractures, and a typical frog-like position of the

legs are the main features.³ Pulmonary and/or cardiac insufficiency, as well as infection during the neonatal period, usually results in death.³ The hallmarks of osteogenesis imperfecta type II include shortened and deformed limbs in the presence of normal birth height and weight.³

Type III occurs in approximately 20% of all patients with osteogenesis imperfecta.⁴ All infants born with fractures and deformity who survive the perinatal period belong to this group. This type is usually recognizable at

Fig 3. Post-pin removal PA (A) and lateral (B) views of the hand demonstrate healing of the fractures of the third and fourth proximal phalanges. Flexion deformity of the second and third digits is best appreciated clinically, but can also be seen on the lateral view.

Fig 4. Lateral femur view demonstrates a spiral fracture of the right proximal femoral diaphysis at 2 years of age.

birth because intrauterine fractures produce deformities of the long bones and severe skeletal changes. Most patients have intrauterine growth retardation, and further progressive growth failure continues during childhood as a result of long-bone deformations and spinal involvement.² Many of these patients exhibit large and asymmetric heads, whereas the face appears triangular. The maxilla is frequently posteriorly

Fig 5. Anteroposterior humerus view shows healing of an oblique fracture of the humeral diaphysis in the patient at 11 years of age. Note the slight varus angulation across the fracture site.

Fig 6. Bilateral anteroposterior views of the elbows demonstrate a healing fracture of the right (A) proximal ulna and a healed fracture of the left (B) proximal ulna. Note the residual bowing of the left proximal ulna.

inclined, and most craniofacial size measurements are reduced.⁵ Dental malocclusion occurs in 80% of patients, with primary teeth more affected with discolorations and abrasions.³ Furthermore, about 30% of patients experience recurrent abdominal pain due to chronic constipation and pelvic deformity with acetabular protrusion.⁶ Occasionally, congenital cardiac malformations, hemihypertrophy, papillary calcifications or kidney stones, and hypercalciuria are seen.⁷ The development of all motor milestones is significantly delayed compared with types I and IV; and most

children who do achieve independent ambulation have poor joint alignment, poor balance, and low endurance.³ These patients are at a higher risk of dying prematurely from diseases secondary to their underlying collagen abnormality.

Type IV is a dominantly inherited form, differing from type I by normal or slightly grayish sclerae, shorter stature, and common presence of dentinogenesis imperfecta.³ Although there is no intrauterine growth retardation, postnatal growth is diminished.⁷ Craniofacial size, dental malocclusion, and hearing loss are all possible affects as well. The highest frequency of fractures is typically seen between childhood and puberty and then again in older age.³ In about one third of patients, progressive kyphoscoliosis occurs, leading to severe cardiopulmonary and neurologic complications.³ Basilar impression and neurologic symptoms are also a common finding. However, life expectancy is only minimally affected.⁸

Radiographic findings

A decrease in osseous density is a characteristic finding on radiographs of patients with osteogenesis imperfecta.⁹ The severity of osteopenia varies, and some patients exhibit no decrease in osseous density on plain films. Rapid bone loss may be observed in middle-aged patients.^{10,11} Fracture after minimal trauma is the most universally observed manifestation of osteogenesis imperfecta.^{11,12} Falvo et al¹² measured a fracture rate of 10.6 per year in patients 1 to 16 years of age who had the congenital form of osteogenesis imperfecta. Fractures occurred in the lower extremities 3 to 12 times the frequency of upper extremities. Fractures are typically transverse, occur on the convex side of a bowed long bone, and occur in the femur more often than the tibia.¹² Fractures occurring in utero may result in limbs that are short and curved.¹¹

Resnick¹³ and Greenfield¹⁴ make reference to the common occurrence of hyperplastic callus formation after trauma or surgical rodding. This finding must be carefully distinguished from osteosarcoma; biopsy may be needed to establish the differential diagnosis.¹⁵ Other differential diagnoses of exuberant callus formation include steroid therapy, neuropathic arthropathy, nonaccidental injury, paralysis, renal osteodystrophy, and multiple myeloma.

An additional finding in patients with osteogenesis imperfecta is the presence of wormian bones. These will usually number more than 10, be arranged in a mosaic pattern, and measure greater than 6 mm by 4 mm.¹⁶

It is important for health care providers to recognize the differences between osteogenesis imperfecta and nonaccidental injury, especially considering the ramifications of allowing an abused child to be returned to their abuser or removing a child with undiagnosed osteogenesis imperfecta from their parents under a false assumption of child abuse.¹⁷ Paterson reported 12 cases of children with osteogenesis imperfecta who were initially diagnosed with nonaccidental injuries. Seven of the children were removed from their parents. Wormian bones were present on radiographs of 4 of the 12 children. It should be noted that nonaccidental injury in children is considerably more prevalent than osteogenesis imperfecta, and it is the leading cause of death among infants 6 to 12 months of age in the United States.¹⁸ If child abuse is suspected, a radiographic survey of the entire skeleton is warranted; individual films should be taken because single, full-body films are not adequate for diagnostic purposes.¹⁹ Rib fractures, especially paraspinal in location, are highly suggestive of nonaccidental injury.²⁰ Acute bilateral fractures, fractures in the lower extremities of children who are not yet walking, and bilateral skull fractures that cross a suture are additional radiographic findings that should incite consideration of nonaccidental injury.²¹⁻²⁴ When considering the diagnosis of nonaccidental injury, care must be taken to evaluate the clinical and family histories, physical examination, and radiographic examination to exclude the possibility of osteogenesis imperfecta.

Conclusion

Although osteogenesis imperfecta is not seen in everyday practice, it is a relatively common disorder, which chiropractors and other providers of manual therapy need to be mindful of. This report reviewed 4 primary classifications, of which chiropractors are most likely to encounter type I. Radiographic features of osteogenesis imperfecta include osteopenia and the tendency of patients to experience fractures related to relatively unremarkable trauma. Osteogenesis imperfecta and nonaccidental injury should both be considered in a differential diagnosis when evaluating fractures in children.

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