S CASE REPORT

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Primum non nocere: a case of a humeral fracture in a patient with fibrodysplasia progressiva ossificans

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Abstract

Fibrodysplasia progressiva ossificans (FPO) is an extremely rare condition characterized by abnormal heterotopic bone formation. The condition is eponymously known as 'stoneman' disease because patients can become effectively entombed within abnormal heterotopic bone. We present the first known case of a diaphyseal humeral fracture managed conservatively in an adult patient with this condition. This patient already had a pre-existing bony bar from a fusion mass involving the thoracic spine, scapula and ribs to her proximal humerus splinting the arm in a position of adduction with the palm of her hand facing towards her groin. This patient also suffered a concomitant unstable cervical spinal fracture for which full spinal precautions were needed. As a result of the rapid bone forming nature of her condition, the humeral fracture was placed in a position where the hand would face outwards to make perineal care and personal hygiene easier in that the fracture was expected to unite in this new position as a result of her FPO.

Keywords

case report, conservative treatment, fibrodysplasia progressiva ossificans, humeral fracture

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Introduction

Fibrodysplasia progressiva ossificans (FPO) is an extremely rare autosomal dominant condition with a worldwide prevalence of approximately one in two million live births.[1] No ethnic, racial or geographical predisposition has been described. It is considered the most catastrophic congenital disease of progressive heterotopic ossification in humans. Heterotopic ossification usually appears in childhood either spontaneously or after minor trauma presenting as swellings in soft tissues, which progressively transform into ectopic bone, leading to severe restrictions of joint movement. Eventually, complete ankylosis of the widespread joints and muscles occur and effectively turn the patient into a 'stone man' or a 'living statue'.[2] By the second decade of life, most patients become progressively more immobile, usually wheelchair bound, and death often results from cardiopulmonary failure secondary to involvement of the thoracic muscles.[3] Iatrogenic harm has been described from attempts to surgically excise heterotopic bone or to perform surgical procedures.[4,5] We present a case of a humeral fracture treated conservatively in an immobile patient with FPO that was allowed to malunite to facilitate her nursing care.

Case report

A 45-year-old Caucasian lady with a known diagnosis of FPO was referred to our hospital following a fall from her wheelchair. Prior to presentation, she was living independently in the community with the help of carers for all activities of daily living but had limited movement of all four limbs and was a wheelchair ambulator. She was admitted to our hospital following the fall

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having sustained an unstable cervical spine fracture at the C6/C7 level, and a closed midshaft right diaphyseal humeral fracture. Prior to her injury, she had some altered neurology in the left arm, which was unchanged following the fall. Because of her medical comorbidities, both injuries were treated conservatively.

She required a hard collar for her spinal fracture but was having difficulty in expectorating and clearing her secretions such that she became dyspnoeic and progressed into respiratory arrest and subsequent cardiac arrest. She was resuscitated and had a surgical tracheostomy.

She was admitted to our intensive care unit for close ventilatory monitoring and complained of pain in her arm. It was noted by the patient that, prior to her admission, her arm was normally held in an adducted position close to her chest wall and that her palm normally faced inwards and lay over her groin. In this position, her hand function was very poor because she was unable to hold anything. This was because of a pre-existing bony bar from her sixth rib and thoracic spine. This can be seen clearly on her plain radiographs from casualty (Fig. 1.)

Because she required full spinal precautions for her unstable cervical spinal fracture, it became apparent that perineal hygiene and general nursing care would be more difficult if an attempt to restore the initial pre-injury position of the humerus was made. Surgical intervention was contraindicated because, in patients with FPO, surgical trauma stimulates more abnormal heterotopic bony ossification in the remaining soft tissues, thereby exacerbating the underlying condition.

Following an uneventful stay on the intensive care unit, the humeral fracture went on to unite and she was transferred to a rehabilitation unit. Clinically and radiologically, she had united her humeral and cervical spinal fractures at 6 weeks (Figs 2 and 3). On this occasion, supervised neglect led to an excellent outcome for this patient.

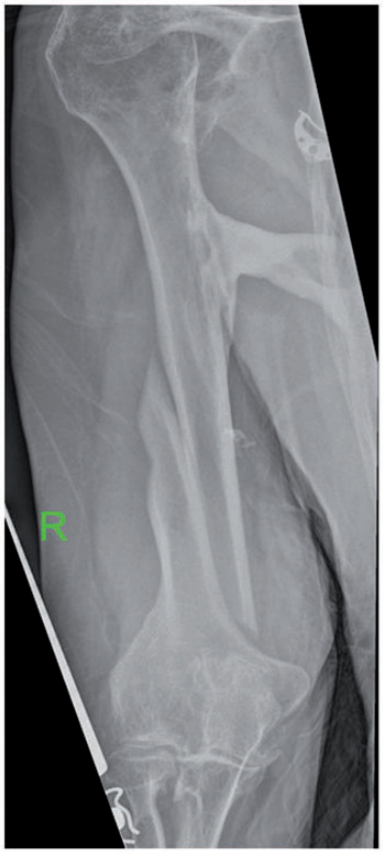

Figure 1. Plain ap humeral radiograph showing pre-existing bony bar proximal to the humeral fracture.

Discussion

FPO is considered to be caused by a mutation in the ACVR1 gene that codes for a type of bone morphogenetic protein type I receptor. The ACVR1 protein is found in many tissues of the body, including skeletal muscle and cartilage. It helps to control the growth and development of the bones and muscles, including the gradual replacement of cartilage by bone (ossification) that occurs in normal skeletal maturation from birth to young adulthood.[6]

In this case, the diagnosis was already known, although the clinical manifestations of FPO are characteristic.[7] Congenital malformations of the great toes and progressive heterotopic ossification that forms qualitatively normal bone in characteristic extraskeletal sites. Children who have FPO appear normal at birth except for congenital malformations of the great toes. During the first decade of life, sporadic episodes of painful soft tissue swellings (flare-ups) occur, which are often precipitated by soft tissue injury, intramuscular injections, viral infection, muscular stretching, falls or fatigue. These flare-ups transform skeletal muscles, tendons, ligaments, fascia and aponeuroses into heterotopic bone, rendering movement impossible.

Preventative management is based on prophylactic measures against falls, respiratory decline and viral infections. The median lifespan is approximately 40 years of age.

This case report aims to raise awareness of this rare condition so that surgeons inexperienced in dealing with it will not be tempted into performing surgical procedures. We consider that this case highlights the need to have a fully informed discussion with the patient and the various members of the multidisciplinary team involved in their care. In this case, a sound knowledge of the natural history of this condition allowed the treating surgeons to effectively make use

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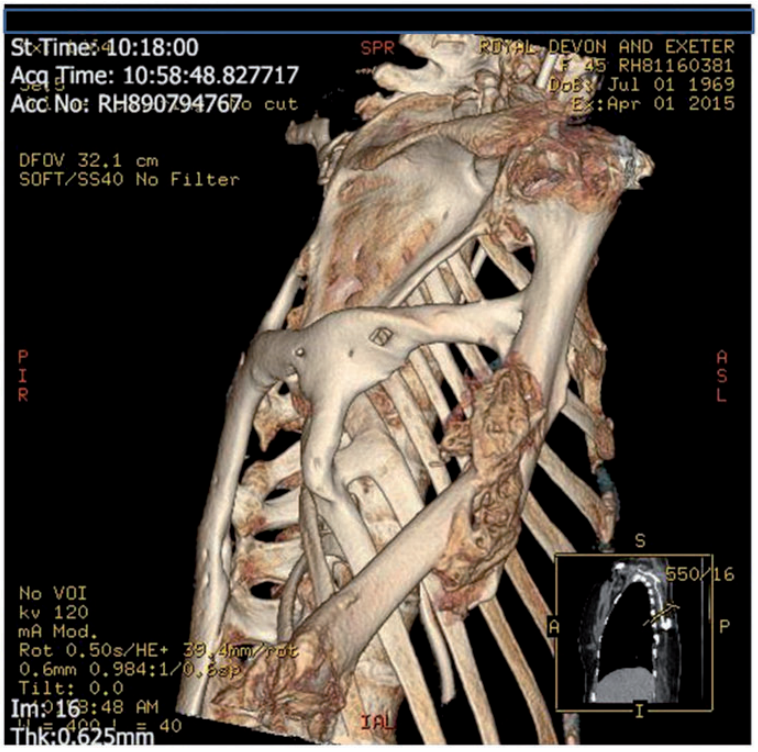

Figure 2. Three-dimensional CT reconstruction showing humeral fracture and pre-existing bony fusion mass from thoracic spine, scapula and ribs to humerus.

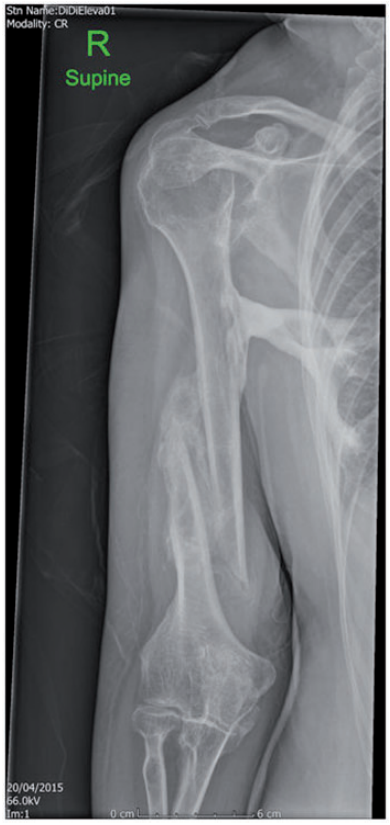

Figure 3. AP radiograph showing bony union at 6 weeks.

of a serendipitous osteotomy, meaning that nursing care becoming easier for the patient because bony union of the fracture was virtually guaranteed.

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