

Bilateral Asynchronous Humeral Shaft Fractures in a Patient with Autosomal Dominant Osteopetrosis Type II (Albers-Schonberg Disease)

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Abstract

Osteopetrosis is a rare metabolic disease which presents with fragile osteosclerotic bone. A thirteen-year-old girl with an underlying autosomal dominant osteopetrosis (ADO) type II who sustained a left oblique humeral shaft fracture after trivial trauma is presented. Radiographic results showed many characteristic features of osteopetrosis, including incomplete remodeling of a previous fracture of the contralateral humeral shaft. The authors obtained good healing and alignment of the left humeral shaft fracture by sugar tong slab.

Key word : Osteopetrosis, Albers-Schonberg Disease, Humeral Shaft Fracture

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Osteopetrosis is a rare inherited skeletal condition characterized by increased bone density, caused by failure of bone resorption due to a functional deficiency of osteoclasts⁽¹⁾. Calcified chondroid and primitive bone persist, leading to osteosclerosis and increased brittleness of the bones. Osteopetrosis was first described by a German radiologist, Heinrich Albers-Schonberg in 1904⁽²⁾. Thereafter, the condition was named Albers-Schonberg disease and later as marble bone disease.

The eponym Albers-Schonberg disease had been used as a generic term for the disease, despite only a single case reported 100 years ago, almost certainly having an autosomal dominant inheritance⁽³⁾. The prevalence of osteopetrosis in Denmark is 5.5 : 100,000 with the male to female ratio of 1 : 1⁽⁴⁾. There are three clinical groups of osteopetrosis: 1) infantile malignant autosomal recessive, the clinical manifestations of which appear at birth or in early infancy with a bad prognosis 2) intermediate auto-

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somal recessive 3) mild type, autosomal dominant osteopetrosis (ADO). The age at presentation for the mild type is usually considerably older than that in the malignant type - the youngest case being a 3 month old infant and the oldest a 76 year old patient. This last form is compatible with a normal life span and usually presents to orthopedists with recurrent long bone fractures and deformity(4).

The clinical diagnosis of ADO is based on characteristic radiological findings: diffuse osteosclerosis, primarily involving axial skeleton, with symmetrical involvement of the long bones. Other bone diseases or conditions known to interfere with bone or calcium metabolism or malignant conditions should be excluded(5).

CASE REPORT

A thirteen year old girl presented to the Orthopaedic Department of Queen Sirikit National Institute of Child Health with swelling and tenderness over her left upper arm after slipping down the stairs. She was the first-born of two, her sibling being normal. Radiographic finding showed an oblique fracture of



Fig. 1. The X-ray shows a short oblique fracture of the proximal humeral shaft with increased bone density.



Fig. 2. Several radiographic examples of the appearance of osteopetrosis in autosomal dominant type II are shown. (A) Lateral radiograph of the lumbar spine shows the bone-within-a-bone phenomenon or "Rugger-Jersey" appearance of the vertebral bodies. (B, C) Radiographs of the lower extremities show obliteration of the medullary canal with flaring of the metaphyseal part of the distal femur or "Erlenmeyer flask" deformity. (D) Endobones in the metacarpals of the hands. Note the transverse striation in the distal radius.

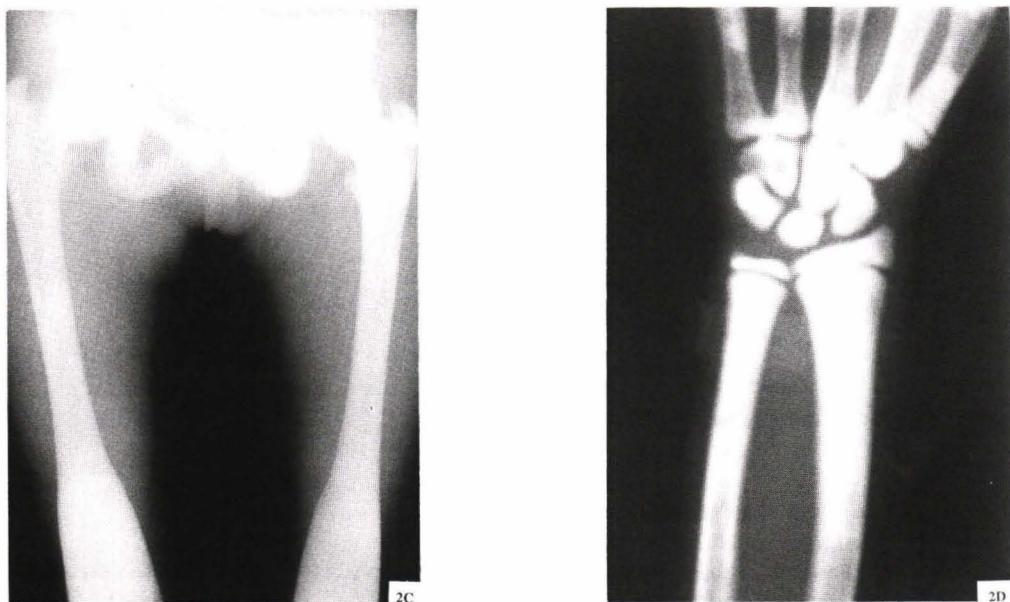


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the left proximal humeral shaft with minimal displacement (Fig. 1). She was admitted for further investigation because of abnormal osteosclerosis seen on the radiograph.

There were no abnormalities with her hearing or visual functions. No liver or spleen enlargement was present on the examination.

The skeletal survey showed many features of osteopetrosis (Fig. 2A-2D); generalized sclerosis of the long bones with partial obliteration of the intra-medullary canal, metaphyseal flaring, "Rugger-Jersey" appearance of the spine, longitudinal and transverse striation on the distal radius, metacarpal bones and distal tibia and fibula. The skull film showed sclerosis on the base of the skull (Fig. 3).

Laboratory results showed mild anemia without any other significant abnormalities: Hb 9.4 g/dl, Hct 31.6 per cent, red cell count 4,160,000/mm³, platelet count 339,000/mm³, white cell count 10,600/mm³, serum calcium 2.15 mmol/L, serum phosphorus 3.85 mmol/L, BUN 2.83 mg/dl, creatinine 0.39 mg/dl, SGOT 96 Unit, SGPT 10 Unit, alkaline phosphatase

212 U/L, total protein 7.10 g/dl, albumin 4.24 g/dl, globulin 2.86 g/dl and normal urinalysis.

Four years earlier, the patient had sustained a fracture of her right upper arm following a fall from a one-meter-high tree. She was advised to have surgical treatment from a provincial hospital which her family refused. Ultimately, the fracture was treated by a local healer without any recognized complication. Radiograph of the right humerus, as a part of the skeletal survey, on this admission revealed continuity of the cortex with mild angulation of the proximal shaft with a sharp oblique line (Fig. 4).

The fracture of the left humeral shaft was treated with sugar tong slab for 7 weeks. Clinical union was attained 3 weeks after immobilization. No delayed union or significant deformity was present at the last radiographic follow-up, which was 3 months after the injury (Fig. 5). Clinical evaluation at the end of the treatment showed no deficit of function or apparent deformity.

Four months after the left humeral shaft fracture, the patient again sustained a left intertrochanteric fracture after falling from a bicycle (Fig. 6).



Fig. 3. Lateral view of the skull showing a normal calvarium and increased density at the base of the skull, consistent with ADO type II.



Fig. 5. Three months after the fracture, the left humeral shaft has healed with a mild degree of angulation.



Fig. 4. Mild angulation of the right humeral shaft with an obvious oblique line at the angulated site.



Fig. 6. The AP view of both hips shows a transverse fracture of the left intertrochanter of the femur.

DISCUSSION

Forty per cent of patients with ADO are often asymptomatic and are usually incidentally diagnosed following radiographic examination⁽⁴⁾. Generally, they have normal physical, mental health and lifespan⁽⁵⁾.

Anemia, caused by displacement of bone marrow by osteosclerotic bone, is an uncommon finding in this type of osteopetrosis⁽⁴⁾. Based on standard radiographs, Bollerslev et al⁽⁶⁾ divided ADO into 2 sub-

types with different clinical, biomechanical and histological manifestations. Subtype I is radiographically characterized by pronounced osteosclerosis of the cranial vault, whereas subtype II has end-plate thickening of the vertebra (Rugger-Jersey spine) and endo-bones in the pelvis.

In subtype II, the risk for fracture is highly increased and delayed healing also seems to be more frequent⁽⁷⁾. Delay in recognition of fractures often leads to extensive periosteal elevation and extracortical new bone formation⁽⁸⁾.

In the presented patient, there were no clinical manifestations of marrow space obliteration. There was no myelophthisic anemia or hepatosplenomegaly. Based on Thai children growth data⁽⁹⁾, she had a slightly short stature. Her height (141 centimeter) was within the range of -2 SD to -1.5 SD. She had a head circumference of 51 centimeter diameter without any dysmorphic features.

She had recurrent pathologic fractures on bilateral humeral shafts and left intertrochanter of the

femur on different occasions caused by her underlying osteopetrosis.

Although the authors could not obtain medical records and films of the previous right humeral shaft fracture, the finding of angulation on the right humeral shaft with an oblique line was assumed to be an old fracture with incomplete remodeling of the fracture site; based on the histopathologic study by Bollerslev et al⁽¹⁰⁾.

According to Armstrong et al⁽¹¹⁾, most diaphyseal long bone fractures and subtrochanteric fractures can be successfully managed with conservative treatment. On the contrary, an intertrochanteric fracture should be treated with internal fixation, despite technical problems with drilling. Conservative treatment for a femoral neck fracture inevitably results in progressive coxa vara requiring surgery.

The authors obtained a good result by treating the oblique humeral shaft fracture in the presented patient with a simple coaptation splint without any problem of healing or disability.

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กระดูกตันแข็งหัก 2 ข้างต่างวาระกัน ในผู้ป่วยออสต์โอลิสต์ ชนิดอัลเบอร์ส-ชอนเบิร์ก

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โรค osteopetrosis เกิดจากความผิดปกติของการทำงานของ osteoclast ทำให้กระดูกเกิดความเปราะแตกหักได้ง่าย คณะผู้วิจัยรายงานผู้ป่วย autosomal dominant osteopetrosis type II ในเด็กหญิงอายุ 13 ปีที่มีกระดูกตันแข็งหักภายในหลังได้รับอุบัติเหตุที่ไม่รุนแรง ได้ทำการรักษาโดยวิธีการใส่เม็ดออกซิทิด sugar tong slab ซึ่งกระดูกติดติดโดยไม่มีภาวะแทรกซ้อน และได้นำเสนอภาพรังสีแสดงถึงลักษณะเฉพาะของโรค รวมทั้งร่องรอยการหายจากการหักของกระดูกตันแข็งข้างตรงข้ามเมื่อหลายนปักก่อน

คำสำคัญ : ออสต์โอลิสต์, อัลเบอร์ส-ชอนเบิร์ก, กระดูกตันแข็งหัก

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