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Bony deformities in a child with untreated osteogenesis imperfecta type III

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Keywords: skeletal dysplasias; radial head dislocation; bone mineralization disorder; fragility fractures; multidisciplinary care.

Case description

Osteogenesis Imperfecta (OI) is a skeletal dysplasia that affects the cross-linking of Type 1 collagen leading to a wide spectrum of manifestations including bony abnormalities, scleral abnormalities, dentinogenesis imperfecta, and hearing abnormalities [1].

Orthopedic manifestations of OI include basilar invagination, kyphoscoliosis, codfish vertebrae, bowing deformities of the long bones, coxa vara, protrusion acetabuli, pathologic fractures, and radial head dislocations [1]. The Sillence Classification, originally designed to describe Types I – IV, has been expanded upon to includes Type V, VI, VII, which do not have a Type I collagen mutation, and instead have other genetic explanations (CRTAP, LEPRE1 genes) for the abnormal bone seen on microscopy [1,2]. Type I, IV represent milder forms of disease, Type V-VII represent milder forms of disease, Type III is the most severe survivable form, and Type II is lethal at birth [1,2].

We present a case of Sillence Type III OI, diagnosed by genetic analysis at birth, with significant bony deformities. She did not have appropriate multidisciplinary medical management of her condition and presented to our clinic at age 14. Radiographs of the spine and extremities demonstrated kyphoscoliosis, bilateral protrusio acetabuli, multiapical bowing bony deformities of femora, tibia, humeri, forearm, radial head dislocations, and short stature. This case highlights the importance of multidisciplinary management for children with OI. This includes physical rehabilitation [3], bisphosphonates [3,4] and more recently

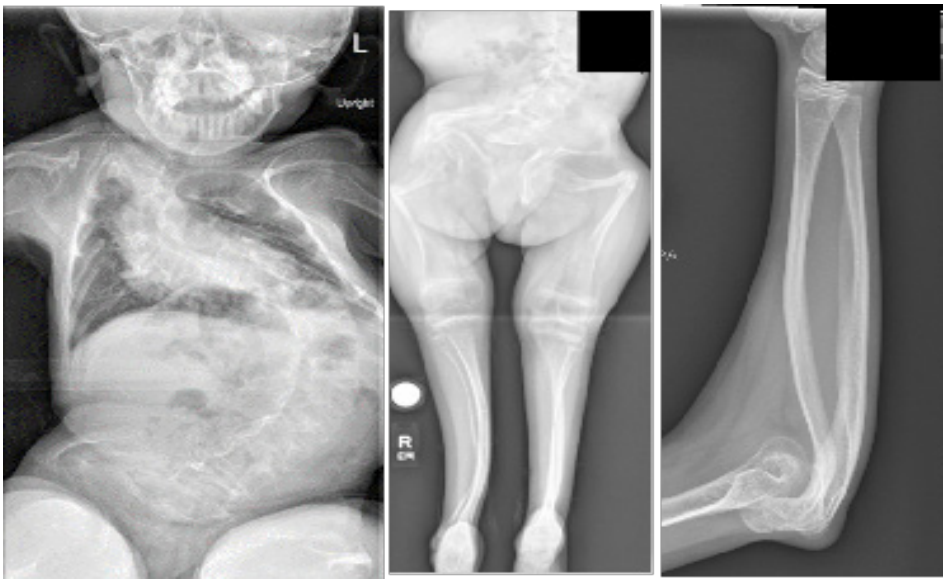

Figure 1: Chest, lower extremity, and anteroposterior forearm radiographs in a patient with Osteogenesis Imperfecta III demonstrating scoliosis, bilateral protrusio acetabuli, anterolateral bowing of the femur in the setting of pathologic fractures, bilateral anteromedial bowing of the tibia and fibula, and a dysmorphic proximal radius and ulna with posterior dislocation of the radial head.

gene therapies [3,4], and stem cell transplantation therapies [4]. Patients with Osteogenesis Imperfecta Type III have a limited lifespan [5] and it is important to maximize the advantage of medical therapies. In the absence of the appropriate medical care, osteogenesis imperfecta can develop progressive deformities in the long bones, a high risk of fragility fractures, and progressive kyphoscoliosis that can be difficult to manage surgically due to underlying osteopenia.

Final diagnosis: Osteogenesis Imperfecta Type III

Three differential diagnosis:

1. Idiopathic Juvenile Osteoporosis
2. Hypophosphatasia
3. Menkes disease

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